Identifying the benefits and disbenefits of Clinical Genetic Services: A framework for Economic Evaluation

Christalla Pithara

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ABSTRACT

A number of methodological considerations have been discussed in the area of economic evaluation of Clinical Genetic Services (CGSs) including the limited knowledge of psychosocial consequences of these services. This study aims to address this gap by identifying tangible and intangible benefits and disbenefits of CGSs and presenting these within a framework to assist in the design of a comprehensive welfarist economic evaluation.

Mixed methods of data collection were adopted and a UK medical genetics service was used as a case study. Face-to-face interviews with genetic service providers were undertaken in Phase One to explore patient pathways and the perceived role of the service. Focus groups and face-to-face interviews with service users explored the perceived benefits and disbenefits of the CGS in Phase Two. Phase Three comprised a pilot study of using Audience Response Systems (ARSs) for exploring stakeholder preferences and tackled issues of respondent validation and transferability.

Both process-related attributes and psychosocial outcomes emerged as utility-bearing for service users. Patient pathways i.e. patient experience, were found to be influenced by factors associated with the genetic condition and with individual patient/family characteristics and needs. The overall (dis)benefits of the service however were found to be common across conditions. The concept of Perceived Familial Control is proposed as a suitable outcome which encompasses the psychosocial dimension of CGSs.

This study has demonstrated the use of qualitative methods in the context of health economics and economic evaluation. It has specifically demonstrated the use of various levels of qualitative analysis for obtaining attributes and outcomes of CGSs and has incorporated these within a framework directed towards the design of a welfarist economic evaluation. The use of ARSs was also tested for their usefulness as a method of establishing preferences and exploring the opinions of CGS stakeholders.

Further research is required to establish whether the emerging (dis)benefits represent the experiences of users of other UK clinical genetic centres. Subsequently, user preferences for the identified (dis)benefits could be explored as a step towards the design of a welfarist economic evaluation. Further research is also required to develop the concept of Perceived Familial Control into an appropriate outcome measure for CGSs.
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AUTHOR'S DECLARATION

I declare that the work in this thesis was carried out in accordance with the Regulations of the University of Glamorgan/Prifysgol Morgannwg. The work is original except where acknowledged or indicated by special reference in the text. No part of this thesis has been submitted for any other degree.

Any views expressed in the thesis are those of the author and in no way represent those of the University of Glamorgan/Prifysgol Morgannwg.

The dissertation has not been presented to any other University for examination in the United Kingdom or overseas.

SIGNED

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List of Abbreviations

ARS= Audience Response System
BMD = Becker Muscular Dystrophy
BMJ= British Medical Journal
CBA = Cost-Benefit Analysis
CCA= Cost-consequences analysis
CEA= Cost-effectiveness analysis
CF = Cystic Fibrosis
CGSs = Clinical Genetic Services
CUA= Cost-utility analysis
DALYs= Disability Adjusted Life Years
DBMD=Duchenne and Becker Muscular Dystrophy
DCEs= Discrete Choice Experiments
DMD = Duchenne Muscular Dystrophy
DoH= Department of Health
DRPC= Devolved Research Programmes Committee
EE = Economic Evaluation
GP= General Practitioner
HADS= Hospital Anxiety and Depression Scale
HBOC = Hereditary Breast and Ovarian Cancer
HNPCC= Hereditary non-polyposis colorectal cancer
LREC= Local Research Ethics Committee
MAP= MUTYH associated polyposis
NHS EED = National Health Services Economic Evaluation Database

NHS = National Health Service

NICE = National Institute for Health and Clinical Excellence

PFC = Perceived Familial Control

PPC = Perceived Personal Control

QALYs = Quality Adjusted Life Years

TS = Tuberous Sclerosis

TSD = Tay-Sacs condition

VHL = von Hippel Lindau disorder

WTP = Willingness to Pay
CHAPTER 1

BACKGROUND AND AIMS OF THE THESIS
1.1. Overview of chapter

This chapter will provide an overview of the context and background to the thesis leading to the presentation of its aims and objectives. This chapter will:

1. Provide a guide to terminology,
2. Introduce UK Clinical Genetic Services (CGSs) and their distinguishing characteristics,
3. Introduce the concepts of scarcity, health care and technology assessment and the need for economic evaluation,
4. Introduce cost-benefit analysis (CBA) as the only technique of economic evaluation able to encapsulate the overall impact of CGSs,
5. Discuss the limited knowledge around the overall consequences of CGSs and the calls for further research in order to increase their understanding,
6. Discuss the issues around existing outcomes and the turn towards the service users' perspective of health services,
7. Present the aims and objectives of the thesis, and finally
8. Present a summary of the remaining chapters.

1.2. Guide to Terminology

In this thesis the terms consequences, impacts, outcomes, process attributes, benefits and disbenefits, costs, and outcome measures will be used. These terms tend to be used interchangeably within the literature and as Donaldson and Shackley (1997) state, often it is difficult to distinguish between what is process and what is outcome. In order to avoid confusion, the way these terms will be used is clarified here. Consequences
and impacts will be used interchangeably and refer to any impact on the service user resulting after referral and use of a CGS. These can be positive or negative, tangible or intangible. Outcomes will refer to any measurable consequence of CGSs by using an outcome measure. Within economics any intangible attributes valued by consumers whether related to process or outcome are also referred to as benefits and disbenefits, while tangible consequences can be also regarded as positive or negative costs. In this sense, a benefit or disbenefit is a broad concept encompassing both process and outcome.

1.3. Background

Clinical Genetic Services (CGSs) are a specialised component of the health care service that has experienced rapid expansion over the past 30 years. The advances made in molecular genetics have enabled the identification of a large number of genes which are responsible for different genetic conditions. These advances have been transferred from the laboratory into clinical practice through the establishment and growth of CGSs as a result of private and government funding, as well as due to the “technological imperative”, i.e. the perception that a technology should be offered to the public only because it exists (Hall, Viney and Haas, 1998).

Currently in the UK there are more than 20 Regional Clinical Genetic Centres (The British Society of Human Genetics, 2010) and the methods and models of service provision differ from one centre to another. This variation has come to depend on commissioning, available resources and local specialisation within the medical staff, but the aims and core functions of clinical genetics remain the same for all centres. The
function of CGSs is to “inform, guide and support individuals and couples as they prepare for parenthood and family life” as well as deal with “conditions of adult life in which there is a significant heritable component, including a number of common cancers” (Fryer and Cheese, 1998). In this way, the core function of the service is based on the provision of information and not on clinical care as other parts of the health care service.

CGSs are distinguished from other health care services by a number of characteristics which are important when considering the methodological approaches utilised for their evaluation. Firstly, CGS interventions are qualitatively different from other health care interventions. Genetic conditions are found to entail more far-reaching consequences than other “apparently ordinary” conditions. For example, when an individual undergoes genetic testing the result is not only relevant to that individual but it also reveals probabilities of risk status for other family members who might not desire to know that information (Douglas, Hamilton and Grubs, 2009; Dancyger et al., 2011; Vos et al., 2011). The family nature of CGSs is exemplified in the designation of the family unit as the unit of currency of these services (Hughes et al., 1998).

In addition to the wider family impact of genetic services, there are complex psychological and social implications associated with genetic conditions and the receipt of genetic test results. Procter (2002) lists a number of psychological and social implications, such as the influence on major life decisions because of the fear of carrying a genetic disorder or because of the results of genetic testing. One example of this is avoiding close relationships and parenthood because of a hereditary condition present in the family. Another implication is the impact of having genetic investigations
on an individual's ability to obtain insurance, buy a house and choose a career (Procter, 2002). Even though other health care interventions might entail implications for individuals other than the one undergoing the intervention, no other health care programme can be considered to have potentially such a wide psychosocial impact as CGSs. CGSs are by definition a family service, because of the cascading effect of risk on other family members.

A second distinguishing characteristic of CGSs is that in contrast to other health care programmes, the aims of CGSs do not include direct changes in the health status of the individuals referred to the service, even though improved health outcomes might result because of targeted and co-ordinated surveillance following risk identification or diagnosis. Based on current available biotechnologies, CGSs are able to identify the presence or absence of a gene mutation in the individual which predisposes them to illness and to offer genetic testing to other family members. Depending on the condition and the life stage that the individual or family is at, the service can also offer advice on reproductive choices, prenatal genetic testing and referral to screening services in order to improve prognosis, or assist in the management of the condition through either directly managing the symptoms or through the co-ordination of the input of different specialists in the individual's care. Because the available biotechnology can only detect the presence of a gene mutation either after symptoms of the condition become apparent or before they appear, without being able to cure the condition, the value of the service is brought into question. The majority of consequences resulting from CGSs are intangible and consequently less measurable than health outcomes.
The developments taking place within medical genetics (encompassing both laboratory and clinical genetics) need to be viewed in the context of the reality of the health care service, where the health care budget has been under enormous pressure from increasing costs (Catala-Lopez et al., 2011). The economic reality of finite resources i.e. scarcity, when needs are virtually infinite means that the developments in the provision of genetic services need to be assessed using a more critical eye. Decision-makers are now being faced with inadequate resources to meet demand, and attention has turned to health care evaluations and health technology assessments for guidance on which interventions should be given priority (McDaid and Cookson, 2003; Drummond et al., 2005). In the present climate, health care and technology assessment have become essential in guiding decision-making in the National Health Service (NHS).

Health care interventions can be evaluated based on a number of alternative viewpoints. CGSs can be evaluated from a psychosocial perspective, where the focus of the investigation is the impact of the genetic service and its numerous constituent parts i.e. genetic counselling, genetic risk assessment and/or genetic testing on psychosocial outcomes such as anxiety, depression and personal control (van Oostrom et al., 2007a; Vodermaier, Esplen and Maheu, 2010; Hart et al., 2011). Other approaches have included a service-provider perspective comparing the impact of different models of service provision on specific outcomes which have also tended to be psychosocial in nature (Brain et al., 2002; Forrest et al., 2008; Graves et al., 2010; Roussi et al., 2010; Culver et al., 2011). A third perspective involves the economic evaluation of healthcare interventions (Holland, Huston and Noyes, 2009; Mvundura et al., 2010; Perez et al., 2011). In the present climate of evidence-based medicine and
health technology assessment, the UK National Institute of Health and Clinical Excellence (NICE) has incorporated economic evaluation information assessing cost-effectiveness (CEA) alongside assessments of quality, safety and efficacy (McDaid and Cookson, 2003; Rawlins and Culyer, 2004). Evaluations based on a health economics and/or the decision-makers’ perspectives have examined the costs and benefits involved in providing a particular genetic intervention e.g. genetic testing for a specific disorder (Holland, Huston and Noyes, 2009; Mvundura et al., 2010; Wordsworth et al., 2010; Perez et al., 2011).

In the case of CGSs, the recommended practice of CEA is brought into question because of the distinguishing characteristics of CGSs. Based on existing methodological knowledge in the field of health economics, a cost-benefit analysis (CBA) is the only type of economic evaluation that incorporates spill-over effects by default, whereas evaluations such as cost-effectiveness analysis (CEA) where investigators could potentially incorporate these effects tend not to (Basu and Meltzer, 2005). Even though CBA is the prevailing method of economic evaluation in other areas such as transport economics (Byford and Sefton, 2003), it has not been favoured by health economists due mainly to its requirement for attaching monetary values to outcomes (Drummond et al., 2005) and its focus on utility as the only indicator of value (Brouwer and Koopmanschap, 2000).

The main difficulty in designing a CBA in health care involves the requirement to identify and value all related costs and benefits from a societal perspective. Identifying all benefits related to a complex health care intervention is difficult (Byford and Sefton, 2003), particularly in the case of CGS where the impact relates not only to the individual
but, as discussed previously, diffuses to other family members and society in general. Even if all consequences are identified, there remains the issue of valuation (i.e. attaching monetary values to these benefits), which is a controversial issue in itself (Drummond et al., 2005). Decision-makers and other stakeholders find the subject of openly valuing health and human lives controversial, despite the implied valuations involved in all health care decisions for resource allocation (Mooney, 2003). In addition to the valuation difficulties inherent in the design of a CBA, the ad hoc organisation of CGSs has resulted in insufficient knowledge of both the models and patient pathways of service provision, something that is required for the design of a CBA (Boardman et al., 2001).

Despite these difficulties, it is the most comprehensive technique of economic evaluation (Drummond et al., 2005), and the only existing methodological approach allowing for all benefits, both tangible and intangible, to be incorporated. A CBA therefore has the potential to influence policy decision-making in a manner representative of the nature of CGSs. Acknowledgement of the superiority of the CBA perspective has begun to emerge in the genetic services literature, with economists arguing for moving away from traditional methodological approaches used in health economics such as extra-welfarism and cost-effectiveness analysis, and towards traditional approaches i.e. welfarism, with their operationalisation through the CBA approach and/or Discrete Choice Experiments (DCEs) (a preference-based approach which can also be used as part of a CBA) (Grosse, Wordsworth and Payne, 2008). The concepts of welfarism and extra-welfarism will be discussed in more detail in Chapter 2.
To date, comprehensive economic evaluations have not been undertaken to assess whether the overall benefits outweigh the overall costs associated with providing CGSs. This is partly the result of limited knowledge of the consequences both tangible (e.g. cost savings) and intangible (i.e. psycho/social), of providing CGSs (Hall, Viney and Haas, 1998; Lenaghan, 1998; Burke et al., 2002; Cohen, Barton and Brain, 2004; Griffith, Edwards and Gray, 2004; Wang, Gonzalez and Merajver, 2004; McAllister et al., 2007b; Payne et al., 2007). Investigators from different branches of research have voiced concerns about the existing methods for the evaluation of CGSs and in particular the outcomes upon which the service has been evaluated. Since existing health-related outcome measures are inadequate to capture the overall impact of genetic services, researchers, economists and non-economists alike have encouraged further understanding of the psychosocial consequences of CGSs (Griffith, Edwards and Gray, 2004; Wang, Gonzalez and Merajver, 2004; Carlson et al., 2005; Skirton, Parsons and Ewings, 2005) as well as the identification of more appropriate outcome measures (Payne et al., 2007; Payne et al., 2008; McAllister, Dunn and Todd, 2011).

At the same time, decision makers and service providers attempting to allocate limited resources in a more efficient manner have turned to the users of health care services for guidance on what their needs are, in order to ensure that the services provided are based on their needs. Patient-reported outcomes have now become a requirement when conducting audits (Black and Jenkinson, 2009) and service users’ preferences are reflected in health care evaluations through preference-based outcome measures e.g. the EQ-5D (Feeny, 2005). Inclusion of service users’ perspectives in evaluations has also been evident within the broader genetics literature (e.g. McAllister, Dunn and Todd,
2011). This reflects an acknowledgement that the patients’ experiences and what service users perceive as important consequences of genetic services need to be better understood (Skirton, Parsons and Ewings, 2005) in order for evaluations to accurately represent the true outcomes of the service (Skirton, 2001; McAllister et al., 2007a).

Current methodological advances and trends have made the design of a CBA a more acceptable task to health economists. For example, the inclusion of service user perspectives and patient-reported outcomes has transferred methodologies from other disciplines into the area of health care evaluation. Qualitative methodologies, once associated with sociological research are now widely used by health care researchers to explore patient views and experiences (Pope and Mays, 2006). Qualitative methods enable the identification of user-derived consequences thereby offering a potentially more accurate description of the wider impact of CGSs. Methodologies derived from welfarist economics have also brought to the fore approaches of valuing and ranking outcomes based on consumer preferences, with the DCE approach gaining in popularity, particularly when evaluating complex health care interventions (Ryan and Farrar, 2000; McIntosh, 2006; Ryan et al., 2006). These developments in the area of health care evaluation make this an ideal time for taking steps towards extending the current knowledge of the full consequences of CGSs. By better understanding these consequences welfarist approaches to evaluation could be further developed for a more comprehensive evaluation and more informed decision-making by stakeholders.
1.4. Aims and objectives

The main aim of this PhD is to identify the benefits and disbenefits (from now on referred to as (dis)benefits) of CGSs whether tangible or intangible. These will be presented within a framework which may be used as a guiding tool by health economists and other stakeholders embarking on a welfarist evaluation of genetic services indicating factors to be taken into consideration during the design of an economic evaluation. This framework will illustrate the following:

- Aims and objectives of genetic services and the patient journey or pathway followed by those referred to a CGS;
- Psychosocial (dis)benefits resulting from the consumption of CGSs;
- Tangible impact to other agencies (NHS or otherwise) where access is facilitated by CGSs and where the patients and their families would not be guaranteed access if they did not receive the services of the clinical genetic service;
- An example of how these costs and consequences would look in a CBA, and within a Discrete Choice Experiment (DCE), a welfarist technique allowing for (monetary) welfare values to be attached on the identified (dis)benefits

The specific objectives of the PhD towards achieving these aims are:

1. To map the patient pathways through the specific CGS;

2. To identify services outside the CGS utilised by those being referred to the service;
3. To identify relevant (dis)benefits of the CGS from the professionals’ perspective;

4. To identify relevant (dis)benefits of the CGS from the service users’ perspective;

5. To identify the most important impacts as these are perceived by the stakeholders of the service;

6. Consider the methodological implications of incorporating these outcomes in economic evaluations.

Commonalities in (dis)benefits across various pathways and conditions were explored by focusing data collection on specific conditions chosen because their individual characteristics (e.g. genetic, clinical, social) and service provision models were varied enough to cover the broader range of conditions and pathways associated with a UK CGS. This issue is discussed in more detail in Chapter 4 of the thesis where methods and research design are presented.

1.5. Structure of the thesis

The remainder of the thesis comprises seven chapters:

Chapter 2:
This chapter will set the theoretical context to the thesis by providing an overview of the principles of economic evaluation i.e. normative economics, and introduce the three techniques of economic evaluation, namely cost-benefit analysis, cost-effectiveness analysis and cost-utility analysis. This chapter will argue that the CBA approach is the most suitable one for the evaluation of increased resource allocation within the context of CGSs and will present the stages towards its design. Within the context of these
stages, the aims and structure of UK CGSs will be described along with the outcomes used so far for the economic evaluation of these services which will be argued to be of more limited value in the case of CGSs.

**Chapter 3:**

This chapter will provide an overview of the psychosocial literature and the outcomes used within this literature for the assessment of psychosocial consequences of CGSs. The appropriateness of these outcomes to be included in an economic evaluation will then be discussed. This chapter will argue that existing psychosocial outcomes are not suitable to be incorporated in an economic evaluation.

**Chapter 4:**

This chapter will provide an overview of the research design and methods used in this thesis in order to achieve the given aims and objectives.

**Chapter 5:**

Chapter 5 will present the procedure and findings of Phase One of data collection. This chapter will provide an overview of the aims and objectives of Phase One and describe the methods and recruitment techniques utilised. Phase One involves face-to-face interviews with genetics professionals in order to map patient pathways and service utilisation within and outside the chosen CGS and explore the perceived role and impacts resulting from service provision. This chapter will present the patient pathways followed by service users for each of five chosen conditions and present the perceived aims, consequences and outcomes of the service as these were discussed by service providers.
Chapter 6:
Chapter 6 will present the procedure and findings of Phase Two of data collection. It will provide an overview of the aims and objectives of Phase Two and describe the methods and recruitment techniques utilised. During Phase Two data from service users of the chosen CGS were collected in order to understand the impact of the service on service users and their families, and identify suitable (dis)benefits for inclusion in an economic evaluation. It will then present and discuss the main findings of the study which include: a descriptive report of the narratives for each of five chosen conditions; main themes representing five service attributes and one conceptual outcome of CGSs incorporating four dimensions.

Chapter 7:
Chapter 7 will present the procedure and findings of the Phase Three of data collection. This chapter will provide an overview of the aims and objectives of Phase Three and describe the methods and recruitment techniques utilised. Phase Three incorporated the quantitative component of this mixed methods research. This phase addressed validity issues and tested the usefulness of Audience Response Systems (ARSs) in preference research. It will then provide a descriptive report of the findings and discuss the methodological implications of this phase and how this knowledge can be used to further the design of an economic evaluation.

Chapter 8:
This chapter is the final chapter of the thesis and incorporates an overall discussion, conclusions, and directions for further research. Chapter 8 will provide an overview of the background and specific objectives of the thesis, the methodological approach
adopted in order to fulfil these objectives and how each one relates to the overall aim, this being the proposition of a framework which can assist in the design of a welfarist economic evaluation. It will then provide a summary of the research findings pulling together the findings from each phase to present a framework for the economic evaluation of CGSs. Findings in terms of (dis)benefits and costs will be presented in the context of a cost-benefit analysis and a discrete choice experiment. A methodological evaluation will then follow discussing the advantages and drawbacks of the combined approach to research adopted in this thesis and discuss how the approach chosen can be used for furthering economic evaluation design within health economics.

1.6. CONCLUSIONS

Rapid advances in biomedical knowledge and the fast rate of transfer of genetic knowledge and technology into clinical practice have made the second half of the twentieth century the “golden scientific age” of medical genetics (Steele, 1992), while the completion of the Human Genome Project has meant that the NHS was unable to provide for the increased demand created from the fast transfer of new genetic knowledge and technologies into medical practice (DoH/Scientific Development & Bioethics Division, 2008). The Department of Health in the 2003 Genetics White Paper and its 2008 update review reaffirms its commitment to the effective and ethical application of new genetic knowledge and technologies as soon as these become available based on its belief that the application of new technologies will bring about a revolution in health care and lead to improved health outcomes in the population (DoH, 2003; DoH/Scientific Development & Bioethics Division, 2008).
To date, despite early concerns of uncontrolled and rapid transfer of technological advances into clinical practice, the rate of development of genetic services has not reflected the rate of expansion in genetics knowledge and available technologies (Donnai and Elles, 2001). However, it is inevitable that genetic services will continue to expand, transferring from the laboratory into clinical practice more genetic tests for more genetic conditions. Investments made in CGSs involve the use of resources which could have been used for the provision of other (health care) services, an economic concept known as opportunity cost (Mooney, 2003). No health care system can fully satisfy all the needs of its users and it is therefore imperative for decision makers to choose which services they should provide, in what quantities and to whom (Cohen, Barton and Brain, 2004) based on the principles of evidence-based care.

In the present climate of budgetary pressures and increasing costs, healthcare and technology assessments have become essential in guiding decision-making in the NHS (Emmerson, Frayne and Goodman, 2000; Catala-Lopez et al., 2011). One aspect of health technology assessment is the economic evaluation of such technologies to assess their effectiveness, an economic “tool” which helps decision-makers assess the costs and benefits of a number of alternative uses of scarce resources (Mooney, 2003; Drummond et al., 2005). Despite the calls for evaluation, there has been a lack of high quality economic evaluations mainly due to the lack of comprehensive understanding of the overall costs and benefits of CGSs, the methodological limitations of popular techniques of economic evaluation, and the lack of an appropriate outcome measure upon which to evaluate such services (e.g. Carlson et al., 2005; Grosse, Wordsworth and Payne, 2008; Payne et al., 2008).
Chapter 1

The current thesis aims to identify the (dis)benefits of CGSs both tangible and intangible and present a framework which may be used as a guiding tool for health economists and other stakeholders embarking on an evaluation of genetic services.

The following chapter will provide an overview of the principles of economic evaluation in order to set the theoretical background to this approach.
CHAPTER 2

ECONOMIC EVALUATION OF CLINICAL GENETIC SERVICES
2.1. Overview of chapter

This chapter follows on from the brief description of the background to the thesis and the presentation of the aims and objectives to begin considering the literature relevant to the aimed framework of (dis)benefits of CGSs. This chapter will:

1. Set the theoretical context to the research by providing an overview of the principles of economic evaluation i.e. normative economics,
2. Introduce the three full techniques of economic evaluation which form the applied side of normative economics, namely cost-benefit analysis (CBA), cost-effectiveness analysis (CEA) and cost-utility analysis (CUA),
3. Propose the CBA approach as the most suitable for evaluating developments in CGSs and present the stages towards its design,
4. Present a description of the aims and structure of UK CGSs,
5. Present a review of the outcomes used in the economic evaluation of CGSs.

2.2. Normative economics

This section will provide an overview of normative economics, the two theoretical frameworks guiding research in normative economics and economic evaluation, and the methodological approaches available for the economic evaluation of healthcare programmes. An overview of this section is given in Figure 2.1.
Figure 2.1: Diagrammatic overview of the principles of economic evaluation

**ECONOMIC PROBLEM**
- Finite resources
- Infinite needs
- Opportunity cost

**NORMATIVE ECONOMICS**
*How should resources be allocated in order to achieve maximum societal well-being (utility) considering scarcity?*

**THEORETICAL WAYS OF APPROACHING THE PROBLEM**

**Welfarism**
- Grounded in economic principles of consumer behaviour and decision-making
- Understanding of utility (well-being) as derived from consumption of goods and services, the process of consumption and in some cases through the consumption (utility) of others
- Acknowledging costs and benefits falling onto society is a prerequisite

**Extra-welfarism**
- Offers an alternative definition of utility allowing for individual characteristics i.e. capabilities and functionings, to act as intermediaries to individual perceptions of well-being
- Introduces the concept of equity as a factor within economic evaluations
- Has adopted health or health-related quality of life as the main outcome of interest

**OPERATIONALISING NORMATIVE ECONOMICS**

**CBA**
Compare between costs and societal benefits

**CUA**
Compare between costs and one multi-dimensional outcome

**CEA**
Compare between costs and one health outcome

**CAPABILITIES APPROACH**

As a starting point to this discussion, a definition of economics is given. Economics is:

“the study of how men and society end up choosing, [with or without the use of money], to employ scarce productive resources that could have alternative uses, to produce various commodities and distribute them for consumption, now or in the future, among various people and groups in society. It analyses the costs and benefits of improving patterns of resource allocation”.


The above definition encompasses concepts which form the basis of health economics from both positive (the description and explanation of economic phenomena) and normative (making value judgements about “what ought to be”) economics. Normative economics form the basis of economic evaluations and thus of essence to this thesis are the concepts of finite resources available for production of any good, including health care services, and the choices that need to be made as a result, guided by the “analyses of the costs and benefits of improving patterns of resource allocations”. Since each resource has a number of alternative uses, limited resources should be utilised in areas where the impact to those involved results in the maximum possible benefit from the given resources.

Economic evaluations provide a way of applying the principles of economics in order to assist decision makers in the private or public healthcare sector to make choices about the best way to use available resources. Drummond et al. (2005)
define an economic evaluation as “the comparative analysis of alternative courses of action in terms of both their costs and consequences” (p. 9). Different types of economic evaluation provide a framework upon which to describe a given project in terms of its inputs (costs) and outputs (consequences), thus helping stakeholders to choose which alternative action to take.

Economic evaluations are able to answer the following questions:

1. Is it worth providing a specific health procedure, service or programme, when this is compared with other interventions we could be providing using the same resources?

2. If the answer to the above is yes, then what is the best way of providing the specific intervention given a certain number of options?

Depending on which of the two questions needs to be answered, different types of evaluation exist to help answer the question. In the case of clinical genetic service provision, the first question is the one that has been least addressed and is in greatest need of addressing (Griffith, Edwards and Gray, 2004; Carlson et al., 2005).

The theoretical basis of economic evaluation lies in the principles of normative economics. This branch of economics deals with value judgements and assessments about which is the best way to organise the economy in order to improve the wellbeing of society (Begg, Fischer and Dornbusch, 2005).

Traditionally, the dominant approach to normative economics has been that of “welfarism”, which forms the theoretical backbone of economic evaluation in
other areas of applied economics. Extra-welfarism is an approach to normative economics developed as an alternative to welfarism (Sen, 1979; Culyer, 1989; Sen, 1991; Sen et al., 1996; Brouwer et al., 2008). The operationalisation of normative (welfare) economics has been through CBA which forms the traditional approach to economic evaluation. Theoretical discussions of extra-welfarism have not been originally associated with an operationalising framework, yet the theory has been influential within healthcare and has led to the development of distinct techniques of economic evaluation, namely cost-effectiveness and cost-utility analysis, as well as to propositions of new approaches to economic evaluation such as the capabilities approach (Anand and Dolan, 2005; Cookson, 2005; Coast, Smith and Lorgelly, 2008a; Coast, Smith and Lorgelly, 2008b).

2.2.1. Theoretical approaches to normative economics

2.2.1.1. Welfarist approaches

Welfare economics involves assessing how well the economy works, rather than how it works, and is divided into theoretical and applied branches with CBA forming the applied form of this theory (Little, 2002). Theoretical welfare economics deals with issues of efficiency, i.e. getting the most out of the resources available (Mooney, 2003) as well as the conditions that need to be satisfied in order for the economy to be working at maximum efficiency. This refers to utilising the resources available so that maximum welfare for the population is achieved. All relevant alternatives therefore need to be ranked based on their impact on utility or well-being. The term utility is a cornerstone for economics and has come to be defined as “the satisfaction or pleasure that an
individual derives from the consumption of a good or service” (Pass, 2005).
Assessing the desirability of given alternatives based on their impact on utility is
guided by the following viewpoints or principles (Morris, Devlin and Parkin, 2007;
Brouwer et al., 2008).

Individualism: Individuals are taken to be the best judges of their own utility and
their behaviour is guided by the single objective of maximising their utility, or well-
being. When decision-makers need to make social choices about the allocation
of resources, only the individuals themselves can judge the impact of these
social choices on their wellbeing, the impact being measured through changes in
their utility. The change in individual utility can be assessed through the
measurement of individual preferences for different bundles of goods or services,
since preference of one bundle over another implies that consumption of the
chosen bundle carries more utility. Since welfare economics is individualistic in
nature, social welfare is taken to be the sum of all individual utilities.

The original idea of measuring utility cardinally (i.e. using a unit measurement of
utility) was abandoned in favour of ordinal utility, the belief that it is not possible
to measure utility accurately, but it is possible to rank levels of utility based on the
preferences of individuals (Morris, Devlin and Parkin, 2007). Utility revealed
through a person’s choices, or decision utility (Dolan and Kahneman, 2008), is
based on traditional microeconomic theory where consumers are assumed to be
rational beings and the best judge of their own utility. These choices ultimately
reveal what they will actually enjoy – choices which maximise their expected
utility. If a bundle of goods and services is preferred over another then the first
bundle has more utility attached to it than the second. It is worth mentioning that policy makers do not talk about individual utility but of collective utility (the sum of individual utilities) or the general welfare of the population. Welfare economics deals with the well-being or welfare of individuals which act as the basic unit of measurement, and it implicitly or explicitly uses the concept of collective well-being. Social welfare refers to the overall utilitarian state of society. Even though a number of issues arise from this assumption, policy makers often have no other measure of well-being for the population than the preferences and therefore utility expressed by individuals (Little, 2002).

**Consumer Choice Theory (Consequentialism):** Under consumer choice theory, all individual consumers are guided in their behaviour by the single objective of maximising their utility. The only variable influencing individual utility under traditional economic theory is consumption of goods and services. Processes themselves or the intention for consumption are not taken to result in any increases in utility. Developments in welfare economics however have allowed for other arguments to feature in the utility function in addition to the consumption of goods and services, such as health (Culyer, 1991), and process utility (Donaldson and Shackley, 1997; Mooney, 1998). In the real world the presence of externalities is also known to exist, where consumption of a good by one consumer influences the utility of another. Considerable externalities are known to be present in the case of healthcare, and two types of external benefits have been discussed: (1) interdependent utility, which incorporates “caring externalities” i.e. B’s health status or utility enters A’s utility function; and (2)
option demand (uncertainty over future need for healthcare leads current non-users willing to pay an amount in the present to ensure that the service will be available at a later time) (Labelle and Hurley, 1992).

Economic Efficiency and the Pareto principle: The decision-rule in welfare economics for judging the desirability of resource-allocation decisions has been the Pareto principle where an option is desirable if no individual can be made better off without another being made worse off. Under Pareto efficiency or optimality, the point achieved is one of allocative efficiency where it is no longer possible to increase the output of one good without reducing the output of another (production efficiency). At the same time the produced output combination is efficiently allocated among the consumers i.e. it is impossible to make one individual better off without making another individual worse off (exchange efficiency) (Nas, 1996; Boardman et al., 2001).

Efficiency involves getting the most out of the resources available (Mooney, 2003). Within a perfect market economy this state is (theoretically) achieved through the price mechanism which enables the market to reach a level of equilibrium in resource allocation where no further improvement in social welfare is possible through the powers of supply and demand. The market equilibrium achieved in the perfect market is efficient since due to the forces of supply and demand, the price of a good equals the marginal cost to the supplier of producing that item and at the same time is equal to the benefit obtained by the consumer of consuming that same item.
Even though Pareto efficiency provides a criterion to judge whether the economy has achieved maximum welfare, in real life it is rarely the case that a change will result in no losses in welfare for any individual, or that all the criteria for perfect markets will be present at once resulting in market failure. In the case of Pareto efficiency, usually it is the case that improvements in welfare take place at the expense of at least one individual. This has led to the concept of “potential Pareto improvement” (also called the Kaldor-Hicks criterion) where changes in production still represent Pareto optimality, as long as the individuals becoming better off are able to compensate those who become worse off. In real life the compensation need not be carried out, only that it would be possible. The Pareto optimality criterion allows economists to distinguish between efficient and inefficient utility distributions and to rank those distributions thus making possible the choice of altering the balance of resources to production in a way that results in maximum utility. In the case of market failure, with the absence of natural market mechanisms to guide reallocation of resources towards a more efficient state, economic analysis provides an alternative framework for guiding resource allocation.

A final point is that in most cases involving choices in service provision, discussions related to choices do not involve the choice of the production or supply of one service or another as in real life most services are provided to some degree. Rather all choices are made at the “margin” (Mooney, 2003), meaning that producers or suppliers are deciding not whether to start providing a given service, but rather how much of that service to produce, or how many
additional units of each service to produce so that maximum utility is achieved for both the supplier and the consumer.

This section introduced the basic concepts of welfare economics. Within this framework the concepts of utility and Pareto efficiency have been the cornerstone and the basis for the design of economic evaluations to assist decision-making. The same concepts however have been controversial within health economics, leading into the proposition of extra-welfarism as an alternative framework for economic evaluation to welfare economics. Extra-welfarism in turn has influenced the design of techniques of economic evaluation argued to be more suitable to health care and more acceptable to health care decision-makers (Brouwer et al., 2008; Coast, Smith and Lorgelly, 2008b).

2.2.1.2. Extra-welfarist approaches

Even though welfarism has been the dominant approach in most areas of economics, in health economics other normative, non-welfarist approaches have been applied with extra-welfarism being the most popular (Brouwer et al., 2008). Whereas welfarism has been defined as “the systematic analysis of the social desirability of any set of arrangements, for example a state of the world or allocation of resources, solely in terms of the utility obtained by individuals” (Morris, Devlin and Parkin, 2007: p.210), extra-welfarism relaxes this restriction placed on utility being the only determinant of social welfare, so that other aspects of each social state are also included in the utility function (Culyer, 1989).
Both “welfarism” and “extra-welfarism” were terms described by Amartya Sen (Sen, 1987; Sen et al., 1996) to criticise the existing approach to economic evaluation, namely welfarism, based on welfarism’s focus on utility as the only measure of individual well-being which he perceived as flawed. Sen commented that individuals differ in their ability to convert commodities into well-being, and their assessment of their well-being and utility are affected by their actual and potential achievements which he termed “functionings” and “capabilities” respectively. Sen also criticised welfarism for not taking into consideration the nature of actions and possible violations of human rights and freedoms limiting the ethical perspective of social decisions based on the welfarist approach (Sen, 2000) [for a discussion on equity and welfarist approaches see Dolan 1998]. Sen’s proposed framework focused on the issue of equity and evaluating policies based on their ability to provide opportunities or capabilities for all individuals to achieve their desired level of functioning (Coast, Smith and Lorgelly, 2008a; Coast, Smith and Lorgelly, 2008b).

Brouwer et al (2008) list four general ways in which extra-welfarism differs from welfarism:

1. it permits the use of outcomes other than utility to be taken into consideration, such as health;

2. it permits the use of sources of valuation other than the affected individuals, such as decision-makers, doctors and other health care providers;
3. it permits the weighting of outcomes (whether utility or other) according to principles that need not be preference-based; and finally
4. it permits interpersonal comparisons of well-being in a variety of dimensions thus enabling movement beyond Paretian economics.

Extra-welfarism is appealing to decision-makers because it speaks a language they can understand as it has come to associate benefit of health care provision to health-related benefits (Morris, Devlin and Parkin, 2007). According to extra-welfarists the welfarist approach of basing social decision-making solely on individual utility is very restrictive, and non-utility information should be allowed in social comparisons (Culyer, 1989; Brouwer et al., 2008). Social comparisons should be made based on “functionings” and “capabilities” of individuals. More important for the evaluation of social states Sen believed to be the capabilities of individuals which involve “the extent to which a person is able to function in a particular way, whether or not he or she chooses to do so” (Coast, Smith and Lorgelly, 2008b: p.1191). For most extra-welfarists however, comparison of social states has come to rely on health as the only outcome of importance for health care provision.

Extra-welfarist economics have been influential in health economics with Culyer taking the notion of something “extra” as significant for social welfare and focusing on one particular characteristic pertinent to health care decision-making, that of health (Culyer, 1991). The concept of “health” as a suitable outcome has given rise to the two most popular economic evaluation techniques in health care, the cost-effectiveness and cost-utility approach. Both approaches are
based on a provider perspective and compare costs to health-related outcome measures such as lives saved, cases detected or Quality Adjusted Life Years (QALYs) (Drummond et al., 2005). As mentioned in Chapter 1, the UK National Institute for Health and Clinical Excellence (NICE) has included the use of CEA and QALYs in its requirements for health technology assessment in the UK. QALYs will be further discussed in the following section.

Even though Sen’s ideas of extra-welfarism have been much broader than simply replacing utility with health (e.g. Cookson, 2005; Coast, Smith and Lorgelly, 2008a; Coast, Smith and Lorgelly, 2008b), so far in health economics extra-welfarism has been synonymous with Culyer’s ideas of health as the most important outcome of health care. The capabilities approach is a recent addition to health economic evaluation (Coast, Smith and Lorgelly, 2008a) despite its extended use within other areas of economics such as developmental economics, and is evidently gaining momentum within health and social care research (e.g. Cookson, 2005; Grewal et al., 2006; Coast, Smith and Lorgelly, 2008a; Coast, Smith and Lorgelly, 2008b). The capabilities approach is arguably more grounded in the original extra-welfarist framework proposed by Amartya Sen and it bases evaluation of programmes on their ability to promote certain abilities or capabilities in individuals, so that they are able to function in a certain way regardless of whether the individual chooses to (Coast, Smith and Lorgelly, 2008a).

This approach has benefits over the existing popular methods of health care evaluation in that it is able to take account of more outcomes than simply health,
as well as tackle issues of equity which so far neither the welfarist nor extra-welfarist approaches have managed to deal with. Coast and colleagues (2008a) present a number of studies which have utilised this approach for developing methods for health care evaluation either using an existing capabilities list (Nussbaum 2003; Lorimer 2007) or by attempting to develop new lists relevant to particular populations (Grewal et al 2006; Kinghorn et al 2007). A number of issues however exist when coming to apply this approach in health care evaluation including the fact that Sen has not incorporated suitable capabilities for evaluation in his theory which leaves the issue of identifying suitable capabilities (Coast, Smith and Lorgelly, 2008a). Other problems related to this approach include the absence of consumer focus when investigating preferences of capabilities within the capabilities approach and how this “expert-centred approach” would fit in the consumer-oriented focus present in health care evaluation; and finally, utility or health maximisation is problematic in the context of the capabilities approach since it is not possible to transfer capabilities from one individual to another, thus the concept of maximisation needs to be reconsidered in the light of equity which is the primary focus of the capability approach (Coast, Smith and Lorgelly, 2008a).

The idea of measuring experienced utility (utility as hedonic experience) as opposed to decision utility (utility as a representation of preference) has also been proposed as an alternative to the QALY (Dolan and Kahneman, 2008). This is because of the impact of condition adaption on utility. Condition adaption refers to “the process of adjustment to new or changed
circumstances” (Dolan and Kahneman, 2008). Dolan and Kahneman (2008) question the validity of QALYs and other techniques which assign utility weights based on hypothetical scenarios, to truly capture these weights because they don’t take into consideration the presence of adaptation. The presence of adaption is argued to bias responses of preferences so the utilities obtained are not accurate. Their critique is based on findings that quality of life does not differ significantly between disabled individuals and control groups since despite their condition individuals rank highly their well-being mainly because they have adapted to their situation (Dolan and Kahneman, 2008). The authors argue that giving lower priority to those who have adjusted to their situation is unjust but at the same time it makes sense to give greater priority to those who have not adjusted to their situation as they will experience higher increases in utility from allocating more resources. This issue of experienced utility is argued to be more relevant when allocating resources amongst patients after a health care budget has been agreed (Dolan and Kahneman, 2008).

2.2.2. Operationalisation of Normative economics

At present a number of economic evaluation techniques can be used for health care evaluation, both full and partial, distinguished by (a) whether they include a comparison of two or more alternatives and (b) whether both costs and consequences of the alternatives are examined. Full economic evaluations incorporate both a comparison between two or more alternatives, and the costs and consequences of these alternatives. Partial economic evaluations on the other hand incorporate one or the other and involve no comparison, therefore no
efficiency questions can be answered by these analyses (Drummond et al., 2005).

Partial economic evaluations are descriptive in nature and may involve: 1) outcome description where only the consequences of the service or programme are incorporated; 2) cost description where only the costs are examined; 3) a cost-outcome description where both costs and consequences are described but no comparison is made with another programme; and finally, 4) evaluations where the costs or consequences of two or more alternatives are compared but not at the same time. Efficacy or effectiveness evaluations compare only the consequences of chosen alternatives whereas cost analyses compare only the costs (Drummond et al., 2005).

Traditionally, normative economics has been operationalised through the design of a cost-benefit analysis (CBA). As discussed in the previous section, within health economics a second theoretical framework i.e. extra-welfarism, has gained in popularity among health economists as the main framework guiding economic evaluation. This framework has been operationalised through two economic analysis techniques popular in health economics, namely cost-effectiveness analysis (CEA) and cost-utility analysis (CUA) even though they adopt only part of this framework’s ideas. These three types of economic assessment are the only types which result in a full economic evaluation where both costs and consequences of two or more alternatives are compared. They are therefore the only types which are able to answer efficiency questions.
2.2.2.1. Cost-effectiveness and cost-utility analyses

Cost-effectiveness analysis is a popular economic evaluation technique in health care evaluation where both costs and consequences of health programmes, interventions or services are assessed. CEAs have received much more acceptance by policy makers and decision-makers than CBAs. One reason for this is that CEAs base their evaluation on the comparison between costs and a single health outcome such as life years saved, cases detected, blood pressure or cholesterol reduction (Drummond et al., 2005), aiming for maximising health impact based on a limited budget. This conceptual basis is arguably more meaningful to decision-makers than monetary valuations of all relevant tangible and intangible benefits for society. For a decision-maker working on a limited budget, maximisation of health impact for the patients concerned is of high importance. CEAs are useful when comparing between interventions resulting in the same health outcome e.g. lowering blood pressure, but are unable to capture any impact of the interventions on the well-being of patients. When evaluating interventions it is important that the appropriate outcome is used for the evaluation and in the case of modern medical and pharmaceutical interventions where health outcomes might be similar, capturing changes in well-being is essential for arriving at a valid conclusion of appropriateness.

Objective health outcome measures incorporated in CEAs have been extensively used in health services research but their adequacy has been questioned (Mooney, 1998; Drummond et al., 2005) as they cannot incorporate changes in a number of health dimensions (Mooney, 1998), do not reflect the preferences of
patients (Petrou and Henderson, 2003) and doctor-patient discrepancies exist in the way health benefit is perceived and reported (Bowling, 2001). Awareness that impact on both length of life and quality of life of patients are of interest has led to the development of health-related quality of life outcome measures which enable the comparison of the overall costs to one outcome measure combining information on quantity and quality of life (Drummond et al., 2005; Kind, 2005). For this purpose, cost-utility analysis (CUA) is used where, similar to a CEA, all relevant costs are compared to a single outcome measure which in this case is multi-dimensional and able to capture changes in well-being as well as changes in physical status (Drummond et al., 2005). Outcome measures used include Healthy Years Equivalent (HYEs), (Gafni, Birch and Mehrez, 1993; Gafni and Birch, 1997) Disability-Adjusted Life Years (DALYs) and the most popular outcome measure used in health care evaluation, the Quality Adjusted Life Year (QALY) (Broome, 1993; Culyer and Wagstaff, 1995),

QALYs recognise that most treatments impact on both the length and the quality of life and reflect the change in survival with a weighting factor for quality of life (Bowling, 2001). The attractiveness of QALYs lies on the fact that unlike physical health outcomes like survival, they reflect preferences for particular health states by attaching utility weights to these states. Since they are generic and a single unit of measurement is derived, QALYs can be used across conditions and interventions in economic analyses. Its health focus appeals to decision-makers who aim to maximise health-related benefits from the use of limited resources, at the same time appealing to health economists because of their preference-based
utility rankings of different health states (Gafni, 2006). At the same time, the CEA/CUA avoid the distributional problems associated with the CBA approach, making the QALY an "income-free" outcome measure (Gafni, 2006).

QALYs are now being used in most clinical trials along with objective health outcomes to capture the patient’s perspective of the impact of treatment provided (Drummond et al., 2005). The most popular tool used to derive QALYs is the EQ-5D, a multi-dimensional generic health and well-being measure encompassing five single-item dimensions: mobility, self-care, usual activities, pain/discomfort and anxiety/depression (Drummond et al., 2005).

Subjective preference-based multidimensional outcomes of health-related quality of life are becoming increasingly popular in health services research as they can identify both changes in health state and quality of life as well as reflect preferences (Drummond et al., 2005). Preference-based measures have received increasing interest from health economists and decision-makers as a result of increasing understanding that incorporating solely health outcomes in evaluations of interventions does not fully capture the impact this intervention might have on patients e.g. (Mooney, 1998). Despite the stated distinction between CEA and CUA in the type of outcome measures used, with CEA using health-related outcomes while CUA incorporating utility information, the guidance offered by NICE is for cost-effectiveness analysis to be a prerequisite of all health technology assessments with QALYs to form the single preference-based outcome measure used in this analysis.
2.2.2.2. Cost-benefit analysis

Cost-benefit analysis is the only economic evaluation technique with its theoretical roots based in welfare economics. It adheres to the principles of utility maximisation and the Pareto principle where all (incremental) costs and benefits (utility) associated with providing the intervention and affecting society (as opposed to the individuals affected or the provider) are taken into consideration, and this means that all sources of utility including externalities can be taken into consideration. An intervention is considered as appropriate only if the costs are less than the benefits, or in other words if it is not possible to make one individual better off without making another worse off (or there is the potential of compensation – Kaldor-Hicks criterion).

CBA was developed especially for the evaluation of public policy issues (Nas, 1996). It involves the identification of all potential costs and benefits, both financial and non-financial, and the comparison of these using decision rules to determine whether the policy or programme under consideration is worthwhile from the perspective of society. This technique offers a framework for answering the question: is it worth providing a given service or an additional unit of a given service, taking into consideration the limited resources available to our disposal?

The broad purpose of CBA is to assist in social decision-making and is specifically aimed to facilitate the more efficient allocation of society’s resources (Broadman et al., 2001). The specific features of CBA are:

- The systematic cataloguing of impacts as benefits (pros) and costs (cons)
The valuing in monetary terms of these pros and cons (assigning weights)

And determining the net benefits of the proposal relative to the status quo and/or other alternative programmes

The main difference between CBA and the other economic evaluation techniques lies in the valuation of benefits. CBA requires that both costs and benefits are valued in common units, thus allowing a direct comparison between the marginal costs of an intervention with its marginal benefits (Drummond et al., 2005). Since a CBA takes a societal perspective, this involves the identification and valuation of all relevant costs and benefits whether these are tangible, as in the case of health outcomes, or intangible, as in the case of psychosocial outcomes such as anxiety or process related outcomes such as satisfaction. For the economic evaluation of goods and services provided through the presence of a market, valuations of costs and benefits are usually calculated based on market prices or the prices consumers are willing to pay for individual impacts, a technique known as “revealed preferences”. In the case of health care services however which are provided by the government, as is the case of CGSs in the UK this technique is not possible due to the absence of market prices (Drummond et al., 2005).

In this case, the most relevant technique to health care evaluation has been contingent valuation most commonly through Willingness to Pay (WTP) which is the main technique used for valuing benefits in a CBA (McIntosh, Donaldson and Ryan, 1999; Sen, 2000; Olsen and Smith, 2001). Contingent valuation uses survey methods to present respondents with hypothetical scenarios about a programme or health benefit and then asks participants to state how much they
would be willing to pay in order to obtain that programme or benefit (Drummond et al., 2005). The theoretical basis of WTP again lies in welfare economics and the preferences revealed by participants through the value attached to different options are taken to represent the utility offered by these options.

A number of benefits have been cited for this approach, such as being sensitive to individual preferences and managing relative weights (Sen, 2000), being grounded in theoretical foundations of welfare economics, imposing no restriction on the number and nature of attributes (e.g. health versus psychosocial attributes) or characteristics of a programme included in the valuation, and finally its ability to explore allocative efficiency improvements through the valuation of benefits in the same unit as costs and subsequent design of CBA (Olsen and Smith, 2001).

A number of drawbacks however have also been cited such as the inability of WTP to take into consideration differences in income (Bala, Mauskopf and Wood, 1999), equity and individual freedoms (Sen, 2000), including the observation that actual WTP studies have failed to match the theoretical benefits professed for this approach (Olsen and Smith, 2001), making this approach at risk of being overshadowed by more recent approaches such as the “capability approach” discussed in previous sections (Smith and Sach, 2009) and the Discrete Choice Experiment (DCE).

DCEs, are an attribute-based stated preference elicitation technique (Ryan, 1999; Ryan and Farrar, 2000; Ryan and Gerard, 2005) which can be used as an
alternative to the traditional methods of valuation. The distinguishing characteristic of this approach is that it allows for the valuation of non-health outcomes and process attributes by breaking down the characteristics of an intervention or programme into a number of attributes described at different levels. These are used to elicit a number of hypothetical scenarios and participants are then asked to choose between scenarios in order to estimate the relative importance of individual attributes (Ryan and Gerard, 2005).

This approach has several advantages: it can estimate whether particular characteristics of a commodity are a predictor of choice; it can estimate the relative importance of different attributes; estimate the demand for a given commodity with specific attributes; and also estimate marginal WTP values for individual attribute levels (McIntosh, 2006). DCEs are also able to present attributes which are relevant to the respondents (Green and Gerard, 2009) as well as attributes related to non-health outcomes and process-related outcomes (Ryan and Gerard, 2005). As with WTP, DCEs have the theoretical advantage of assisting in the design of a CBA (Borghi and Jan, 2008). They have also been praised for being flexible within applied CBAs, but still the increase in DCEs has not been matched by an increase in CBAs (McIntosh, 2006; Grosse, Wordsworth and Payne, 2008). DCEs are also believed to overcome cognitive issues arising when making choices related to the standard gable and time trade-off approaches which are used to estimate quality weights between attributes (Ryan et al., 2006). DCEs however are only able to incorporate a small number of
attributes due to cognitive limitations of human working memory (Smyth et al., 1994).

DCEs have been gaining in popularity and are expected to be used more in health care research (Ryan and Gerard, 2005), even though some methodological drawbacks still exist (e.g. Lancsar and Savage, 2004; Ryan and Gerard, 2005; van Helvoort-Postulart et al., 2009a).

The methodological approaches discussed have made the design of a CBA a more manageable task, since they assist in overcoming the major methodological issues of benefit valuation (Drummond et al., 2005). As was discussed in the section introducing extra-welfarism, CBAs have also been criticised for a number of theoretical issues, specifically for dealing only with efficiency and for not making equity considerations or taking into account characteristics such as health, wealth, handicap, and social isolation which might not be related to utility (Brouwer et al., 2008). It is concerned only with the consequences of the policy or intervention valued by those concerned.

Despite these drawbacks, CBA still presents certain advantages over other economic evaluation techniques. For example, it is the only technique which is able to answer the question of whether a specific goal is worth achieving given the opportunity cost involved (Drummond et al., 2005). With this approach it is possible to calculate the net benefit of each option since all cost and benefits are measured in the same (monetary) unit (Bala, Mauskopf and Wood, 1999). In this sense, CBA has a broader scope than other economic evaluation techniques, as
valuing outcomes in a standardised fashion makes it possible to compare interventions from different policy areas, for example health care interventions with interventions in education (Drummond et al., 2005). In addition, it is possible to incorporate the benefits from an intervention derived by both family, friends, as well as society (Bala, Mauskopf and Wood, 1999) which is now understood to be essential for health care interventions which have broad consequences, such as public health (Borghi and Jan, 2008), as well as CGSs (Hall, Viney and Haas, 1998; Wang, Gonzalez and Merajver, 2004). Finally, it is the only economic evaluation approach founded in traditional economic theory (Drummond et al., 2005).

2.3. Stages to the design of CBA

A number of broad steps are involved when conducting a CBA and these are listed in Box 2.1 (Broadman et al., 2001). Here only steps one to three will be discussed in detail since these are the ones tackled by this thesis.
Box 2.1: Stages to the design of a CBA

1. Specifying the set of alternative projects
2. Deciding whose benefits and costs count (standing)
3. Cataloguing the impacts
4. Selecting measurement indicators (units)
5. Predicting the impacts quantitatively over the life of the project
6. Monetising (attaching monetary values to) all impacts
7. Discussing benefits and costs in order to obtain present values
8. Computing the net present value (NPV) of each alternative
9. Performing sensitivity analysis
10. Making a recommendation based on the NPV and sensitivity analysis

Source: Broadman et al (2001)

2.3.1. Specifying the set of alternative projects

In the early sections of this chapter it was stated that an economic evaluation is a "comparative analysis of alternative courses of action" (Drummond et al., 2005). Decision makers most often have to decide between alternative ways of allocating the resources available to them as each course of action involves an opportunity cost. The set of alternative courses of action need to be specified and one compared in terms of its consequences against the other.

Later in this chapter the available information on the aims and structure of CGSs will be discussed which would be the first step in setting the context for the intervention to be assessed within a CBA. As a second step towards the design
of a comprehensive economic evaluation, this thesis will attempt to further understand the workings and patient pathways within a CGS by using the case study approach.

2.3.2. Deciding whose benefits and costs count (standing)

When implementing one project, the consequences can be varied and impact on a wide range of phenomena. For example, building a hospital does not only impact on the patients that will be cared for by that hospital and the medical professionals that will be employed by the hospital, but it also affects other sectors of the economy that will not have the resources to implement priorities of their own. It can have an effect on the relatives of the patients and health professionals, and might also have an environmental impact in terms of new roads that might need to be built or the population that might be created by that hospital. The economic analyst when starting a CBA needs to decide whose benefits and costs they are going to take into consideration. For example, it could be decided that only the benefits and costs to the government will be taken into consideration (provider perspective). Or that a global perspective needs to be taken, taking into consideration the consequences on all individuals directly or indirectly affected by the project (societal perspective).

In the case of a CBA the preferred approach is the societal perspective as it is based on the welfare foundations of this technique. This study explores consequences other than the ones relevant to the provider which have been the
focus of most existing investigations by turning to the service users themselves following the welfarist framework of evaluation.

2.3.3. Cataloguing the impacts

After the analyst has decided the perspective of the CBA, i.e. which agent’s impacts are going to be taken into account, these need to be identified and listed as costs and benefits and their relevance to the project needs to be justified. Both tangible e.g. the use of healthcare professionals and intangible e.g. increases or decreases in the well-being of the population, benefits need to be taken into consideration when undertaking a CBA. Within an economic evaluation, costs are the opportunity cost of employing various factors of production in the specific project, and these can differ from financial costs.

The gap in knowledge of the precise (dis)benefits of CGSs and in particular the psychosocial (dis)benefits of the service has been discussed by a number of investigators in the literature (Cohen, Barton and Brain, 2004; Griffith, Edwards and Gray, 2004; Carlson et al., 2005; Jarrett and Mugford, 2006) and forms the focus of the current thesis.

2.3.4. Contribution of the present research on other stages of CBA

Even though not directly tackled by the present research investigation, this research will make a contribution to future valuation studies. When all costs and benefits of the projects being considered in a CBA are identified (the focus of this thesis) and their impacts over time are specified, the analyst needs to attach monetary values to them. The reason for this is that in order to calculate what
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the net costs and net benefits are for each project and therefore decide on the best alternative, all costs and benefits need to have a common value attached to them. A monetary value allows the analyst to reach a net figure enabling meaningful decisions to be made. Valuation issues in the context of health care economic evaluation were discussed in section 2.2.2.2. After identifying the (dis)benefits of CGSs the present research will attempt to investigate the preferences of service users aiming to identify which are more important to them, thus setting the parameters for subsequent valuation studies.

2.3.5. DISCUSSION

In the presence of a rapidly developing health care market with increased genetic technologies being available for use in clinical practice, commissioners and service providers need to make decisions as to what is appropriate to be delivered within clinical genetic services in the context of finite budgets and infinite demand or need. Economic evaluations provide a tool for decision making through their grounding in normative economics, a broad theoretical platform focused on how the economy should be organised in order to achieve maximum benefit. The two frameworks under the normative economics umbrella, namely welfarism and extra-welfarism, provide alternative approaches to economic evaluation but only welfarism and its operationalisation through the cost-benefit analysis technique is comprehensive enough to incorporate all costs and all benefits resulting from providing a specific service or technology and encapsulates a societal perspective to economic evaluation where costs and benefits befalling on society rather than on one entity e.g. the service provider,
are incorporated. Most importantly in terms of economic theory, welfarism is the only framework with overall consensus about its grounding in traditional economic theory through its focus on utility, consumer preferences and its addressing of allocative efficiency. Specifically in the case of CGS, the CBA is arguably the most appropriate technique of economic evaluation because of its ability to incorporate both tangible and intangible (dis)benefits as well as its ability to take externalities into consideration, both of which form distinguishing characteristics of these services compared to other less complex health services.

A number of obstacles make the design of a comprehensive cost-benefit analysis difficult, including the knowledge gaps in the event pathways through a clinical genetic service, a lack of understanding of the tangible impact on other services, the overall benefits and disbenefits of clinical genetic services including psychosocial implications and what the service users themselves perceive as benefit from accessing the service.

This thesis will attempt to address these gaps, starting with an overview of the existing knowledge of the structure of CGSs.
2.4. Structure of Clinical Genetic Services

2.4.1. Introduction

The nature of CGSs needs to be described including patient pathways, aims, objectives and activity of the service as well as the resulting outcomes, in order to establish indicators of quality, effectiveness or success, ensure for a comprehensive and valid evaluation of interventions (Wang, Gonzalez and Merajver, 2004) and finally as a first step to conducting a CBA (Broadman et al., 2001). Evaluation of a service whose activity is not clearly known would not reflect the actual implications of that service. In the case of CGS this task is made difficult by the fact that across the European Union there are different practices and genetic services are differently organised (Wonderling et al., 2001; Godard et al., 2003). This to some extent is true for genetic centres in the UK whose level of activity and organisation differ to varying degrees (Donnai and Elles, 2001). This section will present an overview of the developments and nature of genetic services in the UK based on published guidance and service model descriptions.

2.4.2. The development of Medical Genetic Services

Distinctions between medical and clinical genetics are not always clear, with the terms being used interchangeably to indicate the exploitation of biomedical technologies and the application of these technologies within clinical practice, with or without the presence of genetic counselling. Based on the way medical genetics has evolved, services tend to be comprised of clinical genetic services
and laboratory genetic services which may be understood as distinct entities both in terms of service provision and in terms of funding and commissioning (Hughes et al., 1998). Clinical genetic services (CGSs) work closely with laboratory genetic services in order to provide the most appropriate tests for families and ensure the correct analysis of these tests. Laboratory genetic services accept referrals from a number of sources outside CGSs, such as obstetricians, neurologists, paediatricians and other medical professionals. Strictly speaking however, CGSs would not be able to operate the way they do today without the presence and advances made within laboratory genetic services.

This research focuses on the (dis)benefits related to a referral to CGS, rather than the (dis)benefits of the overall medical genetic services i.e. clinical and laboratory services. This is because of the numerous clinical departments linked to laboratory genetic services and which are involved in the genetic testing of patients, the breadth of which would make any investigation of their consequences significantly more complex. CGSs also provide a structured approach to service provision, using protocols of genetic counselling, something that is not controlled for in the provision by other health care specialists. In addition, funding for laboratory genetics services is distinct from the funding allocated to CGSs, specifically in Wales (Hughes et al., 1998), meaning that different quality assessment procedures would be involved for the two parts of the service. This discussion first examines the evolution taking place in the overall medical genetic services before focusing on CGSs because of the link between developments in each of these branches.
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2.4.3. Aims and objectives of Medical Genetic Services

Medical genetic services in the UK are part of multidisciplinary regional centres incorporating a laboratory and a clinical service (Donnai and Elles, 2001). The aims of genetic services have changed through the years, from a more eugenic direction in the early 20th century, to a more individual and family oriented perspective, aiming to respond to the need of its users in identifying risk status for themselves or their offspring (Godard et al., 2003). The activity of genetic services involves assisting families with a genetic risk to live as normally as possible (Donnai and Elles, 2001).

According to the UK Specialised Services National Definitions for Medical Genetics Services (DoH, 2007), a “core genetics service” is defined as:

“a specialised clinical genetics service integrated with a laboratory service which is provided for individuals with/concerned about an inherited disorder with a significant genetic component, and their families” (DoH, 2007: p.2). This definition is an adaptation of the previous 1998 definition, and makes a distinction between the core medical genetics service which is described to be clinically-led, and the genetics services which are a collaboration between laboratory services with or to support other specialties e.g. paediatrics, fetal medicine etc. A core specialised medical genetics service comprises (DoH, 2007):

- Clinical geneticists and genetic counsellors:
  - Offering clinical consultations for diagnosis, counselling and follow-up to patients and their families;
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- Referring appropriate samples to laboratory genetic services for analysis and reporting;
- Communicating genetic testing results to patient/families and referring clinicians;
- Providing education in genetics to other healthcare professionals.

Population screening, prenatal diagnosis and foetal medicine, gene therapy and pharmacogenomics are not included in the responsibilities of genetic services, even though all presumptive positives from population and prenatal screening need to be followed by a genetic test and genetic counselling which fall within the remit of the genetic services (DoH, 2007). Mutation testing for breast, bowel and rare cancers is among the core functions of laboratory genetics services, and some genetic centres now provide dedicated services in the form of Cancer Genetics Services (Wonderling et al., 2001). Mutation testing for some cardiological diseases where there is a significant family history in addition to symptoms is now also included in the core functions of laboratory genetics services, something not present in the previous definition (see DoH, 2007).

2.4.4. Clinical Genetic Services

Clinical Genetic Services are involved in (DoH, 2007):

1. The provision of genetic diagnosis [aided where appropriate by genetic testing provided by the genetics laboratory services];
2. Genetic counselling, being defined as “the process by which patients or relatives at risk of a disorder that may be hereditary are advised of (a) the
consequences of that disorder, (b) the probability of developing and transmitting it, and (c) ways in which this might be prevented or ameliorated”; and

3. Surveillance that can involve continuing contact with the genetic service so that support can be offered, family members counselled and information updated when advances in genetics allow refinement of risks or new alternatives for the family to consider.

After referral of an individual and their family (families are the unit of currency for genetic services) to see a consultant geneticist, the family may first be contacted by a genetic counsellor who will collect background information and in some circumstances also provide genetic counselling at the hospital or in the home. Services are delivered in clinics in the specialist centres, in outreach clinics, in local hospitals, ward or hospital department consultations and in visits to the family’s home.

After the initial contact with a genetic counsellor, an appointment with a clinical geneticist or genetic counsellor is organised for most families, as well as follow-up appointments in order to allow for examination, investigations, diagnosis and counselling to take place. After each session a detailed summary letter is sent to the family along with a copy to the hospital and primary care doctors. In some situations and where it is deemed appropriate, long-term contact with the family is maintained. For late onset disorders the service might make provisions for follow-up of children in these families until the age of consent where genetic testing might be offered. Clinical genetic services are also offered to the
extended family as necessary. Figure 2.2 illustrates the known patient pathway through CGSs.

### 2.4.4.1. Cancer Genetic Services

Following the advances in cancer genetics and particularly the link between certain gene mutations and some common cancers such as hereditary breast and ovarian cancer, demand for genetic counselling and risk identification have increased (Sivell et al., 2007). The Department of Health (DoH) has supported the development of cancer genetics services and now most UK genetic centres offer specific services directed to familial cancers consisting of genetic counselling, genetic risk assessment, genetic testing and cascade screening of other at-risk family members (Wonderling et al., 2001; Sivell et al., 2007).

Specialised cancer genetic services mainly deal with high-risk patients to whom they provide face-to-face genetic counselling and genetic testing if the individual chooses to do so (DoH, 2007), while written information and advice might be provided to medium- and low-risk individuals. Specialised genetic centres work closely with cancer units in order to receive and make referrals for prophylactic, screening or treatment interventions (DoH, 2007).

### 2.4.4.2. Specialised services for women’s health (prenatal genetics and foetal medicine)

The early activity of genetic services has been around obstetrics departments (Biesecker and Marteau, 1999), focused on the genetic counselling of couples
regarding their risk status and subsequent impact on their reproductive choices (Reed, 1980). At present, specialised services for women’s health provide prenatal monitoring, screening and diagnosis of foetuses for a number of genetic conditions, linking with laboratory genetic services for all related tests (DoH, 2007). When a genetic condition is diagnosed or suspected a referral is made to genetic services in order for genetic counselling to be offered to the couple involved and later on genetic testing if that is appropriate (DoH, 2007).
Figure 2.2: Overview of patient pathways through UK genetic centres

- Family referral to clinical genetics team
- Contact by genetic counsellor for background information
- Genetic counselling session in hospital or home
- Clinical appointment with clinical team for examination; investigations; diagnosis; counselling
- Detailed summary letter to family; hospital; primary care physician
- Follow-up appointments (if needed)
- Services offered to extended family
- Long-term contact with family (if-needed)
- Follow-up of children from families with late-onset disorders until age of consent

*Arrows illustrate the sequence of events within the patient pathway.*
2.4.5. DISCUSSION

Clinical Genetics along with Laboratory Genetics are the two parts of an overall Medical Genetic Service. Even though clinical and laboratory genetic services are part of a wider service, they are distinct entities both in terms of service provision and in commissioning. This research will focus on the (dis)benefits of CGSs. In the UK, CGSs aim to provide genetic diagnosis including genetic testing, genetic counselling, and surveillance which may involve different levels of continuity in contact with the service for both the individual and the family unit. Cancer Genetic Services are a recent addition to UK CGSs and have been set up in order to meet the increased demand for genetic counselling and risk identification following the advances in cancer genetics. CGSs also work in collaboration with Specialised Services for Women’s Health which deal with prenatal genetics and foetal medicine. The role of the CGS is to offer genetic counselling to the family and later on genetic testing if that is appropriate.

So far a considerable number of different pathways through the CGS have been reported (Griffith et al., 2005) and the journey through the service does not appear to be homogeneous for all centres (Brain et al., 2003). Broad aims and objectives of the service have been given but the exact model followed by each centre and for each condition appears to be more related to the resources and expertise available rather than to any established guidelines impacting on resource use and referrals to other services, health or otherwise. A more detailed understanding is required therefore of the different alternatives and intervention options available in different circumstances.
2.5. **Review of the economic literature**

2.5.1. **Introduction**

Economic evaluations for genetic technologies have been taking place since the 1970s and these early studies were mainly interested in the costs averted from implementing such programmes. Early economic evaluations focused on the available genetic technologies which at the time focused mainly around prenatal screening and testing for detection of hereditary conditions (e.g. Nelson, Swint and Caskey, 1978; Vintzileos et al., 1998a; Vintzileos et al., 1998b; Vintzileos et al., 1999). Based on their review of economic evaluations in the area of cancer genetic services, Griffith and colleagues (2004) report that the main questions posed in the literature include: 1) The nature of the outcomes or the advantages and disadvantages of cancer genetic services; 2) The nature of the financial costs of conducting testing; 3) The value placed by patients upon genetic services; and finally 4) Identifying the most efficient method of providing genetic services. These research questions can be extended to the wider clinical genetics literature. This review will attempt to present the outcomes or advantages and disadvantages investigated in the literature. A list of these outcomes is given in Table 2.1.

2.6. **Aim and Method of Search Strategy**

The aim of this literature review was to identify the published economic evaluations conducted in the area of CGS and list the outcomes used to evaluate the service. The databases searched for published economic evaluations (last
update September 2011) were CINAHL, EconLit, Medline, EMBASE, PubMed and the Cochrane Library which includes the NHS Economic Evaluation Database and the Health Technology Assessment Database. GoogleScholar was also searched for articles and Google for relevant webpages. Contents of the following economic journals were also searched: *Health Economics, International Journal of Evidence-Based Healthcare, American Journal of Evaluation, Value in Health, Health Services Research, Journal of Health Economics*. Reference lists of retrieved articles were also read for relevant articles and citations were searched using ISI Web of Knowledge. Search terms used included: “economic evaluation” or “cost-effectiveness” or “cost-benefit” or “cost-utility” or “outcome” or “consequence*” + “genetic*” or “genetic service*” or “genetic testing” or “genetic counsel*ing”. A detailed list of all search terms is included in Appendix 2.

A review of the literature was conducted searching for economic evaluations published between 2005 and September 2011. A limit to studies published after 2005 was set as there were three literature reviews of the area published already, with the last one published in 2006 (Jarrett and Mugford, 2006). Only economic evaluations which compared costs to specified benefit(s), discrete choice experiments and contingent valuations/willingness to pay were included. These had to assess some aspect of a clinical genetic service, excluding any evaluations of population screening, prenatal or neonatal screening and pharmacogenomics which are not included among the responsibilities of a CGS.
Overall 1802 papers were retrieved. After excluding all duplicates, papers not in English, commentaries, papers that did not describe an economic evaluation, reviews, cost analyses, economic evaluations of screening programmes, pharmacogenomics and personalised medicine, 206 abstracts were read. Out of these 17 papers fulfilled the search criteria and are included in this review.

2.6.1. A review of the literature

Economic evaluations of genetic services including genetic technologies have been criticised for having considerable methodological drawbacks (Carlson et al., 2005; Jarrett and Mugford, 2006) as well as adopting a eugenic stance. In an early study, Nelson et al (1978) state their aim as being to conduct a cost-benefit analysis of a genetic screening programme for Tay-Sachs disease (TSD), comparing prevention costs and the costs involved in the TSD screening programme, with the medical care costs normally associated with Tay-Sachs babies. Benefits of the programme were described to be humanitarian in nature and associated with reducing the number of Tay-Sachs babies. These benefits were not taken into consideration as the authors describe these consequences as being intangible thus unsuitable to be incorporated in an economic evaluation (Nelson, Swint and Caskey, 1978).

Objective health-related outcomes such as numbers of mutations detected, survival, numbers of pregnancies avoided are common outcomes used in the literature (Griffith, Edwards and Gray, 2004; Carlson et al., 2005; Jarrett and Mugford, 2006). The popularity of objective health-related outcomes is reflected
in the numbers of cost-effectiveness analyses published in the literature. Four literature reviews of economic evaluations in the area of medical genetics have identified cost-effectiveness analysis as the most popular technique of economic evaluation (Griffith, Edwards and Gray, 2004; Carlson et al., 2005; Jarrett and Mugford, 2006; Djalalov et al., 2011). The latest review (Djalalov et al., 2011) reported on the quality of published economic evaluations but did not comment on the suitability of the outcome measures used to capture the overall consequences of genetic technologies/genetic services.

Outcomes used by cost-effectiveness analyses published after 2005 include life-years saved (Phillips et al., 2005), survival and life years gained (Breheny et al., 2006; Ozanne et al., 2009; Wordsworth et al., 2010; Ingles et al., 2011), and cases detected (Cooper et al., 2008).

The large majority of studies evaluated the cost-effectiveness of genetic testing whether compared to other clinical strategies or to the status quo (Phillips et al., 2005; Breheny et al., 2006; Nielsen et al., 2007; Cooper et al., 2008; Wordsworth et al., 2010; Ingles et al., 2011; Perez et al., 2011) or compared different criteria for genetic testing (Kwon et al., 2010). The study by Balmana and colleagues was one of the few studies which conducted a cost-effectiveness study using decision analysis for a genetic counselling and screening programme (Balmana et al., 2004), while only two of the CEA studies published after 2005 took into consideration genetic counselling costs (Kwon et al., 2010; Wordsworth et al., 2010). Only one study has evaluated the impact of genetic testing of female ovarian cancer patients on the life expectancy of their female first degree
relatives (Kwon et al., 2010) despite the considerable spill over effects evident in clinical genetics. The study by Kwon and colleagues has focused on the benefits for first degree relatives even though the authors acknowledge that genetic testing in index cases could potentially have significant benefits for the patients themselves.

Despite calls for moving away from cost-effectiveness analyses and towards other methods of evaluation better able to encapsulate tangible and intangible benefits and externalities of CGSs (Grosse, Wordsworth and Payne, 2008), there has been no increase in the number of DCEs published between 2009, a year after the publication of the paper by Grosse and colleagues calling for moving beyond cost effectiveness analysis, and 2011. On the contrary, there has been an increase in the cost-effectiveness studies which have incorporated a QALY outcome measure. Specifically, one cost-utility study has been published between 2005 and 2009 which evaluated some aspect of CGSs, whereas 6 CUAs had been published between 2009 and 2011, four of which were CEAs incorporating a QALY measure (Ozanne et al., 2009; Kwon et al., 2010; Ingles et al., 2011).

Cost-utility evaluation has been identified as the second most popular technique of economic evaluation in medical genetics evaluations with Quality Adjusted Life Years or QALYs being the most popular outcome (Griffith, Edwards and Gray, 2004; Carlson et al., 2005; Jarrett and Mugford, 2006; Djalalov et al., 2011). QALYs, as discussed previously, are also a requirement of NICE for all technology assessments. In a cost-utility analysis Nielsen et al (2007) assessed
Chapter 2

the societal cost-utility of genetic screening in MUTYH associated polyposis (MAP), which is an autosomal recessive inherited condition by constructing a decision analytic model incorporating QALYs as the measure of benefit. The authors describe their analysis as taking a societal perspective by incorporating health-related non-medical costs including loss of productivity and patient’s time and travel costs associated with health care. Even though the provider perspective is more popular in health care economic evaluations than the societal perspective (and is a requirement of NICE), one issue when adopting a societal perspective in a CUA is the absence from the benefit side of societal benefits, since the QALY reflects the health and well-being of the individual being tested rather than those of the wider family being affected.

Welfarist techniques of economic evaluation such as CBA and discrete choice experiments (DCEs), have the advantage of being able to incorporate benefits relevant to more than one individual while DCEs could be used as a step towards the design of a CBA when they incorporate a willingness-to-pay (WTP) element (Drummond et al., 2005). Existing literature reviews of the area have reported on a number of CBAs and DCEs and some of these have incorporated a WTP element without however comparing the overall costs with the overall benefits between two or more alternatives i.e. being full economic evaluations (Griffith, Edwards and Gray, 2004; Carlson et al., 2005; Jarrett and Mugford, 2006). Three DCEs have been identified through an update of the literature review published between 2005 and 2009. Two of the DCEs used willingness-to-pay as a method of attaching monetary values to the benefits most valued by
participants (Hall et al., 2006; Regier et al., 2009). Both of these studies however, focused on the utility obtained from genetic carrier screening using outcomes or attributes related to the process such as waiting time for results. Hall et al (2006) elicited preferences from the general population about factors that influence willingness to participate in genetic testing for Tay Sachs condition and cystic fibrosis. Regier et al (2009) on the other hand elicited preferences from actual users of a genetics service, these being families with children diagnosed with idiopathic developmental disability. Families were asked to value attributes relevant to a genetic testing programme for identifying genetic causes of this condition. These attributes were “number of children tested whose genetic condition is identified with this test”, “time waiting for results”, and “cost to you”. Neither of these two studies included any psychosocial attributes which might be relevant to a family or individual considering genetic testing, within, or outside the context of CGSs.

One study reporting on a CBA has been identified which compared costs to annual earnings of foetuses born with CF or who were born healthy and has adopted an insurer perspective (Davis et al., 2010). Two DCEs with a WTP attribute and one contingent valuation study have also been identified which incorporated attributes of genetic testing such as test accuracy (Mohamed, Johnson and Knight, 2011; Neumann et al., 2011) and attributes of different models of cancer service provision (Griffith et al., 2009).

Only three studies were found to take into consideration outcomes other than health-related outcomes. One of these studies (the TRACE project – Trial of
Genetic Assessment in Breast Cancer) incorporated psychosocial outcomes in the evaluation of genetic services (Brain et al., 2000a; Cohen, Barton and Brain, 2004) while two studies (one study publishing findings in two papers) explored user preferences (Wilson, Ryan and Haites, 1999; Apicella et al., 2006; Peacock et al., 2006). In the TRACE study a cost-consequences evaluation was designed where costs and benefits were identified for two alternative cancer genetic service provision models incorporating the outcomes of general anxiety, breast cancer worry, perceived risk of breast cancer, knowledge of familial breast cancer and patient satisfaction (Brain et al., 2000a; Cohen, Barton and Brain, 2004). A cost-consequences analysis has been proposed as a better alternative to the other types of economic evaluation because of its ability to reflect more comprehensively the benefits of a service and by being more accessible to decision-makers (Coast, 2004). This technique however, is not a comprehensive technique of economic evaluation since it does not compare the value of costs and benefits and therefore does not address allocative efficiency. In the case of CGSs however, the inadequate knowledge of the overall benefits of genetic services and the inability of the existing outcome measures such as the QALY to encompass the overall psychosocial benefits of such a service mean that a cost-consequences analysis was the most appropriate for the given purpose (Brain et al., 2000a).

Wilson et al (1999) conducted a cost-utility analysis using conjoint analysis (an earlier name for a DCE) of genetic counselling for familial cancer risk. These authors assessed the preferences of patients for process attributes of genetic
counselling in their attempt to evaluate the Scottish model of cancer genetic services. These attributes were: staff seen at the appointment; waiting time till appointment; distance to appointment; and duration of appointment. This study has been cited in the literature as providing useful information on the utility offered by competing models of service provision and the trade-offs of patients between service attributes (Griffith, Edwards and Gray, 2004), but has not incorporated any outcomes more relevant to the nature of these services.

Only the third DCE study (Peacock et al., 2006) was found to incorporate attributes more relevant to the actual aims and objectives of CGS. The focus of this study was on the utility offered to users of genetic counselling by exploring the preferences for different genetic counselling scenarios of Jewish women who had received a BRCA 1 and BRCA 2 genetic test result through an Australian cancer genetics service. The study focused on attributes related to the function of genetic counselling rather than service-related outcomes, and these were identified through reviews of peer reviewed and policy/practice literature on genetic counselling, consultation with clinical geneticists, genetic counsellors, heads of genetic services and psychologists, and also interviews with participants (Apicella et al., 2006). These attributes are outcomes well researched in the genetic counselling literature, namely: providing genetic and cancer risk information; preparation of clients for genetic testing; offering surveillance advice; and help in deciding whether or not to have a genetic test.

Table 2.1 lists the outcomes used in the health economic literature to evaluate CGSs so far and which have been discussed in this section. The studies which
have used these outcomes are mentioned only when a small number of studies have incorporated these in evaluations. In the case of health outcomes and health and quality of life outcomes where the majority of studies have utilised these, no specific reference is given.
# Table 2.1: Outcomes used in the economic literature

<table>
<thead>
<tr>
<th>Process/ function/disease related outcomes</th>
<th>Health outcomes</th>
<th>Health and Quality of Life</th>
<th>Psychosocial outcomes</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Time waiting for results (Regier et al 2009; Griffith et al 2009)</td>
<td>• Number of children tested whose genetic condition is identified with the test</td>
<td>• QALYs</td>
<td>• General anxiety (Brain et al 2000)</td>
</tr>
<tr>
<td>• Providing and explaining cancer, gene and risk information (Peacock et al 2006)</td>
<td>• Cases detected</td>
<td></td>
<td>• Breast cancer worry (Brain et al 2000)</td>
</tr>
<tr>
<td>• Explaining options and giving advice about appropriate surveillance for breast and ovarian cancer (Peacock et al 2006)</td>
<td>• Chance of affected child experiencing mild versus severe symptoms</td>
<td></td>
<td>• Perceived risk of breast cancer (Brain et al 2000)</td>
</tr>
<tr>
<td>• Preparing for the outcomes of genetic testing (Peacock et al 2006)</td>
<td>• Cancer-free years gained</td>
<td></td>
<td>• Knowledge of familial breast cancer (Brain et al 2000)</td>
</tr>
<tr>
<td>• Receiving help in deciding whether or not to have a genetic test (Peacock et al 2006)</td>
<td>• Survival</td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Proportion of people who have been tested/test availability (Hall et al 2006; Griffith et al 2009)</td>
<td>• Life-years saved / life years gained</td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Place of testing (Regier et al 2009)</td>
<td>• Carriers detected</td>
<td></td>
<td></td>
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<tr>
<td>• Staff seen at appointment (Wilson et al 1999; Griffith et al 2009)</td>
<td>• Fetuses detected</td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Waiting time till appointment/distance/duration (Wilson et al 1999; Griffith et al 2009)</td>
<td>• Births averted</td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Who sees results (Mohamed et al 2011)</td>
<td>• Events prevented</td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Cost</td>
<td>• Mutations detected</td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Patient satisfaction (Brain et al 2000)</td>
<td>• Cases prevented/averted</td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Disease risk (Neumann et al, early view; Mohamed et al 2011)</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>• Accuracy of test (Neumann et al, early view; Mohamed et al 2011)</td>
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<td></td>
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<tr>
<td>• Annual earnings (Davis et al 2010)</td>
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</tbody>
</table>
2.6.2. DISCUSSION

Through a review of the literature it appears that no economic evaluation has been published which has incorporated the overall consequences of CGSs and this is mostly due to the inadequate knowledge of the broader psychosocial consequences of CGSs (Cohen, Barton and Brain, 2004; Griffith, Edwards and Gray, 2004). The majority of studies have used health-related outcomes to evaluate the service, with the exception of Wilson et al (1999) and Griffith et al (2009) who assessed the preferences of service users for a number of process attributes of genetic counselling and cancer genetic services respectively, the TRACE study (Brain et al., 2000a; Cohen, Barton and Brain, 2004) which assessed the impact of a multidisciplinary specialist cancer genetic clinic on a number of psychological outcomes and finally Peacock et al (Peacock et al., 2006) who conducted a DCE with attributes related to the actual aims of genetic counselling. The large majority of studies have investigated cancer genetic services and this is reiterated by two literature reviews of the area (Carlson et al., 2005; Jarrett and Mugford, 2006), while one systematic review has been published actually reviewing economic evaluations in the area of cancer genetic services (Griffith, Edwards and Gray, 2004).

Cost-effectiveness analysis has been appealing to decision-makers because of its use of comparison between costs and a single health-related outcome. This has direct relevance to the concerns of decision-makers who aim to maximise health benefits on a finite budget (Drummond et al., 2005). The use of the QALY has gained great momentum in healthcare, being the most widely used outcome measure for economic
evaluations in healthcare and a requirement of NICE, despite its limitations. QALYs present intuitive appeal to healthcare researchers because they encapsulate the apparent essence of healthcare delivery, namely health and well-being. Since they are generic they can be used across conditions and interventions in economic evaluations, making it easy to make comparisons between evaluations (Drummond et al., 2005). They are relevant to decision-makers and health economists alike, since their conceptual background is close to decision-makers’ aims of maximising health in the population, as well as measuring preference-based utility (Mooney, 2003; Gafni, 2006). The preference of CEA/CUA and subsequently of the QALY is also based on the distancing of this approach from monetary valuation and distributional issues central to the CBA approach, making the QALY an “income-free” measure of health consequences (Gafni, 2006).

Despite the apparent appeal of CEA/CUA and the QALY, it has been concluded that overall this is not a superior approach to economic evaluation compared to the CBA (Gafni, 2006), especially in the case of genetic services (Grosse, Wordsworth and Payne, 2008). A focus on health-related outcomes does not do justice to these services, particularly when the health-related outcomes are compared to those of other health care services since genetics professionals do not deal directly with treating the condition (Clarke, Parsons and Williams, 1996). At the same time CGSs do not primarily deal with producing health outcomes and it is arguable whether the overall consequences such as reassurance and information might be adequately captured by a QALY (Shackley and Cairns, 1996; Hall, Viney and Haas, 1998; Wang, Gonzalez and Merajver, 2004). CGSs are in essence a family service, with genetic diagnosis in one
individual having consequences on the well-being of other family members (Procter, 2002) but QALYs do not capture anything beyond the quality of life of the individual being investigated raising the question of whose benefits should be taken into consideration (Shackley and Cairns, 1996; Hall, Viney and Haas, 1998). In addition, evaluations which measure health benefits in terms of a single physical or natural unit are not able to capture the several impacts or health changes that might result on a user of the service within that single outcome, or capture individual preferences (derived utility) for those changes (Petrou and Henderson, 2003).

These findings raise certain concerns about the conclusions of these studies whose evaluations are not wholly valid representations of the cost-benefit aspect of the service since neither the overall costs nor the benefits of these services are currently known (Cohen, Barton and Brain, 2004; Griffith, Edwards and Gray, 2004). Investigators have repeatedly criticised current research and repeated the same concerns about the appropriateness of used outcomes for evaluating these services (Hall, Viney and Haas, 1998; Cohen, Barton and Brain, 2004; Griffith, Edwards and Gray, 2004; Wang, Gonzalez and Merajver, 2004; Carlson et al., 2005; Jarrett and Mugford, 2006; Grosse, Wordsworth and Payne, 2008). In an early critique of the literature Hall et al described the tendency to use health outcomes as a simplistic approach to a complex issue (Hall, Viney and Haas, 1998) since the nature of the service makes the majority of consequences resulting from its use psychosocial in nature. The authors comment that the use of traditional economic evaluation techniques in genetic testing for multifactorial conditions and less severe or late onset conditions fails to incorporate the wider effects for individuals, families and the wider social impact since information is the immediate
outcome and not health changes (Hall, Viney and Haas, 1998). Reviews have commented on the low methodological quality of most economic evaluations and the absence from these evaluations of psychosocial outcomes related to genetics. Both Griffith et al (Griffith, Edwards and Gray, 2004) and Jarrett and Mugford (Jarrett and Mugford, 2006) call for economic evaluations that go beyond health outcomes, while Jarrett and Mugford call for economists to develop methods which encompass psychosocial outcomes of genetic services (Jarrett and Mugford, 2006).

These limitations make the existing evaluations of limited use to decision-makers (Jarrett and Mugford, 2006), and evidence already shows that despite the “mainstreaming” of economic evaluations with support by NICE and the UK Department of Health (Schlander, 2008), the usefulness of economic evaluation data for decision makers has been limited (Hoffmann et al., 2002; Williams et al., 2008). Recently a number of economists have advocated the use of measurement techniques more relevant to a cost-benefit analysis such as DCEs (Grosse, Wordsworth and Payne, 2008) and contingent valuations (Borghi and Jan, 2008), as well as the idea of measuring experienced utility rather than decision utility (Dolan and Kahneman, 2008).

2.7. CONCLUSIONS

So far, the Department of Health has expressed its intentions of incorporating into clinical practice all new genetic technologies and making these available to the wider population. In the present climate of finite budgets, the increasing number of biomedical technologies coming into the market put pressure on commissioners as to what should be provided in clinical practice. Economic evaluations provide a tool for decision
making through their grounding in normative economics, a broad theoretical platform focused on how the economy should be organised in order to achieve maximum benefit. Within health economics, cost effectiveness and cost utility analyses have been the two most widely used economic evaluation techniques but these have so far been methodologically limited in encapsulating the overall impact of clinical genetic services due to single (health-related) outcomes incorporated in these techniques whereas CGS have a wider, psychosocial impact. These techniques are also linked to the extra-welfarist framework of normative economics which does not abide to traditional economic principles. The cost-benefit analysis technique is therefore argued to be the most appropriate technique of economic evaluation because of its ability to incorporate both tangible and intangible (dis)benefits as well as its ability to take externalities into consideration, both of which form distinguishing characteristics of these services compared to other less complex health services.

So far economic evaluations have used mainly health-related outcome measures or process attributes and these do not encapsulate the mainly psychosocial impact of CGSs which has been extensively discussed in the literature. In addition, existing variation in event pathways followed by users of UK CGSs presents difficulties when attempting to map the different alternative pathways through these services, a prerequisite of all economic evaluations when trying to identify resource use and impact on other services.

The following chapter will present the literature on the psychosocial outcomes used to evaluate CGSs in an attempt to identify the broader consequences of the service and assess their appropriateness to be incorporated in an economic evaluation.
CHAPTER 3

PSYCHOSOCIAL IMPACT OF CLINICAL GENETIC SERVICES
3.1. Overview

This chapter will identify the psychosocial outcomes used to assess the impact of Clinical Genetic Services (CGSs) by non-health economists. More specifically it will:

1. Present an overview of the psychological literature and the outcomes used to assess CGSs;
2. Give a discussion of the appropriateness of these outcomes for use in an economic evaluation.

3.2. Aim and Method of Search Strategy

The aim of this literature review was to identify the psychosocial outcomes used to evaluate CGSs. In order to identify studies which have used psychosocial outcomes to evaluate some aspect of CGSs the following databases were searched: CINAHL, MEDLINE, Embase, PsychInfo, Cochrane Library, ScienceDirect, and Google was also searched for relevant WebPages. Non-economics journals individually searched included: the BMJ, Journal of Genetic Counselling, British Journal of Cancer, Social Science and Medicine, American Journal of Medical Genetics, Health Policy, Genetics in Medicine, Journal of Health Psychology. Reference lists of retrieved articles were also read for relevant articles and citations were searched using ISI Web of Knowledge and Google Scholar. Search terms for the psychosocial literature included: “genetic service*” or “genetic* testing” or “predictive testing” or “genetic counsel*ing”+ “psychosocial” or “emotional” or psycho*” or consequence*” or “impact”. A detailed list of search terms used in included in Appendix 2.
This review will focus on the psychosocial implications of undergoing genetic counselling or cancer genetic risk assessment, pre-symptomatic and predictive genetic testing of adults following genetic counselling, and the implications of a genetic diagnosis of children on families and adult patients. Literature on the implications of population, prenatal and neonatal screening was excluded from this review because these services are provided outside CGSs. Literature on the implications of predictive genetic testing of children was also excluded because of the on-going debates on its appropriateness (Clarke and Gaff, 2008). Only studies which evaluated a specific outcome before and after genetic service provision or which explicitly assessed the impact of genetic service provision on a specific outcome e.g. behaviour, were included.

A limit to studies published after 2005 was set because a systematic review investigating the outcome measures used for the evaluation of clinical genetic services had already been published in 2007 (Payne et al., 2008). In addition to this comprehensive review, numerous literature updates and systematic reviews have been published providing an overview of the outcomes and psychosocial impact of clinical genetic services on its users. Reviews have been published between 2007 and 2011 reviewing risk perception (Smerecnik et al., 2009; Tilburt et al., 2011); outcomes of cancer genetic risk assessment on women (Sivell et al., 2007) as well as on their male partners (Sherman, Kasparian and Mireskandari, 2010); the outcomes of genetic counselling (Aalfs, Smets and Leschot, 2007; Kasparian, Wakefield and Meiser, 2007; Smerecnik et al., 2009); genetic testing (Cohn et al., 2008; Douma et al., 2008; Heshka et al., 2008; Hamilton, Lobel and Moyer, 2009; Picot et al., 2009; Vansenne, Bossuyt and de Borgie, 2009); screening participation of high risk individuals (Rees, Martin and
Macrae, 2008); and overall clinical genetic services (Payne et al., 2008). Over 3000 papers were retrieved based on the literature search terms and 240 papers were found to be relevant for the review. Twenty-one of these studies were randomised studies and 22 were literature or systematic reviews.

3.3. Psychosocial consequences of Clinical Genetic Services

3.3.1. Introduction

The psychosocial literature investigating the consequences of CGSs is far broader than that of health economics. Whereas economic evaluations have focused on the cost-effectiveness of genetic testing using mainly objective outcome measures (Griffith, Edwards and Gray, 2004; Jarrett and Mugford, 2006), the psychosocial literature has assessed several aspects of genetic services including genetic testing, genetic counselling, and cancer genetic risk assessment using a variety of subjective, patient-reported psychosocial and behaviour outcome measures (Payne et al., 2008). Psychosocial assessments have also focused on: genetic services for specific hereditary conditions, e.g. cancer genetics, single gene hereditary conditions; and on the life stage of the family or individual when contact with the service is initiated e.g. reproductive genetics, diagnostic or predisposition genetic testing in childhood or adulthood. The majority of studies have focused on cancer genetic services evaluating the impact of genetic risk assessment (e.g. Bennett et al., 2008; Maheu et al., 2010); communication of risk information within families following genetic counselling (e.g. Hopwood, 2005; Forrest et al., 2008; Balck et al., 2011); benefit of surveillance
programmes (e.g. Reis et al., 2009) and changes in health behaviours (e.g. Bennett et al., 2007b; Dorval et al., 2008; Wevers et al., 2011).

Despite the many facets of the service undergoing evaluation, common outcomes have been used. So far the trend has been for studies to use mainly non-genetics outcome measures, even though several genetics specific outcome measures have been designed (Payne et al., 2008). The remainder of this review will present the outcomes that have been of interest to researchers. These outcomes are categorised into the following groups (see Table 3.1. for a breakdown of common outcomes in each category):

- Emotion-related outcomes
- Cognitive outcomes
- Behavioural outcomes
- Adjustment-related outcomes
- Social outcomes
- Satisfaction
Table 3.1: Outcomes used in the psychosocial literature

<table>
<thead>
<tr>
<th>Emotion-related outcomes</th>
<th>Cognitive Outcomes</th>
<th>Behavioural outcomes</th>
<th>Adjustment-related outcomes</th>
<th>Social Outcomes</th>
<th>Satisfaction</th>
</tr>
</thead>
<tbody>
<tr>
<td>Psychological distress</td>
<td>Education focus e.g. Information recall</td>
<td>Breast screening uptake</td>
<td>Coping</td>
<td>Family implications e.g. family functioning, family communication</td>
<td>Satisfaction with genetic counselling</td>
</tr>
<tr>
<td>Anxiety</td>
<td>Knowledge</td>
<td>Uptake of prophylactic surgery</td>
<td>PPC</td>
<td>Insurance implications</td>
<td>Satisfaction with decision-making</td>
</tr>
<tr>
<td>Depression</td>
<td>Risk perception</td>
<td>Use of menopausal therapy options</td>
<td>Self-efficacy</td>
<td></td>
<td>Satisfaction with informed choice</td>
</tr>
<tr>
<td>State-specific worry</td>
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</table>

### 3.3.2. Emotion-related outcomes

Emotional outcomes of genetic services constitute the majority of outcome measures used for evaluating CGSs. The volume of the literature is reflected in the numerous literature reviews which aim to bring together findings from the published studies (e.g. Bredart et al., 1998; Broadstock, Michie and Marteau, 2000; Butow et al., 2003; Braithwaite et al., 2004; Schlich-Bakker, ten Kroode and Ausems, 2006; Heshka et al., 2008; Payne et al., 2008; Hamilton, Lobel and Moyer, 2009; Picot et al., 2009; Vansenne, Bossuyt and de Borgie, 2009). Literature reviews, reflecting the divisions within the literature, have tended to focus on particular aspects of service provision e.g. genetic testing, genetic counselling and genetic risk assessment, as well as on specific types of genetic conditions, usually focusing on cancer related conditions (e.g. Bredart et al., 1998; Meiser and Halliday, 2002; Braithwaite et al., 2004; Schlich-Bakker, ten Kroode and Ausems, 2006; Heshka et al., 2008; Hamilton, Lobel and Moyer, 2009;
So far, only one systematic review has brought together validated outcome measures used across the broad range of services provided by CGSs (Payne et al., 2008).

The broad focus of evaluation studies makes attempts to draw conclusions regarding the positive or negative implications of a CGS problematic. Reviews report that the majority of studies have used a prospective design, measuring a number of emotional outcomes usually before the first genetic counselling session and at different time points following DNA testing or disclosure of genetic test results (Broadstock, Michie and Marteau, 2000; Schlich-Bakker, ten Kroode and Ausems, 2006), even though only a part of all service users will undergo genetic testing.

Emotional well-being has been assessed using a variety of outcome measures related to emotional constructs such as anxiety, depression, worry and mood (Kasparian, Wakefield and Meiser, 2007; Heshka et al., 2008; Payne et al., 2008). Meiser and Halliday (2002) identified generalised distress and anxiety in addition to accuracy of perceived risk as the most researched outcomes, while Braithwaite et al (2007) reported that the outcome investigated most often was cancer worry. This overview will be structured around the main themes and outcomes present in the literature which are:

- Psychological distress
- Anxiety and Depression
- State-specific worry
3.3.2.1. Psychological distress

Findings from literature review studies have concluded that genetic testing does not appear to lead to any long-term increases in psychological distress in either carriers, non-carriers, or patients (Heshka et al., 2008; Hamilton, Lobel and Moyer, 2009; Vansenne, Bossuyt and de Borgie, 2009), individuals with inconclusive test results (Hamilton, Lobel and Moyer, 2009) or breast cancer patients (Schlich-Bakker, ten Kroode and Ausems, 2006). It is pointed out that within the literature the dimensions of anxiety, depression and worry are sometimes incorporated within the construct of psychological distress, reporting the implications for these dimensions together (e.g. Broadstock, Michie and Marteau, 2000; Schlich-Bakker, ten Kroode and Ausems, 2006). Here, these dimensions are discussed separately, even though it is acknowledged that reports of distress in the literature sometimes incorporate measures of anxiety, depression and worry.

Long term follow up (between 2.5 and seven years following genetic test result disclosure) of women with inconclusive, positive and true negative genetic test results for hereditary breast and ovarian cancer (HBOC) showed that genetic test results were found to result in decreased levels of cancer-related distress despite the genetic test outcome (van Dijk et al., 2008; Hamilton, Lobel and Moyer, 2009) suggesting that genetic test result disclosure might not be the main cause of distress. Studies have shown that the strongest predictor of emotional distress following genetic testing appeared to be pre-testing levels of distress (van Oostrom et al., 2007b; Graves et al., 2011; Rosenblatt et al., 2011). Other predictors of distress include complicated (i.e. problematic) grief, number of affected first-degree relatives and strong emotional illness
representations (van Oostrom et al., 2007b), marital status (Condello et al., 2007) and health status (Keller et al., 2002; Condello et al., 2007; Keller et al., 2008; Ertmanski et al., 2009), risk perceptions (Christiaans et al., 2009) and emotional or coping responses (van Oostrom et al., 2007b; Dougall et al., 2009; Murakami, 2010).

Pre-testing risk status awareness (Broadstock, Michie and Marteau, 2000; Rosenblatt et al., 2011) and being part of a high risk family with at least one family member diagnosed with cancer was also found to be related to increased psychological distress (van Dooren et al., 2005; Rabin et al., 2007; van Oostrom et al., 2007b). Other factors gaining evidence on their impact on psychological distress are the loss experiences of Hereditary Breast and Ovarian Cancer (HBOC) high risk individuals, where women who have lost a relative from cancer have been found to experience higher levels of anxiety (Hopwood et al., 2001; van Oostrom et al., 2003; Hopwood, 2005; van Oostrom et al., 2007b); receiving uninformative genetic test results (O'Neill et al., 2009); having small children (van Oostrom et al., 2003); and lack of social support (Lammens et al., 2010). Time appeared as an important variable for changes in emotional outcomes (Hamilton, Lobel and Moyer, 2009) with emotional responses right after testing found to differ from those at various points of follow-up.

Service delivery models appear to have an ameliorating effect on negative emotional outcomes of genetic testing with genetic counselling positively affecting genetic testing outcomes (e.g. Broadstock, Michie and Marteau, 2000; Hamilton, Lobel and Moyer, 2009; Ashida et al., 2010; Hooker et al., 2011), even though one study reported increased levels of distress after oncogenic counselling for cancer genetic test disclosure (Condello et al., 2007). This study however did not have a follow-up period
for assessing the long term impact of counselling. The effects of genetic counselling were not found to be affected by differences between service delivery models, but notably it appears that information given during genetic counselling within UK genetic centres is broadly similar (Hopwood et al., 2004; Hopwood, 2005).

Studies have found a positive impact of genetic counselling on levels of distress, with individuals at high cancer risk reporting decreased distress following genetic counselling (Codori et al., 2005; Mikkelsen et al., 2009; Roussi et al., 2010; Hasenbring et al., 2011). Roussi and colleagues report that genetic counselling appears to be more beneficial for carriers in terms of decreases of generalised distress compared to non-carriers (Roussi et al., 2010) suggesting a more targeted approach to genetic counselling following result disclosure. Emotional distress following counselling has again been found to be positively correlated with baseline levels of distress and anxiety symptoms (Schlich-Bakker, ten Kroode and Ausems, 2006; Hasenbring et al., 2011) and to be inversely correlated with a tolerance for ambiguity (Codori et al., 2005). Age, social circumstances, personality and coping characteristics were all found to be associated with emotional well-being following genetic counselling (Schlich-Bakker, ten Kroode and Ausems, 2006; Hart et al., 2011; Hasenbring et al., 2011) and with health status (Keller et al., 2008). Decreases in psychological distress have also been linked to increases in self-confidence and sense of control following genetic counselling (Keller et al., 2002). Cancer genetic risk assessment has also been found to help reduce distress (Sivell et al., 2007).
3.3.2.2. Anxiety and Depression

The dimension of depression tends to be incorporated with that of anxiety and it is rare that studies have investigated this dimension in isolation, most probably because used outcome measures tend to measure the two dimensions together e.g. Hospital Anxiety and Depression Scale (HADS) (see: Schlich-Bakker, ten Kroode and Ausems, 2006; Payne et al., 2008; Hamilton, Lobel and Moyer, 2009). For this reason these two dimensions are discussed together.

Individuals undergoing genetic testing for HBOC are found to experience somewhat elevated levels of anxiety during the process of blood sampling where higher levels of anxiety have also been found to be a predictor of long-term distress (van Oostrom et al., 2003; Rosenblatt et al., 2011). Cancer patients have also been found to experience higher levels of anxiety during genetic testing (Hasenbring et al., 2011). Based on the literature review by Heshka and colleagues (2008), the majority of studies identified found no change between pre- and post-testing anxiety levels in HBOC gene carriers 12 months after testing, with similar results observed for non-carriers. Lack of a negative impact on anxiety levels as a result of genetic testing has also been concluded in the literature review by Schlich-Bakker et al (2006). Hamilton et al (2009) found significant changes in magnitude of anxiety experienced between carriers, non-carriers and those with inconclusive test results at short-term follow-up studies (0-4 weeks), but no difference in the magnitude of such changes after a moderate (5-24 weeks) or long period of time (25-52 weeks). One of the few studies with a follow-up of more than one year had found that five years after genetic testing for BRCA1/2 both carriers and non-carriers showed a significant increase in anxiety and depression, this being associated
with increased visits to professionals for psychosocial support (van Oostrom et al., 2003). A more recent study which investigated long-term psychological distress at onset of regular surveillance for HBOC high risk women and at five-eight years follow-up found no decreases in anxiety and depression, even though this study was only published in abstract form and does not describe provision of genetic counselling (Den Heijer et al., 2011).

Again, similar to psychological distress, factors other than having genetic testing appear to influence the levels of anxiety and depression in service users. Increased anxiety has been linked to baseline levels of distress, family experiences and having children (van Oostrom et al., 2003; Rosenblatt et al., 2011). Other predictors to such changes in pre- and post-testing anxiety levels were found to be time, gender and age among men and women testing for HNPCC (Hasenbring et al., 2011). Depression has been linked to individuals declining genetic testing, where higher levels of depression have been found in those women deciding to decline cancer genetic testing than those who have gone through the process (van Oostrom and Tibben, 2004).

Individuals who share their genetic test results with health professionals and friends have also been found to have decreased anxiety and depression levels one year after genetic testing (Ashida et al., 2010), suggesting inclinations to face up to the genetic risk. A passive coping style has been found to be correlated with high anxiety levels even one year after disclosure of genetic test result (Claes et al., 2005; van Oostrom et al., 2007b) and five to eight years after onset of regular surveillance for HBOC (Den Heijer et al., 2011). Coping styles refer to habitual preferences of the ways one approaches or deals with problems, with passive coping referring to a passive or
submissive way of dealing with problems such as praying, hoping or generally submitting control to external sources (Lazarus and Folkman, 1984; Folkman et al., 1986). Despite hope being considered an aspect of passive coping, increased levels of hope before genetic testing have been found to be a determinant of lower levels of anxiety and depression post-genetic testing (Ho et al., 2010). Research however has illustrated that at different time points individuals may adopt different coping techniques in order to deal with specific stressors (Phelps et al., 2010). Family and personal experiences with the genetic condition emerge as important for experiences of negative emotional outcomes (van Oostrom et al., 2003; Den Heijer et al., 2011; Rosenblatt et al., 2011) and how threatened people feel from the genetic condition appears to mediate the impact of the genetic test result on anxiety levels (Michie, French and Marteau, 2002). Genetic counselling in general, models of genetic counselling and cancer genetic risk assessment have also been found to decrease anxiety and depression levels (Brain et al., 2000a; Bowen et al., 2004; Keller et al., 2008; Graves et al., 2010; Hasenbring et al., 2011).

3.3.2.3. State-specific worry

Several studies have measured state-specific worry mainly of cancer-related conditions in individuals at high risk, and worry is among the domains included most often in outcome measures assessing emotional well-being in cancer high risk individuals (Payne et al., 2008). Comparisons of state-specific worry have shown that cancer worry may differ between carriers and non-carriers (Heshka et al., 2008; Metcalfe et al., 2010; Fantini-Hauwel et al., 2011; Graves et al., 2011) but not between conditions (Fantini-Hauwel et al., 2011) or between men and women (Graves et al., 2011). Long-term
follow-up (between 2.5 and seven years following genetic test result disclosure), has found levels of worry to be significantly lower in women with inconclusive, positive and true negative genetic test results for HBOC (van Dijk et al., 2008). Genetic counselling (Graves et al., 2010; Roussi et al., 2010) and cancer genetic risk assessment (Sivell et al., 2007) have also been found to lead to decreased cancer worry.

Cancer worry, again, appears to be associated with pre-testing levels of distress (Graves et al., 2011; Hart et al., 2011), and risk perceptions (van Oostrom et al., 2003; Cabrera et al., 2010; Den Heijer et al., 2011), with lower pre-test levels of distress and lower perceived risk for cancer being associated with lower post-testing levels of cancer worry. Coping responses have also been found to affect cancer worry with emotional coping styles to be associated with increased state worry in mutation carriers compared to non-carriers (Den Heijer et al., 2011; Fantini-Hauwel et al., 2011). Social factors have also been found to affect cancer worry levels including experiences with affected relatives (Vodermaier, Esplen and Maheu, 2010), family communication patterns and social networks (Lammens et al., 2010; Graves et al., 2011). Women who had lost one or more close relatives from HBOC tended to report higher levels of cancer worry at five-year follow-up, and so did those who utilised a less open manner of communication about genetic test results with relatives (van Oostrom et al., 2003). Vodermaier and colleagues conclude that past and on-going experience of cancer in the family may have a stronger influence on cancer specific distress than other factors such as time point of carrier notification (Vodermaier, Esplen and Maheu, 2010).
3.3.3. Cognitive outcomes

Cognitive outcomes relate to thought processes such as knowledge, understanding and decision-making and have been used to evaluate changes in these outcomes following genetic testing (Broadstock, Michie and Marteau, 2000), genetic counselling (Braithwaite et al., 2004; Kasparian, Wakefield and Meiser, 2007; Smerecnik et al., 2009), and cancer genetic risk assessment (Sivell et al., 2007). These outcomes are linked to the educational aspect of genetic counselling and CGSs and its goals of informing and educating service users in order to facilitate decision-making. Cognitive outcomes have been linked to emotional outcomes such as psychological distress and anxiety (Butow et al., 2003; Hopwood, 2005; Tilburt et al., 2011) as well as to health behaviours (Sivell et al., 2008; Tilburt et al., 2011) (a more detailed discussion about the associations between these three categories will be given in the section reviewing behavioural outcomes).

3.3.3.1. Early focus on education

In early reviews of the literature, the major outcomes used to evaluate genetic counselling have been reported to be educational effectiveness, reproductive intentions and behaviour and risk assessment, while the most common outcome used was the amount of diagnostic and risk information patients can remember i.e. information recall (Michie and Marteau, 1996). Several issues are associated with these outcomes in relation to their appropriateness. Assessing the effectiveness of CGSs based on decision-making and its impact on decreasing affected cases is highly controversial because of the associations to eugenics (Pilnick and Dingwall, 2001), while information recall might be inconclusive because of the role of the researcher in determining what
should be remembered (Michie and Marteau, 1996). More recently, cognitive outcomes have been limited to knowledge and risk perceptions because of their assumed link to informed decision-making.

### 3.3.3.2. Knowledge and risk perceptions

Accuracy of perceived risk and assessments of risk perceptions are among the most common outcomes used to evaluate genetic counselling and cancer genetic risk assessment in the literature. (e.g. Codori et al., 2005; Schlich-Bakker, ten Kroode and Ausems, 2006; Smets et al., 2006; Heshka et al., 2008). This is not surprising considering that informing individuals of their or their children's risk for a hereditary condition in order to support decision-making is one of the earliest stated aims of genetic counselling (Kessler, 1979). Facilitating decision-making has been described as the “core” of genetic counselling with genetic risk accuracy argued to be a central concept for measuring its effectiveness (Smerecnik et al., 2009).

Genetic service provision is known to impact on levels of perceived risk through genetic testing and genetic counselling (Meiser and Halliday, 2002; Butow et al., 2003; Smerecnik et al., 2009; Tilburt et al., 2011). Genetic counselling can result in improvements in knowledge with those receiving counselling having higher levels of knowledge (Randall et al., 2001; Cabrera et al., 2010), and lower or more accurate risk perceptions (Matloff et al., 2006; Halbert et al., 2010) than individuals not receiving genetic counselling. Cancer genetic risk assessment services have also been found to improve the accuracy of perceived risk and increase knowledge about breast cancer and genetics (Sivell et al., 2007).
The focus on risk perceptions has been supported by suggestions that risk perceptions influence decision-making and behaviours, with higher risk perceptions being associated with inappropriate screening behaviours (Sivell et al., 2008; Den Heijer et al., 2011) and adopting a more healthy lifestyle in the case of familial cardiovascular disease (Claassen et al., 2010). Findings have shown that inappropriate screening behaviours associated with unrealistic cancer risk perceptions persist even after negative genetic testing results (Duprez et al., 2010). Risk perceptions have also been associated with emotional outcomes such as breast cancer anxiety (Cabrera et al., 2010; Den Heijer et al., 2011) and distress (van Dooren et al., 2005; Cabrera et al., 2010) with inaccurate risk perceptions of high risk being associated with increased anxiety. Evidence has shown that the understanding of risk is a complex process influenced by many facets in an individual’s experience (Sivell et al., 2008; Tilburt et al., 2011). Individual factors such as family experience with breast cancer or other genetic disease also influence risk perceptions with higher perceived risk being associated with more members of the family being affected (Hopwood et al., 2001; Claassen et al., 2010).

3.3.4. Behavioural outcomes

Health psychologists have developed a special interest in behaviours and specifically health behaviours because of their link to health status (Conner and Norman, 1995; Manderbacka, Lundberg and Martikainen, 1999). A health behaviour is “any activity undertaken by a person believing himself to be healthy for the purpose of preventing a condition or detecting it at an asymptomatic stage” (Kasl and Cobb 1966: cited in Conner and Norman, 1995); as well as by individuals with existing conditions with the
objective of self-management, delaying condition progression and improving general well-being (Conner and Norman, 1995). Behaviour change within health psychology has been mostly assessed using intentions and motivation to change as well as attitudes towards the desired target behaviour (Ogden et al., 2007), with the most frequently used measure of motivation being intentions to change (Marteau and Lerman, 2001). Conditions which have seen an exponential increase in frequency of onset in recent years and which have been linked to genetic inheritance such as coronary heart condition and cancer are linked to behavioural patterns such as diet and exercise (e.g. Trichopoulou et al., 2003; Hung et al., 2004; Leitzmann et al., 2007).

With the ability to test for conditions where patient behaviour plays a role in health outcomes, the impact of CGSs on specific behaviours such as screening practices and adopting healthy lifestyles is now under assessment. Providing genetic risk information to these individuals might have an impact on health outcomes by setting up individually targeted and more appropriate interventions (Fisher et al., 2011) such as lifestyle modification and attending screening programmes. While some commentators have argued the special nature of CGSs where health outcomes are not the primary objective (Clarke, Parsons and Williams, 1996), others have claimed that genetic services should ultimately aim for an improvement in long-term health status and public health (Wang, Gonzalez and Merajver, 2004). Wang et al (2004) have claimed that genetic services should be evaluated upon their impact on modifiable risk factors. Genetic services should provide support to individuals in order to help them reduce the risk of yet untreatable conditions by adopting certain health behaviours such as attending
screening programmes and having prophylactic surgery (Wang, Gonzalez and Merajver, 2004).

Behavioural outcomes have been investigated to assess the impact of genetic counselling (Hayat Roshanai et al., 2009) and genetic risk assessment (Sivell et al., 2007) on behaviour change. Sivell et al (2007) identified changes in health behaviours as an outcome used to evaluate cancer genetic risk assessment services while genetic counselees at risk for genetic conditions have reported adherence to surveillance programmes (Hayat Roshanai et al., 2009) or to make life-style changes (McKinnon et al., 2007). A family history of genetic disease has been found to be associated to a higher likelihood of adopting a healthy lifestyle in the case of both cardiovascular disease (Zlot et al., 2010) and diabetes (Pijl et al., 2009). Heshka et al (2008) identified preventive behaviours such as mammography and breast exams, prophylactic surgery, diet and exercise, as outcomes used to evaluate genetic testing in the literature. Individuals undergoing genetic testing were reported to have high uptake levels of both prophylactic surgery and report engagement in prophylactic activities (Heshka et al., 2008). More recent studies have suggested that high risk for genetic conditions or a familial history of genetic conditions such as cardiovascular disease and diabetes may prompt patients to adopt healthy lifestyles or screening behaviours (Pijl et al., 2009; Claassen et al., 2010; Zlot et al., 2010) or express intentions for such (Ramsey et al., 2010; Sutphen et al., 2010). Chao and colleagues in the context of a randomised clinical trial assessed the impact of Alzheimer’s disease genetic risk information provided to asymptomatic patients on health behaviours and found that those receiving positive genetic test results were more likely to make positive changes to their health
behaviours than those receiving negative test results one year after disclosure (Chao et al., 2008). Even if intentions do not always lead to actual behaviour, studies have found that having positive attitudes towards prophylactic practices make it more likely to adopt such practices in the future (Julian-Reynier et al., 2010; Landsbergen et al., 2010). Intentions to adopt or change behaviours rather than actual changes have been more often investigated in the literature (Bennett et al., 2007b; Sutphen et al., 2010). Genetic counselling has been found to have a positive impact on the intentions of high risk women to engage in cancer risk management behaviours (Sutphen et al., 2010).

Cognitive and emotional outcomes have also been found to be associated with preventive behaviours (Codori et al., 2005; Antill et al., 2006; De Leeuw, van Vliet and Ausems, 2008; Den Heijer et al., 2011; Schwartz et al., 2011) and have been used as a proxy to screening behaviours after findings that inaccurate risk perceptions and high levels of distress are correlated with lower levels of cancer screening uptake (Codori et al., 2005). Breast cancer worry has been found to be positively associated with screening behaviours, with more breast cancer worry being associated with increased likelihood to engage in screening behaviours (Hay, McCaul and Magnan, 2006), as well as higher intentions for adopting screening behaviours (Sherman et al., 2009). Pre-counselling anxiety in carriers has been found to be associated with subsequent uptake of risk-reducing mastectomy (Schwartz et al., 2011). In a review of the literature on the psychosocial predictors of adopting screening behaviours and prophylactic surgery within high risk women for HBOC, DNA test results, cancer worry and anxiety, age, perceived risk, recommendations from physicians and family history are reported to be predictors of cancer screening practices (De Leeuw, van Vliet and Ausems, 2008).
Family history has also been found to be associated with perceived risk for developing cardiovascular disease and the adoption of a healthy lifestyle in individuals who have been diagnosed with Familial Hypercholesterolemia through DNA testing (Claassen et al., 2010). Claasen and colleagues report that only about half of those included in the study reported adopting advice concerning healthy diet and exercise uptake (Claassen et al., 2010). In a different study in the context of a PhD thesis Claasen and colleagues conclude that family disease history might be a more important predictor than DNA information in explaining perceptions of and responses to risk (Claassen, 2011).

The impact of genetic services on behaviour has only recently received attention from researchers compared to the psychosocial consequences of the service, most importantly because of their relevance to conditions where successful interventions can be designed for improving health outcomes and which have only recently entered the realm of genetic testing. Research studies investigating behaviour change or intentions for behaviour change are limited and existing results are contradictory making any conclusions difficult to make (Marteau and Lerman, 2001; De Leeuw, van Vliet and Ausems, 2008). Commentators of the literature have also pointed out on methodological drawbacks of the literature with absence of prospective longitudinal studies and inconsistent definitions of high risk between the studies (De Leeuw, van Vliet and Ausems, 2008). Behavioural outcomes have been proposed as suitable outcomes of CGSs because of the potential of CGSs to impact on health outcomes in the case of conditions where such intervention is possible. The direct link of health behaviours to psychosocial variables however suggests that broader constructs might be required to capture the complex ways in which CGSs affects its users.
3.3.5. Adjustment-related outcomes

One of the main and widely cited goals of genetic services and specifically genetic counselling is to assist families to deal with and adjust to their genetic condition (Resta et al., 2006). Evaluating CGSs based on its ability to promote adaptation and adjustment is now emerging as an accepted approach among researchers particularly after qualitative research evidence has shown emotional adjustment as an outcome of the service (Skirton, 2001). So far, one outcome measure has been specifically designed to address service users’ Psychological Adaptation to Genetic Information (PAGIS) and has been psychometrically assessed (Read, Perry and Duffy, 2005). In the majority of research studies however, emotional adjustment has been operationalised using non-validated item scales (e.g. van Dijk et al., 2008), emotion-related constructs (e.g. Vodermaier, Esplen and Maheu, 2010) and more recently broader concepts which capture both emotional and cognitive dimensions acknowledging the psychosocial perspective of CGSs such as coping, perceived personal control and self-efficacy (e.g. Pieterse et al., 2005b).

The ability to cope and perceived personal control are related aspects of psychological adjustment and emotional well-being. More specifically, personal control and self-efficacy have been described as personal coping resources (Thoits, 1995). Coping, self-efficacy and personal control are constructs emerging from health psychology and utilised within clinical genetics research in order to assess whether specific situations are stressful and appraise the process of dealing or coping with the situation (Lazarus and Folkman, 1984). Before moving on to the review of the literature, these three concepts will first be defined.
**Coping** refers to the ability of an individual to deal with the stresses and pressures resulting from a situation assessed as such, in this case the presence of a genetic condition. In order to deal with the various stressors presented during one’s lifetime, several coping strategies or styles have been identified which one may utilise depending on the assessment of the situation and one’s decision about which is the best choice in order to deal with the specific stressor. These may consist of behaviour-focused and/or cognitive-focused strategies including dealing with: the stressor itself i.e. problem-focused strategies, the emotional reactions resulting from the stressor i.e. emotion-focused strategies (Lazarus and Folkman, 1984). Active coping styles tend to involve strategies where one decides to take certain actions towards alleviating the stressor whereas passive styles of coping might involve strategies such as avoidance or hoping (Thoits, 1995). Reviewing the literature shows that coping as a concept has been operationalised in different ways. Generic coping-specific measures, adaptation to genetic information, personal control and adjustment are all concepts used to operationalise coping within the literature (Kasparian, Wakefield and Meiser, 2007; Payne et al., 2008; Shiloh et al., 2008). Within the context of clinical genetic services, Phelps and colleagues have designed and tested a cancer genetics-specific measure of coping with the risk assessment process which they named GRACE (the Genetic Risk Assessment Coping Evaluation) (Phelps et al., 2010).

**Sense of control** or mastery over life has been described as a personal coping resource which influences the coping strategies individuals utilize in order to deal with the stressors in their lives (Thoits, 1995). Coping with health threats has been found to be influenced by perceptions of control over the health-related stressor and to adapting
to different health problems. Individuals high in perceived control tend to utilize active, problem-focused strategies while those with low perceived control tend to use passive or avoidance strategies (Thoits, 1995). An outcome measure has been developed designed to capture perceptions of personal control, namely Perceived Personal Control (PPC) (Berkenstadt et al., 1999). PPC refers to the belief of an individual that they have the resources available to deal with negative effects of a stressful event (Davey et al., 2005). PPC encompasses three dimensions, namely behavioural control, cognitive control and decisional control which according to Berkenstadt and colleagues relate to most of the goals of genetic counselling (Berkenstadt et al., 1999). These dimensions have also been found to be appropriate outcomes for evaluating genetic counselling in a study investigating the beliefs of genetic nurses on suitable outcomes of CGSs (Williams et al., 2001).

**Self-efficacy** refers to an individual's belief that they are able to perform a particular action (Bandura, 1977; Bandura, 1982; Ozer and Bandura, 1990). Self-efficacy has been discussed in the literature as a personal coping resource along with a sense of control and has featured as a critical concept within adjustment to life events research (Bjorvatn et al., 2008). This construct has not received as much attention as the ones of coping and perceived sense of control. It has been investigated however as one of the constructs in the Psychological Adaptation to Genetics Information outcome measure, defined as “the perception of control over the consequences of the condition-related gene” (Read, Perry and Duffy, 2005:p.204 ). More recently, an outcome measure assessing the self-efficacy of genetic counselees to deal with a number of tasks and
challenges arising during and after genetic counselling has been designed and used (Bjorvatn et al., 2008).

### 3.3.5.1. Coping

Contact with CGSs is associated with considerable distress during the time individuals await for their genetic risk assessment (Bennett et al., 2007a; Hilgart et al., 2010; Phelps et al., 2010) and genetic testing results (O’Neill et al., 2009; Ashida et al., 2010). Distress is also linked to different types of stressors for each specific situation e.g. dealing with concerns from other family members and anxiety over the genetic test result among others (Phelps et al., 2010). In order to assist women in dealing with the uncertainty and associated distress linked to waiting for genetic risk assessment results, Phelps and colleagues designed a genetic risk assessment-specific measure (the GRACE) aimed to assess levels of distress during the risk assessment process and identify coping strategies adopted by individuals to deal with their stress levels (Phelps et al., 2010).

Coping style within psychological research has been found to be a determinant of emotional reactions (van Oostrom et al., 2007a; Geirdal and Dahl, 2008; Shiloh et al., 2008), with the emotional impact of mutation status being moderated by the individual’s coping style (Shiloh et al., 2008; Den Heijer et al., 2011; Fantini-Hauwel et al., 2011). Coping styles that have been associated with positive as well as negative emotional outcomes following genetic risk disclosure include monitoring coping style (Shiloh et al., 2008), emotional coping style (Geirdal and Dahl, 2008; Fantini-Hauwel et al., 2011), passive and palliative coping styles (van Oostrom et al., 2007a; Den Heijer et al., 2011) and avoidant coping (Dougall et al., 2009). The relationship between coping style and
negative emotional responses has been found to differ depending on risk status, with higher distress and depression levels to be found in those with a positive or indeterminate test result and who adopt a monitoring coping style (Shiloh et al., 2008). On the contrary, emotion-focused coping strategies have been associated with higher anxiety in women with indeterminate cancer genetic test results (Geirdal and Dahl, 2008). Carriers adopting both a problem-focused strategy approach (Geirdal and Dahl, 2008) and emotional, palliative and passive coping styles were found to be associated with increased levels of anxiety following genetic test disclosure (Geirdal and Dahl, 2008; Fantini-Hauwel et al., 2011) and in the long term (Den Heijer et al., 2011).

3.3.5.2. Perceived Personal Control

Perceived Personal Control (PPC) is a concept which has gained in popularity within the literature and has been used to evaluate genetic counselling (Wang, Gonzalez and Merajver, 2004; Kasparian, Wakefield and Meiser, 2007; Payne et al., 2008). PPC was proposed as an appropriate outcome for evaluating genetic counselling by Berkenstadt and colleagues based on the convergence between the dimensions proposed for PPC by Averill and the suggested goals of genetic counselling by Fraser (Berkenstadt et al., 1999). The three dimensions for PPC were: 1) Behavioural control, which refers to the availability of an instrumental response able to directly influence or change the physical characteristics of a stressful event; 2) Cognitive control, which refers to the processing of information and subsequently make a potentially threatening situation less stressful; and 3) Decisional control, which is the opportunity to choose among various courses of action (Berkenstadt et al., 1999). Fraser similarly described the goals of genetic counselling as “a communication process meant to help an individual or family
comprehend medical facts including diagnosis, probable cause, and available management of a disorder, to understand how heredity contributes to the disorder and the risk of recurrence in relatives, to choose and follow the most appropriate course of action in view of risks and family goals, to make the best possible adjustment to a disorder in an affected family member and to deal with the risk of recurrence of that disorder” (in Berkenstadt et al., 1999: p.82).

Perceived Personal Control has been found to increase significantly after counselling. Research investigating PPC in HBOC affected and non-affected genetic counselees has shown that levels of perceived control are higher following genetic counselling in both groups and these levels appear to be maintained over time (Pieterse et al., in press in press). Levels of uncertainty have been found to influence perceived control and higher levels of control were found among those given more definite information in terms of diagnosis and risk status. Uncertainty in parents whose child was diagnosed with a rare disorder, was found to be significantly associated with lower levels of PPC and perceived helpfulness of the genetic counselling (Lipinski et al., 2006). Davey et al (2005) reported differences in post-counselling perceived control and satisfaction between users of three types of genetics clinics, namely familial cancer clinics, general genetics clinics including antenatal and paediatric clinics. Participants in the general genetics clinic reported highest scores in satisfaction and perceived control followed by those attending the paediatric and finally the familial cancer clinics (Davey et al., 2005). Arguably more uncertainty is involved in familial cancer genetics where the majority of patients would receive an inconclusive test result (van Dijk et al., 2008).
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PPC was also found to correlate with knowledge, satisfaction, counselling evaluations and expectation fulfilment (Berkenstadt et al., 1999; Pieterse et al., in press). PPC is also reported to be related with emotional outcomes like anxiety (Davey et al., 2005; Pieterse et al., 2005b) with PPC being inversely related to levels of anxiety after genetic counselling (Pieterse et al., 2005b). What is important with PPC is that individuals do not need to exert direct control over a situation in order to feel able to cope with the situation. Rather it is the perceived ability that they can effectively manage the situation that is key for coping (Davey et al., 2005).

More recently qualitative research by McAllister and colleagues has broadened the PPC framework to incorporate other benefits of CGSs including family impact (McAllister et al., 2008a; McAllister et al., 2008b; McAllister, Dunn and Todd, 2011), a dimension inherent in genetic service provision yet one that has been largely ignored in the literature (Payne et al., 2008). McAllister and colleagues have called their broadened framework “empowerment” which they defined as “a belief system that allows a person to feel in, or take control of their lives and have responsibility or autonomy over decisions and choices” (McAllister et al., 2008a; p.898). This construct is made up of four belief-related dimensions, namely that one: 1) is able to make important life decisions in an informed way (decision-making); 2) has sufficient information about the condition, including risks to oneself and one’s relatives, and any treatment, prevention and support available (knowledge and understanding); 3) is able to make effective use of the health and social care systems for the benefit of the whole family (instrumentality); and 4) can look to the future having hope for a fulfilling family life, for
oneself, one’s family and/or one’s future descendants (future orientation) (McAllister et al., 2008a).

Original findings were based on focus groups with stakeholders of CGSs (patients, health professionals and support group representatives) who were asked to describe the perceived role of the service and what the service needs to provide in order to maximise benefits, “what patients want from clinical genetic services” (McAllister et al., 2008a, p. 896). In this sense, discussions tapped into the expectations of service users rather than what they had actually received. More recent research however has tested the validity of the construct through interviews and focus groups with service users, health professionals and patient representatives where they explored the actual benefits of the service as well as their views on the representativeness of the empowerment construct to actual benefits (McAllister, Dunn and Todd, 2011). In their restructured model of empowerment, the authors renamed some of the dimensions of the construct with three of them adopting terms from the PPC outcome measures and a new dimension was added. Namely the new five-dimensional outcome of empowerment was stated to include: cognitive control (previously knowledge and understanding), behavioural control (previously Instrumentality), and decisional control (previously decision-making), hope (previously future orientation) and emotional regulation (previously not encapsulated in the model). This construct has recently been developed into an outcome measure for evaluating genetic counselling (McAllister et al., 2011).

3.3.5.3. Self-efficacy

Not only actual coping style but also perceived self-efficacy around coping with a positive test result has been found to be associated with negative emotional reactions...
More specifically, lower perceived self-efficacy in coping with a positive genetic test result was found to be associated with higher levels of cancer worry, and higher levels of decisional conflict (Peterson et al. 2008). Higher levels of perceived self-efficacy concerning tasks and challenges arising during and after genetic counselling (e.g. recall of genetic information, dealing with difficult emotions, communicating risk information to family members) have also been associated with lower levels of anxiety and depression (Bjorvatn et al. 2008). Self-efficacy has been suggested to be a moderator of the relationship between cancer-related distress and mental health, with higher levels of perceived self-efficacy being associated with improved states of mental health (Carlsson et al., 2004). Other research has investigated perceptions of self-efficacy in relation to behaviour change and feeling able to make the appropriate behaviour changes to decrease risk associated with receiving risk information for a genetic condition but no impact was found of risk information on perceived self-efficacy (Sanderson, Persky and Michie, 2010).

3.3.6. Social Outcomes

3.3.6.1. Family Implications

In addition to psychological changes observed in the individual as a consequence of the genetic service, the nature of hereditary conditions means that implications diffuse to the wider family environment in terms of both emotional implications such as distress (Lammens et al., 2011) as well as the communication and relationships between patients and other family members (Koehly et al., 2008; Vos et al., 2011). One of the
reasons for choosing to contact clinical genetic services has been found to be worry for other family members (Brain et al., 2000b; Kausmeyer et al., 2006b; Schlich-Bakker et al., 2007). Undergoing genetic testing may impact on relationships within families, for example, among siblings, parents and offspring, or affect the partners of those who undergo testing (Lim et al., 2004; Manne et al., 2004; Lammens et al., 2011). The cascading fashion in which genetic testing of family members takes place may also have emotional implications for the members involved (Hadley et al., 2010). In one study Hadley and colleagues investigated the impact of having family members utilising genetic services in the past, on members subsequently accessing the service. The authors found that in the cases where the index case belonged to the same generation as the individual being subsequently tested, cascade screening resulted in increased genetic test-related distress and cancer worry especially when testing took place at increasing distant time intervals (Hadley et al., 2010).

Communication of genetic risk status and genetic test results have been investigated in the context of genetic counselling and genetic risk assessment because of the importance of informing family members of their family’s genetic situation, something that genetic counsellors actively encourage. Genetic counselling has been suggested to play a role in supporting service users with the process of risk disclosure by discussing family reactions and options for dealing with difficult situations (Hopwood, 2005). Research on family communication has been mainly limited to those families with hereditary cancers (Kasparian, Wakefield and Meiser, 2007) with very few studies focusing on more traditional Mendelian conditions (Gaff et al., 2007; Maxwell et al., 2009; McConkie-Rosell, Heise and Spiridigliozi, 2009). In the case of hereditary
cancer, identifying and recording family history is a prerequisite for assessing individual risk to a genetic condition. Genetic testing of family members requires the genetic testing of an affected individual creating interdependency between family members in terms of establishing carrier or risk status. Following the identification of risk status or diagnosis irrespective of condition the probands shoulder the responsibility of informing their family of their genetic status and provide information about the genetics of the condition and the possible risk to other family members (Gaff et al., 2007). This is a complex process and research has illustrated how decisions to contact the service and receive genetic counselling depends on the ability, knowledge levels and confidence of the proband in providing appropriate information to other family members (Nycum, Avard and Knoppers, 2009; Cheung et al., 2010; Vos et al., 2011). Targeted genetic counselling addressing communication of results to other family members has been found to result in increased levels of communication in those receiving enhanced counselling (Forrest et al., 2008) and this approach can result in greater satisfaction in the way individuals communicate their genetic results to others (Roshanai et al., 2009).

Only a minority of members are often informed of the identification of a genetic mutation in the family (Balck et al., 2011) and informing other relatives is often associated with family history and family context, and the proband’s perceptions of family duty (Hopwood, 2005; Stoffel et al., 2008; McCann et al., 2009; Nycum, Avard and Knoppers, 2009). Family cohesion or degree of the relationship between family members may influence decisions to inform other family members (Stoffel et al., 2008; Nycum, Avard and Knoppers, 2009) and not surprisingly informing others may be seen as a way of receiving social support (Roussi and Miller, 2005). Communication patterns
have also been found to be influenced by socioeconomic status, with individuals from a lower socioeconomic class being less likely to communicate their genetic status with their family members (Cheung et al., 2010).

Despite the lack of importance attributed to family communication in the past by genetics providers (Michie et al., 1998; Williams et al., 2001), it is now realised that this is an important aspect of genetic service provision (Gaff et al., 2007). In their systematic review of the process of communication of genetic information, Gaff and colleagues (2007) propose that it might be more appropriate for researchers to assess the process of counselling about family communication which involves dealing with family dynamics and relationships during the counselling process rather than the more quantitative variables used in the literature.

3.3.6.2. Insurance implications

Research has also investigated the implications of accessing CGSs on obtaining health and/or life insurance. Qualitative studies have identified the theme of insurance in discussions with users of genetic services. Participants in research based in the USA have expressed concern that genetic testing might result in stigma and discrimination in employment and insurance (Lewis, Konda and Rubin, 2009; Shostak, Zarhin and Ottman, 2011) while Australians who were offered genetic testing for colorectal cancer in the context of the Victorian Colorectal Cancer Family Study (VCCFS) reported insurance implications as one of the reasons for declining genetic testing (Keogh et al., 2011). The VCCF study also found that participants who were informed of possible insurance implications were more than twice as likely to decline genetic testing for colorectal cancer as those who were not informed (Keogh et al., 2009). In one of the
few studies which quantitatively assessed genetic discrimination, 46.2% of respondents from HD families who tested either positive or negative in Australia, the USA and Canada reported genetic discrimination (Erwin et al., 2010). Of the overall incidence of discrimination 25.9% was found to be in the area of insurance and 6.5% in employment (Erwin et al., 2010).

Life insurance has raised more interest in the UK than health insurance, most probably because of the established National Health Service. Steps have been taken internationally to protect consumers and ensure they have the ability to obtain life insurance with the Genetics and Insurance Committee being established in 1999 (see Morrison, 2005). In the UK after the setting up of an insurance moratorium in 2001 consumers are comprehensively protected against genetic discrimination. Based on this moratorium (Association of British Insurers, June 2008):

1. Insurance companies are not allowed to ask consumers to take any genetic test in order to be insured;

2. Consumers can apply for up to £500,000 for life insurance and £300,000 for critical illness insurance without having to inform the insurance company of the results of any genetic test already taken;

3. Above these amounts consumers only need to inform the insurance company of the results of genetic tests that have been approved by an independent Government Committee and the only genetic test that has been approved up to 2008 is the genetic test for Huntington’s condition.

Most UK studies on insurance implications following genetic testing have taken place in the 1990s, with very few studies investigating insurance implications in the 21st century.
Foster et al (2007) reported that 40% of female BRCA1 carriers had problems obtaining life and/or health insurance while Watson et al (2004) reported that only 20% of female BRCA1/2 carriers had such problems. Because of the limited evidence, further research is required to investigate the real impact of genetic services on obtaining insurance in the UK.

3.3.7. Satisfaction

Due to the complexities in achieving consensus on what is a suitable outcome upon which to evaluate genetic services, it has been suggested that outcomes which assess the process of genetic services such as satisfaction with the service received, might be a more suitable approach than single outcomes such as emotional outcomes (Clarke, Parsons and Williams, 1996). Satisfaction has been linked to quality of care and patient-centred care (Zellerino et al., 2009). Satisfaction with a broad range of elements of service provision has been assessed, including satisfaction with genetic counselling, decision-related outcomes such as satisfaction with decision-making and informed choice (Kasparian, Wakefield and Meiser, 2007).

Patient satisfaction with different aspects of service provision has been widely used as a “complementary” outcome along with emotional or cognitive outcome measures when evaluating the impact of genetic service provision (e.g. Brain et al., 2000a; Davey et al., 2005; Smets et al., 2006; Schwartz et al., 2009; Sutphen et al., 2010; Meropol et al., 2011). Payne et al (2008) list two general and four genetics-specific outcome measures used in the literature to measure satisfaction with CGSs. Two of these genetics specific outcome measures assess satisfaction with decision-making and two with genetic
counselling. A fifth outcome measure designed to assess patient satisfaction in a clinical genetics setting has focused on technical aspects of care as well as interpersonal relationships (Zellerino et al., 2009).

Measures assessing patient satisfaction have been designed around the genetic counselling goals and process and most consist of dimensions relevant to the abilities, skills and quality of service provided by the health professional, the emotional interest expressed by the health professional, procedural aspects of the process (Shiloh, Avdor and Goodman, 1990; Tercyak et al., 2001; DeMarco et al., 2004) and time (Tercyak et al., 2001; DeMarco et al., 2004).

Rather than focusing on satisfaction with the overall service, investigators have urged for a measure of satisfaction with specific aspects of the service (Clarke, Parsons and Williams, 1996). This is arguably more meaningful since it is more clear which aspects of the counselling process service users are satisfied with leading researchers to conclude that satisfaction is not an actual outcome measure, but rather a proxy outcome measure (Payne et al., 2008). Higher levels of satisfaction have been found to correlate with increased levels of PPC (Berkenstadt et al., 1999; Davey et al., 2005) and decreased levels of anxiety (Tercyak et al., 2001; Hamang et al., 2011) while those more satisfied with counselling were more likely to communicate risk results to other family members (Cheung et al., 2010). Hopwood et al (2004) found satisfaction to differ between five UK genetic centres in terms of satisfaction with communication and some aspects of satisfaction with the service, even though high satisfaction was reported overall. Davey et al (2005) found that both satisfaction and PPC varied depending on the genetic service accessed, with higher satisfaction scores obtained from individuals
seen at a general genetics service, followed by those visiting a paediatrics service and lowest satisfaction scores obtained from individuals seen at a cancer genetics clinic. Higher satisfaction has been associated with the communication of medical information during genetic counselling (Pieterse et al., 2007a) and with being contacted by a genetic counsellor over the phone prior to the genetic counselling appointment (Davey et al., 2005).

Satisfaction with the genetic counsellor has also been found to be positively correlated with the levels of certainty obtained by parents whose child has been diagnosed with a genetic condition. (Lipinski et al., 2006). In this case, individual characteristics such as coping style might play a role in the reported levels of satisfaction. A tolerance for ambiguity might be an important advantage for individuals referred to the service, since genetic services tend to be associated with high levels of uncertainty (Bish et al., 2002). Increased levels of uncertainty following genetic counselling were found to be associated with lower levels of satisfaction with genetic counsellors (Lipinski et al., 2006). This reality might be in conflict with the value placed on the certainty offered by CGSs (Lim et al., 2004).

Satisfaction is a popular outcome measure proposed for the evaluation of CGSs as an alternative to outcomes which are difficult to measure or capture (Clarke, Parsons and Williams, 1996; Aalfs et al., 2007). Client satisfaction with information and services received as part of genetic counselling is one way of measuring the quality of genetic counselling focusing on the process rather than the outcomes (Clarke, Parsons and Williams, 1996). Even though satisfaction with procedural aspects of CGSs is an attractive outcome of the service, several problems exist with its use (Kasparian,
Wakefield and Meiser, 2007; Payne et al., 2008). Research investigating satisfaction with genetic services has indicated high levels of satisfaction reported by the clients (e.g. Metcalfe et al., 2010; Hamang et al., 2011; Meropol et al., 2011) but satisfaction has also been found to be associated with the content of the session and studies provide limited information about the specific aspects of the service that contribute to satisfaction (McAllister et al., 2007a). Satisfaction has been proposed to be more of a proxy rather than an outcome measure of clinical genetic services (Payne et al., 2008).

3.4. DISCUSSION

The appropriateness of existing outcomes for the evaluation of CGSs

The aim of this chapter was to discuss the literature evaluating the psychosocial implications of CGSs, list the outcomes used for these evaluations and discuss their suitability to be included in an economic evaluation. A considerable number of outcomes have been identified and these have been categorised in the following domains: emotional outcomes, cognitive outcomes, behavioural outcomes, adjustment-related outcomes, social outcomes and satisfaction.

3.4.1. Outcome-specific limitations

3.4.1.1. Emotion-related outcomes

Emotional consequences of CGSs have received most attention from researchers so far. Depression, anxiety, state-specific worry and distress are common constructs used to evaluate CGSs but evidence has shown that the levels of these outcomes might be more associated with individual, social and temporal variables rather than actual service
Research has shown that psychological well-being depends mainly on the emotional state of the individual before accessing the CGS rather than on the actual services received (van Oostrom et al., 2007b; Graves et al., 2011; Rosenblatt et al., 2011) as well as on prior expectations (Hilgart et al., 2010).

Researchers have also begun questioning these consequences and whether they are able to encapsulate both the overall impact of CGS and the overall experience of service users (Wang, Gonzalez and Merajver, 2004; Skirton, Parsons and Ewings, 2005; Payne et al., 2008). Even though genetic risk assessment has been found not to lead to any negative emotional outcomes for the majority of those referred, studies have also shown that for a small minority of individuals, this process may lead to some negative emotions such as distress during the process of risk assessment (Fry et al., 2003) or anxiety about the individual's ability to cope after receiving a risk assessment (Phelps et al., 2006).

The emotions experienced during the risk assessment process can be both positive and negative (Bonadona et al., 2002; Lynch et al., 2006) and researchers have suggested that research assessing this process should move away from the pathological perspective adopted so far through the use of outcomes like depression and anxiety (Phelps, Bennett and Brain, 2008). Genetic cancer risk assessment can also elicit emotions of hope, gratitude, relief and tranquillity and a predominance of optimistic expectations in addition to negative emotions like anxiety and fear (Phelps, Bennett and Brain, 2008). Claes et al (2005) found that even though no negative overall psychological impact was found in individuals after testing for BRCA1/2 genetic mutations, results from open-ended questions identified a number of positive as well as
negative implications particularly from carriers. Relief in the case of non-carriers and relief from uncertainty for carriers was part of the advantages reported by participants. Having to deal with negative consequences of prophylactic surgery, uncertainty, negative changes in body image and changes in the relationships with relatives were some of the negative aspects of undergoing genetic testing (Claes et al., 2005). Kausmeyer and colleagues (2006b) also found that even though 72% of participants choosing to have cancer genetic testing reported to be glad about their decision while waiting for their test results and more than half felt empowered by their decision, almost half reported feeling anxious during that time. The same study also found that following testing, 95.7% of participants felt glad about deciding to have genetic testing, and equally large numbers reported being satisfied, very satisfied or extremely satisfied with the Cancer Genetics Programme. These findings indicate that outcome measures focusing only on experienced anxiety fail to capture the broader benefits of using a CGS.

Outcome measures should be able to go beyond the depiction of unilateral concepts such as depression which do not reflect the actual experiences of service users and be able to encapsulate the nature of the user experience. These findings suggest that other emotional consequences are present which are not yet known or captured by the existing outcomes research. Further investigation is required therefore in order to identify and subsequently incorporate the comprehensive spectrum of emotional consequences in an economic evaluation.
3.4.1.2. Cognitive outcomes

Cognitive outcomes which involve one of the main objectives of CGSs, namely the provision of information in order to support decision-making, have shown that access to CGSs improves the knowledge and risk perceptions of individuals undergoing genetic counselling and genetic testing (Smerecnik et al., 2009; Roussi et al., 2010; Wevers et al., 2011). Qualitative research findings have shown that the need for facts and information is one of the most important expectations from genetic counselling in mothers of children diagnosed with a genetic condition after birth (Collins et al., 2001; Tluczek, 2006) as well as a major benefit following genetic counselling (Bernhardt, Biesecker and Mastromarino, 2000). Collins et al (2001) however have also shown that even though all women participants reported learning something from genetic counselling, the amount and type of information differed between women, with three out of four women reporting “information overload”. The issue here is whether the information assessed by genetic service providers as important is the same as the information needs of patients and families. In the above research, mothers reported differing information needs, ranging from information about the condition and its implications as a way of coping with the diagnosis, to requesting no information at all (Collins et al., 2001).

Quantitative research studies focus on evaluating the cognitive impact of counselling in terms of accurate risk perceptions and knowledge of genetics and inheritance, but qualitative research has illustrated that service users attribute value on more subjective aspects of the process. These include: integrating lay and scientific information around genetics (Skirton, 2001), and receiving complicated information in layman’s terms
leading to informed decision-making (Macleod, Craufurd and Booth, 2002). In some cases individuals seek this information for more than decision-making (Lim et al., 2004), but also as a means to better coping with their situation (Collins et al., 2001; Skirton, 2001; Barr and Millar, 2003; Lim et al., 2004). In the light of these findings cognitive outcomes as a single outcome measure appear inadequate in capturing the real impact of CGSs and the perceptions of service users and therefore unsuitable for an economic evaluation.

3.4.1.3. Behavioural outcomes

Studies investigating the impact of CGSs on behaviours have focused on conditions where appropriate interventions can be designed in order to improve the health outcomes of conditions, such as cancer and coronary heart disease. These studies are yet sparse with several methodological drawbacks and conflicting evidence and it is not yet clear whether individuals choosing to access genetic services are already engaged in health behaviours or whether behaviours might change following their access to the service. Evidence has shown that following access to genetic counselling and genetic risk assessment or genetic testing individuals are more likely to engage in prophylactic practises such as a healthy diet and exercise (McKinnon et al., 2007; Pijl et al., 2009; Zlot et al., 2010) and risk management activities (Heshka et al., 2008; Claassen et al., 2010).

Assessing health behaviours rather than behaviour change might be a good outcome to incorporate in economic evaluations in addition to assessments of other psychosocial outcomes but this is arguably relevant for a small group of multifactorial conditions such
as cancer, diabetes or coronary heart condition where such behaviours could potentially in the long term affect health outcomes.

### 3.4.1.4. Adjustment-related outcomes

Perceived control is one outcome which has received much attention in the literature and refers to the perceived ability of an individual that they have the resources needed to deal with a stressful situation. The importance of this outcome is that it does not require that individuals actually control the situation, but that they perceive to be able to control it (Davey et al., 2005). Research has shown that access to CGSs raises levels of perceived personal control (Pieterse et al., in-press) and that this outcome correlates positively with satisfaction, knowledge, counselling evaluations and expectation fulfilment (Berkenstadt et al., 1999; Davey et al., 2005; Pieterse et al., in-press) and negatively with anxiety (Davey et al., 2005; Pieterse et al., 2005b), thus indirectly touching upon these “unidimensional’’ constructs. Skirton (2001) in a grounded theory qualitative study identified the client’s psychological adaptation to the genetic situation as the overall outcome of genetic counselling. Skirton’s findings are relevant to the psychological construct of PPC in that her participants reported actually experiencing few practical changes following genetic counselling in terms of reproductive plans, insurance, employment and family relationships. Changes experienced were reported to be psychological changes most often referred to as “peace of mind” (Skirton, 2001). This outcome was reported to be influenced by the satisfaction of the need for certainty of the client, the integration of lay knowledge and new information provided during genetic counselling, and the quality of the relationship between the client and the genetic staff (Skirton, 2001).
PPC so far appears to be a promising construct to be used as an outcome for evaluating CGSs, albeit one with several limitations. The most important limitation in the context of CGSs might be its individualistic focus. McAllister and colleagues (2008a) have already pointed out the family nature of CGSs and have proposed a related concept to that of PPC which they have called “empowerment” which addresses impact to other family members. This model has adopted some of the PPC dimensions while adding additional ones to make this concept broader thus more representative of actual implications. This five-dimensional outcome of empowerment included the following dimensions: cognitive control, behavioural control, decisional control, hope, and emotional regulation (McAllister et al., 2011).

3.4.1.5. Social Outcomes

The family impact of CGSs has also been investigated. CGSs have been evaluated for their impact on family communication (McCann et al., 2009; Balck et al., 2011; Vos et al., 2011), disclosure of genetic information (Gaff et al., 2007) and the emotional impact on family members from a genetic diagnosis and the process of genetic testing (Dinc and Terzioglu, 2006; Hadley et al., 2010; Lammens et al., 2011). Having to communicate genetic information to other family members has been found to be a stressing experience for individuals even though at the same time information is disclosed because of a sense of obligation to inform other family members (Hopwood, 2005; McCann et al., 2009; Nycum, Avard and Knoppers, 2009). Service users have been reported to attribute higher significance on this group of consequences compared to genetics professionals (Michie et al., 1998; Payne et al., 2007). At the same time, the
complex and sensitive nature of the family communication process has raised questions about the appropriateness of genetic services to tackle this process (Gaff et al., 2007).

Insurance implications which are of particular interest to health economists have been investigated only by a few studies within the UK which have focused on HBOC. These studies show that less than half of women experienced genetic discrimination (Watson et al., 2004; Morrison, 2005; Foster et al., 2007). The presence of a UK government moratorium (Morrison, 2005) put in place to protect consumers from such discrimination arguably explains the lack of significant insurance problems in the UK. Because of the limited evidence on actual insurance implications, further research is required to understand the actual experiences of individuals affected by genetic conditions when trying to obtain insurance coverage. In addition, attention should be given not only to health or life insurance coverage but also to other types of insurance such as travel insurance, and to conditions other than cancer.

3.4.1.6. Satisfaction

Satisfaction is another outcome used to evaluate the effectiveness of CGSs and specifically of genetic counselling. This outcome however is not an appropriate outcome for the evaluation of CGSs particularly for the purposes of an economic evaluation since it does not constitute a direct consequence of the service and relies heavily on contextual factors such as genetic test results.

3.4.2. Methodological limitations of the wider literature

In addition to the problems specific to each category of outcomes, a number of issues related to the broader literature have been identified which limit the ability to draw
conclusions as to the nature of consequences of CGSs. Methodology has been a major drawback in the research of psychosocial outcomes of genetic testing. A few papers have been published discussing the methodological issues around measuring the psychosocial consequences of CGSs (Timman, Stijnen and Tibben, 2004; Vadaparampil, Ropka and Stefanek, 2005; Payne et al., 2008). Vadaparampil and colleagues (2005) list three main issues with the existing studies. Firstly, there is limited documentation of psychometrics for the used outcome measures with instrument selection being arbitrary or based on tradition rather than to suit the unique needs of a given study population. Secondly, the variability of instruments used by existing studies makes any comparisons across studies difficult and drawing conclusions about the psychosocial implications of CGSs impossible. The same criticism of the psychosocial literature on CGSs was made by a more recent systematic review by (Payne et al., 2008). Criticisms have also been made on the methodology and statistics used by follow-up studies where the majority were found to apply statistical techniques that were less suitable or less efficient for the data available to the researchers (Timman, Stijnen and Tibben, 2004). Timman and colleagues point out that the psychological implications of genetic testing are stronger right after the test but tend to stabilise over time so they recommend follow-up studies that are continued over several years (Timman, Stijnen and Tibben, 2004).

Another problem that has limited the meaningfulness of the findings is the lack of coherence and agreement in what the provision of CGS is trying to achieve. So far there appears to be a lack of coherence and agreement in what the provision of CGS is trying to achieve (Wang, Gonzalez and Merajver, 2004) or how the service should be
provided. Studies may refer to different models of service provision (Brain et al., 2005; Smerecnik et al., 2009), for example genetic testing with or without genetic counselling, and these may not be clearly described and may result in different psychological outcomes and decision making e.g. to undergo genetic testing or not (Hopwood et al., 2004; Wang, Gonzalez and Merajver, 2004; Matloff et al., 2006). Smerecnik and colleagues (2009) through their literature review provided a description of the counselling session content included in the 19 reviewed studies and report that only four studies mentioned using a genetic counselling protocol while twelve did not mention the use of any protocol, standardised script or audio- or videotapes as a content check, and four studies did not provide a description of the counselling session at all.

A related problem is the lack of clarity of definitions of “genetic services” and “genetic counselling”. It seems that authors often refer to the term “genetic services” to refer to the provision of genetic counselling even though a “genetic service” comprises many aspects of service provision (Sivell et al., 2007). This makes it difficult to understand which particular aspect of service provision is being evaluated and how specific findings can be compared with the wider literature. Different models of service provision and genetic counselling protocols make it difficult to draw conclusions about the consequences of each aspect of the service, with some form of genetic counselling being reported to be provided in the majority of studies on the consequences of genetic testing published in the past 10 years. Genetic counselling models appear to differ according to country and genetic centre (Brain et al., 2003; Godard et al., 2003; Brain et al., 2005), with some focusing on educational aspects of counselling and others on psychotherapeutic aspects (Biesecker and Peters, 2001) even though within the UK
information provided during genetic counselling appears to be similar (Hopwood, 2005).
There is worldwide difference in the professional identify and training of genetic counsellors, with the professionals placing their practice at any point on a continuum between an educational focus and a therapeutic focus (Biesecker and Peters, 2001).

3.5. CONCLUSIONS

The psychosocial impact of clinical genetic services has been extensively investigated, and the large amount of studies is reflected in the number of literature reviews published so far. Studies have investigated the impact of CGSs on emotional, cognitive, behavioural, adjustment-related, social outcomes, and satisfaction. Satisfaction has been stated to be an inappropriate outcome upon which to evaluate clinical genetic services because high levels of satisfaction are generally recorded in research studies irrespective of the variables assessed, while it is not always clear which aspects of service provision the user is satisfied with. Overall, genetics services may impact on all other outcomes to some degree even though in general emotional outcomes are linked to a number of other variables other than service provision including other main outcomes, such as perceptions of risk and levels of emotional distress prior to genetic counselling or genetic testing. Quantitative research has focused on the assessment of a small, predetermined number of unidimensional outcomes while qualitative and mixed methods research has illustrated how CGS implications are multidimensional, complex and affect more than the individual referred to the service. There appears to be growing consensus in the quantitative and qualitative literature on the role of Perceived Personal Control (PPC) as an appropriate framework to guide evaluation and the concept of “empowerment” has been proposed
as an extension of the PPC framework more appropriate to the genetic services context. What is now needed is research explicitly linking overall genetic services with what the users perceive as benefits or disadvantages within the context of health economics.
CHAPTER 4

METHODS AND RESEARCH DESIGN
4.1. Overview

This chapter follows on from the overview of outcomes used in the economic and psychosocial literature to:

1. Reiterate the aims and objectives of the thesis;
2. Provide an overview of the theoretical approaches to research design;
3. Discuss the research design and methods adopted.

4.2. Aims and objectives

The main aim of this thesis is to identify the (dis)benefits of CGSs and present a framework which may be used as a guiding tool by health economists and other stakeholders embarking on a welfarist evaluation of genetic services. In order to achieve this aim the following objectives will be dealt with:

1. Map the patient pathways through the chosen CGS (in order to gain an understanding of the processes of CGS provision);
2. Identify services outside the CGSs utilised by those being referred to the service (in order to gain understanding of possible resource implications for other services);
3. Identify relevant (dis)benefits of CGSs from the professionals’ perspective (in order to explore any conflict between service user and provider);
4. Identify relevant (dis)benefits of CGSs from the service users’ perspective (the main focus of the research);
5. Identify the most important impacts as these are perceived by the stakeholders of the service (to gain an understanding of user preferences and what features in their utility function); and finally

6. Consider the methodological implications of incorporating these outcomes in economic evaluations.

4.3. Epistemological approach to research design

This PhD set out to explore the overall (dis)benefits of CGSs and map the patient pathways through a CGS and possible use of other health services. This objective was guided by reports on the difficulty faced by health economists when attempting to conduct a comprehensive cost-benefit analysis due to the largely unknown impacts of CGSs (Cohen, Barton and Brain, 2004). Through the findings of the literature review discussed in the previous chapter, it was noted that even though the psychosocial impact of genetic services has been extensively investigated, research studies suffer from a number of drawbacks which limit the possibility of incorporating the existing findings into the design of economic evaluations. In order to assess the impact of the service therefore, the broader experiences of families need to be explored by turning to the users of the service for obtaining their perspective.

Research design in this PhD was objective-driven, with the methods being guided firstly by the aims and objectives of the research; and secondly by the theoretical foundations underpinning economic evaluations, i.e. welfare economics. As a paradigm to guide research design the pragmatic paradigm (Murphy, 1990; Cherryhomes, 1992; Patton, 2002) was chosen as the most appropriate for the problem-focused approach of the
thesis. The maxim or rule of pragmatism states that “the current meaning or instrumental or provisional truth value of an expression is to be determined by the experiences or practical consequences of belief in or use of the expression in the world” (Murphy, 1990 in Johnson and Onwuegbuzie, 2004: p. 16). Pragmatism forms an alternative epistemological framework to that of positivism/post-positivism and constructivism and it attempts to provide a middle ground where philosophical dogmatisms can meet (Johnson and Onwuegbuzie, 2004). Pragmatism is also the dominant paradigm guiding the use of both qualitative and quantitative methods within a mixed methods approach (Tashakkori and Teddlie, 1998).

Qualitative and quantitative methodologies are separated by the way each one chooses to understand the world and create knowledge, or in other words by their epistemology (Creswell, 2002). Quantitative methodologies are adopted by all the natural sciences and until recently by some social sciences, and they are found to align to a “positivist” or scientific approach in order to produce knowledge. Positivism believes that phenomena in the world exist irrespective of individuals having extensive knowledge of them, and the only way to produce valid knowledge is through the scientific approach and the validation of theories (Green and Thorogood, 2004). More recently positivism has been replaced by post-positivism, following the recognition that being “positive” about what constitutes reality or the absolute truth when studying the behaviour and action of humans is unrealistic (Creswell, 2002). Despite this acknowledgement, post-positivism remains deterministic and reductionist in its philosophy towards knowledge, and investigations are based on careful observation and measurement of the objective reality (Creswell, 2002).
Qualitative methodologies on the other hand, adopt a constructivist and interpretative approach where reality is believed to be socially constructed and the aims of research are not to explain human behaviour but instead to understand people’s interpretations of their social environments and experiences (Green and Thorogood, 2004). Qualitative research “is concerned with the meanings people attach to their experiences of the social world and how people make sense of that world […] [and] tries to interpret social phenomena in terms of the meanings people bring to them…” (Pope and Mays, 2006).

Pragmatism rejects the dogmatism attached to these traditional approaches and in a sense frees researchers from the restrictions placed by epistemological paradigms. The focus is now turned to the problem and whether a given piece of knowledge addresses and proposes solutions to this problem. Within pragmatism, knowledge claims arise through actions, situations, and consequences but not through antecedent conditions as is the case with positivism (Creswell, 2002). Pragmatism believes that the only sensible way to judge a piece of knowledge is through its consequences and the usefulness of these consequences for a practical purpose (Cornish and Gillespie, 2009). In this sense, the established concern of whether a given knowledge accurately reflects the underlying reality is now replaced by the question of whether this knowledge serves a given purpose (Cornish and Gillespie, 2009).

In a world guided by different and sometimes conflicting types of knowledge and practical problems that require solutions (Cornish and Gillespie, 2009), the focus on research which adopts a single paradigm might be restrictive. Both the post-positivistic/positivistic paradigm and constructionist paradigm which are the common paradigms in health care research present drawbacks as to the nature of knowledge
produced by each one (Creswell, 2002; Johnson and Onwuegbuzie, 2004). The pluralistic nature of pragmatism however allows for the use of multiple methods, different world views and assumptions and also allows for the use of different methods of data collection and analysis (Creswell, 2002). For this reason, pragmatism has been proposed as the theoretical backbone of the mixed methods approach, a third strategy of enquiry along with quantitative and qualitative research strategies (Tashakkori and Teddlie, 1998; Creswell, 2002; Johnson and Onwuegbuzie, 2004; Creswell and Clark, 2007; Cornish and Gillespie, 2009).

4.4. Research Design

A mixed methods approach to data collection is adopted within an exploratory design (Creswell and Clark, 2007). When using a mixed methods approach, the researcher is utilising research methods from both quantitative and qualitative methodologies in order to answer the research questions set by the investigation. An exploratory design involves the consecutive use of the two methods, where the findings from the first (qualitative) method are used to develop or inform the data collection within the second (quantitative) method (Creswell and Clark, 2007). The purpose of this design is to explore a phenomenon as well as identify important variables which are currently unknown, generalise to other groups, or measure the explored phenomenon’s prevalence (Creswell and Clark, 2007).

Mixed methods design means the utilisation of an approach which combines elements or techniques from both quantitative and qualitative approaches (Lingard, Albert and Levinson, 2008). Johnson and Onwuegbuzie (2004) have defined mixed methods
research as “the class of research where the researcher mixes or combines quantitative and qualitative research techniques, methods, approaches, concepts or language into a single study” (Johnson and Onwuegbuzie, 2004: p.17). Even though mixed methods can be used in the design of different research projects, the weight that is assigned to the two methods of data collection can vary between projects, with more weight being given to data collected through qualitative methods or vice versa (Creswell, 2002).

In recent years when qualitative methods began to infiltrate health care research, researchers have started to increasingly adopt both methods of research (O'Cathain, Murphy and Nicholl, 2007), especially due to the interest in the psychosocial determinants of health and illness and understanding aspects of medical care (Lingard, Albert and Levinson, 2008), and the turn towards practical rather than scientific research that is used to inform policy (Brannen, 2005). The choice of research methods utilised in a research investigation should be guided by the kind of questions posed (Brannen, 2005), and the variety of questions posed within healthcare research which might call for both the in-depth understanding of an experience and the setting of priorities, evaluation, and how interventions work (O'Cathain and Thomas, 2006), calls for the use of a “complex of methods” (Brannen, 2005) rather than a single technique. By using both quantitative and qualitative methods of inquiry and taking advantage of characteristics of each approach, a researcher may expand the scope of enquiry (O'Cathain and Thomas, 2006) in a way that each approach complements the other to achieve more valid data (Creswell, 2002).

The two methodological approaches can be combined in a variety of scenarios based on the aims of the research. A common feature in the use of mixed method studies is
the use of both approaches within the context of method triangulation (Creswell and Clark, 2007), even though mixed methods are now used to achieve objectives beyond triangulation (Tashakkori and Teddlie, 1998). The purpose of using mixed methods in this PhD was to use the findings from the qualitative part to firstly explore respondent validation and transferability of findings within the context of triangulation and secondly to pilot the use of a new technological tool, namely Audience Response Systems (ARSs), as an approach for exploring the preferences of stakeholders about which (dis)benefits they considered most important and thus featured more strongly in their utility line. Establishing preferences is a necessary step in valuation of (dis)benefits so they can be incorporated in an economic evaluation. In this sense, an exploratory design is adopted where the findings of the main qualitative phase are used to inform the design of a quantitative phase incorporating respondent validation and transferability of findings, and pilot testing a new technology for its suitability in exploring preferences.

In conclusion, data collection in the present thesis takes place within three phases, where phases are consecutive to each other and deal with specific objectives (see Table 4.1). Specifically, Phase One and Two adopt a qualitative approach to data collection with the findings from Phase One informing data collection in Phase Two, while findings from this phase feed into the design of Phase Three which constitutes the quantitative part of the research.
4.4.1. Data Collection

4.4.1.1. A case study approach to data collection

One UK CGS, was used as a case study for exploring the (dis)benefits of genetic services. Case study design is characterised by the intense focus on a single phenomenon within its real-life context (Yin, 2003). Case studies are a useful method for evaluations in healthcare settings, where the nature of the intervention may not be well understood and the identification of its consequences is therefore particularly difficult. They are often concerned with understanding the nature of the intervention or policy in question, as well as with establishing its costs and effect (Keene, 2006). Using a case study approach for this research was particularly suitable because of the variation in service organisation between different UK genetic centres and by extension the possible differences in (dis)benefits. Focusing on one genetic centre allowed the detailed understanding of the processes involved in service provision, i.e. the patient pathways or the patient needs. Moreover it allowed for exploring the possibility that different ways of using the service might lead to different (dis)benefits. The detailed understanding of how one UK genetic centre is organised would enable future comparisons with other UK genetic centres to test further the research outcomes.

Variability within the chosen CGS exists as in other UK genetic centres, through the diversity in genetic conditions seen by genetic professionals and the multiplicity of experiences related to these conditions. In order to explore this variability and identify commonalities between different experiences as well as differences, the present
research directed data collection to specific conditions. This data collection strategy is
discussed in the following section.

4.4.1.2. Condition focus of the study

The focus of the study was limited to specific genetic conditions. An initial overview of
the literature showed that studies belonged to two major camps, those which
concentrated on the experiences of patients from particular condition groups e.g.
cancer, and those which used patients from a number of genetic conditions without
distinguishing between their experiences. Research investigating suitable outcomes for
the evaluation of genetic services has adopted a broad perspective on the overall
service and studies have not compared outcomes between conditions (e.g. Macleod,
Craufurd and Booth, 2002; McAllister et al., 2007b). The possibility that outcomes might
be condition specific has received limited investigation and calls for further exploration
(Wang, Gonzalez and Merajver, 2004; McAllister et al., 2007a).

In order to explore whether and how patient pathways, and therefore (dis)benefits,
might differ between different conditions, categories representing modes of genetic
inheritance and other characteristics of genetic conditions and genetic service provision
were listed and conditions associated with these categories were identified (see Figure
4.1). This formed a genetic framework aimed to enable targeted condition inclusion and
data collection using conditions which were sufficiently different from each other to
enable comparison between the patients’ experiences, as well as identification of
consequences which were common regardless of condition and at the same time
representative of the wider spectrum of hereditary conditions seen by a genetics
service. This genetic framework was developed with the scientific advice and support of a consultant geneticist from the chosen CGS who acted as the clinical adviser to the project, and using input from geneticists and genetic counsellors of the service.

The genetic framework was made up of the following categories of characteristics which may influence the experience of a family or individual affected by a genetic condition.

Genetic condition characteristics:

1. Mode of inheritance – the genetic mechanisms of inheritance of the gene e.g. dominant, recessive, or X-linked;

2. Onset of the genetic condition – the age of manifestation of condition symptoms;

3. Condition penetrance – the proportion of individuals who carry a particular gene mutation who also exhibit clinical symptoms;

4. Expression of the condition – the presence of qualitative differences in the manifestation of the condition among individuals who carry the same gene mutation;

5. Prevalence – the total number of cases of the condition in the population at a given time;

6. Prognosis – the likely outcome of the condition, influenced by the availability of effective clinical management.

To some degree, condition characteristics may influence some aspects of service provision, which in turn also impact on the CGS experience. The following service characteristics were integrated in the genetic framework:
1. The availability of genetic testing whether diagnostic, predictive, prenatal or neonatal genetic testing;
2. The molecular genetics of a genetic condition, and whether these are easy or complex which might influence the time it takes for the genetic analysis to be complete;
3. The presence of organised surveillance by the health service (whether genetics service or other health care services);
4. The level of involvement of the genetic service into the form of patient and family follow up. Services could provide either proactive or reactive follow up.

The way genetic conditions express themselves e.g. age of onset, prognosis and their impact on activities of daily living, might have implications for the psychosocial well-being of patients and families, which might lead into the need for social services involvement.

Five groups of conditions were finally selected: Cystic fibrosis (CF), hereditary breast and ovarian cancer (HBOC), von Hippel Lindau (VHL) disease, tuberous sclerosis (TS) and the two most common types of muscular dystrophy, namely Duchenne and Becker Muscular Dystrophy (DBMD).

These conditions were presented to members of staff of the CGS in order to obtain their views on the conditions selected. Even though the groups of conditions originally selected were not changed, it was decided to make two amendments as a result of discussions with geneticists. Originally only Duchenne’s muscular dystrophy was included in the five conditions, but it was decided to include Becker’s muscular dystrophy as well. The reason was the very small number of families included in the
Duchenne’s genetic database which would make recruitment extremely difficult, considering that enough individuals were required to form a focus group. Becker’s is similar to Duchenne’s but has a later onset as well as slower and usually milder progression. A second change involved the cancer genetics service where originally it was decided to include families with a link to BRCA 1 and 2, the gene mutations associated with hereditary breast and ovarian cancer (HBOC). Later on however it was decided based on discussions with the geneticist involved in cancer genetics to broaden this group to include all families referred to the service in relation to hereditary breast and ovarian cancer which represented a more accurate name for this group of individuals.
**Figure 4.1: Genetic framework informing condition selection including related elements of service provision**

### CONDITION CHARACTERISTICS

<table>
<thead>
<tr>
<th>Mode of inheritance</th>
<th>Onset</th>
<th>Penetrance</th>
<th>Expression</th>
<th>Prevalence</th>
<th>Prognosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>e.g.</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Effective management</td>
</tr>
<tr>
<td>Recessive gene</td>
<td>Adult In childhood In uterus</td>
<td>High</td>
<td>Variable</td>
<td>High</td>
<td>Little can be done</td>
</tr>
<tr>
<td>Dominant gene</td>
<td></td>
<td>Low</td>
<td>Consistent</td>
<td>Low</td>
<td></td>
</tr>
<tr>
<td>X-Linked</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Multifactorial</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Single-gene</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
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</tbody>
</table>

### SERVICE CHARACTERISTICS

<table>
<thead>
<tr>
<th>Organised surveillance involved</th>
<th>Availability of genetic testing – including diagnostic, prenatal and neonatal diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Not at all</td>
<td></td>
</tr>
<tr>
<td>Extremely</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Level of involvement</th>
<th>Molecular genetics</th>
<th>Molecular genetics</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reactive</td>
<td>Easy</td>
<td>Difficult</td>
</tr>
<tr>
<td>Proactive</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

### SOCIAL CONSEQUENCES

<table>
<thead>
<tr>
<th>SOCIAL SERVICES INVOLVEMENT</th>
</tr>
</thead>
</table>
Chapter 4

The sampling strategy for both Phases One and Two was guided by the choice of these five conditions and by the restrictions placed by individual conditions in terms of the participants that could be recruited in Phase Two.

No individual participants were chosen for Phase Three but rather groups of stakeholders with knowledge of clinical genetic services. Participation in this phase was completely voluntary for all individuals and no data were collected from named participants.

In the following chapters a detailed description of the research methods used in the three consecutive phases will be given.

The following table presents a visual representation of the three phases with the data collection methods used in each phase and the objectives addressed in each phase. The data collection techniques, recruitment and analysis of each phase will be discussed in detail in the chapters that follow.
Table 4.1: Phases of data collection

<table>
<thead>
<tr>
<th></th>
<th>PHASE ONE</th>
<th>PHASE TWO</th>
<th>PHASE THREE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Data collection with CGS professionals – objectives 1&amp;2&amp;3&amp;5</td>
<td>Face to face interviews</td>
<td>Focus groups and face to face interviews</td>
<td>Stakeholder event using ARSs</td>
</tr>
<tr>
<td>Data collection with service users – objective 4&amp;5</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Data collection with service users and other stakeholders – 1. Respondent Validation &amp; Transferability 2. Pilot study (ARSs)</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Objective six is addressed in the overall discussion of the final chapter of the thesis.

Phase One

Here objectives one, two, three and five were addressed. These were:

Objective One: Map the patient pathways through the CGS.

Objective Two: Identify services outside the CGS utilised by those being referred to the service.

Objective Three: Identify relevant (dis)benefits from the professionals' perspective.

Objective Five: Identify the most important impacts as these are perceived by the stakeholders (in this phase, the service providers).
One-to-one semi-structured interviews with medical and non-medical health professionals involved in genetic service provision took place. Results from Phase One were used both to familiarise the researcher to the patient pathways of the service in advance of patient focus groups in Phase Two; and to assist in the mapping of patient pathways of the service. Perceptions of service providers of the (dis)benefits of the service were also explored even though the (dis)benefits proposed in the final framework were identified through service user narratives. The purpose of this was to understand what the providers saw as a benefit because service provision arguably relates to what providers consider to be the role and consequently the benefit of the service to the service users. It also enabled a comparison between service provider and service user perceptions of benefit and thus triangulation of the findings.

**Phase Two**

Here objectives four and five were addressed. These were:

Objective Four: Identify relevant (dis)benefits from the service users’ perspective.

Objective Five: Identify the most important impacts as these are perceived by the stakeholders (in this phase, the service users).

This was the main component of the research which entailed focus groups and face to face interviews with users of the CGS. Results from Phase Two were used to inform the methods used in Phase Three.
Phase Three

This phase addressed validity issues and attempted to pilot the use of Audience Response System technology for use in future preference research. It encompassed a stakeholder event where stakeholders of the CGS i.e. patients, their families and members of support groups for families with genetic conditions, were invited to attend. An Audience Response System was used for simultaneously collating opinions and attitudes of attendees towards the presented (dis)benefits. The design used for Phase Three was dependent on the data collected during Phase Two.

4.5. Ethical considerations

Ethical approval was obtained as per guidelines of the Devolved Research Programmes Committee (DRPC) of the University of Glamorgan (May 2006), the South Wales Local Research Ethics Committee (LREC) (November 2006), the Cardiff and Vale NHS Research and Development Office (October 2006), and the Pembrokeshire Derwen NHS Research and Development Office (October 2007). In order to be able to have access to the families referred to the chosen CGS and conduct research within NHS and university premises, the researcher also obtained an honorary title as Honorary Research Fellow with the university with which the CGS was affiliated (October 2006).

Because of ethical issues the researcher did not access patient records or contact potential participants without the knowledge of the geneticist. All potential participants were identified and contacted by the geneticist, guided by inclusion and exclusion criteria chosen in collaboration with service providers. Inclusion criteria for each
condition are included in Appendix 3, while common exclusion criteria are listed in Chapter 6.

4.6. SUMMARY

This chapter provided a description of the research paradigm and research design adopted in this research investigation. This research was objective-driven and therefore a pragmatic approach to research design and investigation was deemed as the most appropriate approach for the research. A mixed methods design was used in order to meet the research objectives with qualitative methods being the primary approach used and guiding data collection in the first two phases while Phase Three was guided by a quantitative approach.

The following chapters will provide a detailed description of the research methods, analysis and results of the three phases of data collection.
CHAPTER 5

INTERVIEWS WITH SERVICE PROVIDERS
5.1. Overview

This chapter will address the first phase of data collection and will:

1. Provide an overview of the aims and objectives;
2. Describe the methods and recruitment techniques utilised;
3. Present the patient pathways for each of the chosen conditions;
4. Present services utilised by service users outside the chosen CGS;
5. Present the perceived benefits and disbenefits of the service;
6. Present the perceived role of the service;
7. Discuss the findings.

5.2. Aims and Objectives

During this phase the following objectives were addressed. These were:

- To map the patient pathways through the chosen CGS (objective One)
- To identify services outside the CGS utilised by service users (objective Two)
- To identify (dis)benefits as perceived by the service providers (objective Three)
- To identify which (dis)benefits are perceived to be the most important by the stakeholders of the service – in this phase the providers (objective Five)

The (dis)benefits proposed by this research will be based on the service users’ accounts; however, an exploration of the providers’ perspective would enable a comparison between the two i.e. triangulation of the findings, and highlight any disparities between them.
5.3. METHODS

5.3.1. Data collection techniques

One-to-one semi-structured interviews with clinical geneticists, genetic nurses and genetic counsellors involved at different stages of service provision took place aimed to understand the patient pathway and provider perceptions of the service provided. Within qualitative research, describing the context of the research, for example background information about the overall structures, settings and frameworks within which participants are situated, is described as a feature which distinguishes good quality research (Horsburgh, 2003). The effect that these could have on the participants’ actions should be actively acknowledged in order to place the data obtained from them within a wider context (Popay, Rogers and Williams, 1998). Knowing the circumstances under which individuals or families come into contact with the service provides a first glance into the patient experience and gives a platform upon which to start understanding the service user narratives.

A preliminary search on service provision models and patient pathways followed by users of UK CGSs retrieved broad guidelines and protocols informing the delivery of the overall clinical genetic service and guidelines about the overall management of individual genetic conditions where a small role of the CGS was described. This search showed a lack of detailed reports on patient pathways and use of related services which could be used to inform the design of an economic evaluation as per CBA design guidelines (see Chapter 2). It was required therefore to explore further the nature of the service delivered.
Qualitative interviews can allow the exploration of issues that may be too complex to investigate through quantitative means and where gaps, contradictions and difficulties are perceived to exist by the interviewee (Burman, 1994). Face-to-face interviews were useful for exploring issues not made clear by the existing literature and where individual geneticists might hold differing views regarding practice, role and consequences. This phase aims to understand the nature of the services provided to patients from a range of conditions but also enquire on the individual beliefs and perceptions of professionals in terms of the aims and role of the service and the psychosocial or other (dis)benefits resulting for the patients and their families.

5.3.2. Recruitment and procedures

Purposeful sampling was used for guiding participant recruitment. Purposeful sampling is a similar term to selective sampling (Coyne, 1997), meaning the selection of participants based on their characteristics. As Patton (2002) describes, the logic and power of this sampling method lie in the selection of “information-rich cases” for in-depth study. Information-rich cases are those from which the researcher can learn a great deal about issues which are of central importance to the purpose of the research (Patton, 2002). Based on the aims of Phase One, staff members of the chosen CGS were recruited. These included geneticists, genetic nurses, genetic counsellors, and members of the management team of the service.

Recruitment was informed by the level of information needed to be obtained. In order to understand pathways of service provision and user experience associated with referrals from the five groups of conditions, participants were chosen based on the main criterion
of in-depth knowledge of the patient experience and their direct involvement in service delivery. This professional could be a clinical geneticist or genetic counsellor/genetic nurse, as long as they were directly involved in service provision thus familiar enough to offer an informed opinion. Within the chosen CGS, professional involvement varies depending on the condition. For example, some conditions have limited involvement from a genetic counsellor while others are counsellor-led. In order to understand overall aims of the CGS and the long-term strategic direction of the service including issues of service evaluation, funding and commissioning senior members of the service involved in management were recruited.

The researcher was directed to potential participants by the clinical advisor who was the link between the researcher and the CGS. All potential participants were informed of the purpose of the research individually and all members of staff were introduced to the project via presentations delivered by the researcher as part of the centre's lunch time seminars.

One professional directly involved in service provision for each of the five groups of conditions was recruited. If more than one professional was working with a specific condition then one member of the team was selected by the other members to participate. It was also decided to interview a genetic counsellor involved in the prenatal genetic service of the CGS since this service receives referrals from all conditions when the foetus is identified as being at high risk for an inheritable condition either through prenatal screening or through a family history. Issues related to this aspect of the service might not be that familiar to service providers dealing with specific conditions. In order to understand issues of overall organisation, funding and
commissioning the two senior-level genetics professionals involved in service delivery decision-making were recruited.

Acceptance to take part in the research was taken as informed consent for participation. All participants were given information about the purpose of the interview and of the estimated duration (1.5-2 hours). Interviews were conducted between January 2007 and January 2008. All interviews took place either in the health professional’s office, or in a free room on the centre’s premises booked especially for the purpose of the interview. All interviews were tape recorded using an Olympus DSS player and fully transcribed.

Interviews were structured around four main themes:

1. Structure of the service and patient referral pathways;
2. Links and collaborations with other agencies;
3. Benefits and disadvantages of the service;
4. Preferred outcome on which the service should be evaluated.

A detailed interview guide is included in Appendix 4.

5.3.3. Participants

Eight professionals were recruited for Phase One:

- Two professionals were involved in senior-level decision-making about overall CGS provision;
- Two professionals were associate specialists in medical genetics;
- Two professionals were genetic consultants in clinical genetics;
• One professional was a genetic nurse counsellor;
• One professional was a genetic nurse specialist.

5.4. ANALYSIS

Transcripts were analysed using descriptive analysis of the manifest content. Manifest content refers to what the text says rather than what it talks about and therefore it deals with describing the visible components rather than interpreting meaning (Graneheim and Lundman, 2004). Initially the interviews were read repeatedly in order for the researcher to familiarise herself with the content of the interviews and then data were categorised based on the four content areas covered during the interview. For each interview with a geneticist or genetic counsellor in charge of a service, a patient pathway within the genetics service and across services was mapped based on the descriptions of the professional and the perceived aims and objectives. The (dis)benefits on those referred and the perceived main outcome were also recorded. What follows is a description of participants’ narratives, and where information from another source is given the reference of the source is provided.

5.5. RESULTS

5.5.1. Overview of the Clinical Genetic Service processes

Access to the CGS can be instigated through a number of routes. A referral can be initiated either by a general practitioner or other medical specialist based in the catchment area or from outside the catchment area as an “out of area” referral. Cascade screening can also result in the access of individuals who are members of a
high risk family and who are traced after the referral of a patient. Access can, in some cases, be instigated by the individual themselves when the family of the individual is already known to the service.

When individuals or families are referred to the CGS, and within one month after receipt of the referral, they are usually contacted by their local nurse specialist before being seen in clinic. The purpose of this contact is to instigate support for the family and to gather data required for risk assessment and genetic counselling. Depending on the protocols and procedures followed in each genetic condition for confirming risk status, this contact may or may not be present. Once families are referred to the service, information and support is available in the long-term based on family and individual needs.

The CGS, in addition to a general service offered by all regional genetics centres, offers two distinct specialist services: a specialist prenatal genetic service including a clinical and counselling team for dealing with all referrals from specialist obstetrics services, and a cancer genetics service dedicated to all hereditary cancers. The cancer genetics service is the only aspect of the chosen CGS employing dedicated full time nurse specialists.

Of the five groups of conditions identified for this research, four conditions (CF, DBMD, VHL, TS) fall under general genetic services while prenatal genetic services may come into contact with cases from any of the four, with cystic fibrosis cases comprising a significant part of the workload for prenatal genetics. A reason for this is that CF is the most common genetic condition among Caucasians affecting one in 2500 individuals.
For this reason patient pathways for CF and prenatal genetic services will be reported together. The fifth group, hereditary breast and ovarian cancer, falls solely under the cancer genetics service.

As part of standard practice, all individuals seen by a genetic professional are sent a post-consultation letter outlining the discussion that has taken place between the professional and patient, along with any risk information disclosed during the consultation.

The following section will provide a detailed overview of the patient pathways and model of service provision existing for each of the five groups of conditions at the time of the data collection.

5.5.2. Cancer Genetics Service: Hereditary Breast and Ovarian cancer

5.5.2.1. Overview of condition

Breast cancer is the most common type of cancer in women, with 1 in 10 women being diagnosed with breast cancer in their lifetime. Ovarian cancer is less common with 1 in 100 women being diagnosed with the condition. Five to ten percent of cases of breast/ovarian cancers are linked to the presence of mutations in dominantly inherited genes, including BRCA1 and BRCA2. A hereditary predisposition is more likely when there is a cancer family history, with multiple relatives having the same or related cancers from the same side of the family and often at a young age (NHS Evidence, 2005). In individuals identified to be at high risk for HBOC a special management programme is recommended (Gronwald et al., 2008). Management can include prophylaxis (e.g. oral contraceptives, Hormone Replacement Therapy,
chemoprevention and mastectomy), surveillance (e.g. self-examination, mammography and transvaginal ultrasound), and treatment involving different considerations than non-hereditary cancer (Gronwald et al., 2008). Prognosis for HBOC depends on the stage of the cancer at the point of diagnosis.

This genetic condition is different from the others in that in order to be tested individuals need an affected living member of their family to be tested first for the presence of a genetic mutation. When there are no affected family members alive or when affected individuals are unwilling to be tested, this means that a number of individuals cannot undergo genetic testing. At the same time however, the majority of those who can be tested, receive an inconclusive genetic test result. Even if an individual is identified as having a genetic mutation for HBOC, this does not mean that the individual will certainly develop cancer during their lives. Rather it means that the individual has a higher risk than the general population of developing cancer.

5.5.2.2. Patient Pathways

The Cancer Genetics Service receives approximately 60% of referrals of the whole of the CGS and the majority of cases are breast, ovarian and bowel cancer referrals. The majority of referrals are received from oncologists, breast surgeons and GPs. Self-referrals are also possible, although not as frequent. After referral of an individual at suspected risk for a hereditary breast and/or ovarian cancer (HBOC) gene mutation the individual is registered and then it is determined whether the referral is appropriate i.e. whether the individual is indeed at risk. If the referral is not appropriate then a discharge letter is sent to the individual(s) and their GP. If it is considered appropriate (based on a number of criteria such as number of affected family members, age of
cancer onset), then the individual(s) is sent a questionnaire in which they are asked to provide details of their family history. The information provided in the family history questionnaire is used to draw up a family tree to assist in patient risk evaluation.

The process from referral to risk review is led to a large extent by genetic counsellors and can take between six to nine months from the date of referral, depending on the accuracy of information given in the family history questionnaire. Those identified as being at or near population risk are sent a letter informing them of their “average” risk and are referred back to their GP. Moderate risk cases, where an above-average risk is involved are sent a letter containing information about their risk and age-related recommendations for screening and, where appropriate, preventive practices. High risk cases are seen in a genetic counselling clinic where their risks are explained, risk management options and the possibility of genetic testing discussed.

The service is reactive, whereby individuals and families are advised to contact the service when new information becomes available, for example a new cancer incident in the family, or when the individual reaches an age where onset of cancer screening is advisable. If the person does not access the service, no mechanisms are in place to pro-actively contact the individual.

Genetic testing is only possible where a living member of the family with cancer is present and willing to give a blood sample for mutation searching. The average number of times that a high risk patient will be seen by a geneticist is four. Age-appropriate cancer screening is offered to those at moderate or high risk. Breast cancer screening is offered by Breast Test Wales, a population screening service which has formal direct
links with the AWCGS to accept referrals for women at a younger age than the population recommended onset age of 50 years (NICE Clinical Guideline 41, 2006). Ovarian screening is offered within a UK clinical trial evaluating its clinical effectiveness in high risk women aged over 35 years (UK Familial Ovarian Cancer Screening Study: UK FOCSS). Bowel screening may also be recommended for those at increased risk of carrying a bowel cancer predisposing gene mutation.

During the genetic counselling session with the clinical geneticist the option of prophylactic surgery is also discussed. This might involve prophylactic mastectomy and/or oophorectomy depending on the case. If a woman chooses to undergo prophylactic surgery then she is referred to a secondary care specialist who will discuss the available choices with the patient. The cancer genetic service is involved in the process to provide information and advice to both parties.

The interviewee pointed out that up to the point of the interview there are no formal guidelines regarding what specifically should be offered by all UK cancer genetic centres. Guidelines exist regarding screening and management practices for all cancer patients. All centres in the UK provide the same core services including risk assessment, genetic testing and genetic counselling; however differences can exist in the models of service provision followed by each specific centre, for example in the cancer risk assessment models used (Amir et al., 2003) and methods of delivering genetic risk information (Phelps et al., 2004). Other differences include whether “comprehensive mammographic screening” is available to women. As yet not all above-average risk women can access mammographic screening through UK genetic centres since not all centres offer regional wide, comprehensive services. Other UK areas can
be found to have a “post-code lottery” type of service where women have to access a surgeon who would then have to decide whether a referral to screening is appropriate.

**Figure 5.1: Patient pathways for HBOC**
Box 5.1: Use of services other than the cancer genetics service

<table>
<thead>
<tr>
<th>Referrals are made based on appropriateness to:</th>
</tr>
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<tbody>
<tr>
<td>• Breast Test Wales</td>
</tr>
<tr>
<td>• Ovarian cancer screening – UK Familial Ovarian Cancer Screening Study (UK FOCSS)</td>
</tr>
<tr>
<td>• Secondary care specialists for prophylactic surgery</td>
</tr>
</tbody>
</table>

5.5.2.3. Perceived role of the cancer genetics service

The role of the service was perceived as identifying people who are at increased risk of developing cancer and informing them of their risk status so that screening and prevention can be targeted. One important aspect of the cancer genetics service is that any impact on health outcomes depends on outside services directly involved in providing the screening and other health interventions such as surgery, as well as on the uptake by the patients. The immediate role of the genetic service was therefore considered to involve informing, educating and encouraging high risk individuals to attend screening and consequently to allay their fears.

5.5.2.4. Perceived (dis)benefits

The most important benefit of the service as this was reported by the geneticist was the information provided about the meaning and implications of the genetic cancer risk status. More specifically this involved making sure that the family understands the genetic information given and they have the relevant information needed to make informed choices. The following figure (Figure 5.2) illustrates the perceived benefits and disbenefits distinguishing between service-related benefits and psychosocial
outcomes relating to the service provision. Perceived outcomes understood to be a result of contact with the service included reassurance of knowing the individual and family risk status and of being given options about what to do to address the risk. Related to this is the consequence of coming to terms with the family risk status, as this was reported by the participant. In some cases the participant reported that there might be a conflict of expectations where perceptions of low or even high risk status were met with a reality which did not reflect these beliefs, resulting in negative emotional consequences. A negative impact on emotional well-being was also understood to result from the sometimes extended time periods between referral and subsequent contact with the service, depending on the amount of time it takes to collect information from the various sources and confirm family history. Time delays could also result between the time of the blood test and genetic test result depending, for example, on laboratory case load and how easy it is for the laboratory to identify the specific gene mutation. Disturbed relationships between the family as a result of increased risk status in one member and altered and/or negative self and body image were also described, and these were believed to often be present before the referral to the service. However, identification of high risk and subsequent decisions made to manage that risk such as decisions to undergo mastectomy, were believed to potentially have a negative impact on relationships and on the psychological state of the individual and their partner. In this context, the participant saw their role as being to make the couple or the individual aware of the potential implications of being at high risk and especially of the impact of management options such as mastectomy but not to directly deal with the psychological issues which they perceived should be handled by a psychologist in the case of issues
relevant to family history or by surgical services in the case of issues relevant to surgical prophylaxis.

**Figure 5.2: Perceived (dis)benefits of HBOC genetics services**

The left column lists the (dis)benefits related to the process of service provision while (dis)benefits on the right illustrate psychosocial outcomes.

5.5.2.5. **Most important outcome measure**

Satisfaction with the genetics service was identified by the geneticist as the most suitable outcome measure to evaluate the impact of the service. Even though the interviewee recognised the difficulty in measuring satisfaction, it was believed that the
effectiveness of the service rests in making sure that all patients are satisfied with and value the services received.

5.5.3. Cystic fibrosis

5.5.3.1. Overview of condition

Cystic fibrosis (CF) is the most common inherited condition among the Caucasian population. CF is an autosomal recessive condition, meaning that both parents need to carry the mutation in order for their child to be at risk of being affected. It is found to affect 1 in every 2500 newborns. It is a chronic, progressive, and frequently fatal genetic condition of the body’s mucus glands. CF primarily affects the respiratory and digestive systems in children and young adults. Sweat glands and the reproductive system are also usually involved. On average, individuals with CF have a lifespan of about 30 years. The condition course is variable and unpredictable with onset of symptoms in infancy or early childhood. The condition is untreatable and control of symptoms requires physiotherapy, antibiotics and in severe cases lung transplantation (Accurso, 2008).

5.5.3.2. Patient Pathways

5.5.3.2.1. Cystic Fibrosis

A first route into the service is following newborn screening available to all babies in the UK (Southern, 2004). Initially, identified babies are referred and managed by their local paediatricians and there is probably variability across Wales as to the timeframe parents and extended family members are referred to the genetics service. Genetic counsellors
might have very little contact with parents of newly diagnosed children through the newborn screening programme but these families are more likely to access the service when family members reach a reproductive age in order to discuss the impact of the condition on their reproductive choices, or when carriers of the condition want their partner to be tested.

As a result of the neonatal screening, occasionally carriers of the condition are also identified, and parents are then referred to the service for genetic counselling. These referrals could then lead to cascade screening of other family members. Couples, where both individuals are carriers, access the service intermittently to discuss prenatal testing or care pathways for their child if they choose to continue with the pregnancy.

Affected individuals might very rarely be referred to the service for cascade screening and further genetic counselling if no genetic diagnosis or counselling were provided following the clinical diagnosis. Affected males could also be referred to the service to discuss potential infertility options regarding Assisted Reproductive Technologies, as 98% of males with CF are infertile (Lyon and Bilton, 2002).

All individuals seen by the genetic counsellor are sent a letter following their session where the content of their genetic counselling session is summarised. The genetic test result is included in a second letter also forwarded to their GP. In Wales, CF patients are managed predominantly by counsellors working in Cardiff and the district and all genetic counsellors have CF families in their work load. Service provision is reactive, where patients are most often seen a small number of times after initial referral and
access to the service is then initiated by the patient and their families when further questions arise.

One issue with CF is that because of newborn screening where children are referred to paediatricians and because management of CF is carried out by clinicians outside the genetic service, a number of genetic tests are ordered from departments outside the service. In these cases, the clinician is not required to refer the patient and their families to the genetic service. Therefore not all individuals tested for CF receive genetic counselling. In these situations the service might at a later stage be called to “pick up the pieces” in cases where inaccurate information was given, or where a test is difficult to interpret outside the service. A distinct characteristic of the service is its expertise and knowledge, which ensures that the appropriate genetic test is offered to families along with accurate and case-appropriate information about the meaning of the result. In cases where it is not possible to offer a genetic test, the service might contact families again at a later stage to offer the appropriate test when the circumstances change and/or new technology is available.

Families and patients can be referred to health professionals such as paediatricians if there is concern about the care of the patient, and care pathways might be put in place. Information about, and referrals to services able to provide social and practical support to families, are also put in place.

Cystic fibrosis is unique among the chosen conditions in that the CGS has limited involvement with patients. All patient management is done outside the genetic service and families and individuals come into contact with the service following referrals from
Chapter 5

prenatal or neonatal services, or because contact was initiated due to a family history of CF. Contact with the service therefore is short-term and usually only involves genetic counselling.

5.5.3.2.2. Prenatal genetic services

The prenatal genetic service deals with all referrals to the CGS of pregnant women regardless of condition. It is a tertiary referral sector for specialist obstetrics services and the prenatal genetic clinical and counselling team is based on the main hospital site. All referrals from the catchment area are co-ordinated through the centrally-based genetic counsellor team who refer women from the district to district-based counsellors while women who require immediate clinical input are seen by professionals centrally-based. This is a genetic counsellor-led service with geneticist input following the initial visit by the genetic counsellor. The majority of referrals to the prenatal genetic service are related to a family history of CF.

The processes and principles of prenatal referrals are similar to the ones in general referrals with the difference being that everything is compressed in a very small time frame. Women need to be seen very quickly after referrals and decisions need to be made under considerable pressure without the time available to families seen in the general genetic services.

Referrals come from four different sources: 1) Families with a genetic condition known to the CGS tend to self-refer and contact the service soon after pregnancy to ask questions about options during pregnancy and for some to ask for prenatal testing; 2) Families with a family history of a genetic condition but who have had no contact with
the CGS are usually referred by midwives, GPs, obstetricians or sometimes by paediatricians either at the family’s request or because of the professional’s concern; 3) With the availability of prenatal screening to all pregnant women, women are referred to the service after the detection of abnormal results through amniocentesis or CVS, and 4) After the detection of foetal abnormalities during the ultrasound scan.

Following referral, genetic counsellors contact the family via letter or telephone-call on the day the referral is received in order to arrange for a pre-clinic appointment. Families are mainly interviewed over the phone, even though in some cases a home visit or face-to-face clinic appointment can be arranged, based on assessment of the individual case scenario. During the initial phone contact the counsellor discusses the family’s concerns and gives an overview of what the genetic service offers, enquiring about questions the family might have at that point and what their expectations of the service might be. The family’s anxiety levels are assessed in order for the counsellor to make a judgement of how the contact with the family should proceed, e.g. make a house visit, and the specific needs of the family. During genetic counselling appropriate carrier testing might be offered to the couple if possible. This is accepted by some, but not all, with some couples choosing to have testing after their baby is born. A pregnancy outcome form is given to families asking, among others, questions on service satisfaction and whether families would like to be contacted again by the service. Patients are not traditionally followed-up after their care is passed over to either district genetic counsellors or other medical professionals, even though some genetic counsellors choose to contact patients occasionally if they consider it appropriate.
Prenatal genetic services have very close links with other departments and work in a multidisciplinary setting including close collaboration with laboratory genetic services; and multidisciplinary working with foetal medicine, neonatology and specialist obstetrics services. The service also needs to co-ordinate management of the pregnancy when the family decides to go ahead with prenatal testing, by co-ordinating different tests with obstetric services, foetal medicine and the laboratory service and also arrange for termination of the pregnancy with the gynaecology service if this is chosen by the family. The service therefore works as part of a multidisciplinary team involved in the management of the pregnancy, the genetic counselling of the family, education of other professionals and co-ordination of the management of the pregnancy.
Figure 5.3: Patient pathways for cystic fibrosis

Neonatal screening ➔ Known families ➔ Genetic Counsellor

- Family history
- Genetic counselling

Cascade screening ➔ Genetic testing possible ➔ Genetic testing not possible

- Opt for genetic testing
- Opt out of genetic testing

Facilitating access to:
1. specialists (care pathways)
2. social services
3. support groups
Figure 5.4: Patient pathways for prenatal genetics

- Known families
- Family history families
- Amniocentesis/CVS/serum abnormal results
- Ultrasound scan abnormalities

PRENATAL GENETICS

- Genetic counsellor pre-clinic appointment

Clinic appointment

- Parental carrier status (if known family mutation)
- Postpone contact with genetic services after birth

Prenatal testing (~10%)

Care back to district teams, obstetricians, paediatricians
Box 5.2: Use of services other than the cystic fibrosis genetics service

Referrals are made based on need to:
- Secondary care specialists e.g. Paediatricians
- Social services
- Voluntary organisations

Box 5.3: Use of services other than the prenatal genetics service

Referrals are made based on need to:
- Obstetrics and gynaecology departments
- Foetal medicine department
- Paediatricians

5.5.3.3. Perceived role of the service

The most important role of the CF service was perceived to be providing accurate information including clarifying whether someone is a carrier or not in order to facilitate informed decision-making, and if necessary facilitate other, usually prenatal options. By providing relevant and accurate information to patients and their families the service facilitates informed decision-making based on the options available as well as on the family’s views, moral, religious and ethical beliefs. The genetic counsellor’s role is to support the family and the individual through the decision-making process in order for them to achieve their family’s goals.
After a decision has been made, a second role of the service is to facilitate access to other professionals and services whether these are clinical or outside the health service who can provide them with further information and/or practical support in order to help the family deal effectively with the situation.

Similarly, the prenatal service aims to answer questions during pregnancy about a family’s genetic history, chromosome results or scan anomalies; to provide information about genetic inheritance and risks; to inform families about their options for managing their pregnancy; and support them in choosing the right management plans. The genetic service co-ordinates the management plan of the pregnancy if one is chosen by the family. Part of the role of the genetic service is also to allow families to express their needs and expectations of the genetic service, the nature of the information they require in order to make an informed decision and fulfil these needs. Education of the families, health professionals, educators, as well as the public is a third role of the service.

5.5.3.4. Perceived (dis)benefits

The most important benefit of the CF and prenatal genetic service was perceived to be the provision of genetic counselling and associated support with decision making. Numerous other process-related benefits were raised by the participant. In the context of genetic counselling, counselees were offered accurate information on genetic risk status and were given understanding of the genetic condition. They were also given information on their reproductive options as well as other options available to them without being pressured into making decisions perceived or presented as appropriate by the counsellor. Time was also perceived to be an important characteristic of the service.
where individuals and families were given the required time to think through the information they were given and make decisions such as the decision to undergo genetic testing. In order to support families further in their decisions, the participant described how the counsellors made links to other services such as obstetric services or other specialists and support groups. Other characteristics of service provision, which were perceived as a benefit to service users, were the open-access policy of the service where families could contact the service when they needed new information, or in some cases the genetic counsellor themselves contacted the family to check if everything was ok, thus providing follow-up. The service was also described to provide person-centred care, adjusting service provision based on individual family needs. Another benefit raised was its involvement in educating other professionals, thus increasing knowledge of genetic conditions among practitioners and consequently improving service provision.

Several psychosocial (dis)benefits were also raised, and these were linked to the positive aspects of service provision and the implications of genetic disease. Coming to terms with the situation, enabling and supporting individuals and families to make decisions resulting in empowerment, supporting individuals to manage uncertainty and thus promote a sense of control, were raised as benefits of genetic counselling. Helping individuals and families understand the information they were given was described to result in more accurate information being passed on to future generations, and subsequently ensure the access of future generations to CGSs.

As a result of the genetic information, a number of psychosocial implications on the wider family unit were described and these could be associated with negative outcomes
on other family members as well as the individual initially referred to the service. These potential disbenefits included changes in family relationships because of: the identification of a family risk to a genetic condition; implications to other family members who might be at risk; and, implications for the individual's sense of identity and belonging to the family unit. Sometimes individuals who are part of families with a history of a genetic condition, link genetic status to a sense of belonging to the family unit, thus not carrying the genetic mutation might disrupt that sense of belonging.

All (dis)benefits raised by the genetic counsellors can be seen in Figure 5.5.

5.5.3.5. Most important outcome measure

The appropriateness of the information given by the genetic service and the usefulness of that information in terms of both timing and achieving of goals were perceived as an appropriate way to evaluate the service by the district genetic counsellor. A related outcome measure proposed by the second counsellor was the empowerment of families to make choices and take decisions at a bad time in their lives. The following table illustrates the (dis)benefits discussed by the two counsellors distinguishing between service-related benefits and other consequences.
**Figure 5.5: Perceived (dis)benefits of cystic fibrosis and prenatal genetics services**

- Support with decision-making
- Pressure-free decision-making
- Offer options
- Time
- Person-centred care
- Open-access policy
- Follow-up
- Links to other services/specialists and support groups
- Educating other professionals
- Coming to terms
- Empowerment to make decisions
- Managing uncertainty
- Sense of control
- Understanding of information
- Passing on of accurate information to future generations
- Access of future generations to CGS
- Impact on family relationships
- Sense of identity in family unit
- Genetic implications to other family members
5.5.4. Tuberous Sclerosis

5.5.4.1. Overview of condition

Tuberous Sclerosis is a fairly common inherited condition which affects 1 in 6000 newborns. It has an autosomal dominant inheritance pattern with variable expression, which means that the condition is very variable in the severity and diversity of manifested signs and symptoms; and reduced penetrance which means that a low proportion of individuals with the specific gene mutation exhibit symptoms and signs of the condition. Tuberous sclerosis affects many of the body’s systems and involves development of tumours and hardening of organs or tissues in the brain, skin, eye, heart, kidney, bones, lungs and intestines. In 75% of cases epileptic seizures might occur while some degree of learning difficulty is present in 50% of cases. Behaviour problems and autistic tendencies are also common features of the condition.

5.5.4.2. Patient Pathways

Referrals to the CGS come almost entirely from other medical specialties following suspicions of TS. Patients can be referred to the service in a number of ways. Firstly, abnormalities might be present in utero when heart tumours which are a symptom of the condition would be apparent during the prenatal ultrasound. A prenatal diagnosis can be made and the family would be referred to the medical genetics service for genetic counselling. A second way in which families are referred is when the paediatrician suspects tuberous sclerosis, typically during the first year of the baby’s life because of fits or other symptoms of the condition. Clinical problems might also present anywhere
between later childhood and adulthood and these can bring the patient to clinical
attention and eventually referred to the medical genetics service.

The service is a geneticist-led service with input from genetic counsellors in both the
initial contact visit of taking family history and in the subsequent clinics. The clinical
service for tuberous sclerosis is a reactive one, and care depends on the needs of each
individual patient, as opposed to other pro-active services for other conditions. The
service is reactive in that patients and their families contact the service based on need,
e.g. when information is required either on the symptoms of the condition or on
reproductive decision-making. Patients and their families however are life-long clients
of the service as is the case with all other genetic conditions. The service is centred on
providing information on the nature and the symptoms of the condition and the co-
ordination of care for the patient by involving other medical or other professionals rather
than providing clinical care. Prenatal diagnosis and pre-implantation diagnosis can be
organised for the patients and their families.

Services for tuberous sclerosis have evolved around the local clinical and research
interests of the professionals working in the CGS as well as the available resources.
National guidelines for the care of tuberous sclerosis patients are available and the
provision of genetic counselling for all patients is part of those guidelines.
Figure 5.6: Patient pathway for tuberous sclerosis

In utero
- Antenatal Diagnosis

Childhood
- Paediatricians

Adulthood
- Related Medical Specialists: e.g. kidney specialists Lung specialists

Known families
- GPs
- Direct patient contact

Other sources
- Social Services
- Patient societies

MEDICAL GENETIC SERVICES

Family History

Genetic Clinics
- Confirm diagnosis

Care co-ordination

Needs-based follow-up

- Genetic Testing (needs based)
- 1. Genetic Diagnosis
- 2. Prenatal Diagnosis

Known TS families
Box 5.4: Use of services other than the tuberous sclerosis genetics service

Referrals are made based on need to:
- Community paediatricians
- Secondary care specialists for managing symptoms
- Social Services
- Psychologists
- Educational services

5.5.4.3. Role of the service

Three components of the role of the service were described. Firstly, to provide information on the nature of the condition, the inheritance patterns and the implications for the families, information on the symptoms and prognosis and genetic testing if that is appropriate.

A second component is to provide support to families and coordinate their care by involving all the specialists necessary for treating the symptoms of each patient. The family are informed of all the support networks available to them both in terms of specialist medical care as well as outside the medical services in terms of educational or social support. Children are referred to a community paediatrician who will be in charge for their overall care and condition management and who will be the person liaising with other services in the community. Adults are assigned a team of specialists managing the various symptoms of the disorder which would not be treated without some co-ordination from the part of medical genetic services. Finally, the service provides options to patients and their families for genetic diagnosis, prenatal diagnosis and any other reproductive options available to families.
5.5.4.4. Perceived (dis)benefits

The service was described to offer a number of benefits to families and individuals associated to service delivery. According to the participants genetic providers were able to provide an overview of what the condition involved and of the available services and care pathways. Where further management was required, individuals were described to be allowed access to clinics in other healthcare departments making sure that all symptoms were managed by the appropriate professional. Because of the varied characteristics of the condition, the service was also described to ensure that symptoms are treated as part of the condition rather than as isolated issues. In addition to clinical services, the service also enabled access to social and educational services where the family could obtain support in dealing with the implications of the condition. Other important benefits raised which also related to accessing the service were, the opportunity given to parents to have prenatal tests, the confirmation of genetic status in relatives and the decrease in false diagnosis cases as a result of genetic testing. As a result of the above, the participants believed that families were offered certainty about their risk status and supported in making informed decisions. A sense of control over their lives was also believed to ensue from the support provided.

Figure 5.7 illustrates the (dis)benefits raised by the geneticist, distinguishing between service related characteristics and psychosocial outcomes.
5.5.4.5. **Most important outcome measure**

The geneticist interviewed believed that some measure of efficacy of meeting individual needs represented the most important outcome measure of the service. This involved the meeting of expectations but long-term rather than short-term expectations.
5.5.5. von Hippel-Lindau syndrome

5.5.5.1. Overview of condition

Von Hippel-Lindau Syndrome (VHL) is a rare genetic condition which affects a number of body organs through the appearance of angiomas and benign cysts or tumours. The incidence of VHL is estimated to be 1 in 36,000 individuals. VHL is passed on through the generations using an autosomal dominant inheritance pattern even though spontaneous mutations can also occur. Organs most often affected are the cerebellum, the spinal cord, the kidneys, the pancreas and the retina of the eye. VHL is a chronic variable condition, which means that not all individuals carrying the gene are affected to the same degree of severity, and expression of the condition can vary even within families and among those with the same mutation. Complications usually appear around the age of 40 even though for some individuals these may delay until after 50. Childhood onset is rare. It is recommended that all individuals who are at risk for developing symptoms of the condition are offered screening for early detection.

5.5.5.2. Patient Pathways

VHL patients make up a small part of the whole of medical genetics referrals with approximately two to five new referrals a year. The majority of VHL patients seen by the service come from known VHL families that have been followed for some generations. New referrals are mainly made by specialists e.g. ophthalmologists or neurologists, when they come across VHL-like symptoms, and from other UK genetic centres referring relatives of diagnosed VHL patients in order to confirm the diagnosis. All GPs with VHL positive patients on their lists are informed of the diagnosis and the
nature of the condition and are advised of the management and clinical needs of the patients. All patients are registered in the lists of medical specialists for direct access in case of symptom onset so any symptoms can be managed in their early stages.

Diagnostic, prenatal, child and adult pre-symptomatic genetic testing are offered to those seen by the service even though uptake is not compulsory and not taken up by all individuals. Refusal to undergo genetic testing does not exclude the individual from accessing the screening programme. In terms of neonatal screening which is more controversial, according to the geneticist, the time allowed between the first and second clinic appointment during which decisions need to be made has appeared to help couples come to terms with the uncertainty and not go through with testing.

All new patients referred to the service are invited to a clinic appointment with the geneticist. During the first appointment a family history is taken and family concerns are discussed. The issues around a VHL diagnosis and the implications to the family are also raised. The option of genetic diagnosis and the implications for the patient and their families are discussed during a second appointment and the decision to move forward with testing lies with the patient and their family. VHL is explained in more detail along with the scenarios for the progression of the condition and the availability of screening for the symptoms of the condition. The impact on their families and the need to inform their relatives is raised for the possibility of cascade testing. A fourth appointment is arranged at the family’s request for any unanswered questions and concerns after the diagnosis. Following the diagnosis (whether through a clinical or a genetic diagnosis) all registered families attend an annual genetics clinic and annual screening appointments with specialists for screening of VHL symptoms i.e. tumours.
and cysts developing in the kidney, brain, spinal cord, pancreas and retina for example. Even though all patients attend an annual genetics clinic to discuss their screening and general condition, patients are free to contact the service via phone or mail to discuss their concerns relating to the hereditary condition and the results of their screening.

In addition to direct access and referrals to medical specialists, the service will also provide information and education about VHL to patients and their family. This involves emotional and practical support through information regarding which symptoms are likely to appear and their potential physical and psychological impact on the patient. Patients are prepared and warned in advance about the possible impact of the condition.

The CGS provides a geneticist-led service for all VHL families within the catchment area with minimum input by genetic nurses or genetic counsellors. It is a pro-active service, meaning patients and their families are recalled back into the service at regular intervals, the norm being yearly follow-up, for monitoring of condition progression. The service organises and co-ordinates the annual screening for families which takes place in health services outside the CGS. Symptomatic patients will tend to contact the service more frequently depending on the symptoms present and the needs of the patients.

Patients are also referred to other sources of information or support as well as specialists in other genetic centres for second opinions on clinical aspects of the condition. Patients are also informed and enrolled in clinical research taking place in other genetic centres. In cases where the patient is experiencing extreme emotional
strain the service can also refer the patient to counsellors through their GP and also inform about voluntary support organisations.

**Figure 5.8: Patient pathway for VHL**
Box 5.5: Use of services other than the VHL genetics service

<table>
<thead>
<tr>
<th>Referrals are made based on need to:</th>
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<tbody>
<tr>
<td>• Secondary care specialists for symptom screening and management</td>
</tr>
<tr>
<td>o Ophthalmologists</td>
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<tr>
<td>o Neurologists etc</td>
</tr>
<tr>
<td>• Other genetic centres for clinical research</td>
</tr>
</tbody>
</table>

5.5.5.3. **Role of the service**

The role of the service is to facilitate access to screening for VHL families, regardless of whether the individuals decide to undergo pre-symptomatic testing or not. Even though the service is there to support the decisions and needs of the patient, uptake of the screening programme is the only aspect of the service advocated to the patients. This is because participating in screening has a clear medical benefit on the lives of the patients increasing their lifespan by at least 20 years by treating symptoms of the condition as soon as they are detected (Priesemann et al., 2006). A second role of the service is to limit the (psychological) impact of the condition on the patients.

5.5.5.4. **Perceived (Dis)benefits**

The most important aspect of the VHL service was perceived by the participant to be the provision of screening and management of the condition (clinical support), which involved long-term follow-up encompassing psychological and practical support. This was believed to result to the most important consequences of the service, namely
increased life-span, more years with family and children, reassurance of being part of a management programme.

VHL is somewhat different from the other conditions in that there are clear medical benefits from the genetic diagnosis of VHL if that is used to organise and provide medical follow-up. Figure 5.9 illustrates the service-related benefits and psychosocial outcomes reported by the geneticist.

5.5.5.5. Most important outcome measure

When asked what the most important outcome measure would be that the service could be evaluated upon, the geneticist thought that it would be the sense of normality that the service would be able to give to the patients and the feeling they can live a purposeful life. This included the ability to feel “equal and normal human being”, “how good they feel about themselves […] how much of an outcast does VHL make you feel”.

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5.5.6. Duchenne and Becker muscular dystrophies (DBMD)

5.5.6.1. Overview of condition

Duchenne (DMD) and Becker (BMD) muscular dystrophies are the two most common (1 in 3,500) of over 20 types of muscular dystrophies. This group of conditions are neuromuscular disorders involving muscle degeneration or progressive weakness of the muscles. DBMD are inherited with an X-linked recessive pattern and even though women can also be carriers of the condition symptoms are mostly evident in males where one copy of the altered gene is enough to cause the condition.
DMD and BMD differ in the age of onset of symptoms, with the first DMD symptoms evident in toddlers when the baby begins learning to walk whereas symptoms of BMD are often evident later in childhood or adolescence; rate of progression, with DMD progressing rapidly; and their severity, with DMD being more severe than BMD with affected children being confined in a wheelchair by the age of thirteen. Heart and respiratory muscles are also affected and by the late teens affected individuals will suffer from respiratory failure which is fatal within a few months of onset if is not treated. The signs of BMD are usually milder and demonstrate a large range of variation.

5.5.6.2. Patient Pathways

Traditionally most patients are identified and referred to the genetic service after symptoms are observed (around 3-5 years of age for DMD and in late childhood or adulthood for BMD). In Wales however, an “opt-in” neonatal screening service for DMD has been running since 1990 (Parsons, et al 2003), increasing the numbers of cases identified and referred to the service. DBMD make up a significant part of the muscle genetic service. This service, similar to the other services provided by the CGS, is provided to patients “for life”, because of the chronic nature of the condition and the links with the other members of the family.

The way the service is currently structured is a result of both the interests of the geneticists developing the service and of the resources at their disposal. The search for muscle dystrophy genes has led to geneticist-led muscle clinics in addition to the genetic clinics traditionally offered by the service. In recent years the input of genetic nurses has been introduced at different stages of the delivery of the service. This is a pro-active service with an active register where the patients attend the muscle clinics for
monitoring of their condition and referrals for check-ups either 6-monthly or yearly. A register for all female carriers also exists and these women are recalled every 5 years for heart monitoring, since recent evidence suggests that female carriers can be at increased risk of heart conditions (Mirabella et al., 1993).

**Duchenne Muscular Dystrophy**

*Referral to the CGS via the Newborn Screening Programme*

Since 1990 screening for DMD is routinely offered as part of the Guthrie test. This is an opt-in test, meaning that parents need to consent for their child to be tested for DMD. The child is tested for high levels of the CPK enzyme which is an indicator for DMD diagnosis. Results are given to parents whose child has raised CPK enzyme levels at week 6 along with a referral to a paediatrician or paediatric neurologist. The paediatric neurologist collects blood samples for genetic diagnosis and refers the family to the CGS where the family is visited by a genetic nurse for a family history and genetic counselling. The family is explained the nature of the condition, the way it is inherited in families, its progression and the implications to the family members of their family. Further genetic counselling is provided by the geneticist when the patient attends the genetic clinics. The management of the condition in children is the responsibility of the paediatrician. The service is reactive but still available to families “for life” and can be accessed at any point.

*Referral to the CGS after the child develops symptoms*

A child might also be referred to the CGS by a paediatrician after the child develops symptoms of DMD around 18 months to 6 years of age. The same procedure is followed in that the child is tested for the presence of the gene or in some cases a
muscle biopsy would be performed. The family is again visited by a genetic nurse followed by an appointment with the geneticist at the genetics clinic. Again the management of the condition is the responsibility of the paediatrician.

**Known DMD families**

All families who have accessed the service because of DMD in a member of their family are part of the service “for life”. When a patient with DMD reaches adulthood they attend a muscle clinic once or twice a year where their condition is monitored and the results of tests are discussed with the patient. The geneticist goes through all the medical needs of the patient as well as discusses social and family needs. The patient might be informed of services they could receive from outside agencies, for example social services for disability benefits, adaptations to their houses and access to physiotherapy and Occupational Therapy. The needs of their families are also discussed and opportunistic genetic counselling could also take place. Arrangements for prenatal genetic testing or pre-implantation diagnosis are also made for the families of patients.

All female members of known DMD families will be tested for being carriers of the DMD gene i.e. mothers and sisters of the patients, aunts and other more extended relatives. All carriers will be part of a register and would be recalled every five years via a letter for heart problem check-ups.

Milder symptoms of DMD might also appear in female carriers who would also attend the muscle clinics for monitoring of their symptoms.
Becker Muscular Dystrophy

BMD is a milder form of DMD where symptoms appear in male carriers in late childhood, adolescence and adulthood. Access to the service might be through two different routes: either when symptoms appear in a patient with no prior family history of the condition and the individual is referred via their GP or neurologist; or because of a known family history of the condition.

New referrals are again visited by a genetic nurse for family history and providing information of the condition, followed by a visit to the genetics clinic for taking blood samples for genetic testing. All patients are regularly seen in the muscle clinic and the procedure would be the same as in DMD. Prenatal diagnosis, pre-implantation diagnosis and pre-symptomatic testing mainly for BMD can be arranged for all families. Referral to the service for carrier testing for the at-risk individual or their families can be postponed if that is required by the families.

5.5.6.3. Role of the service

The role of the service is firstly, to ensure the patients and their families have accurate genetic information based on the results of their genetic testing, and secondly, that the patients are getting the management and support they require. Providing the correct genetic information involves making sure the right diagnosis is communicated to the patient i.e. X-linked condition, and the appropriate options are offered to the families. Providing genetic diagnostic services and giving the correct genetic advice to families is considered to be the most important role of the service.
Figure 5.10: Patient pathways for adults referred to DBMD genetics services

New Referrals for BMD
GP
Neurologist

Referral to the CGS

Family Tree and Genetic Counselling (genetic nurse)

Visit to genetic clinics – genetic counselling (geneticist)

Genetic Testing

BMD positive

Cascade screening for Female members of family

BMD negative

Adult Muscle Clinic for management of condition (x1 or x2 per year)

Follow-up of female members of the family

Known BMD families

Presymptomatic testing for children (less often for adults)

Known DMD families

Prenatal Testing

Pre-implantation diagnosis

Regular recall for heart monitoring
Figure 5.11: Patient pathways for under-age referrals to DBMD genetics services

**DMD**

**Pre-symptomatic**

- Newborn Screening Service

- Newborn identified with high CK levels

- Referral to Paediatrician Paediatric Neurologist

**Child has symptoms**

- Neurologist/Paediatrician

- Genetic Testing through Laboratory Genetic Services

- Genetic testing identifies DMD

- Referral of family to Clinical Genetic Services

- Family Tree and Genetic Counselling (genetic nurse)

- Visit to genetic clinics – genetic counselling (geneticist)

- Child referred back to paediatrician/neurologist for management of condition

- Mother/family tested for gene

- Mother is positive

- Cascade screening of female members of the family

- Follow-up

- Annual recall service for female carriers

- Pre-implantation diagnosis

- Prenatal Testing
Box 5.6: Use of services other than the DBMD genetics service

<table>
<thead>
<tr>
<th>Referrals are made based on need to:</th>
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</thead>
<tbody>
<tr>
<td>• Social services</td>
</tr>
<tr>
<td>• Occupational Therapists</td>
</tr>
<tr>
<td>• Physiotherapists</td>
</tr>
<tr>
<td>• Voluntary organisations/support groups</td>
</tr>
</tbody>
</table>

5.5.6.4. Perceived (dis)benefits

The participant distinguished between (dis)benefits resulting from accessing the CGS and those which were perceived to be a consequence of the disease diagnosis. Important attributes of the service were perceived to be the time available to families during sessions where they can be given appropriate information about their risks and the nature of their condition, they are prepared for the prognosis of the condition and given practical support such as enabling access to Social Services and voluntary organisations. The service was described to offer patient-centred care and forming long-term relationships with families which were also perceived as benefits of the service. Psychosocial outcomes described to result from being part of the service were the supportive relationships formed and the associated psychological support given to families and reassurance.

Implications described to result from obtaining a diagnosis were the communication difficulties which can possibly follow within families, psychological harm such as depression resulting from multiple prenatal testings, as well as depression from having a chronic degenerating condition.
All (dis)benefits mentioned by the geneticist distinguishing between service-related benefits and outcomes can be seen in the figure 5.12.

5.5.6.5. **Most important outcome measure**

The most important outcome measure upon which the service could be evaluated, according to the geneticist, was the understanding of patients and families of the correct and appropriate information: “…everybody should be given the right information and understands it…” The families should be given the information when they want it and the geneticist should make sure this information is understood, as the stress and anxiety individuals experience when given this diagnosis can impede on their understanding. Accurate understanding of the situation can also enable the families to make the right decisions for them.
5.5.7. Laboratory genetic services

The Genetic Diagnostic Laboratory service of chosen medical genetics service is comprised of three sections: cytogenetics, molecular genetics and molecular cytogenetics. Laboratory genetic services are involved in technical analysis of samples and interpretation of results, quality assessment and accreditation processes, teaching and education, advice on the availability and appropriateness of specific genetic tests and interpretation of the analysis results.
The laboratory service is linked both to NHS services and to Academic departments of the affiliated medical school through the joint appointments of members of its staff, thus allowing research findings to be incorporated into clinical practice, and at the same time the education and training of students. Laboratory services are linked to clinical genetic services in that the head of laboratory genetic services reports through their line management procedures to the clinical director of genetics rather than the clinical director of laboratory medicine.

5.5.7.1. **Referrals to the service**

Laboratory and clinical genetic services are distinct from each other but also linked, as is the case with all UK medical genetics centres. The laboratory genetic service is the second arm of the chosen CGS and receives referrals from both clinical genetics and from other secondary care specialists. In that sense, laboratory genetic services work independently of clinical genetics therefore do not depend on geneticists and genetic counsellors for their existence.

The majority of referrals for genetic testing are from sources outside the clinical genetic service. The service does not accept self-referrals from individuals. Laboratory genetic services are a “consultant-led service” since they are acting as a consultant to the specialists referring samples for genetic testing. This involves advising clients on the appropriateness of their requests, where a request for genetic testing is considered by the laboratory based on its appropriateness and a decision is based on the patient’s interest. In this case a secondary care specialist might be advised that a test that has been requested is not appropriate for the specific case or that a genetic test might not be needed. For a number of genetic conditions clinicians outside the clinical genetic
service are advised that a referral for a genetic test can only be made through the clinical genetic service. After a genetic test is completed, the result of that analysis is given back to the specialist making the referral, along with an interpretation of that analysis.

Referrals are also made to the clinical genetic service in cases of abnormalities found during prenatal amniocentesis testing, or clinicians might be advised to refer patients to the service before a genetic test is done when that is deemed appropriate. Some of the demand for clinical genetics therefore is created by the laboratory service which is trying to link the patient with the appropriate person or service for their situation. In the same way, clinical genetic services act as a filtering system for patients referred to the laboratory service for genetic testing, ensuring that only appropriate cases are referred on for genetic testing.

### 5.5.7.2. Role of the service

The service aims to provide a quality service. This involves the provision of comprehensive, accurate analysis with an interpretation in a timely fashion, on as many condition conditions possible, at a cost within the resources allocated by the NHS. The laboratory service provides services to other secondary care specialists and not directly to the patients. In that sense, it aims to provide as much assistance as possible to clinicians through the provision of the appropriate genetic test and its interpretation, in order to enable them to provide the best possible treatment to their patients.
5.6. DISCUSSION

This chapter presented the methods and results of the first phase of data collection which aimed to map out patient pathways, use of resources and the perceived role and benefits of the service. A figure mapping out the patient pathway through the service was given separately for each condition, along with the referrals to services outside the CGS. An overview of the use of resources other than of the CGS is given in Box 5.7 while a detailed table incorporating all perceived aims and objectives of CGSs, perceived (dis)benefits and outcomes is provided in Appendix 5. A discussion of methodological limitations of this and of the following two phases is given in the final chapter.

The models of service provision described in this chapter apply to the period during which data collection took place (between January 2007 and January 2008). At that time a service audit was being undertaken were patient pathways were being formally recorded. Based on discussions with senior members of the CGS, current models of service provision were being reconsidered in the light of current developments and the expanding role of genetic counsellors. Since this process of reassessment was ongoing and no formal changes had taken place in the models of service provision at the time the data were collected, the model of service provision given by individual service providers is reported. This is also the service model provided to the service users taking part in the current research; therefore (dis)benefits described in the focus groups correspond to this model of service provision.
Box 5.7: Use of resources other than of the Clinical Genetic Service

Secondary care specialists (HBOC; VHL; TS; CF and prenatal genetics)
Social Services (TS; DBMD; CF and prenatal genetics)
NGOs and support groups (TS; DBMD; CF and prenatal genetics)
Occupational Therapists (DBMD)
Physiotherapists (DBMD)
Community specialists (TS)
Psychologists (TS)
Educational services (TS)
Cancer screening services (HBOC)
Other genetic centres (VHL)

Based on the descriptions of service providers, service provision models appear to be influenced by the needs arising from the individual condition characteristics and from the available condition management options. For example, the availability of screening for both VHL and HBOC means that high risk individuals as well as carriers of genes of these conditions are referred directly to established screening programmes in the case of HBOC, or a screening programme is put in place for each patient and coordinated by the genetics service as in the case of VHL where no national programme is in place. The involvement of the genetics service with VHL families was more pro-active compared with HBOC, arguably because of the benefit gained by the participation in a
screening programme of these individuals and the absence of such a programme in
general healthcare for VHL as well as the very small numbers of VHL families when
compared to cancer. Least involvement of the genetics service was described in the
case of cystic fibrosis, where quite an organised care programme is established outside
the genetics service so its role was limited to the provision of genetic counselling
concerning reproductive options and following a diagnosis in a child when a referral was
made by other healthcare professionals. CF is the most common genetic condition in
European populations and the wide knowledge among health care providers of this
condition might explain the limited role of the service.

What service providers perceive to be the role of the service is another important factor
influencing service provision models. The two most frequently cited aims of the CGS
were firstly to provide accurate information in order to ensure informed decision-making
and secondly to ensure that patients get the clinical management and social or other
support needed. Genetic services appear to be closing a gap in the care of individuals
affected by a genetic condition to ensure that appropriate treatment of these individuals
is in place. Impact on health outcomes in this sense takes place indirectly and is a
result of the expert understanding of genetic conditions and the needs of patients for
managing their symptoms.

The first of these components i.e. provision of accurate information and informed
decision-making accurately corresponds to the stated functions of CGSs as these were
discussed in Section 2.4. The second stated aim, i.e. establishing patient pathways and
ensuring that patients get the clinical interventions required in order to minimize
negative health implications, is not an explicitly stated aim of the service, yet is an
important one to service providers. Even though it is not as yet clear in what ways service provision differs between UK genetic centres and how carriers, patients and high risk individuals are dealt with by the healthcare service including the CGSs, research has suggested that practices may vary depending on the area (Foster et al., 2007). Participants in the current research perceived considerable variability present among centres in terms of providing follow-up, support and co-ordination of health surveillance. This variability seems to depend on manpower and financial resources (BSHG, 2000) and arguably on the outlook on the service i.e. the perceived goals of the service.

Based on the current service provision models and perceived aims of the service, participants described a number of service related and psychosocial benefits. Table 5.1 lists all the process and outcome benefits raised by participants. Attributes of service provision as benefits of the service were described by all participants and these were given more readily than psychosocial consequences. Two categories of process benefits were mentioned by all participants. Firstly, the information provided to families regarding their risk, the nature of the condition and the options available to deal with the condition, and secondly the access to surveillance, screening and management of symptoms. These benefits correspond to the two most important perceived aims of the service. Other categories of benefits raised by the majority of participants were the individualised approach to service provision e.g. taking concerns seriously and allowing adequate time for all users to address their concerns, and providing a long-term relationship with families through follow-up. Even though information and surveillance were raised by all participants, when asked to state what they perceived as the most
appropriate outcome measure upon which to evaluate the services they provided, not all perceived knowledge as the most appropriate outcome. Outcome measures raised by five participants fell under the theme of information, knowledge and decision-making whereas outcome measures linking to adjustment were mentioned by two participants. One participant perceived satisfaction as the most appropriate outcome measure and meeting of long-term expectations was raised by another, even though conceptually meeting of expectations is associated to satisfaction (Zellerino et al., 2009).

Despite the importance of perceived aims and goals of genetic services for service provision, research on the perceptions of service providers on what CGSs aim to provide and what their perceived impact is on service users, is scarce. Few studies were identified which aimed to compare the perceptions of service providers and service users on the outcomes of genetic counselling (e.g. Bernhardt, Biesecker and Mastromarino, 2000; Aalfs et al., 2007; Payne et al., 2007), or have focused on the perceptions of service providers (e.g. Michie et al., 1998; Williams et al., 2001).
Table 5.1: Process and outcome benefits raised by participants

<table>
<thead>
<tr>
<th>Process benefits</th>
<th>Outcome benefits</th>
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<tbody>
<tr>
<td>Time available</td>
<td>Reassurance</td>
</tr>
<tr>
<td>Access to screening and management interventions</td>
<td>Adjustment/coming to terms</td>
</tr>
<tr>
<td>Overview of condition and care</td>
<td>Informed decision-making</td>
</tr>
<tr>
<td>Effective symptom management</td>
<td>Understanding of information</td>
</tr>
<tr>
<td>Open-access policy</td>
<td>Certainty</td>
</tr>
<tr>
<td>Follow up of families</td>
<td>Sense of Control</td>
</tr>
<tr>
<td>Patient-centred care</td>
<td>Sense of normality</td>
</tr>
<tr>
<td>Long-term relationships</td>
<td>Limit the impact of the condition</td>
</tr>
<tr>
<td>Addressing concerns</td>
<td>Access of future generations to CGSs</td>
</tr>
<tr>
<td>Preparing for prognosis and psychological support</td>
<td>Increased lifespan</td>
</tr>
<tr>
<td>Accurate information about risks and genetic condition</td>
<td></td>
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<tr>
<td>Pressure-free decision-making</td>
<td></td>
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<tr>
<td>Provide options for decision-making</td>
<td></td>
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<tr>
<td>Decrease cases of false positives</td>
<td></td>
</tr>
<tr>
<td>Access to social and educational services and support</td>
<td></td>
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<tr>
<td>Education of other professionals</td>
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Outcome disbenefits
Anxiety due to waiting time

One of the few studies that investigated the perceptions of genetic counsellors as well as counselees explored what providers and users perceived to be the goals and outcomes of genetic counselling (Bernhardt, Biesecker and Mastromarino, 2000). Many counsellors in this study framed their perceptions of goals and outcomes in terms of
process, something that was also evident in the present study. Bernhardt and colleagues report that counsellors participating in their focus group were apprehensive in describing goals for a genetic counselling session, because they thought this implied that there was a “predetermined agenda” (Bernhardt, Biesecker and Mastromarino, 2000). Interviews with providers in the present study mainly focused around the patient pathways and service delivery models while the aims and psychosocial outcomes of the service were explored following discussions of these models. One might argue therefore, that in this case the focus of participants on service-related benefits might be a result of that initial conversation and no firm conclusions can be drawn as to the meaning behind this observation and thus make comparisons with the Bernhardt and colleagues findings. It appears however, that a clinical genetics session is overall adjusted to the individual needs of the service user, as these are perceived by the provider, since both participants in the Bernhardt and colleagues study and participants in the present study emphasised the individual needs of service users and how they try to meet these needs. The fact that data from the interviews in the present study were not thematically analysed, but had only undergone descriptive analysis, limits the ability to draw conclusions about the meaning behind participant responses.

In terms of provider-perceived benefits of a CGS, information-related benefits, decision-making, practical and psychological support, sense of control and empowerment, and preparing families for future eventualities are all outcomes reported in the literature (Bernhardt, Biesecker and Mastromarino, 2000; Payne et al., 2007). Benefits mentioned by genetic counsellors by Bernhardt and colleagues were: provision of information and acquisition of knowledge; listening and validating concerns;
empowerment; supportive counselling with regard to decision-making; and, anticipatory
guidance for medical aspects of the condition and psychosocial aspects i.e. feelings that
might come up in the future (Bernhardt, Biesecker and Mastromarino, 2000). In a
Delphi study where genetic professionals and service users were asked to prioritise
commonly used outcome measures used in CGS evaluation, Payne and colleagues
(2007) reported that decision-making, satisfaction, knowledge of the condition, coping,
Perceived Personal Control (PPC), accuracy of diagnosis, risk perception, meeting of
expectations and quality of life were ranked as the most suitable outcome measures of
CGSs. Knowledge and decision-making received the highest levels of consensus
among providers. This reflects the outcomes and benefits raised by the participants in
this study where knowledge and decision-making were raised by the majority of
participants while meeting perceptions, adjustment and satisfaction were raised by one
or more participants.

Service delivery models also depend on the views of commissioners and the amount of
funding allocated and it remains to be explored how funding and commissioning
influences user outcomes. What can be assumed however is that, what commissioners
understand to be the role of CGS is vital to what is made available to the service users.
This is because the aim of the service is central to the key issues of concern to
commissioners which in turn determine what kind and at what level (i.e. how much)
services will be commissioned (Lenaghan, 1998). Allocated funding determines the
choices made available to service users regarding the way professionals and
specifically genetic counsellors are able to contact them.
For example, based on discussions with a high-ranked member of the CGS, service home visits made by genetic counsellors are a controversial issue and depend on the time available to counsellors. Home visits are dependent on available funding and would be a candidate for removal from service provision if funding were reduced when the visits were not seen as an important aspect of the role of the service. Time given to patients therefore and the number of times each individual or family can be seen by the service before decision-making and after a decision is made, is also dependent on available funding and perceptions of what is appropriate service provision. In addition, things like follow-up which at the moment of data collection were provided to families would also be stopped based on funding allocation and on the perceptions of decision-makers of what is the role of the service. Based on the discussion with a director of the CGS, the chosen service might be perceived more as a counselling service compared to other UK genetics centres because of the difference in how much time is allocated for meetings with families.

5.7 CONCLUSIONS

Patient pathways between the five condition groups were found to differ depending on the nature of the condition and the specific needs resulting from the individual characteristics of these conditions. Genetic counselling and offer of diagnostic genetic testing were shared features of the journey for all condition groups as was the referral to other specialists or services for managing either the symptoms of the condition, offering screening services or for providing social support to patients and their families. Patient follow-up and proactive or reactive care varied between conditions again based on the condition characteristics and on the means available to the service for supporting
families dealing with the impact of the condition. Facilitating understanding of relevant information in order for clients to make informed decisions was the most important outcome measure mentioned by participants, while satisfaction, meeting of expectations, empowerment to make decisions, sense of control and sense of normality or adjustment were also discussed. Reported aims and consequences concur with formal frameworks and guidelines for service provision but very few studies exist on provider perceptions of service provision with which to compare findings.

The following chapter will describe the findings from interviews with users of the service about their perceptions of the (dis)benefits of the CGS.
CHAPTER 6

PHASE TWO: PERCEIVED (DIS)BENEFITS OF ACCESSING THE CGS
6.1. Overview

This chapter will:

1. Provide an overview of the aims and objectives of the second phase of data collection;

2. Describe the methods and recruitment techniques utilised;

3. Provide a descriptive report of data collected from each condition concentrating on the distinguishing characteristics of each group compared with the rest;

4. Discuss the common themes emerging through the analysis of focus groups and semi-structured interviews and present an overall conceptual outcome;

5. Discuss the findings in the context of existing literature.

6.2. Aims and Objectives

During this phase the following objectives were addressed:

- Identify the relevant benefits and disbenefits of the CGS from the service users’ perspective (objective Four)

- Identify which impacts are perceived as important by the service users (objective Five)
6.3. METHODS

6.3.1. Data collection techniques

Focus groups with individuals and families referred to and having made use of one UK clinical genetic service (CGS) were the primary source of data used in order to meet the aim of exploring experiences and identifying relevant benefits and disbenefits of the CGS. Focus groups were deemed appropriate for this phase because of the interaction that takes place between the discussants, a characteristic that distinguishes this technique from that of interviews (Barbour and Kitzinger, 1999; Pope and Mays, 2006). The purpose of the research was not to explore in-depth individual experiences, but to identify benefits that are common among service users.

Focus groups are ideal when exploring shared experiences, opinions, wishes and concerns. The method is particularly useful for allowing participants to generate their own questions during the discussion and pursue their own priorities through the interaction with individuals of the same peer group. The researcher is also able to examine the different perspectives arising among the group as the participants interact within a social network (Barbour and Kitzinger, 1999). The social interaction inherent in focus group sessions often stimulates data that may not arise if the participants are interviewed individually (Freeman, O’Dell and Meola, 2001). The researcher can observe the initial opinions and perspectives of the specific phenomenon and the ways in which these opinions may change during the discussion and whether and how agreement is reached among the group about the particular issue.
Because of these characteristics focus groups would allow for issues like individual (dis)benefits to be raised, discussed and debated between participants within the focus groups leading to a consensus on important (dis)benefits of a CGS.

Even though focus groups were originally developed and used in market research (Barbour and Kitzinger, 1999), they have increasingly come to be used in healthcare (Webb and Kevern, 2001). Their use has been advocated as an appropriate method for conducting health research into sensitive issues and for investigating people’s experiences of illness and their use of health services (Kitzinger, 2006). With focus groups the moderator can pace the discussion accordingly and participants are more likely to share their experiences among people of the same group. On the other hand, it might also be the case that certain individuals might not want to share their experiences with others and this is acknowledged. In health research focus groups have been used in a number of settings, for example in assessing health education messages; public understanding of illness and health behaviours; people’s experiences of conditions and health services (Barbour and Kitzinger, 1999).

In addition to focus groups, face-to-face interviews were also utilised in cases where the use of focus groups was not viable. This was the case where there were small numbers of patients in specific conditions or the small numbers of individuals agreeing to take part would not allow for a focus group to be organised, or where individuals would explicitly express the need to discuss their experiences in private.

It was recognised that the use of both techniques might have implications for the comparability of the data collected through each technique. Interview data tend to be
more in-depth on the experience of one individual while focus group data might cover a broader range of issues being of interest to the group without going in-depth into individual experiences and usually participants are observed to be influenced by the discussion and change their mind throughout the discussion (Reed and Payton, 1997) until a consensus or shared opinion is reached. These methodological issues were considered when arranging the interviews, which all took place after the focus groups thus allowing for initial analysis to take place. Interviews were handled keeping in mind the themes emerging from the focus groups in order to identify the commonalities in the (dis)benefits discussed and also the differences in perspective. Consensuses reached during the focus groups were posed as ideas and questions during the interviews to probe discussion. In this way the two methods can actually be used for triangulation, through testing the ideas emerging from the focus groups during the interviews.

6.3.2. Recruitment and procedures

Purposeful sampling was used to guide participant recruitment in the second phase of data collection (A description of purposeful sampling was given in Chapter 5). Based on the aims of Phase Two, individuals who had been referred to the chosen CGS and were registered in the CGS database were chosen as potential participants for this phase of data collection. This included patients and/or carriers of a genetic condition, parents, carers and other members of families who have utilised the service.

A number of exclusion and inclusion criteria guided recruitment, and for some conditions these were partly determined by the prevalence of the condition and subsequently the numbers of patients from each condition registered with the service.
For example, for VHL there were around 60 individuals registered with the service while the Cancer Genetics Service had several hundred registered service users. Conditions had distinct inclusion criteria whereas a set of common exclusion criteria were set in place. The inclusion criteria for each condition can be seen in Appendix 3 while the common exclusion criteria were the following:

- Terminally ill patients
- Taking part in any concurrent study
- Not being able to communicate in the English language
- Extreme levels of anxiety as judged by the consultants

Recruitment of participants took place through the CGS. Recruitment packages for each condition included information sheets, invitation letters signed by the consultants and a consent form. These were prepared by the researcher and sent by the consultants to potential participants identified by geneticists and/or genetic counsellors from the databases of the CGS based on the agreed upon inclusion and exclusion criteria. Potential participant identification and distribution of recruitment packs were made by the CGS staff because the researcher was not employed by the CGS and for ethical reasons was not allowed access to patient records.

After the recruitment packs were dispatched by the CGS, all interested individuals returned their consent forms directly to the researcher who contacted them in order to arrange for suitable dates. Focus groups were arranged to take place in CGS premises, while interviews were arranged to take place at a venue chosen by the participant. During the majority of the focus groups two moderators were present, one being the researcher and the second being one of the research supervisors who is an
expert in qualitative methods. During focus groups it is recommended for two researchers to be present where one moderates the discussion and the second acts as an observer taking notes on group interaction (Morgan, 1998). The two can also act as co-moderators with some level of agreement being present on the level of input from each moderator (Gibbs, 2007). During the focus groups where both moderators were present the main moderating role was held by Christalla Pithara.

Discussion was structured around the following topics (a copy of the discussion guide is included Appendix 7):

- First contact with the CGS including patient journey and experiences with the service

- Needs and expectations from the CGS

- The resulting benefits and disbenefits following their contact with the CGS

The same topics were covered in both the focus groups and face-to-face interviews, with the focus group guide being adjusted for individual interviews e.g. excluding the introductory and group summarising aspects of the focus group and going straight to the questions of interest. During the focus groups “activity-oriented questions” (Morgan, 1998; Colucci, 2007) were prepared as a way to deal with problems of focus during the discussion. These activities were aimed to re-focus the discussion back to the research questions when the group became unmanageable. These included individual participants writing down on paper their individual experiences of genetic services and the benefits or disadvantages resulting; and writing down how their lives would be
different if they hadn’t accessed genetic services. These were used when and as needed to re-focus discussion.

Table 6.1 shows the number of recruitment packs given to each professional. Packs for DBMD, tuberous sclerosis and cystic fibrosis were sent twice because of low initial response. In the same table the number of consent forms returned for each condition and the number of participants for each focus group are included. For cystic fibrosis and tuberous sclerosis it was not possible to arrange for focus groups because of the very low response so interviews were scheduled instead. Table 6.2 illustrates the information on the genetic status and family history of participants.

Focus groups lasted approximately two hours while interviews lasted between 45 minutes and 1.5 hours. Focus groups and interviews were conducted between January 2007 and November 2008. All focus groups and interviews were tape-recorded using an Olympus Digital voice recorder. All focus groups and interviews were transcribed in verbatim and transcripts were imported in QSR NVivo 8.0 to be analysed.
## Table 6.1: Numbers of disseminated recruitment packs

<table>
<thead>
<tr>
<th>Focus Group</th>
<th>No of packs</th>
<th>Replies</th>
<th>Confirmed attending</th>
<th>Participants</th>
</tr>
</thead>
<tbody>
<tr>
<td>VHL</td>
<td>25</td>
<td>4</td>
<td>3</td>
<td>5</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>3 males (patients)</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>2 spouses</td>
</tr>
<tr>
<td>DBMD</td>
<td>2x50</td>
<td>7</td>
<td>3</td>
<td>3 (mothers of patients)</td>
</tr>
<tr>
<td>HBOC*</td>
<td>100</td>
<td>11</td>
<td>Group1 - 7 members</td>
<td>3 (all female)</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Group2 – 5 members</td>
<td>5</td>
</tr>
<tr>
<td>TS</td>
<td>2x40</td>
<td>2</td>
<td>2 couples</td>
<td>2 interviews took place</td>
</tr>
<tr>
<td>CF</td>
<td>2x40</td>
<td>2</td>
<td>2 (1 was a couple)</td>
<td>1 interview took place</td>
</tr>
</tbody>
</table>

* Some individuals who were expected to attend the first focus group did not attend and then confirmed attending the second one.
### Table 6.2: Information on genetic status and family history of participants

<table>
<thead>
<tr>
<th>Focus group</th>
<th>Participant Number</th>
<th>Gender</th>
<th>Genetic status</th>
<th>Family history</th>
</tr>
</thead>
<tbody>
<tr>
<td>VHL</td>
<td>1</td>
<td>Female</td>
<td>Wife of P2</td>
<td>No known history</td>
</tr>
<tr>
<td></td>
<td>2</td>
<td>Male</td>
<td>Carrier</td>
<td>Possibly father was carrier</td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>Male</td>
<td>Carrier</td>
<td>Family history</td>
</tr>
<tr>
<td></td>
<td>4</td>
<td>Female</td>
<td>Wife of P3</td>
<td></td>
</tr>
<tr>
<td></td>
<td>5</td>
<td>Male</td>
<td>Carrier</td>
<td></td>
</tr>
<tr>
<td>DBMD</td>
<td>1</td>
<td>Female</td>
<td>BMD carrier</td>
<td>2 sons, one with BMD</td>
</tr>
<tr>
<td></td>
<td>2</td>
<td>Female</td>
<td>BMD carrier</td>
<td>2 daughters, 1 is carrier</td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>Female</td>
<td>DMD carrier</td>
<td>1 son with DMD; 1 daughter who is carrier</td>
</tr>
<tr>
<td>HBOC FG1</td>
<td>1</td>
<td>Female</td>
<td>Not tested</td>
<td>Gene identified in family – chosen to have prophylactic mastectomy</td>
</tr>
<tr>
<td></td>
<td>2</td>
<td>Female</td>
<td>Not tested</td>
<td>Mother in the process of being tested</td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>Female</td>
<td>Not tested</td>
<td>Mother has been tested - result not known</td>
</tr>
<tr>
<td>HBOC FG2</td>
<td>1</td>
<td>Female</td>
<td>Not tested</td>
<td>Sister in the process of being tested</td>
</tr>
<tr>
<td></td>
<td>2</td>
<td>Female</td>
<td>Tested non-carrier* (population risk)</td>
<td>Extended family history</td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>Female</td>
<td>Not tested</td>
<td>Extended family history but cannot be tested</td>
</tr>
<tr>
<td></td>
<td>4</td>
<td>Female</td>
<td>Carrier</td>
<td>Extended family history</td>
</tr>
<tr>
<td></td>
<td>5</td>
<td>Female</td>
<td>Not tested</td>
<td>Cannot be tested</td>
</tr>
<tr>
<td>TS</td>
<td>Interview 1</td>
<td>Female</td>
<td>Not a carrier</td>
<td>2 sons: one severely affected with TS</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Male</td>
<td>Not a carrier</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Interview 2</td>
<td>Female</td>
<td>Not a carrier</td>
<td>2 daughters: 1 affected by TS but not severe</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Male</td>
<td>Not a carrier</td>
<td></td>
</tr>
<tr>
<td>CF</td>
<td>Interview 1</td>
<td>Female</td>
<td>Not a carrier</td>
<td>Suspected CF-affected foetus (false positive)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Male</td>
<td>Not a carrier</td>
<td></td>
</tr>
</tbody>
</table>

* Even though being at high-risk was the main inclusion criterion for this group, a recruitment pack was sent to this participant who was referred to the service because of her high risk. Despite this, no major differences between her and the other participants’ perspectives emerged during the focus group.
6.4. ANALYSIS

Transcripts were analysed using thematic analysis, defined as “a method for identifying, analysing and reporting patterns or themes within data” (Braun and Clarke, 2006: p.79), and the constant comparative approach. Thematic analysis is a shared approach to analysis among qualitative researchers and can be used both within an overall theoretical or epistemological framework which may guide data collection, such as grounded theory, and independently of such a framework (Braun and Clarke, 2006). The use of thematic analysis was believed to be the most relevant technique for the pragmatic objectives and methods associated with this research, as well as the aims of healthcare evaluation in general. Focus groups, like thematic analysis, are not bounded to any theoretical or epistemological approach, allowing for the analysis of data collected to follow the theoretical framework adopted by the researcher (Wilkinson, 2008). The present research was guided by the pragmatic paradigm as well as a welfarist approach to economic evaluation where service consumers are the only experts appropriate for evaluating their own utility (Morris, Devlin and Parkin, 2007).

The constant comparative method was used as a framework for the process of analysing categories and themes, where each new transcript from subsequent focus groups or interviews was coded using the existing codes and each data item was checked and compared against the existing codes in order to refine their fit to underlying concepts (Green, 1998) and establish analytical categories (Pope, Ziebland and Mays, 2006). The constant comparative method is a technique originally described in grounded theory (Pope, Ziebland and Mays, 2006), where data is constantly
compared with data in order to identify similarities and differences thus prompting the researcher to think analytically about the data coded (Charmaz, 2008). This technique was used as a tool to assist in establishing themes and categories, without however being bounded by any theoretical connotations associated with grounded theory. Braun and Clarke (Braun and Clarke, 2006) suggest a process alluding to the constant comparative method where, in order to review the preliminary themes identified, the researcher goes through all the coded extracts for each theme to decide whether they form a coherent pattern and then repeat the same process for individual themes in relation to the entire data set. In the context of this research the technique was used to give a systematic approach to the steps taken in analysing the data because of the breadth that tends to be associated with the term “thematic analysis”.

6.5. RESULTS

6.5.1. Overview of findings

During the group and individual discussions, several (dis)benefits were raised by participants some of which related to their overall experience with the genetic condition and the diagnosis, of which the CGS was a part, and some were directly linked to the CGS experience. These constituted the initial categories upon which further in-depth analysis was based. These categories were used in the third and final research phase, in which they were presented to stakeholders of the service in a methodological exercise testing the use of ARSs for exploring preferences and also testing the credibility of these categories. Table 6.3 in page 248 lists all the initial categories of
(dis)benefits raised by participants during the discussions and the final themes as these were formed during analysis.

The benefits and disbenefits identified that were linked to both CGS experiences and to the genetic condition were:

- Insurance problems
- Changes in family relationships
- Certainty about genetic risk status
- Support in dealing with social effects of genetic condition
- Inform families of possible insurance problems
- Support in communicating genetic risk to other family members
- Continuity of care
- Research updates
- Clinical (health) outcomes
- Access to other NHS services
- Clinical support
- Decision-making support
- Being offered choices
- Levels of knowledge and education
- Access to genetics experts
- Reassurance and relief from anxiety
- Empowerment in making decisions
- Feelings of personal control over the situation
- Adjustment to the genetic condition
Overall, important themes emerging from the analysis appeared to be common among conditions, even if some differences existed when categories were either more prevalent within discussions for certain disease groups, or were isolated to some groups but still did not emerge as prominent as the common themes. It is noted that clinical or health outcomes were discussed by some participants and were especially relevant to participants in the VHL group and to a lesser degree to a few participants in other focus groups. Further analysis of the transcripts focused on the intangible (dis)benefits which are the focus of this thesis while further consideration of the importance attached to health outcomes is given in the next and final phase of data collection.

Benefits and disbenefits discussed by participants and listed above were re-grouped under three categories representing (1) process attributes, (2) psychosocial outcomes of the CGS, and (3) impacts relating to the genetic condition rather than the use of the CGS e.g. insurance problems and changes in family relationships.

Analysis suggested that impacts relating to the genetic condition were altered in their direction by the CGS, for example genetic counselling could help alleviate certain of the negative consequences on family relationships and assist families prepare and deal with insurance problems. Process attributes related to characteristics of the process of service delivery and were readily discussed when participants were asked to think of the benefits resulting from utilising the CGS. These attributes were:
- **Expertise**
  This attribute refers to the accurate, up to date, and knowledgeable information given to service users, resulting from the expertise of genetic service providers in medical genetics.

- **Individualised approach to service provision**
  This attribute refers to the individualised approach with which service users were treated by the CGS professionals, taking into consideration individual needs and adjusting service provision. Participants were treated as an individual rather than as impersonal service users.

- **Interdisciplinary problem-based care**
  This attribute refers the capacity of the CGS to work as part of a multidisciplinary environment, make links with, and refer families to other health and non-health e.g. social, educational, psychological, services based on the individual issues faced by service users in order to support them to deal with the implications of the condition.

- **Accessibility and continuity over time**
  This refers to the open-access policy of the CGS where individuals and families were either given long-term follow-up or were able to re-access the service themselves in the short- and long-term when there was a need both for them and for other family members.
- **Family centeredness**

  This attribute refers to the family-oriented care provided to service users. This means that the service was available to all family members who had concerns related to the genetic condition, allowed for other family members to access the service at the same time as the individual who was initially referred, and enabled access to non-genetics services to other family members.

Psychosocial outcomes emerged following further analysis of the transcripts and the meaning behind the participants’ narratives. These outcomes were:

- **Knowledge and understanding**

  This refers to obtaining accurate and up-to-date information appropriate for the particular family, which was understood and consolidated with previously held knowledge of the genetic condition. This incorporates information on the genetic condition and inheritance patterns, prognosis and symptoms of the condition, available options to deal with tangible and intangible consequences of the condition.

- **Informed and shared decision-making**

  This refers to feeling able to make decisions based on accurate and up-to-date information, and these decisions are in agreement with the individual beliefs and goals of that family without feeling forced to take decisions related to the goals of the service provider.
- **Enablement**

  This refers to feeling enabled to actively cope with the tangible consequences of the condition including symptoms, onset of symptoms and social implications of condition through the facilitation of the CGS to access and use needed health and social services. This includes the establishment of care pathways, information about and access to new healthcare treatments, advice and practical assistance in using the social and education services.

- **Reassurance**

  This refers to the experience of positive emotional outcomes because of the presence of the CGS in the lives of families. Peace of mind because of knowing the service was accessible to self and family when needed, and that the service was there in both the clinical and research dimensions focusing on the wellbeing of patients and their families.

These psychosocial outcomes formed dimensions of a conceptual theme, which was labelled Perceived Familial Control. The broad term “familial” was chosen because it was relevant to members of the immediate and extended family both within and across generations i.e. intra- we well as inter-generational.

Before the proposed themes are explored, an overview of the background to the discussions and of the discussion with each condition is given, stressing the distinguishing characteristics of each group.
6.5.2. Overview of narratives based on condition

6.5.2.1. Von Hippel Lindau syndrome

The VHL focus group was the first to be arranged even if VHL constitutes one of the smallest patient groups, with around 60 people on the database at the time the focus groups took place. The impression of the researcher was that this group of patients appeared to have a tighter bond with the service compared to the other groups, firstly because of the level of on-going contact between families and the CGS, and secondly because of the tangible health benefits observed due to the condition monitoring programme set up and overseen by the service. Participants were very vocal about their satisfaction with the service and held in high esteem the geneticists and genetic counsellors involved in service delivery. In this focus group all individuals reported participating in the focus group because they wanted to “give something back” to the genetic service. In a way, this group was an exemplar of how the service benefited the service users both in terms of health and psychosocial outcomes. The undeniable improvement in health outcomes was the most distinctive difference between this group and the rest, since none of the other groups enjoyed such a direct improvement as a result of the presence of the CGS.

This group depended extensively on the genetic service for monitoring their symptoms through yearly planned check-ups and ad hoc investigations. For this group, seemingly harmless symptomatology could indicate the presence of cysts or tumours which could be life-threatening and which were very difficult to identify by health professionals not familiar with VHL. With the CGS, patients felt that they had a point of contact where
they could investigate concerns involving any newly arising symptoms and also believed that the expertise of the service could inform them of new medical developments which could be relevant to them.

Issues of recruitment and possible bias are discussed in Chapter 8, but it is briefly pointed out that since suitable participants were chosen by the geneticists based on the inclusion and exclusion criteria, and because of the very small patient group, it is possible that these participants were the ones perceived by the geneticist to be the most “appropriate” to discuss the service biasing in a way the results. At the same time, again because of the very small numbers of affected families it might be possible for family experiences to be more similar than for other conditions where a larger number of families are attached to the service.

6.5.2.2. Hereditary Breast and Ovarian Cancer

This condition had the largest database with the largest number of recruitment packs being sent to this group (100 recruitment packs) and two focus groups were organised. Participants were women who had extended family histories of breast and ovarian cancer. The majority of women had not undergone genetic testing but all were assessed as being at high risk, while one woman had chosen to undergo prophylactic mastectomy as soon as the HBOC gene had been identified in her family. Women reported to access the service with preoccupations and concerns for themselves as well as other female family members, or described their mothers accessing the service having the same concerns. The value placed on the familial element of the service was apparent, and women expressed their satisfaction in being allowed to attend clinics with
other female members of their families, or being part of the active involvement of service providers in cascade screening of other family members.

The majority of women expressed a desire to undergo genetic testing if it was possible, because of their belief that it’s better to know and were very motivated in dealing with their risk. The need for empowerment to take control was especially strong in this group, arguably related to the uncertainty associated with the condition and the need to access risk management interventions. Women had expectations of risk management options, especially access to breast screening, when accessing the Cancer Genetics Service.

Because of the distinct characteristics of this genetic condition, uncertainty about whether or not one is at high risk or will develop the disease, and familial implications such as the willingness of affected members of the family to get tested and thus allow access to genetic testing for other members, were inherent in this situation. In this group there was a high value placed on the individualised counselling approach of the CGS where women felt taken seriously and provided with all the relevant information about their risk and options of how to deal with that risk.

6.5.2.3. **Duchenne and Becker Muscular Dystrophies**

Two of the women participating in this focus group had BMD in their families with one woman caring for a teenage son who was affected and the other having two daughters one of whom was a gene carrier. The third woman had DMD running through her family and her affected son had recently died from the condition while her young adult daughter was found to be a carrier. This group appeared to be most isolated from the
CGS and all women described in some way that attending the focus group was an attempt from their part to regain access to the genetics service. Two of the women explicitly acted as representatives for their carrier daughters. In the two cases where women were or had been carers of affected sons, their main contact was with secondary specialists, physiotherapists/occupational therapists, NGOs and voluntary organisations providing services to and supporting families with DBMD but these women described how the genetic aspect was lacking from their care. All three women were in search of genetic counselling either for new information available on the condition and its treatment, or for reproductive counselling for their daughters.

In a sense this group was on the other end of the continuum from VHL since contact with the service was infrequent and unstructured, and some of their needs, namely the psychosocial ones, appeared to be lagging behind. This group was also seen as a “negative case” since the benefits discussed by other groups were not shared by this one. Instead of disproving the categories identified so far however, this group verified what other groups had already proposed as benefits of the CGS. This group of participants expressed their satisfaction with the service which they felt met their needs at the time of the referral i.e. genetic testing and diagnosis for themselves and their children, but they felt that their long-term needs were not being met. Accessibility over time was what these women were in need of and placed value on in order to obtain the knowledge and information they required which would help them and their children to feel in control of the condition. These narratives confirmed the importance of such process characteristics.
6.5.2.4. Tuberous Sclerosis

Two couples responded to the recruitment invitations. One couple were the parents of a daughter with TS who had been diagnosed in childhood due to the presence of seizures and behavioural problems. They also had a younger daughter who was not a carrier. Their affected daughter was described to lead a normal life and at the time of the interview she was studying at a UK university. She was able to take care of herself and lived in student accommodation, but experienced infrequent seizure attacks and was taking medication to control her symptoms. These parents described themselves to be actively involved in their daughter's life by being involved with the CGS and have supported her throughout her schooling and education.

The second couple were the parents of a son with TS who was diagnosed in infancy and was severely affected. Their son required 24 hour care but was described to have a good quality of life. This couple was also actively involved in CGS research and the Tuberous Sclerosis Association and described their involvement as benefiting their son. They also had another son who was not a carrier.

The experiences shared by these individuals represented the views of individuals who actively sought sources of information and support, a prime example of such source being the CGS.

6.5.2.5. Cystic Fibrosis

Only one couple responded to the two invitations for participation. This couple had been referred for genetic counselling following a false positive screening result during
routine prenatal screening tests. The couple saw the invitation to participate as an opportunity to share the negative experiences they had with the obstetrics and gynaecology department and reported no knowledge of the CGS even though they had received genetic counselling from this service. Following explanations by the researcher of what the CGS is and what services they would have received from the department the couple acknowledged their contact and went on to discuss and compare their experiences between these two departments. Based on these comparisons, the individualised counselling approach and informed and shared decision-making emerged as particularly valued by these participants.

6.5.2.6. Summary

Comparisons between the (dis)benefits raised by each group were made in order to explore for any differences. Analysis indicated that there was homogeneity in the discussed benefits and disbenefits between the groups. This finding is of importance to the evaluation of CGSs since it indicates that (dis)benefits emerging through the analysis can be included into the economic evaluation of genetic services provided for most if not all conditions. Where there appeared to be a difference, that was on the value attached to each (dis)benefit depending on the condition group. This is because the needs of each group may vary, making one (dis)benefit more important than another. The identified (dis)benefits are discussed in the following section.
6.5.3. Benefits and disbenefits of the Clinical Genetic Service

6.5.3.1. Process Attributes

Focus groups and interviews with service users of the Clinical Genetic Service (CGS) explored the (dis)benefits experienced by families following their referral to the centre. The following process attributes were identified:

1. The expertise of the centre in medical genetics and genetic conditions;
2. An individualised approach to service provision;
3. Interdisciplinary problem-based care;
4. The accessibility of the service to families over time; and finally
5. The family-centeredness of the service.

6.5.3.1.1. Expertise

Families referred to the CGS were looking for accurate and up to date information which would provide them with some level of certainty. Participants sometimes felt that this information was not provided by health professionals outside the genetic service who they felt were often unaware of the genetics or the clinical characteristics of the condition. The CGS was credited with high levels of expertise in genetics and genetic conditions by all participants. Participants expressed the value of this expertise often through comparisons between the level of information obtained outside the CGS and the level of information obtained from genetic professionals. In some cases the CGS was the only place where families could obtain such information.
Mother:
I think probably the genetic counselling were the only sessions we were going along to at the time where we thought right we are with somebody who is going to give us

Father:
Give us information

Parents of girl with TS

The expertise was also perceived by some participants to have health implications since the ability of the genetic professionals to recognise and distinguish between symptoms which were associated with the condition from those which were not, meant that patients could be advised and directed to the appropriate intervention.

if I saw (the genetics) professor and said I’ve got a headache, he’d deal with it seriously, if I saw my GP and said I’ve got a headache he’d give me some anadin, or something like that. Even though he knows I’ve got VHL.

Participant 3, VHL focus group

6.5.3.1.2.   An individualised approach to service provision

Throughout their contact with the CGS participants acknowledged the ability of professionals to make them feel at ease and that their opinions and needs are taken into consideration. This difference in attitudes between the CGS and the rest of the NHS was something that the majority of participants raised during the discussions. Participants described how they were “treated as an individual” by the geneticists and genetic counsellors during their sessions where “no question is ever, ever stupid”.

Women in the HBOC focus groups where particularly vocal about this aspect of the service.

I think the whole service really put you at ease and they make it quite, I was quite comfortable with it all, because they don’t put any pressure on you, they just leave the decision up to you really, and they talk through any questions that I had, and I took my friend with me as well, and they just left the decision up to me, and then I decided..

Participant 2, HBOC focus group 2

Putting no pressure on counselees but allowing them the time they needed to think through the information they were given was acknowledged to be a significant aspect of the individualised counselling approach.

...you never ever felt that, you know, right, you got three minutes starting now! You know, however long that appointment was, [...], and (the geneticist) would have come out and apologised on a couple of occasions [...]. But that gave you a degree of confidence that he wasn’t saying well I’m gonna give each person 20 minutes exactly and if they are not out of my office I’m going to blow their brains out (laughter) you know. He gave everybody as long as they needed, and again, that’s the individual, not a one size fits all but it’s the individual tailoring...

Participant 1, HBOC focus group 1

Allowing people time to come to terms with the information given and providing information in a way they can understand was a shared need among participants. Most participants described the ability of genetics professionals to provide only the
information needed at the time by assessing the psychological state of the family and how much information they were able to take, allowing for multiple sessions with different levels of information being given during sessions.

I think when people are given a diagnosis they do need time to come to terms or get used to that diagnosis and I think it’s important for the support services that are in place be responsive to each individual, and it’s quite a task on those counsellors sort of identifying what people need and when they need it or how much information they need and how it’s delivered

_Mother of boy with TS_

The CGS was flexible in the way it delivered services in order to provide them in a way that met the individual needs of patients and families, addressing problems as they arise. The hub and spoke model of the service allowed for families to be seen at their area without inflicting unnecessary travelling on the families. Family needs and circumstances were also taken into consideration when organising clinical interventions.

...because my wife has [neurological condition] when I went in to have my adrenal glands taken out I think normally they [the healthcare providers], for example feed you blood pressure lowering tablets and they send you home [...] but because (of our circumstances) they said let’s not prolong it, stay in hospital, ...

_Participant 3, VHL focus group_


Chapter 6

6.5.3.1.3. Interdisciplinary problem-based care

In addition to the gaps present in knowledge, a further difficulty faced by families in their efforts to cope with the medical, emotional and social problems related to the condition. Difficulties in managing their contacts with the social as well as health care systems were quite common among participants.

We see a professor of neurology […] and unfortunately […] he has far too many people to see, and it’s not his fault but we end up having to fight for an appointment, […] it’s not to do with the genetics or anything but I’m having to fight the system

*Mother of girl with TS*

Genetic professionals were viewed as being at the centre of a group of services i.e. health services, social services and other voluntary and non-governmental organisations, establishing patient pathways, informing the families of different services relevant to their problems, enabling access and putting them in contact with support services.

*(the genetic providers are) in the best position to give us all the relevant information and tell us all about he’s an expert, so he put us in touch with the TS Association, which is again very valuable and we’ve become members of that, so we get their quarterly magazines, so we keep up to date with things and that’s quite handy,*

*Mother of girl with TS*

Interventions made by the CGS were valued by all participants who were anxious to deal with the clinical and social implications of the condition. Women with a high risk of HBOC viewed the links to oncologists for preventative cancer procedures and the
screening programs open to them because of their access to genetics as an important part of their struggle against developing the condition.

You’ve got choices then if you want to… like decide to have your breasts off removed as well, to make sure or not if you don’t want to then it’s up to you, you can check, be extra careful

Participant 2, HBOC focus group 1

6.5.3.1.4. Accessibility and continuity over time

Because genetic conditions have continuity in both within-generational and inter-generational aspects, the needs of families are on-going and do not end at the genetic counselling session of one family member. Ease of access for both current and future or potential service users was a benefit discussed by the majority of participants.

But I’ve had good support from them, but the thing that bothers me now is, well not bothers me, is as you said [daughter] is going to need support, you know she’s 18 now, she’s got her first you know, real boyfriend, not that that’s a, but you start thinking […] it’s eh, making sure she’s got support when the need arises, and knowing where to go

Participant 3, DBMD focus group

Participants raised their evolving needs following the initial counselling session, where unanswered questions or emerging new ones called for re-accessing the service.

The fact that you had (the geneticist’s) phone number, you know, he could throw this information at you, […] but when you went back home it was then that the brain cells started to (yes, that’s right) take over, […] and from my point of
view the fact that I could go back and phone, ok he might not have been there
there and then, when I phoned, but [...] he’d come back

Participant 1, HBOC focus group 1

Accessibility over the long term was essential particularly in the case of family members
who were concerned about the implications of the genetic condition on them or their
children, as well as for individuals in whom new needs had arisen.

I just felt, a diagnosis you get so carried along with getting involved with all the
services, that stopping and giving yourself time to get used to all these, you
need, you know I just felt sort of, 6 months into it, was when I needed a bit of
support...

Participant 3, DBMD focus group

Some participants reported how the CGS was the only constant source of support and
information through their or their children’s lives.

(Our son) has over the years seen around a hundred professionals from
medical, social services and health teams, but we felt that the main stay over
the years has been the genetic service, [...] when health professionals come
out and say who do you see, I always say we’ve got the link with (the CGS),
because we felt it’s been important for us and for them to recognise that as well
and quite a few have said oh you see genetic services it’s a one off, and we say
no not in our case it’s not a one off there is continued support there

Mother of boy with TS
Facilitating accessibility and continuity in the contacts between the CGS and service users meant that in many cases families had contact with the same geneticist or genetic counsellor throughout their genetic service experience. This made them feel more connected with the service and more confident about accessing the professional when they had the need.

Participant 2:
I will say it’s been very nice to have the same doctor from… (yes) because I had expected 13 years down the line that (talking over)

Participant 3:
That’s one thing, yes, [the geneticist] from when [my son] was diagnosed till now

Participant 2:
And she remembered me (talking over)

Participant 1:
She does remember you

Participant 3:
She’s the one constant name all the way along

Participant 2:
Yeah, she remembers aspects of our situation and that’s really important

6.5.3.1.5. Family-centeredness

Because of the hereditary nature of genetic conditions, all diagnoses have a subsequent impact on other family members. The CGS was not only open to current service users, but also to all family members concerned about the implications. Several
participants, especially women at risk for HBOC discussed how they initiated access to the CGS in order to open the gates for their daughters or other family members.

it was on my mother’s side, the breast cancer and the ovarian cancer, and the history was quite horrendous really, and it was just myself and one cousin left, and I think it was only because I had three boys and then I had a granddaughter that I decided to find out, because we thought that by reaching the age of 50, myself and my cousin, that we were ok, that we missed it, but then she developed it, she was age 54, and in the meantime then my granddaughter was aged 2, so for her sake I think more than anything, that’s why I decided to go ahead with the genetics […]

Participant 2, HBOC Focus group 2

This was facilitated by the outlook of the service as a “family service” which allowed for other members of the family to gain access through the referral of one individual.

the reason I went really was because I had a daughter, and my sister as well has a daughter, and they were marvellous, because when I came down to see that chap here, I brought my sister with me

Participant 3, HBOC focus group 2

The service did not only provide counselling services to families but also facilitated access to care for all family members. Mothers of sons with DBMD and their carrier daughters were also part of screening programmes arranged by the CGS because of their high risk for heart-related conditions. In other genetic conditions, children in families carrying a genetic condition were also followed up along with their parents or other siblings in order to screen them for potential symptoms of the condition.
[The genetics service has] looked after my family, we have two sons as well. They've been scanned annually and looked after as well, and it's been an excellent service to me.

Participant 2, VHL focus group

6.5.3.2. Psychosocial outcomes

Iterative comparisons between the emerging categories led to the identification of a number of psychosocial consequences linked to the process attributes discussed and also others linked to the genetic diagnosis. This thesis deals with the (dis)benefits of CGS and thus reporting of findings focuses on the outcomes of the service. It should be pointed out however, that through reading and re-reading the transcripts it became apparent that the psychosocial consequences directly linked to the genetic diagnosis, e.g. changes in family relationships, were influenced by involvement of the CGS. The proposed psychosocial outcomes are:

1) Knowledge and understanding;
2) Informed and shared decision-making;
3) Reassurance; and
4) Enablement

6.5.3.2.1. Knowledge and understanding

All participants accessed the CGS with the expectation of obtaining more information about the genetic condition and its implications on the life of patients and their families. During the initial stages of a diagnosis families could feel very isolated from the time they were given the diagnosis up to their genetic counselling session, and the
fragmented or inaccurate way they were being given information aggravated their feelings of frustration.

**Mother:**

Because we had not heard anything about the condition when we were given the diagnosis... we didn’t realise the consequences of the disorder, but what we were aware of immediately was the fact that they said that it was genetic and I can remember feeling not only concern about [son] but the knock on effects on all of the family as well [...] 

**Father:**

...there was then in terms of information, a guy did come from the genetics department took a family history and went, and it was like, we’ve had this diagnosis, we are giving this information and there was nothing back, and it was many weeks then when we just felt isolated you know and no one really talking to us and no one giving us any information other than what you could get from the library which is all doom and gloom, and it was quite a sad time really, and it doesn’t have to be, it’s changed a lot since then hasn’t it

*Parents of boy with TS*

A distinction emerged between simply imparting information and knowledge and understanding. Information which was not accurate, appropriate or which was not given in a way that reflected the needs and requirements of that individual or family could result in information overload and increased anxiety.

To be honest it was too much in one go, and my younger daughter [...] she was frightened because she had to have blood taken and all that and then she just started to cry because the information, it was too much for a young person
Participant 2, DBMD focus group

Participants valued their knowledge and understanding of the risks and implications of the genetic condition and it was “best to know rather than not know”. Knowledge in itself was valued for its own right and for the empowerment it brought to counselees.

I would rather know everything really, and sort of make decisions based on that, and yeah, I felt as though I was in control. I’d rather know exactly what chances, what the chances are.

Participant 3, HBOC focus group 1

Information needed to be accurate, up-to-date and based on the needs and requirements of that family. Within the context of the CGS expertise and the individualised approach of providers enabled that understanding.

6.5.3.2.2. Informed and shared decision-making

The most obvious implication of knowledge and understanding was the ability of participants to make informed decisions. Being knowledgeable of the inheritance patterns of the condition, the prognosis of the condition and the various options open to them for dealing with the condition meant that participants had the tools to make decisions appropriate to them.

Because we are proactive in that way, and we need information, I think it’s helped us regain a little bit of control, and because we are aware of the research going on we feel that if ever we need to use the information that we have to (our son’s) benefit then that’s the best way we can help him, [...] we take one day at a time but in regaining some control over that as well the more information that we have, that
we are making informed decisions whenever we have to make important decisions about medication or any programs we want to try with (son) or the way we deal with difficulties that arise

_Mother of boy with TS_

The individualised counselling approach meant that individuals were not pressured into making decisions which the professionals considered appropriate. On the contrary their decisions appeared to be informed, because of the information presented to them, and also shared, because they were made in partnership with the genetic professionals input but based on their own perceptions and attitudes. In this sense participants did not feel that their treatment or relationship with the CGS was threatened by the decisions they made.

…we’ve got a 24 year old son, and a 21 year old son, [...] (The geneticist) did come back last year and said, look, we think we probably could do (a genetic test for the boys) but they didn’t really want to know, and we discussed it with them, and we thought well nothing is going to change if they know so it’s better if they don’t know, so if something happens, she said you have your scans every year, you are checked every year, you have your urine tests, she said you are men now, it’s totally up to you, but she said I leave it up to you.

_Participant 1, VHL focus group_

Some participants discussed their appreciation of the informed and shared decision-making facilitated by the CGS and made comparisons with other NHS specialists. The pressure placed on families by healthcare providers coercing them into making decisions was expressed very vividly in the case of the couple referred to the service
because of a false-positive CF screening result during prenatal screening. In the following excerpt the couple are discussing their experience with the obstetrics and gynaecology department.

**Husband**

we had a terrible time, we didn’t like the way the hospital treated us or anything because they kept telling us to abort the baby, we didn’t in the end we’d gone for nearly 40 weeks then we’d gone (wife: stage 2) she (the obstetrician) advised us to abort then basically [...] we found her bed side manners very bad, and because we were very emotional at the time we thought you know, you’ve got to treat people a bit kinder, because we don’t know what’s going on at all, we only know what the doctor tells us

**Wife**

It was very quick wasn’t it, [...] and we were thinking all sorts, you know, [...] and I was going, well can you tell me what this means and she said “can you stop there, can you just not interrupt me, because I lose my train of thought”, well you can imagine I was, “what??”, and then when they went on about abortion I just filled up then

**Couple referred because of abnormal CF prenatal screening results**

Here the couple is comparing their experience with the O&G department with the one from the genetic counselling session.

**Wife**

The lady down [at the genetics service] we found

**Husband**
The hospital down here we found far better than the Heath in Cardiff, we found far more personal, far more approachable and basically better bed-side manners if you want to call it that but be it the doctor that the people we dealt with in Cardiff we found just treat us like a slab of meat like… didn’t treat it us really did they, it was just, they weren’t, there were no people skills

Couple referred because of prenatal screening abnormal CF results

6.5.3.2.3. Reassurance

A number of participants described the CGS to give them “peace of mind”. This was the result of receiving explanations about their risks, the genetics of the condition and the subsequent implications, and also being given options of how they can deal with this risk.

Participant 1:
I think with us we just don’t, we don’t live with it day to day, we forget about it

Participant 2:
That’s very much so

Participant 1:
We really do forget about it, I think we are not the type of people that think, when [husband] got a headache, ok, slightly you think oh Gosh

Participant 2:
That’s too many glasses of wine

Participant 1:
As the consultant says everybody gets them, you haven’t got to live your life with worrying about, and really we don’t, we do tend to put it to one side and resurrect it every time we go to the hospital

Participant 2:
Because we know about it we don’t worry

VHL Focus Group

It was also linked to the knowledge that “somebody was there for you”.

It's been great support for us, a lot of support over the years, sometimes it can be a little bit emotional, it gets a little bit frightening, but at least there’s somebody there for you. For us a great help.

Participant 1, VHL focus group

This was particularly important for women living with a high risk for HBOC where there was increased uncertainty of whether these women would develop cancer.

...there’s peace of mind I think they give you as well, if you can get your mammograms done and... which you wouldn't have had, every 18 months I get them they would have been every 3 years, well 3 year can be a lot

Participant 2, HBOC focus group 2

Reassurance was relevant to all the attributes of the genetic service, from the knowledge gained, to the knowledge that the professionals in charge of their condition were experts aware of any new interventions, and to the easy access to these professionals.

Interviewer

What is the service adding to your life?

Wife

Well, we would not have had the support we had from them probably, and the knowledge... genetics wise we needed to know for the future as I said about,
that’s a big thing for [daughter] (husband: yes) knowing that she might have a child with, that’s a huge eh…. Sort of… problem looming possibly

Husband

Yes, but knowing that the service is there and understanding what may happen, you know, how do we tackle this

Parents of girl with TS

6.5.3.2.4. Enablement

Access to the genetic service for some participants was associated with efforts of actively dealing with the implications of the condition and the uncertainty involved in their perceptions of risk.

Participant 3:

You know, you get this devastating emotional news and you’ve got to take, but at the time… […]

Participant 3:

…but at the time you are so busy with everything else…

Participant 1:

I think your head is reeling with all the things…

Participant 3:

…you get them social workers and geneticists, and physiotherapists and occupational therapists you get carried along in the (P1: yeah…) … and you’ve got no time, and then probably two, three years later, you’re thinking well why am I feeling like this, it should have been, but it’s only then when things have calmed down and everything is in place, that it all (not clear)

Participant 1:
Chapter 6

I didn’t find out until [son] was actually 3 that there was a children’s centre for instance, or anything, and they said well why haven’t you been here before and I said well I didn’t know there was all these I had no idea…

*DBMD focus group*

Following access, the genetic service was found to provide practical support to families through acting as mainly the middleman or link between the family and a number of other health, social or voluntary/NGO organizations or by simply providing advice on how to deal with the hurdles involved. In many cases the CGS was found to intervene between the patient and other medical disciplines in order to establish patient pathways, make necessary referrals to specialists, and arrange for emergency treatments to patients who required them.

in the past [the geneticist] has helped me get appointments at the epilepsy clinic because they are very overstretched and very busy […] and in desperation I did ring him one day and said what can we do about this […] so he managed to sort of get us seen

*Mother of daughter with TS*

The practical involvement of the CGS promoted feelings of enablement, an important dimension in perceptions of being able to cope with the condition. Enablement emerged through the perceptions of families that they have information about the different services existing in the health, social, educational or voluntary/NGO sectors and that they have or can obtain the links which enable them to navigate and use these systems effectively.
6.5.3.3. Overall conceptual outcome of CGSs

6.5.3.3.1. Perceived Familial Control

Perceived Familial Control is proposed as the overall conceptual outcome of CGSs. Perceived Familial Control is related to the family unit rather than the individual, making a direct link to the unit of currency of the service which is the family. It is also an outcome which relates to familial beliefs of past, present and future ability of coping with the condition. This characteristic reflects the substantial concerns expressed by participants about future generations and the hereditary nature of genetic conditions. A CGS therefore is proposed to result in Perceived Familial Control over a situation that is highly uncertain and in many cases with limited options available to families.

This conceptual outcome is presented to be the result of a process consisting of the five process attributes which contribute towards the outcomes of knowledge and understanding, informed & shared decision making, reassurance and empowerment which are key dimensions of Perceived Familial Control.

The following figure illustrates the relationships between the attributes and outcomes described.
Figure 6.1: Attributes and outcomes of the Clinical Genetic Service

- Expertise
- Individualised approach to service provision
- Interdisciplinary problem-based care
- Accessibility over time
- Family-centeredness
- Knowledge & understanding
  - Informed & shared decision making
  - Reassurance
  - Enablement
- Perceived Familial Control
Table 6.3: Initial coding categories and final analytical themes

- Inform families of possible insurance problems
- Support in communicating genetic

- Clinical support
- Enablement

- Individualised approach to service provision

- Continuity of care
- Obtaining research updates

- Accessibility and continuity over time

- Access to other NHS services

- Interdisciplinary problem-based care

- Decision-making support
  - Being offered choices
  - Certainty about genetic risk status
  - Empowerment to make decisions

- Informed and shared decision-making

- Access to genetics experts

- Expertise

- Levels of knowledge and education

- Knowledge and understanding

- Reassurance and relief from anxiety

- Reassurance

- Support in dealing with social consequences
  - Clinical support

- Enablement

- Feelings of control
  - Adjustment to the genetic condition

- Perceived Familial Control

- Improved clinical outcomes

- Family centeredness
6.6. DISCUSSION

Thematic analysis was used to analyse data collecting from focus groups and semi-structured interviews with service users of one UK clinical genetic service. Analysis of transcripts showed that service users overall valued the same process and outcome attributes. There were differences however in the importance attributed to each (dis)benefit based on the condition group, something that warrants further investigation through preference measurement. A methodological exercise aiming to explore differences in preferences and ranking of (dis)benefits was undertaken and is discussed in the following chapter. Five key attributes of service provision were identified based on participants’ reports of what they perceived as (dis)benefits of the CGS and these were associated to four psychosocial consequences resulting from making use of CGSs. Figure 6.1 illustrates the five key attributes and associated dimensions of Perceived Familial Control. Perceived Familial Control was proposed as the main outcome of the service, a theoretical model that describes the intangible consequences of the service.

Recent methodological criticisms of the outcome measures used to evaluate CGSs, have led into research on the outcomes and attributes which best reflect effectiveness for CGSs (Bernhardt, Biesecker and Mastromarino, 2000; Skirton, 2001; Macleod, Craufurd and Booth, 2002; Skirton, Parsons and Ewings, 2005; Apicella et al., 2006; Peacock et al., 2006; McAllister et al., 2008a; McAllister et al., 2008b; McAllister, Dunn and Todd, 2011; McAllister et al., 2011). McAllister and colleagues have conducted similar research to the one described in this thesis and have identified attributes and
psychosocial consequences of CGSs which could be used for the evaluation of the service.

There are methodological differences between the present study and past investigations, including the focus on the overall CGS rather than on one aspect of the service such as genetic counselling (e.g. Bernhardt, Biesecker and Mastromarino, 2000; Skirton, 2001; Macleod, Craufurd and Booth, 2002; Apicella et al., 2006; Peacock et al., 2006); and the use of current users of the service aiming to identify (dis)benefits of the CGS as opposed to using stakeholders of the service and exploring the needs or expectations from CGSs (e.g. McAllister et al., 2008a; McAllister et al., 2008b). In addition, none of these studies have looked into the identification of (dis)benefits from an economic evaluation perspective, and more specifically from the welfarist perspective. Despite these differences, the similarities in the (dis)benefits identified between the current and past research studies will be discussed, before focusing on the implications for the economic evaluation of CGSs discussed in the final chapter.

6.6.2. Attributes of CGSs

Provision of information is an attribute of the service appearing throughout research on what makes CGSs effective. Within the current research, information was a theme running throughout narratives while the need for information was found to be a driving force behind participants’ choice to access the CGS. Current findings have shown that even though information is actively sought by participants, the service is valued because it offers more than simply information. If the genetic information is not understood and contextualised within the personal circumstances of families,
Information overload can result in negative psychosocial outcomes. Other research has also shown that both the quality of the information ("certainty of information") and its understanding ("assimilating information") are attributes which make genetic counselling effective (Macleod, Craufurd and Booth, 2002) and are linked to satisfaction (Lipinski et al., 2006).

Finding answers as to why a genetic condition has occurred has been found to be integral to subsequent adaptation following the genetic diagnosis (Barr and Millar, 2003; Lipinski et al., 2006). The search for information therefore appears to be a coping mechanism facilitating positive effects (Pain, 1999) on emotional outcomes such as adjustment. Both current and past findings point to instigating access to CGSs as part of a coping mechanism to deal with the perceived risk and the uncertainty involved. A passive coping style depicted by feelings of resignation and acceptance over their risk status accompanied by perceptions of having no control over the cancer have been shown to be associated with increased anxiety and depression (Bennett et al., 2008).

Research has shown that information about risk status carries more utility than other aspects of genetic counselling including advice about breast and ovarian cancer screening (Peacock et al., 2006). The need for information about available risk management options is an important reason why women attend cancer genetic services and women anticipate discussing their risk status (Hallowell et al., 1997; Brain et al., 2000b; Brandt et al., 2002; Holloway et al., 2004) because they expect this information to facilitate decision-making such as whether to undergo prophylactic surgery (Hallowell et al., 2002).
The value attached to risk-management options has led Hallowell et al (2004) to propose that multidisciplinary models of service provision should be organised offering both clinical genetic and surgical services. The “interdisciplinary problem-based care” has emerged as an important element of service provision. This involves referrals to other specialists and establishing care pathways, enabling access to services, links to social and educational services and tangible support to enable access to needed benefits, and information about support organisations. Knowledge that the CGS was able to facilitate these links and the information obtained which allowed for long-term ability to use the services enabled service users to deal with the implications. The ability of the CGS to transcend the boundaries of the service and make links with health and non-health services in order to allow for service users to obtain the required support was what appealed to participants. Problems, whether health or social, were reported to and identified by genetic professionals, who then referred families on to other professionals able to deal with these problems.

This aspect of service provision has not been widely discussed within the literature, and services have not tended to be evaluated upon their ability to refer families on to other professionals. Recent research however has also identified a “co-ordinated and tailored family care” as an attribute of service provision even thought this attribute does not fully reflect the definition of “interdisciplinary approach” (McAllister et al., 2008b) (Table 6.4 lists the attributes proposed by the McAllister team along with the attributes proposed in this study in order to make explicit comparisons between the two. A more detailed comparison between these two studies is given in the next section where the theoretical model of PFC is discussed p.258).
### Table 6.4: Comparison between proposed and McAllister et al attributes

<table>
<thead>
<tr>
<th>Attributes by McAllister team</th>
<th>Attributes by Pithara</th>
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<tbody>
<tr>
<td><strong>Local and accessible services</strong></td>
<td><strong>Expertise</strong></td>
</tr>
<tr>
<td>• Peripheral clinical in local areas</td>
<td>• Professionals experts in genetics</td>
</tr>
<tr>
<td>• Often unclear where the genetics service fits into overall care</td>
<td>• Able to provide accurate and up-to-date information</td>
</tr>
<tr>
<td><strong>Open-access and follow-up</strong></td>
<td>• Research involvement</td>
</tr>
<tr>
<td>• Additional appointments after enough time has elapsed for the information to sink in</td>
<td>• Aware of all new treatments and developments in genetics</td>
</tr>
<tr>
<td>• Named person they can get in touch with</td>
<td>• Comprehensive and holistic knowledge of all aspects of a genetic conditions e.g. symptoms, inheritance patterns, prognosis</td>
</tr>
<tr>
<td>• Yearly contact letter with care updates and reminders</td>
<td><strong>Individualised approach</strong></td>
</tr>
<tr>
<td>• Updates about research and possible new treatments</td>
<td>• Take into consideration individual needs when scheduling service provision e.g. home visits</td>
</tr>
<tr>
<td><strong>Co-ordinated tailored family care</strong></td>
<td>• Adapt level of communication to the individual’s existing knowledge, background, emotional state and needs</td>
</tr>
<tr>
<td>• Provide for the long-term needs of the whole family</td>
<td>• Respect all questions and concerns</td>
</tr>
<tr>
<td>• Contact named person when children reach reproductive age</td>
<td>• Treated as an individual</td>
</tr>
<tr>
<td>• Service directed towards whole families</td>
<td><strong>Accessibility and continuity</strong></td>
</tr>
<tr>
<td>• Provide information to family members about risks, family communication, up-to-date information</td>
<td>• Open access policy</td>
</tr>
<tr>
<td>• Telling family members about risks</td>
<td>• Able to access the service when there is a need like additional concerns after information is provided</td>
</tr>
<tr>
<td><strong>Quality of the relationship</strong></td>
<td>• Follow-ups by named genetic counselor including phone calls</td>
</tr>
<tr>
<td>• Positive interpersonal interaction</td>
<td>• Service is open to all family members without the need for initiating another referral</td>
</tr>
<tr>
<td>• Social skills and flexibility</td>
<td>• Yearly follow up letters</td>
</tr>
<tr>
<td>• Listen and empathise</td>
<td><strong>Family centeredness</strong></td>
</tr>
<tr>
<td>• Compassion</td>
<td>• Family at the centre of service provision</td>
</tr>
<tr>
<td>• Expertise</td>
<td>• Available to all family members</td>
</tr>
<tr>
<td><strong>Time to talk</strong></td>
<td>• Family needs taken into consideration</td>
</tr>
<tr>
<td>• Time available for appointments</td>
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</table>
In another study participants perceived the ability of the geneticist to do something about their genetic situation as an important benefit (Macleod, Craufurd and Booth, 2002). Access to condition or risk management options as well as health services in general have been raised as benefits of CGSs especially within the cancer genetics literature (Hallowell et al., 1997; Atkin and Ahmad, 2000; Brain et al., 2000b; Brandt et al., 2002). Even though health surveillance co-ordination has not been used as part of CGSs service evaluation, this aspect of service provision has been a stated role of a clinical geneticist (Godard et al., 2003).

The quality of the relationship between counselee and counsellor is an attribute repeatedly presented among outcomes of genetic services (Bernhardt, Biesecker and Mastromarino, 2000; Skirton, 2001; Macleod, Craufurd and Booth, 2002; Skirton, Parsons and Ewings, 2005; McAllister et al., 2008b). Present findings showed that the good relationship which developed between genetic professionals and service users was a result of the individualised approach adopted by professionals which allowed the individual to feel comfortable, being listened to, valued, and allowed to express their concerns without making them feel rushed to fit during a pre-determined time slot. Time therefore is important, and this is another characteristic emerging within the literature (Bernhardt, Biesecker and Mastromarino, 2000; McAllister et al., 2008b).

Attributes raised so far appear to have considerable similarities with what patients may value in health services other than CGSs. Genetic conditions are also lifelong conditions, and in that sense the needs of families remain the same regardless of the services they access at different times. Research investigating the coping needs of parents whose child has been diagnosed with a chronic genetic condition reports that
ability to cope is greatly influenced by access to services, health or social, as well as the quality of the contact and these can promote or hinder the family’s coping abilities (Atkin and Ahmad, 2000; Tong et al., 2008; Kepreotes, Keatinge and Stone, 2010). The professionals’ level of expertise of the condition, the abilities as well as willingness of professionals to support families and their personal approach are all attributes perceived as important for promoting a sense of control (Atkin and Ahmad, 2000; Kepreotes, Keatinge and Stone, 2010). Information that families expected to receive from services and most often didn’t, included information about the condition and tangible support available (Atkin and Ahmad, 2000; Kepreotes, Keatinge and Stone, 2010). These characteristics reflect the (dis)benefits associated with the CGS.

It appears that CGSs might not be all that different from other health services in terms of what attributes service users expect and value and the outcomes of that contact. Other attributes however appear to distinguish CGSs from the rest of the NHS. Where CGSs differ is on their implications for the whole family unit, intra- and inter-generationally. The family nature or “family centeredness” of the CGS was a very strong theme running through all (dis)benefits identified in the current research and through all conditions. Individuals pursuing genetic services and choosing to have genetic testing have reported that they needed to know their cancer genetic status because this information was perceived important for their children and other family members to help them make important decisions in the future (Kausmeyer et al., 2006b; MacDonald et al., 2007). This attribute has also considerable economic implications since it illustrates the particular case of CGSs involving the presence of considerable externalities, arguably
more than in the majority of healthcare services, something that will be discussed in the final chapter.

An aspect of CGSs not featuring as much among literature has been that of the longitudinality and continuity of service provision, here termed as “accessibility over time”. One possible reason might be that research has tended to evaluate outcomes of CGSs using short- to medium term follow up, i.e. some weeks to up to 1 year follow up within most research. Qualitative research has also tended to assess perceptions of individuals following genetic counselling therefore this aspect of service provision might not have emerged that strongly (e.g. Skirton, 2001). In the study by Bernhardt et al participants valued their long-term relationship with genetic counsellors through phone-calls or letters and this focused mainly on psychosocial support (Bernhardt, Biesecker and Mastromarino, 2000). McAllister and colleagues identified “open access and yearly follow-ups” as an attribute of service provision (McAllister et al., 2008b).

In a systematic review, accessibility, continuity of care, consultation time and doctor-patient relationships were found to be associated with patient satisfaction in the context of family medicine (Sans-Corrales et al., 2006), findings which reflect the current ones in terms of the benefits resulting after access to the CGS. Continuity of care, doctor-patient relationships and consultation time were also found to be related with health improvements (Sans-Corrales et al., 2006). Even though continuity of care has more frequently been associated with primary care (Freeman, Olesen and Hjortdahl, 2003; Sans-Corrales et al., 2006; Adler, Vasiliadis and Bickell, 2010) having a stable point of contact and a close relationship associated with that established contact with the service is obviously important for all patients. Continuity of care has also been found to
encourage more regular attendance to cancer screening programmes (O’Malley et al., 1997; Schueler, Chu and Smith-Bindman, 2008).

Expertise is another attribute emerging strongly in present findings but not commonly identified as a benefit of the service. Other research has shown that outside clinical genetic services, there is great variability in what healthcare professionals from various specialties know about genetic conditions (Abramsky et al., 2001; Van Riel et al., 2010). This aspect of the service arguably warrants further consideration, particularly in the context of investigations of genetic counselling being offered outside CGSs.

6.6.1. Theoretical Framework of Perceived Familial Control

As a conceptual outcome of CGSs, this research has proposed the outcome of Perceived Familial Control which relates to a process of empowerment taking place longitudinally throughout the family’s contact with the service rather than an end point resulting following contact with CGSs. This outcome has been defined as the belief of family members that:

- They and their family members have the tools or know how to obtain the tools to be able to deal with the tangible and intangible consequences resulting from having a genetic condition in the family.

This conceptual outcome encompasses the following four dimensions: (1) knowledge and understanding, (2) informed and shared decision-making, (3) enablement, and (4) reassurance. These dimensions were defined in p.222, section 6.5.1.
The present findings partially fit two existing frameworks proposed as outcome measures for CGSs, namely the concept of Perceived Personal Control by Berkenstadt (Berkenstadt et al., 1999; Smets et al., 2006) and the concept of empowerment by McAllister et al (McAllister et al., 2008a; McAllister et al., 2008b; McAllister, Dunn and Todd, 2011). Current and past findings will be reviewed.

As discussed in Chapter 3, the outcome measure of PPC encompasses three dimensions, namely behavioural control; cognitive control and decisional control. This outcome measure asks participants their perceptions of how much control they believe they have over the genetic condition in terms of understanding the genetic condition (cognitive control); knowing what to do in order to deal with the implications of the condition (behavioural control); and finally their perceptions of their ability to make decisions crucial for the outcomes of the genetic condition (decisional control).

One problem of the PPC theoretical model is that it involves an individualistic perspective of control, rather than the familial sense of control emerging through the current findings. The current research also illustrates how achieving a sense of control is a life-long process for the families referred to CGS rather than an end-point resulting after genetic counselling. Even though a positive impact on the feelings of control might result after using the service, absence of a long-term relationship between the service and the families might lead to feelings of control falling to the pre-access levels, as was suggested by the current findings. Existing studies using the outcome measure of PPC (Berkenstadt et al., 1999) have tended to measure this outcome pre-and post-counselling (Berkenstadt et al., 1999; Davey et al., 2005; Pieterse et al., 2005a; Aalfs et
al., 2007; Pieterse et al., 2007b), but a long-term follow-up of users of the service might be more appropriate to accurately measure the impact of the service.

McAllister and colleagues have proposed a theoretical framework very similar to the concept of PPC but which is expanded to encompass a number of other dimensions based on their qualitative findings. McAllister and colleagues proposed the concept of “empowerment” as a construct integrating all of the outcomes of CGS emerging through their research (McAllister et al., 2008a; McAllister et al., 2008b; McAllister, Dunn and Todd, 2011). These authors defined “empowerment” as “a belief system that allows a person to feel in, or take control of their lives and have responsibility or autonomy over decisions and choices” (McAllister et al., 2008a: p.898). This construct was introduced in section 3.3.5.2 where the outcome of Perceived Personal Control (PPC) was discussed. Here it is briefly mentioned that the updated construct of “empowerment” was developed and its dimensions validated through a two stage data collection process to incorporate the following five dimensions: cognitive control, behavioural control, decisional control, hope and emotional regulation. This construct is a promising one, and has recently been developed into an outcome measure for evaluating genetic counselling (McAllister et al., 2011). Table 6.5 illustrates the conceptual models proposed in this study and those proposed by the McAllister group to describe the impact of CGSs.
Table 6.5: Comparison between proposed and McAllister et al conceptual models of benefit

<table>
<thead>
<tr>
<th>(McAllister et al., 2008a)</th>
<th>(McAllister, Dunn and Todd, 2011)</th>
<th>Pithara 2011</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Empowerment(1)</strong></td>
<td><strong>Empowerment (2)</strong></td>
<td><strong>Perceived Familial Control</strong></td>
</tr>
<tr>
<td>A set of beliefs that enable a person from a family affected by a genetic condition to feel that they have some control over and hope for the future</td>
<td>A set of beliefs that enable a person from a family affected by a genetic condition to feel that they have some control over and hope for the future</td>
<td>The belief of an individual that they and their family have the tools or know how to obtain the tools to deal with all the consequences resulting from having a genetic condition</td>
</tr>
<tr>
<td><strong>Decision-making</strong></td>
<td><strong>Decisional control</strong></td>
<td><strong>Informed and shared decision-making</strong></td>
</tr>
<tr>
<td>The belief that one can make important life decisions in an informed way</td>
<td>The belief that one can make important life decisions in an informed way</td>
<td>The belief that one can make decisions based on accurate and up-to-date information, and these decisions are in agreement with their individual beliefs and goals.</td>
</tr>
<tr>
<td><strong>Knowledge and understanding</strong></td>
<td><strong>Cognitive control</strong></td>
<td>Knowledge and understanding</td>
</tr>
<tr>
<td>the belief that one has sufficient information about the condition, including risks to oneself and one’s relatives, and any treatment, prevention and support available</td>
<td>The belief that one has sufficient information about the condition, including risks to oneself and one’s relatives, and any treatment, prevention and support available</td>
<td>The belief that one has appropriate, accurate and up-to-date information which was understood, including inheritance patterns, risks, prognosis, and management options</td>
</tr>
<tr>
<td><strong>Instrumentality</strong></td>
<td><strong>Behavioural control</strong></td>
<td><strong>Enablement</strong></td>
</tr>
<tr>
<td>The belief that one can make effective use of the health and social care systems for the benefit of the whole family</td>
<td>The belief that one can make effective use of the health and social care systems for the benefit of the whole family</td>
<td>The belief that one is enabled to cope with the tangible consequences of the condition including symptoms, and social implications accessing using health and social services.</td>
</tr>
<tr>
<td><strong>Future orientation</strong></td>
<td><strong>Hope</strong></td>
<td><strong>Reassurance</strong></td>
</tr>
<tr>
<td>The belief that one can look to the future with hope for a fulfilling family life, for oneself, one’s family and/or one’s future descendants</td>
<td>The belief that one can look to the future having hope for a fulfilling family life, for oneself, one’s family and/or one’s future descendants</td>
<td>The belief that one is able to deal with major (genetic condition) stressors leading to improved emotional outcomes</td>
</tr>
<tr>
<td><strong>Emotional regulation</strong></td>
<td><strong>Enablement</strong></td>
<td></td>
</tr>
<tr>
<td>The belief that one can manage one’s feelings about having a genetic condition in the family</td>
<td>The belief that one is enabled to cope with the tangible consequences of the condition including symptoms, and social implications accessing using health and social services.</td>
<td></td>
</tr>
</tbody>
</table>

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Findings reflect to a great extent the conceptual model of empowerment proposed by McAllister and colleagues (McAllister et al., 2008a; McAllister, Dunn and Todd, 2011), especially the adapted model of empowerment and the addition of an emotional dimension to the model. The most distinct difference however is the familial element inherent in the definition of PFC as opposed to “empowerment” where the impact on family members is incorporated in the distinct dimensions but not the definition. The familial element was inherent in all narrative and it was therefore considered important to incorporate this in any theoretical framework used to describe an outcome of CGSs. The implications of this for operationalising such an outcome are discussed in the final chapter.

The dimension of reassurance did not feature in the initial dimensions proposed by the McAllister team, an outcome found to result following contact with the CGS and a positive emotional outcome emerging in other research where genetic risk information has been found to play a role towards reassurance and peace of mind (Kausmeyer et al., 2006a). During the second round of data collection however, the emotional aspect of CGSs was raised and thus an additional dimension was incorporated. In our model of Perceived Familial Control, peace of mind resulting from accessing the genetic service, and encompassing the two dimensions of hope and emotional regulation of the empowerment model was captured in the theme of reassurance.

Research utilising generic outcome measures has failed to capture conclusive evidence on the emotional impact of genetic testing or genetic counselling on users of the service (e.g. Braithwaite et al., 2006; Hamilton, Lobel and Moyer, 2009). Research utilising qualitative methods however has shown that genetic services can elicit emotions of
hope, gratitude, relief and tranquillity and a predominance of optimistic expectations (Phelps, Bennett and Brain, 2008). Claes and colleagues found that even though no negative overall psychological impact was evident in individuals after testing for BRCA1/2 genetic mutations, results from open-ended questions identified a number of positive as well as negative implications particularly from carriers. Relief in the case of non-carriers and relief from uncertainty for carriers was part of the advantages reported by participants (Claes et al., 2005). Service users may experience conflicting emotions and Kausmeyer and colleagues (Kausmeyer et al., 2006a) found that even though 72% of participants choosing to have cancer genetic testing reported to be glad about their decision while waiting for their test results and more than half felt empowered by their decision, almost half reported feeling anxious during that time. It is evident that an outcome measure which can capture the range of users’ experiences (cognitive, emotional, adaptation-related and behavioural) needs to be utilised.

The construct of empowerment has been taken forward into the design and initial validation of an outcome measure described as “the genetic counselling outcome scale” (McAllister et al., 2011). Findings presented in this paper raise two points for discussion. Firstly, the construct of empowerment is presented as an adaptation of the Perceived Personal Control outcome measure and in accordance to this model the McAllister team have renamed three of their dimensions according to the PPC dimensions of cognitive, behavioural, and decisional control. Based on the distinct characteristics of genetic services and the constituent dimensions of the emerging constructs one might argue that “empowerment” might be a less suitable concept to describe the emerging processes. Empowerment is a construct which tends to be used
in different contexts within the literature. Within a health context it has mainly been connected with patient’s informed decision-making about the treatment and services they receive, even though it has been argued that in order for patients to be empowered a positive provider-patient interaction which promotes trust is integral (Nyatanga and Dann, 2002). At the same time, the universality of this concept i.e. it can be applied to any context of human activity, makes it highly ambiguous (Nyatanga and Dann, 2002). Finally, empowerment within a health context has mainly been understood as an individualistic concept, where the process corresponds to empowering individuals rather than groups of individuals i.e. families. This individuality is also reflected in the definition given to this construct by McAllister and colleagues. It is argued therefore, that Perceived Familial Control might be a more appropriate framework to describe the impact of CGSs than “empowerment”. In this way, the shift from an individualised approach to service provision to recognition of the familial (longitudinal) impact is made explicit as well as making explicit the evolution of the theoretical framework of PPC to encompass the familial element.

Despite their methodological and theoretical differences, both studies provide support for the role of CGS as a family, generation-spanning service.

6.7. CONCLUSIONS

Overall, common benefits and disbenefits were raised by participants even if their individual pathways through the service differed. The presence of a gene mutation which results in the manifestation of symptoms relevant to a specific genetic condition or to an increase of the risks for such manifestation may result in a number of
consequences. These consequences could be linked to the genetic condition itself, such as increased pressures on the family from caring for an affected individual, or to the genetic service, such as accurate information about ways to deal with these consequences. Impacts relating to the genetic condition can be altered in their direction by the CGS, for example genetic counselling could help alleviate certain of the negative consequences on family relationships and assist families prepare and deal with insurance problems. Even though this study was informed by literature which emphasised the need for identifying psychosocial implications of CGSs, process attributes were also found to be of value to service users. The familial element emerged as being of particular importance in the case of genetic services. In order to represent such an emphasis on inter- and intra-generational influences, the construct of Perceived Familial Control was proposed as the theoretical model to describe the overall impact of CGSs. This model reflects similar findings by McAllister and colleagues, even though the two studies differ on the construct chosen to describe their respective findings.

The findings of both studies add considerably to the efforts made to understand the broader implications of CGSs and moving on from that, to identify one outcome which could be operationalised for evaluating the work of the services. Both studies propose similar attributes and psychosocial outcomes and confirm the importance of the accurate information provided and subsequent informed decision-making, the support provided to families in order to deal with the implications of the genetic condition, the respect given to the individual patient and their family’s needs and the sense of control facilitated by the service. Their differences point to areas where future research is
required to untangle the ambiguities which might reflect differences relevant to the different genetic services used for data collection and their respective event pathways, or differences in the value placed on perceived benefits discussed during the focus groups conducted by each study. The following chapter will present the third and final phase of data collection where initial categories of (dis)benefits were presented to stakeholders of the CGS in order to explore which ones they perceived as most important.
CHAPTER 7

Phase Three: Views of stakeholders
7.1. Overview

This chapter will:

1. Provide an overview of the aims and objectives of the third phase of data collection;

2. Describe the methods and recruitment techniques utilised;

3. Provide a descriptive report of the findings;

4. Discuss the methodological implications of this phase and how this knowledge can be used to further the design of an economic evaluation;

5. Discuss the findings in the context of existing literature.

7.2. Aims and Objectives

This phase has two objectives, namely to

1. Establish respondent validation and transferability of findings, and

2. Perform a pilot study to test the use of Audience Response Systems (ARSs) as an approach to exploring the preferences of stakeholders

**Respondent validation**: this is a suggested criterion for ensuring increased validity in qualitative research. Here the researcher's account of the data is compared with the accounts of those taking part in the research in order to establish the rate of correspondence between the two sets of accounts (Mays and Pope, 2006). Incorporating this aspect was considered essential to ensure that proposed (dis)benefits
accurately reflected the experiences of participants and give an opportunity to stakeholders to comment and propose alternative (dis)benefits of the service.

Transferability of findings was also attempted through the inclusion of stakeholders who were asked to contribute by reporting their perceptions about the applicability of findings to themselves. Through this it was possible to test whether individuals other than those taking part in previous phases agreed with what had emerged or whether they had alternative experiences to share. Again, this was made possible by giving an opportunity to those attending to contribute to the research.

This phase also aimed to pilot ARSs for their utility in exploring the preferences of stakeholders about which (dis)benefits they considered important and thus featured more strongly in their utility function, an approach which could then take place on a larger scale and with users from other genetic centres in order to establish generalisability of findings.

7.3. METHODS

7.3.1. Data collection techniques

This phase constituted the quantitative element within the mixed design used in this research.

This phase made use of categories of (dis)benefits identified at the early stages of analysis (see Table 6.3). These categories are listed in Table 7.1 (p.273). Categories were presented to stakeholders during a half-day event using TurningPoint technology. TurningPoint technology is an Audience Response System (ARS) which integrates fully
with PowerPoint presentations thus allowing the audience to actively take part by submitting responses to interactive questions through the use of a hand-held computer device. TurningPoint has the ability to collect, process, and record responses from those members of the audience choosing to respond to the interactive questions or statements presented using the personal hand-held devices. These responses are collected by central software and translated into measurable results which can be presented during the presentation and also stored for later access and use.

This technology has been commonly used within education as an education-enhancing and assessment technique (Miller, Ashar and Getz, 2003; MacGeorge et al., 2008) but has not been traditionally used as a technique for data collection (McCarter and Caza, 2009) even though within healthcare ARSs have been used for developing educational frameworks in the context of nursing (Kirk et al., 2004). ARSs have the advantage of being anonymous; facilitate contribution from all participants and have even been found to encourage participants to overcome their shyness because of the anonymity and unthreatening nature of participation (Graham et al., 2007); and finally allow instant collection, analysis and presentation of responses thus being both a time- and cost-saving option. The quick nature of collecting and analysing responses and incorporating these findings within the presentation thus feeding into and guiding subsequent data collection makes this a useful method to use for collecting audience attitudes, perceptions and consensus development. ARSs could be seen as an alternative to a Delphi study since it can and has been used for establishing consensus and prioritising given alternatives (Kirk et al., 2004).
7.3.2. Recruitment and procedures

This phase focused on stakeholders of the chosen CGS including service users, service providers, friends and family of patients, and representatives of patient support groups.

Stakeholders were invited to attend a free event through e-mail advertisements, posters on web-sites and postal invitations. Posters were advertised on Wales’ genetics-related websites and through circulating advertisements to members of the Genetic Interest Group (Wales). The CGS service users invited to attend the second phase of data collection were also invited to attend this event, regardless of whether they had actually participated in any discussions. Users were sent a leaflet describing the event and inviting the individual and any other family member or friend who might also be interested to attend. A free-post reply slip was included in the invitation to assist with organising the event.

No written informed consent was taken for this phase since participation within the ARS process was voluntary and no personal information was recorded from which participants could be identified, other than demographic information. Agreeing to the process of pressing a button representing a given response to a question was taken as indirect consent.

During the event all participants were given a personal hand-held response device (TurningPoint technology) with which they interacted during a PowerPoint presentation. All participants were given a list with all the terms used during the presentation along with a brief definition of each one based on the analysis of the previous phase. A list of these definitions is included in Appendix 17.
The event took place on CGS premises on the 28\textsuperscript{th} of November 2008 from 12.30pm until 4 pm.

Data collection was organised around two discrete parts. During the first part attendees were given practice questions to familiarise themselves to the ARS technology, followed by demographic questions and the presentation of categories of (dis)benefits to explore attitudes. The final part explored the use of ARS technology for investigating perceived relationships between process-related attributes and psychosocial outcomes.

An introduction to the event was presented by the clinical advisor to the project who acted as the link between the research taking place and the genetic service.

7.3.3. Part One

A PowerPoint presentation was given incorporating practice questions aimed to familiarise participants to TurningPoint technology (see Appendix 16 for a list of all questions). These were followed by demographic questions, and again by the categories of (dis)benefits reported in Table 7.1 (p.273). Participants were asked to think each (dis)benefit in turn and rate each one based on their perceptions of whether each variable was important to them, to the affected member in their family, or to individuals they were involved with who were affected by a genetic disease before giving an answer. Participants could state their opinions using a seven-point Likert scale with one representing “Strongly agree” and seven “strongly disagree” that each variable was an important benefit or disbenefit of the service.
By asking for levels of agreement on the importance of each (dis)benefit the relevance of each one to the participants could be tested, thus establishing some credibility, and also indirectly establish levels of importance i.e. ranking their preferences.

Even though ranking could be achieved by directly asking participants to rank the (dis)benefits, this presented some problems because of the large number of (dis)benefits presented and the cognitive limitations of human working memory. Working memory refers to the structures and processes used for storing and manipulating information for a short period of time (Smyth et al., 1994). Working memory is generally considered to have a capacity of seven items (Miller, 1956), and overall, 19 (dis)benefits were presented. This raised some limitations in the way (dis)benefits could be presented for ranking.

Through presenting each (dis)benefit separately using ARSs and asking participants to state their agreement as to its importance, it would be possible to test this approach for its validity in identifying both consequences that were or were not relevant to participants (through responses falling at the two opposite ends of the scale), and also to rank (dis)benefits indirectly in order of highest relevance or importance.
TABLE 7.1: Categories presented during the half-day event

*This denotes categories presented in Part 2 of the event

<table>
<thead>
<tr>
<th>Elements of service provision</th>
<th>Emotion-related consequences</th>
<th>Social consequences</th>
</tr>
</thead>
<tbody>
<tr>
<td>Support in dealing with social effects of genetic condition*</td>
<td>Reassurance and relief from anxiety</td>
<td>Insurance problems (disbenefit)</td>
</tr>
<tr>
<td>Inform families of possible insurance problems</td>
<td>Empowerment (making decisions)</td>
<td>Changes in family relationships</td>
</tr>
<tr>
<td>Support in communicating genetic risk*</td>
<td>Feelings of control over the situation</td>
<td></td>
</tr>
<tr>
<td>Continuity of care*</td>
<td>Adjustment to the genetic condition</td>
<td></td>
</tr>
<tr>
<td>Obtaining research updates*</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Access to other NHS services*</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinical support*</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Decision-making support*</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Being offered choices*</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Levels of knowledge and education*</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Certainty about genetic risk status*</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Access to genetics experts*</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Improved clinical outcomes*</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

7.3.4. Part Two

The second part was used as a pilot for the methodological utility of ARSs in service user involvement within outcomes research and theory testing. The TurningPoint Ranking wizard enables the researcher to compare and rank one or more items based on chosen criteria and present the findings on a ranking chart (TurningPoint Technologies webpage). This technology enabled exploration of attendees’ perceptions of what aspects of service provision they perceived to contribute towards two popular outcome measures, namely “adjustment to the genetic condition” and “sense of control” These two outcomes were chosen because of their popularity in the literature evaluating CGSs and the absence of process-related attributes from their conceptualisation even
though these types of benefits emerged repeatedly among participants of the present research. The psychosocial literature encompassing these two concepts is reviewed in Chapter 3. Twelve categories linked to service provision were presented to attendees (these are marked with an asterisk in Table 7.1), and each category was attributed levels of importance in relation to each of the two outcomes, which the TurningPoint software later ranked. A decision was taken not to include the category “Inform families of possible insurance problems” in this part because of its overlap with “support in dealing with social effects of genetic condition”.

**7.4. ANALYSIS**

Collation and analysis of the data obtained during this phase was done automatically by the TurningPoint software. This software analysed demographic information and response rates of different groups. Despite advertising of the event only a small number of individuals turned up during the day (see next section for numbers of attendees and demographic characteristics) which prevented statistical analysis from taking place.

**7.5. RESULTS AND DISCUSSION**

Five demographic group options were given to attendees: “user of the CGS”; “geneticist/counsellor”; “working with voluntary groups”; “researcher”; and “other”. Ten individuals identified themselves as “users” of the CGS, four as “other” and two as “geneticist/counsellor” and 1 as “researcher”. The three individuals belonging to the “geneticist/counsellor” and “researcher” categories had been intimately involved in all stages of the research and their responses were excluded from the analysis.
The following table shows the demographic characteristics of “service users” and “others” taking part in the voting session.

Table 7.2: Demographic information of attendees

<table>
<thead>
<tr>
<th>Question</th>
<th>“Service User” responses</th>
<th>“Other” responses</th>
</tr>
</thead>
<tbody>
<tr>
<td>Taken part in previous phases of this research</td>
<td>Yes: 7</td>
<td></td>
</tr>
<tr>
<td></td>
<td>No: 2</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Abstain: 1</td>
<td></td>
</tr>
<tr>
<td></td>
<td>No: 4</td>
<td></td>
</tr>
<tr>
<td>Sex</td>
<td>Female: 7</td>
<td>Male: 2</td>
</tr>
<tr>
<td></td>
<td>Male: 2</td>
<td>Male: 2</td>
</tr>
<tr>
<td></td>
<td>Abstain: 1</td>
<td></td>
</tr>
<tr>
<td>Age</td>
<td>31-45: 2</td>
<td>19-30: 1</td>
</tr>
<tr>
<td></td>
<td>46-65: 5</td>
<td>31-45: 1</td>
</tr>
<tr>
<td></td>
<td>Over 65: 3</td>
<td>46-65: 1</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Over 65: 1</td>
</tr>
<tr>
<td>First contact with the genetics service</td>
<td>Less than a year ago: 2</td>
<td>1-5 years ago: 2</td>
</tr>
<tr>
<td></td>
<td>1-5 years ago: 4</td>
<td>Not relevant: 2</td>
</tr>
<tr>
<td></td>
<td>More than 5 years ago: 3</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Abstain: 1</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Family history of condition</td>
<td>Family history: 7</td>
<td>Family history: 1</td>
</tr>
<tr>
<td></td>
<td>No family history: 3</td>
<td>No family history: 1</td>
</tr>
<tr>
<td></td>
<td></td>
<td>I don’t know/can’t remember: 1</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Abstain: 1</td>
</tr>
<tr>
<td>Availability and uptake of genetic testing</td>
<td>Not offered and not interested: 2</td>
<td>Have not been offered and would not be interested: 1</td>
</tr>
<tr>
<td></td>
<td>Not been offered and interested: 2</td>
<td>Have not been offered and would be interested: 3</td>
</tr>
<tr>
<td></td>
<td>Offered but not interested: 1</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Offered and uptake: 4</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Not applicable: 1</td>
<td></td>
</tr>
<tr>
<td>Reasons for referral</td>
<td>Family history of cancer: 6</td>
<td>Other: 1</td>
</tr>
<tr>
<td></td>
<td>Diagnosed with genetic condition: 1</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Child diagnosed with genetic condition: 1</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Sibling/relative diagnosed with genetic condition: 1</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Not applicable: 1</td>
<td>Not applicable: 3</td>
</tr>
</tbody>
</table>

Fourteen individuals turned up for the event. Most attendees were women (nine individuals) while half of the attendees (seven service users) reported participating in previous phases of the research allowing for respondent validation of the categories identified through the interviews and focus groups taking place Phase Two (Chapter 6).
The majority of attendees had a family history of the condition (seven “service users” and one “other”) and most “service users” were referred to the service because of a family history of cancer. This represents actual referrals to the CGS where cancer referrals constitute the largest group of referrals to the centre. The majority of “service users” had been offered and had undergone genetic testing while three individuals belonging to the “other” category reported being interested in genetic testing even though they had not been offered the test.

7.5.1. Part One

Nineteen categories of (dis)benefits were presented to attendees and these can be seen in Table 7.1 (p.273) (a definition for all categories is given in Appendix 17). Because of the small numbers of individuals attending this final phase statistical analysis could not be undertaken, but some trends could be identified based on the level of agreement of contributors. In order to define what constituted as an “important” (dis)benefit it was decided to focus on those (dis)benefits which the majority of contributors rated as important. This means that only those (dis)benefits which attracted ratings of “strongly agree”, “agree” or “somewhat agree” by the majority of contributors would pass the importance criterion.

Based on this criterion, nine categories of (dis)benefits were identified (see Table 7.3). Eleven out of fourteen individuals strongly agreed that “decision-making support” and “continuity of care” were important benefits of the CGS, even though there appeared to be higher consensus in the case of “continuity of care” where responses were limited to “strongly agree” (11 contributors) and “agree” (three contributors).
Table 7.3: Categories of (dis)benefits accumulating the highest levels of agreement

<table>
<thead>
<tr>
<th>Consequence</th>
<th>Strongly agree</th>
<th>Agree</th>
<th>Somewhat agree</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 Continuity of care</td>
<td>11</td>
<td>3</td>
<td></td>
<td>14</td>
</tr>
<tr>
<td>2 Access to genetics experts</td>
<td>10</td>
<td>4</td>
<td></td>
<td>14</td>
</tr>
<tr>
<td>3 Being offered choices</td>
<td>10</td>
<td>4</td>
<td></td>
<td>14</td>
</tr>
<tr>
<td>4 Research updates</td>
<td>10</td>
<td>4</td>
<td></td>
<td>14</td>
</tr>
<tr>
<td>5 Levels of knowledge and education</td>
<td>5</td>
<td>9</td>
<td></td>
<td>14</td>
</tr>
<tr>
<td>6 Decision-making support</td>
<td>11</td>
<td>2</td>
<td>1</td>
<td>14</td>
</tr>
<tr>
<td>7 Access to other NHS services</td>
<td>9</td>
<td>4</td>
<td>1</td>
<td>14</td>
</tr>
<tr>
<td>8 Empowerment (making decisions)</td>
<td>5</td>
<td>8</td>
<td>1</td>
<td>14</td>
</tr>
<tr>
<td>9 Clinical outcomes</td>
<td>3</td>
<td>10</td>
<td>1</td>
<td>14</td>
</tr>
</tbody>
</table>

All attendees were given the opportunity to comment on the (dis)benefits presented and state if they agreed with what was presented and also propose (dis)benefits if they believed that (dis)benefits existed which were not raised by the researcher. None however commented or proposed new (dis)benefits even though response forms were returned with comments about the overall event, demonstrating respondent validation of these (dis)benefits and transferability of these to those not taking part in the previous phase.

Four out of the eight categories indirectly ranked among the most important categories of (dis)benefits, including the one ranking first ("continuity of care"), were categories which had formed main themes as a result of in-depth analysis highlighting their perceived importance. “Family centeredness” which had formed a main theme but was
not presented as a separate category was a concept that had permeated all (dis)benefits presented during the half-day event as they were described to apply to the individual themselves and all other family members.

A surprising finding was that “clinical outcomes” appeared among the top nine (dis)benefits preferred by attendees, a category not emerging to be widely relevant during the second phase. One possible reason for this is that participants might have been rating categories of (dis)benefits based on the motivating factors prompting them to access the CGS in the first place. Current findings identified an interdisciplinary problem-based approach to genetic service provision as an important benefit of the CGS, where the genetic service provides clinical support and/or access to other healthcare, social or other services. The need for multidisciplinary service provision has been raised by research reporting that women referred to a cancer genetic centre regarded access to other specialists as very important (Brain et al., 2000b; Holloway et al., 2004). Access to screening services for individuals with a cancer family history is a motivating factor for accessing CGSs (Brain et al., 2000b), while access to required healthcare services is one of many needs of families affected by a genetic condition (McAllister et al., 2008a). Even though a direct impact on health outcomes is not the main aim of genetic services (Clarke, Parsons and Williams, 1996), an interest for improving their health outcomes might be what is motivating families to access CGSs.

This might suggest that improved health outcomes might be among the expectations or hopes of service users as a result of them dealing with the situation rather than a perceived direct outcome of the service. Other research has shown that women diagnosed with cancer value their options of cancer genetic testing, regardless of any
anticipated emotional overload as long as the test result would affect their treatment options (Ardern-Jones, Kenen and Eeles, 2005). It is suggested that service users attribute higher importance to more practical aspects of service provision which are perceived to help in the clinical and other practical implications of the condition, and are willing to endure some emotional distress during the process. This arguably explains the low ratings given to emotional outcomes such as “reassurance from relief and anxiety” by contributors in this phase.

7.5.2. Part Two

This part explored the usability of ARSs in outcomes research by exploring which dimensions of service provision attendees perceived as important in achieving two popular outcomes. This technology presents the possibility to include service users in the design of outcome measures by asking them to contribute in validating proposed dimensions of outcome measures and exploring their attitudes towards different types of variables.

During this part attendees were presented 12 categories representing elements of service provision including “improved clinical outcomes” (see categories marked with an asterisk in Table 7.1, p.273) and were asked to rate each attribute according to its perceived importance in achieving either a sense of control or adjustment to one’s circumstances. Definitions to both attributes and the psychological outcomes were provided to attendees (Appendix 17). Table 7.4 lists the seven attributes ranked most important for achieving adjustment and Table 7.5 the attributes related to achieving a sense of control.
‘Access to other NHS services”, “continuity of care”, “certainty about genetic risk status”, “access to genetics expertise”, “knowledge and education” and “support with decision-making” again emerged as the benefits valued most by stakeholders of CGSs. Interestingly, even though “research updates” and “being given choices” were ranked highly in the first part, these did not rank as high during the second part. This was also the case for “improved clinical outcomes”, suggesting that contributors were able to differentiate between the purposes of each part.

Tables 7.4 and 7.5 illustrate the items contributors perceived as more important for promoting Adjustment (Table 7.4) and Sense of Control (Table 7.5).

**Table 7.4: Item rankings for Adjustment**

<table>
<thead>
<tr>
<th>Ranking</th>
<th>Item</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Access to other NHS services</td>
</tr>
<tr>
<td>2</td>
<td>Continuity of care</td>
</tr>
<tr>
<td>3</td>
<td>Access to genetics experts</td>
</tr>
<tr>
<td>4</td>
<td>Certainty about genetic risk status</td>
</tr>
<tr>
<td>5</td>
<td>Knowledge and education</td>
</tr>
<tr>
<td>6</td>
<td>Being offered choices</td>
</tr>
<tr>
<td>6</td>
<td>Clinical support</td>
</tr>
</tbody>
</table>
Table 7.5: Item rankings for Sense of Control

<table>
<thead>
<tr>
<th>Ranking</th>
<th>Item</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Access to other NHS services</td>
</tr>
<tr>
<td>1</td>
<td>Continuity of care</td>
</tr>
<tr>
<td>2</td>
<td>Knowledge and education</td>
</tr>
<tr>
<td>3</td>
<td>Certainty about genetic risk status</td>
</tr>
<tr>
<td>4</td>
<td>Decision-making support</td>
</tr>
<tr>
<td>5</td>
<td>Support in communicating genetic risk status</td>
</tr>
<tr>
<td>6</td>
<td>Access to genetics experts</td>
</tr>
</tbody>
</table>

The items ranked as more important for “perceived control” closely match the dimensions found in the Perceived Personal Control (PPC) outcome measure (Berkenstadt et al., 1999) as well as the model of control proposed in the previous chapter. The outcome measure designed by Berkenstadt et al (1999) encompasses three dimensions of control: behavioural control, being defined as “the availability of an instrumental response that may directly influence or modify the physical characteristics of a stressful event”; cognitive control, which involves the processing of information in order to make a potentially threatening situation less stressful; and finally decisional control, which involves the opportunity to choose among different courses of action.

Identified items could be categorised based on the PPC dimensions: access to other NHS services, continuity of care, and support with communicating genetic risk status under behavioural control; knowledge and education, and certainty about genetic risk status.
status falling within the cognitive control dimension; and finally decision-making support falling within the decisional control dimension. The expertise of the service could be thought of as contributing towards both cognitive control and decisional control.

Psychological adjustment has mainly been investigated using proxy outcome measures even though an outcome measure specifically designed to measure adjustment to genetic information has been designed (Read, Perry and Duffy, 2005). The PAGIS outcome measure has been presented to encompass these dimensions: non-intrusiveness (freedom from uncontrolled spontaneous thoughts and feelings that occur as a result of knowledge of the condition related gene); support (satisfaction with relationships with others that allow for discussion of genetic information and provide the feeling of being cared about); self-worth (overall evaluation of self-esteem given the knowledge of a condition-related gene); certainty (perception of accurate knowledge and understanding of what it means to have the condition-related gene); and self-efficacy (perception of control over the consequences of the condition-related gene).

Conceptually these dimensions are very similar to the dimensions of the PPC outcome measure even though the PPC does not incorporate a dimension of (social) support. Even though the issue of family relationships had been raised during the discussion in Phase Two, in the third phase this aspect did not emerge among those attributed high importance by contributors, suggesting that this implication of genetic information warrants further investigation.
7.6. DISCUSSION

This phase aimed to establish respondent validation and transferability of findings as well as pilot Audience Response Systems (ARSs). All attendees attributed at least some importance to the categories presented suggesting for both respondent validation and transferability within those attending the event but not participating in the previous phase.

With this phase it was aimed to pilot ARSs for their utility in exploring the preferences of stakeholders about which (dis)benefits they considered important and thus featured more strongly in their utility function, an approach which could then take place on a larger scale and with users from other genetic centres attempting to establish generalisability. According to Gafni and Birch (1995) “for an outcome to be consistent with the welfarist approach [...] it must be consistent with a theory of utility [and in a Paretian approach] the method of measuring benefits must accurately reflect the individual’s preferences even if these are not the preferences we feel they should have” (p.768).

ARSs have not been widely used for research data collection or theory testing (McCarter and Caza, 2009) even though they have been used to establish educational competencies in nursing (Kirk et al., 2004). This technology might be preferred over other more widely used techniques, such as questionnaires, because of its time- and cost-saving characteristics. This technology presents several other advantages, such as the ability to feed responses into the ongoing presentation, and incorporate ranking
options. This technology could be used in a future preference-elicitation and (dis)benefit valuation exercise incorporated in the stages of a CBA or DCE evaluation.

Preferences and specifically stated preferences have been traditionally explored using written survey methods such as DCEs. DCEs which have gained increased popularity in health economic research investigate preferences for identified characteristics or attributes of a given phenomenon i.e. healthcare services as well as the strength of preferences in order to investigate what users want more from a given service in addition to the relative value and how much value they place on a number of attributes (Ryan and Gerard, 2005). One problem with this approach however is its limitations in the number of attributes possible to be presented at each experiment due to the cognitive limitations of human memory (Smyth et al., 1994; van Helvoort-Postulart et al., 2009a; van Helvoort-Postulart et al., 2009b). This research attempted to address this issue in the context of ranking by presenting consecutively each category and asking attendees to rate each category in turn, thus allowing for indirect ranking.

Results from the pilot study suggest that by using this technique, attendees are able to differentiate between the categories presented. This conclusion is based on the differences in perceptions as to the importance of each category of (dis)benefit and the differences in the range of responses for each category. Methodological research investigating different approaches for dealing with large numbers of attributes in stated preference investigations, have illustrated that presenting attributes consecutively in the form of a questionnaire has limitations (van Helvoort-Postulart et al., 2009b). Van Helvoort-Postulart and colleagues have compared the use of a questionnaire to that of a DCE using Hierarchical Information Integration (HII) techniques to deal with large
numbers of attributes, and have concluded that a DCE is superior since participants were not able to differentiate between variables in the context of the questionnaire, thus giving similar ratings to all attributes (van Helvoort-Postulart et al., 2009b). Different rankings were obtained by using the DCE technique and the questionnaire (van Helvoort-Postulart et al., 2009b). This points out to the importance of the presentation context i.e. the way an attribute is presented to participants. In the present study categories of (dis)benefits were presented by asking attendees to consider each category as to its perceived importance in relation to using the CGS, to them, their family or to individuals using the service to which they were associated. Van Helvoort-Postulart and colleagues on the other hand, exploring barriers and facilitators to implementing change, translated the attributes used in the DCE into statements expressing preconditions to change and expectations of the realisation of given barriers to change in actuality. Participants were asked to state whether they agreed or disagreed with the given statements. The authors conclude the limitations of using the questionnaire based on the calculated mean and median of responses which was similar for all attributes (van Helvoort-Postulart et al., 2009b). In the present research it was not possible to conduct statistical analysis of results due to very small numbers of attendees. It is acknowledged that given larger numbers of attendees results might have reflected the ones presented in the van Helvoort and colleagues study.

ARSs present possibilities for alternative uses within a stated preference context. The ability for face-to-face interaction with stakeholders might present opportunities for feedback as to why participants have made the given choices, whereas this investigation would have been part of cognitive debriefing at a subsequent stage in the
case of pen-and-paper assessment [debriefing has been described to be part of a “good” contingent valuation study (Carson, 2000) and has been described to take place in DCE studies e.g. Lloyd et al. 2007].

Another issue relevant to the ARS approach is the question of whether automatic feedback and the ability to view or listen to the attitudes of other stakeholders while contributing to the interaction, might affect responses. In this sense ARSs present similarities with focus groups, where group interaction is acknowledged to affect participant responses but this is not perceived as a negative attribute of the method. One advantage of the ARS technique is its anonymous nature, where even though attendees can see responses, they don’t know to whom each response belongs. One implication however is that with large numbers of attendees, elements such as cognitive debriefing might be difficult to manage. In the current study attendees were given the opportunity to comment on the event and one possible solution to this problem might be to ask attendees to comment on their decision-making in a similar comment-sheet following each stated choice.

Several difficulties have been identified however in the organization of such an event. The time saving, fast and interactive character of this venture is offset by the intensive need for preparation. Advertising the event does not make up for direct and face-to-face recruitment through involvement of all service providers and patient support organizations upon which recruitment depends, as was evident through this research. Arguably, the overall process might not offer time-savings over other pen-and-paper methods. However, it offers the possibility of fast data-collection when networking has
been made and within a diverse setting and population, i.e. data collection during patient organization conferences or as part of staff conferences.

Using this technique nine consequences were identified (table 7.4) based on indirect ranking of 14 contributors. These were “continuity of care”, “access to genetics experts”, “being offered choices”, “research updates”, “levels of knowledge and education”, “support with decision-making”, “access to other NHS services” and “clinical outcomes”, and “empowerment to make decisions” relevant to their genetic situation. These categories reflect themes proposed in section 6.5.3. Clinical outcomes, which have been debated to be inappropriate outcomes with which to evaluate CGSs, have emerged in this phase among the (dis)benefits perceived as most important in relation to the service. Findings have led to the suggestion that health improvements are hoped for by service users and drive their search for information and subsequently access to the service. Low ratings of emotional outcomes provide some support to the hypothesis that service users are willing to endure some emotional distress if their access to the service will help them deal with the clinical and other practical implications of the condition (Ardern-Jones, Kenen and Eeles, 2005).

The emerging significance of “continuity of care”, defined as “receiving care over time from a particular service” is an aspect of the service missing from service evaluation but one perceived as most important by participants in the present study. Attributed importance of longitudinality of contact with the service is linked to long-term follow-ups of the service pointing to expectations of service users for service delivery. Preferences for service delivery alternatives might be useful to consider when thinking of appropriate models of service provision which could improve user outcomes. Within the literature it
has been acknowledged that it is important for research to provide empirical evidence about alternative ways of providing genetic services which can inform service development (Holloway et al., 2004).

Debate within the literature about alternative models of service provision for mainly cancer genetic services has been present, (e.g. Holloway et al., 2004; Iredale et al., 2007), but still the main focus has been on the delivery of cancer genetic counselling rather than overall service provision (e.g. Brain et al., 2002; Brain et al., 2005; Apicella et al., 2006). In previous research Brain and colleagues have identified that risk information about self and family along with risk reducing options rank high among women’s expectations of cancer genetic services (Brain et al., 2000b). Arguably, alongside the information and counselling needs of women which need to be taken into account for service development (Brain et al., 2000b), the specific needs in terms of service delivery pathways also need to be taken into account.

Surveillance of families in the form of continuing contact with the service is a stated role of the service (Fryer and Cheese, 1998) and a valued benefit by service users, yet other elements are also valued and expected to be provided such as multidisciplinary working to ensure appropriate care is in place. A multidisciplinary approach to cancer service provision where women access oncologists and cancer screening has been frequently mentioned in the cancer genetics literature (Lynch et al., 1993; Brain et al., 2000b; Holloway et al., 2004), yet features such as continuity of care and access to other healthcare specialists have not been frequently raised for other genetic conditions in the context of genetic services.
“Knowledge” about the genetic condition, “decision-making”-related outcomes and having “choices” are outcomes which have received considerable attention in the literature. In a similar study where the views of service users and providers were compared using a Delphi study, “knowledge about the genetic condition” and “perceived personal control” attracted the highest consensus as suitable outcomes of CGSs (Payne et al., 2007). Other outcomes perceived as suitable by more than 75% of participants in this study were: “ability to cope”, “satisfaction with service”, “meeting of expectations”, “accuracy of diagnosis”, “decision-making”, “quality of life” and “perception of risk”. Similar to the present study, emotional outcomes included in the Delphi study like anxiety levels and depression did not reach the 75% cut off point for establishing consensus even though more service users than providers thought of them as important (Payne et al., 2007).

Despite the fact that emotional outcomes have been the most popular for the evaluation of the study, both past literature and the current study suggest that they might not be the most appropriate outcomes for evaluating genetic services. A review of the psychosocial literature has indicated that emotional consequences such as anxiety and depression are more associated with individual, social and temporal variables than actual service provision. More recent studies suggest that coping-promoting (dis)benefits might constitute a more appropriate way of assessing the service and arguably reflect more accurately its stated role. Findings from the present study also suggest that a more specific description of the role of CGSs might need to integrate the multi-disciplinary approach of the service and its role in establishing patient pathways, involvement in patient care and family follow-up not only for the purpose of disease
prevention but for keeping families informed, addressing new concerns and enabling continuous access to the service for the whole family. The emerging preferences of contributors to this phase suggest that CGS should be understood as more than a short-term genetic-counselling providing service.

7.7. CONCLUSIONS

Fourteen individuals attended a half-day event aiming for respondent validation and transferability of findings and pilot testing ARSs for their use in preference research. There was consensus for all of these categories with the majority of attendees agreeing that these were important to CGSs suggesting respondent validation and some transferability of the (dis)benefits to stakeholders other than the ones taking part in the previous phase. This pilot phase suggested some interesting trends in the preferences of CGS stakeholders. Among the five attributes attracting highest levels of agreement among attendees none were emotional outcomes. “Clinical outcomes”, which is a controversial outcome within the literature, in this phase, emerged among the nine benefits perceived as most important. Even though the direct involvement of the CGS as a whole in producing clinical outcomes is limited, it could be that some users may perceive a relationship between accessing genetic services and clinical outcomes. Further research is needed to better understand the perceptions of service users on what they understand to be the relationship between accessing genetic services and clinical outcomes.

ARSs appear to allow management of a large number of attributes within a stated preference exercise, unlike DCEs, and enable face-to-face interaction for explaining the
process of making choices in the context of such exercises and enabling debriefing. Their flexibility and capability in presenting variables whether by direct or indirect ranking and also their capability for making comparisons between variables contributing to broader concepts calls for further investigation for their use within preference elicitation and outcomes research. Comparisons with other popular techniques such written DCEs will enable further exploration of their validity. However, the presence of a large number of participants might make such endeavours difficult to manage and recruitment problems might prove an inhibiting factor to the success of the event during the day.

The next and final chapter will provide an overview of the findings from the three phases and discuss the methodological implications of incorporating the proposed (dis)benefits in an economic evaluation.
CHAPTER 8

A FRAMEWORK FOR ECONOMIC EVALUATION

CONCLUSIONS
8.1. Overview

This chapter will:

1. Provide an overview of the background to the research problem;
2. Reiterate the aims and objectives with an overview of how these objectives were achieved;
3. Discuss methodological issues emerging from the present research related to the economic evaluation of CGSs;
4. Discuss limitations of the research;
5. Present a framework incorporating knowledge necessary for the first stages of CBA design;
6. Discuss directions for further research.

8.2. Background and aims of the research

The economic problem of limited resources coexisting with virtually infinite demand for health care has resulted in a widespread understanding that only the most efficient as well as effective interventions should be provided to the population. Effectiveness of health care interventions and in particular of pharmaceutical interventions has been a focus of health care providers across the world, with evidence-based practice being an integral part of medical and health care provision. Economic evidence forms one aspect of the evidence required by evidence-based practice and are now common in evaluations of health care interventions.
Economic evaluations have gained in popularity with the two most frequently used techniques being cost-effectiveness (CEA) and cost-utility (CUA) analyses; as opposed to the traditional cost-benefit analysis (CBA) which forms the operationalisation of traditional economic theories and is used in other branches of economics (Drummond et al., 2005). This is mainly because the characteristics of health and health care make the requirements of traditional CBAs, namely to identify all costs and benefits on society and express these in money values, problematic. In the case of healthcare a CBA has been viewed as a time-consuming, complicated and controversial method of assessing efficiency. CEAs and CUAs allow for the evaluation of interventions in terms of their production efficiency and they assess whether interventions manage to achieve a given objective with the least possible use of resources as compared to the status quo. Health-related outcomes such as life years gained are the desired outcome assessed by a CEA while health related quality of life is the objective assessed by a CUA.

Health economists have recently raised questions about the suitability of these two approaches to realistically capture the efficiency of health care services and specifically their allocative efficiency where the issue is not about the most appropriate way to provide a given service, but whether that service should be provided in the first place, given the economic problem of opportunity cost (Mooney, 1998). One major problem with these two approaches relates to the outcome measures they use to capture benefit, while some economists have argued that health-related outcomes are not able to capture the whole spectrum
of benefits from complex healthcare interventions (Coast, 2004), a good example of which is clinical genetic services (CGSs).

The focus on economic evaluations which incorporate health-related outcomes means that consequences which are now understood to be more relevant to the activity of genetic services, such as the value of information, the impact on the broader family, reassurance and more informed decision-making, cannot be taken into consideration (Mooney and Lange, 1993; Hall, Viney and Haas, 1998; Griffith, Edwards and Gray, 2004; Wang, Gonzalez and Merajver, 2004; Jarrett and Mugford, 2006), making these evaluations of limited use to decision-makers (Jarrett and Mugford, 2006). Rapid and ongoing developments in molecular genetics and the subsequent increased allocation of resources for the expansion of CGSs brings the appropriateness of such investments into question, considering firstly that money invested for CGSs is forgone from other services, health or other, and secondly the lack of comprehensive evaluations on the overall benefits of these services on society.

This thesis has argued for the need of a CBA assessing the full impact of CGSs but at the same time recognising the difficulties involved in such an endeavour. One such difficulty is identifying the necessary parameters for future economic evaluations or economic decision models because of the limited understanding of both the precise patient pathways, and consequently, the overall tangible and intangible (dis)benefits of these services. In the UK there are several genetic centres, all of which adhere to specific general guidelines on what should be
provided by a genetics service, but insufficient evidence exists in the literature on the detailed patient pathways and use of resources.

8.3. Framework for the design of an economic evaluation

In order to develop the proposed framework, semi-structured face-to-face interviews and focus groups, alongside a quantitative phase were used within a case study approach. This framework is presented in Figure 8.2 p.328. In the first chapter the following elements were reported to be incorporated in the proposed framework:

- Aims and objectives of genetic services and the patient journey or pathway followed by those referred to the CGS of a UK clinical genetic service;

- Tangible impact to other agencies (NHS or otherwise) where access is facilitated by the CGS and where the patients and their families would not be guaranteed access if they did not receive the services of the clinical genetic service;

- Psychosocial (dis)benefits resulting from the consumption of the CGS;

- An example of how these costs and consequences would look in a CBA and within a Discrete Choice Experiment (DCE), a welfarist technique allowing for (monetary) welfare values to be attached on the identified (dis)benefits.
In order to contextualise economic evaluations in the realm of CGSs, Chapter 2 presented the economic theoretical background to economic evaluation and set the ground for what is required for CBA to take place. This thesis has directly dealt with two prerequisites for CBA, namely the context of CGSs and identifying the (dis)benefits which need to be incorporated in the evaluation.

Based on the preparatory requirement of having clear knowledge and understanding of the structure and context of a given service in order for it to be evaluated, Chapter 2 also presented what is currently known of the aims and patient pathways of UK genetic centres concluding that a detailed understanding of what each patient experiences when referred to a UK genetic centre is lacking.

Benefits used in economic evaluations were reviewed in Chapter 2 where it was concluded that outcome measures used so far have not incorporated the overall benefits of a CGS which have been argued to be mainly psychosocial in nature. Chapter 3 reviewed popular psychosocial outcomes used for the evaluation of CGSs, and concluded that existing outcome measures do not appear to be suitable to be incorporated in a comprehensive economic evaluation. The methods and research design utilised for primary data collection and analysis in the present thesis were described in Chapter 4.

The following section will present the knowledge obtained through the three phases of data collection and the evidence collected for each of the above elements of the proposed framework (Chapters 5 to 7).
8.3.1. Aims and role of the CGS

What service providers perceive to be the role of the service arguably influences service provision. The two most frequently cited aims of the CGS were: 1) to provide accurate information and enable informed decision-making, and 2) to ensure that patients get the clinical management and social or other support required. Genetic services in the case of the chosen CGS appear to be closing a gap in the care of individuals affected by a genetic condition to ensure that appropriate treatment is in place. Impact on health outcomes in this sense takes place indirectly and is a result of the expert understanding of genetic conditions by service providers when trying to meet the needs of patients for managing their symptoms.

Establishing patient pathways and ensuring that patients get the clinical interventions required to minimize negative health implications has not been an explicitly stated aim of the service, even though the new UK Specialised Services National Definitions for Medical Genetic Services has been more clear about the role of CGSs in providing appropriate follow-up and ongoing support (DoH, 2007). This aspect of service provision has not featured in evaluations of CGSs either, with evaluations focusing on genetic counselling or genetic testing. However, this aspect of genetic service provision has emerged as an important one to service providers of the chosen CGS. Even though it is not as yet clear in what ways service provision differs between UK genetic centres particularly in providing disease management support and how carriers, patients and high risk individuals are dealt with by the healthcare service including the CGSs, research
has suggested that practices may vary depending on the area in which they are located (Brain et al., 2003; Foster et al., 2007). Service providers participating in this study perceived considerable variability to be present among centres in terms of providing follow-up, support and co-ordination of health surveillance. This variability seems to depend on manpower and financial resources and arguably on the outlook on the service i.e. the service’s perceived role and service objectives.

8.3.2. Patient pathways within the CGS

Genetic counselling and access to genetic testing formed the core of the service and were common features in the pathway of the five groups of conditions. Over and beyond these elements however the patient journey appeared to be influenced by individual and family characteristics including the nature of the genetic condition and its severity. The degree of involvement of the CGS professionals depended on their personal assessment of the individual needs of families and the availability of clinical support from services outside the CGS. Where individual cases required medical surveillance and this was not offered by mainstream services then this was provided by the CGS. Access to the service was either reactive or proactive and this was based on the needs and demands placed by the condition. Continuity in contact with the service was encouraged by the professionals in order to ensure that the needs of families were met.

Use of resources therefore depended on the individual needs of patients and families to deal with the implications of the genetic condition, the availability of clinical services outside the CGS which could affect patient outcomes and by the
special interests of the service. Linked to special interests are the perceptions of the genetic centre as well as of individual providers of what the service is aiming to provide to the families referred. Based on discussions with the providers and with the director of the CGS, the centre appears to differ from other centres in its emphasis on the counselling aspect of the service in addition to the medical and/or educational aspect i.e. not simply imparting information, but encouraging more personal involvement, striving to support families, and promoting emotional adjustment and a sense of control.

8.3.3. Use of resources other than the CGS

There appeared to be both ad hoc and structured links and referrals from the CGS and other health, social or other services. These services were listed in Box 5.7 in Section 5.6.

The biggest impact from CGS use appears to be on secondary care specialties where patients are referred in order to receive needed clinical treatment. Again, these links can be structured as in the case of prenatal genetics and Obstetrics and Gynaecology departments or ad hoc where women at high risk for cancer are referred to surgical services for prophylactic surgery.

Families are also assisted in dealing with social services and obtaining needed support through being explained the benefits they are entitled to, what services are in place and also with writing letters about their circumstances. Families are also referred to or given information about NGOs and voluntary support groups which are also able to provide practical or emotional support to families.
Secondary care specialists, social services and NGO’s including voluntary support groups are the three types of services discussed to be used most often by genetic professionals.

Other services include cancer screening services, Occupational Therapy, Physiotherapy, other genetic centres, community specialists, psychologists and educational services.

**Box 8.1: Use of resources other than of the Clinical Genetic Service**

| Secondary care specialists (HBOC; VHL; TS; CF and prenatal genetics) |
| Social Services (TS; DBMD; CF and prenatal genetics) |
| NGOs and support groups (TS; DBMD; CF and prenatal genetics) |
| Occupational Therapists (DBMD) |
| Physiotherapists (DBMD) |
| Community specialists (TS) |
| Psychologists (TS) |
| Educational services (TS) |
| Cancer screening services (HBOC) |
| Other genetic centres (VHL) |

**8.3.4. (Dis)benefits of the CGS**

Qualitative research with both service providers and service users has illustrated how service related attributes are also sources of utility since these took up considerable part of the discussions and were what participants came up with when initially asked to describe the benefits of the service. The utility-bearing nature of service attributes was also illustrated in the quantitative part of the
research (Phase Three) where categories representing elements of service provision were rated as more important than other categories of (dis)benefits by participants and non-participants of the qualitative part of data collection.

In-depth analysis of the narratives of the service users led to the identification of process-related attributes and psychosocial outcomes which represent what users perceived as the most important (dis)benefits of the CGS and which contributed towards both their individual and their family’s utility. Process related attributes are shown in Figure 8.1 to contribute towards four psychosocial outcomes which in turn are dimensions of the broader outcome of Perceived Familial Control. These benefits are shown to interact with each other illustrating the long-term process that takes place in order to sustain levels of Perceived Familial Control and the interplay between process and outcome, where families go back in and out of the service as and when they require additional support.
Process attributes which emerged as important sources of utility for participants were:

1. The expertise of the service in genetic conditions,
2. The individualised approach to service provision,
3. The interdisciplinary problem-based approach to care,
4. Accessibility for all family members over time,
5. Family-encompassing focus of the service.
Psychosocial outcomes have also been proposed through the use of qualitative analytic techniques necessary for exploring meaning and interpreting participant narratives. The overall outcome emerging through the analysis was a Perceived Familial Control, an outcome encompassing four dimensions and which is linked to the coping abilities of family members. This outcome represents the psychosocial aspect of CGSs, an element largely missing from considerations of the utility function of CGS consumers.

Themes proposed in the second phase of data collection (Chapter 6) gained validity following respondent validation and investigation for transferability in Phase Three which constituted the quantitative part of data collection (Chapter 7). “Continuity of care”, “access to genetics experts”, “being offered choices”, “research updates”, “levels of knowledge and education”, “support with decision-making”, “access to other NHS services” and “empowerment to make decisions” were categories rated most important by attendees in Phase Three.

### 8.3.5. Some comparisons between findings

Chapters 5 to 7 presented and discussed findings from each consecutive phase of data collection. During data collection in Phase One service providers were asked about their perceptions of benefit resulting from their contact with families. These issues were explored in more depth with service users in the second phase of data collection reported in Chapter 6. Chapter 7 attempted to bring together stakeholders including providers and users in order to explore their preferences in what they perceived to be the most important benefits. Here some tentative comparisons will be made between the findings in the different
stages of data collection, focusing on the data collected in Phases One and Two, using findings from Phase Three to attempt to make some preliminary conclusions. Comparisons are tentative because of the differences in analysis between the two sets of data based on the objectives of each phase, where provider narratives were descriptively reported whereas user narratives had undergone in-depth analysis.

The most striking similarity in benefit perception between providers and users is the central role of information about all aspects of the disease and the understanding by families and patients of this information. Following on from this basic genetic counselling focus, almost equal mention was given by providers and users on the ongoing support in the form of yearly follow-ups, proactive recall of families into the service, management clinics, establishing patient pathways, facilitating contact with other health specialists, and interventions from other support services including social services and educational or psychological interventions. Emphasis placed however on these two benefits appears to differ between users and providers. This is suggested by the importance placed by users on the benefit “continuity of care” in Phase Three, whereas the majority of providers raised outcomes falling within the theme of knowledge, understanding and decision-making as the most appropriate outcome measures upon which to evaluate the service they provided. The contested place of continuity of care, which might include proactive follow-ups, clinical and social support, and occasional phone calls by the service, in the remit of the service, might explain the differences in consensus of what are the important outcomes of CGSs
between providers and users. Even though the vast majority of providers perceived as important the long-term relationship with families, the personal contact and the open-door policy promoted by the service, not all agreed on whether this aspect of service provision was part of the role of a CGS. One provider even suggested that they were providing clinics and clinical monitoring to patients even though they did not think they should be providing such services. The majority of providers raised the point that this specific CGS differed from other genetic centres in the level of proactive follow-up and personal contact with families. One provider thought this is not part of the role of a CGS and it should be ceased in the following years, mainly due to budgetary cuts.

These differences among the important outcomes of the service alluding to possible differences in the perceived role of the service among providers and users was evident in the only study explicitly comparing provider and user perceptions of the outcomes of CGSs. In the study by Payne and colleagues knowledge about the genetic condition had also received equal levels of significance by providers and users as an important outcome of the service whereas health status defined to incorporate the ability to take care of oneself, ability to perform day-to-day activities, anxiety and depression was perceived as far more important by users than providers (Payne et al., 2007). What is interesting is that in the same study the ability to cope with living with a genetic condition was perceived as more important by providers than users even though consensus was achieved in both groups as to its importance for evaluating the service (Payne et al., 2007). These differences arguably reflect the conclusions
suggested in Chapter 7 that users might ultimately aim for improved health outcomes through their access to the CGS.

One benefit which has been attributed more importance by users than by providers was the familial element of service provision. Even though some providers did raise this attribute of the service it was not as highly thought of as by users. Again, the same finding was described in the Delphi study by Payne and colleagues (Payne et al., 2007).

Overall, there was general agreement between the perceived role of CGSs described by providers, the benefits they raised and the experience of CGSs described by users, reflected in the perceptions of provision of information about the condition and about options of dealing with the implications and disease management support as important by providers and service users. What is of interest however, is the ad hoc nature of disease management support provided by the service. A number of studies have so far raised the importance placed by users of CGSs on disease management support through referrals to other professionals, follow-up letters informing of new clinical and genetic advances, as well as supporting them through social implications of the disease (Macleod, Craufurd and Booth, 2002; McAllister et al., 2008b; McAllister et al., 2008a) illustrating the importance of providing multidisciplinary genetic services (Holloway et al., 2004). However, it is not yet clear as to what exactly is the role of the service in supporting families to deal with the condition, other than providing genetic counselling.
Based on the definition of clinical genetic services, genetic counselling is but one of the constituent parts of this service (DoH, 2007), and participants described aspects of service provision that might not traditionally be considered as genetic counselling e.g. intervening in setting up care pathways. It might be appropriate to clearly distinguish between what one refers to by genetic counselling and by genetic service provision. For example, following genetic counselling service users are free to re-enter the genetic service for any concerns, questions or guidance, while the genetic service might offer follow-ups through telephone, mail, or face to face contact. Is this defined as genetic counselling? In addition, the CGS may set up clinical pathways based on individual needs and circumstances, or oversee the clinical pathway and clinical management of individual patients, something that is also stated as part of the clinical geneticist’s role (Godard et al., 2003). Again, is this part of genetic counselling? It is the overall package which has been found to result in Perceived Familial Control, rather than the one or a consecutive series of genetic counselling sessions following referral. In this sense it is difficult to separate process benefits from outcome benefits when the two are interlinked in the users’ experience, and when there is variability in process between genetics centres. Genetic counselling remains the one constant element of CGSs while other elements such as coordination of care vary according to individual circumstances and the outlook of individual centres.
8.3.6. Practical applications of the framework within welfarist methodologies

8.3.6.1. Outline of a cost-benefit analysis

A cost-benefit analysis presents costs and (multiple) consequences of an intervention and involves valuing each in money terms. Presentation of the full range of costs and benefits without benefit valuation – referred to as “costs consequences analysis” – does not allow conclusions about allocative efficiency to be drawn, but still assists decision makers by presenting all natural units which allows them to attach their own values (Greenhalgh, 1997; Coast, 2004). It has been argued that cost-consequences analysis is a more appropriate form of economic study than CEAs and CUAs because it allows for outcomes other than the limited-in-perspective health-related outcomes to be taken into consideration and is arguably more meaningful to decision makers (Coast, 2004). In the case of cancer genetic services, Cohen and colleagues carried out a cost-consequences analysis which they argued to be the most appropriate approach to the economic evaluation of these services. This is because of the limited knowledge of overall implications and the inability of existing outcome measures to capture psychosocial outcomes believed to be associated with service provision (Cohen, Barton and Brain, 2004).

In order to illustrate how the present research has contributed towards knowledge that can be used for a comprehensive economic evaluation, costs of a clinical genetic service for women with a family history of breast cancer as presented in Cohen et al are presented below, in addition to the additional costs
and consequences arising from this research, albeit without an alternative scenario comparison. The identified (dis)benefits can be incorporated in a cost-benefits analysis, following valuation based on the use of approaches which incorporate a willingness to pay (WTP) element. Sources of utility other than outcomes which in this case involve process-related characteristics will be listed in the consequences column, even though “consequences” is not the technical term used in the present thesis to describe these (dis)benefits (look at Section 1.2. for a guide to terminology).
### Table 8.1: Outline of a cost-benefit analysis

<table>
<thead>
<tr>
<th>Costs</th>
<th>Consequences</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Use of CGS resources</strong></td>
<td><strong>Main Psychosocial Outcome</strong></td>
</tr>
<tr>
<td>*Cost per initial consultation per woman</td>
<td>Perceived Familial Control</td>
</tr>
<tr>
<td>*Cost per <em>preliminary clinic</em>/woman</td>
<td>Knowledge and Understanding</td>
</tr>
<tr>
<td>Cost per initial consultation per patient/family</td>
<td>Informed and shared decision-making</td>
</tr>
<tr>
<td>*Cost per mutation search</td>
<td>Reassurance</td>
</tr>
<tr>
<td>*Cost following identification of a mutation in an affected relative</td>
<td>Enablement</td>
</tr>
<tr>
<td>Subsequent costs per family plus other relatives consultations</td>
<td></td>
</tr>
<tr>
<td>Cost per counselling and testing per patient</td>
<td></td>
</tr>
<tr>
<td>Cost per counselling and testing family members</td>
<td></td>
</tr>
<tr>
<td>*Cost of tracing, counselling and testing living affected relatives</td>
<td></td>
</tr>
<tr>
<td><strong>Service user resources</strong></td>
<td><strong>Other consequences</strong></td>
</tr>
<tr>
<td>*Cost per woman (e.g. time, travel)</td>
<td>Insurance issues</td>
</tr>
<tr>
<td>*Subsequent costs to presenting woman plus other relatives (e.g. time, travel)</td>
<td>Health outcomes</td>
</tr>
<tr>
<td>*Cost per family (e.g. time, travel)</td>
<td></td>
</tr>
<tr>
<td>Subsequent costs to presenting patient/family plus other relatives (e.g. time, travel)</td>
<td></td>
</tr>
<tr>
<td><strong>Use of non-CGS resources</strong></td>
<td><strong>Other sources of utility</strong></td>
</tr>
<tr>
<td>Cost of referrals and initial consultations with secondary care specialists e.g.</td>
<td><strong>Process attributes</strong></td>
</tr>
<tr>
<td>Breast/Plastic surgeons</td>
<td>Expertise</td>
</tr>
<tr>
<td>Neurologists</td>
<td>Individualised approach service provision</td>
</tr>
<tr>
<td>Ophthalmologists</td>
<td></td>
</tr>
<tr>
<td>Cost of follow-up care by secondary care specialists</td>
<td>Interdisciplinary problem-based care</td>
</tr>
<tr>
<td>Cost of referrals and use of social services</td>
<td>Accessibility over time</td>
</tr>
<tr>
<td>Cost of surgical procedures</td>
<td></td>
</tr>
<tr>
<td>Cost of screening interventions</td>
<td></td>
</tr>
<tr>
<td>Cost of use of NGOs/voluntary support groups</td>
<td></td>
</tr>
<tr>
<td>Cost of referrals and consultations with non-medical health specialists</td>
<td></td>
</tr>
<tr>
<td>OTs</td>
<td></td>
</tr>
<tr>
<td>Physiotherapists</td>
<td></td>
</tr>
<tr>
<td>Cost of referrals and consultations with other specialists</td>
<td></td>
</tr>
<tr>
<td>Psychologists</td>
<td></td>
</tr>
<tr>
<td>Educational services</td>
<td></td>
</tr>
</tbody>
</table>

Costs labelled with * are costs presented by Cohen et al (2004).
8.3.6.2. Applying the (dis)benefits in a Discrete Choice Experiment

The previous section had presented the identified benefits and costs within the context of a CBA. In order for an actual CBA to be designed however, (monetary) welfare values need to be attached to the identified benefits. These could then be compared to the costs identified and listed in Table 8.1. A DCE is an appropriate vehicle towards the elicitation of welfare and monetary values of a set of attributes, health and non-health outcomes. This section will attempt to present the identified (dis)benefits within a DCE framework.

Lancsar and Louviere describe three main inter-related stages to the design of a DCE, and this section deals only with parts of the first of the three stages stated above, namely defining attributes and levels, and conceptualising the choice process in order to illustrate how the proposed (dis)benefits would look in a DCE (Lancsar and Louviere, 2008). It is acknowledged that the attributes used here need further refinement since they are based on researcher interpretations and the terms or definitions used may not reflect service user interpretations. Further research is required to ensure that the terms used to describe the given (dis)benefits also reflect user interpretations (Coast et al., 2011). An opt-out option rather than the status quo is presented as the alternative to the proposed scenario since in the case of CGSs there is no overall consensus on the way these services should be provided or upon the aims of the service.

Two different choice sets are presented below as examples of a discrete choice experiment, incorporating a closed-ended willingness to pay component (Drummond et al., 2005). Deciding the cost vector requires detailed attention
including the range of bid amounts to include, and in a real WTP study tests of construct validity could be undertaken to provide some validation to the findings (Drummond et al., 2005). For this example four levels to the cost attribute are given even though in a real life DCE a broader range to the cost attribute levels would need to be considered in order to reflect real values.

Even though it is possible to combine process and outcome attributes within a single choice set, here process and outcome are presented in different choice sets. This is because of the interplay between process and outcome in the given model of Perceived Familial Control. Psychosocial outcomes are presented as a result of the process attributes of the CGS, while emerging through the iterative process of analysing and attaching meaning to narratives of CGS experiences. In addition, additional research is required to explore how individuals process choices between process and outcome and if these choices are meaningful given the interplay between the two, i.e. what is the process of decision-making when individuals are given the choice between e.g. expertise and enablement?
Table 8.2: Example of a choice set within a traditional DCE (process attributes)

Which type of service do you prefer?

<table>
<thead>
<tr>
<th>Alternative A</th>
<th>Alternative B</th>
<th>Alternative C</th>
</tr>
</thead>
<tbody>
<tr>
<td>The service provider has some experience in clinical genetics</td>
<td>The service provider is an expert in clinical genetics</td>
<td></td>
</tr>
<tr>
<td>Service provided does not take into consideration the needs and abilities of the individual/family</td>
<td>Service provided always takes into consideration the individual needs and abilities of the individual/family</td>
<td></td>
</tr>
<tr>
<td>Service providers have formal links with services involved in disease management and can set up links with other services if there is a need</td>
<td>Service providers may set up links with services involved in disease management and may indicate other services that can provide support</td>
<td>OPT OUT OF THIS SERVICE</td>
</tr>
<tr>
<td>The service makes contact with you through follow-up phone calls and update letters but you and your family do not have contact details for initiating contact</td>
<td></td>
<td></td>
</tr>
<tr>
<td>When you access the service any services provided to you are also available to your immediate family, for example family members are allowed to attend sessions with you even if they have not been referred and your family's needs are addressed</td>
<td>Genetic services are only available to individuals and other members of your family will need their own referral in order to access the service.</td>
<td></td>
</tr>
<tr>
<td>Cost of the service: £250</td>
<td>Cost of the service: £100</td>
<td></td>
</tr>
</tbody>
</table>

Which alternative service do you prefer? □A □B □C

One disadvantage of this DCE design, is the cognitive burden placed on respondents from having to mentally process and simultaneously make trade-offs between the given levels and attributes (Lancsar and Louviere, 2008) and its inability to address attribute impact issues i.e. differences in utilities between attributes (Flynn et al., 2007).
A new development in DCEs in healthcare evaluation is best-worst scaling (Coast et al., 2006; Flynn et al., 2007; Coast et al., 2008; Flynn et al., 2008). With this approach a number of different scenarios are presented to participants individually, as opposed to the traditional DCE where they involve a comparison between two or more alternatives paired together. Each scenario is made of a set of attributes at different levels and the task is for respondents to choose the best and the worst option (e.g. attribute) for each of the scenarios (Coast et al., 2006). In order to obtain information on the attractiveness of the whole scenario compared to the respondent’s current position, respondents are also asked to accept or reject each scenario (Flynn et al., 2007).

One point to note is not including a cost attribute within the choice set. In the case of the best-worst scaling approach, presenting the willingness-to-pay question separately was decided to be more appropriate for eliciting a monetary value for the presented scenarios. Again, testing the methodological validity of such a design would need to be tested prior to conducting a DCE study.
Table 8.3: Example of a best-worst scaling choice set (psychosocial and health outcomes)

This is a hypothetical scenario following your contact with the CGS. You are asked to choose between these alternatives which attribute is the best and which the worst within this scenario.

<table>
<thead>
<tr>
<th>Best outcome</th>
<th>One example of a hypothetical scenario following your contact with the CGS</th>
<th>Worst outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>You feel that you have been provided with some but not all of the information that you wanted and you fully understand and feel confident to communicate this information to other family members</td>
<td></td>
<td></td>
</tr>
<tr>
<td>The genetics provider directs you towards the “best” options about how to deal with your risk and/or the condition and you follow their advice</td>
<td></td>
<td></td>
</tr>
<tr>
<td>You and your family are confident that you can cope with the tangible (health and social) aspects of the genetic condition and feel able to access needed support</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Thinking of the genetic service, there is peace of mind most of the times and you sometimes feel worried about the implications of the genetic condition</td>
<td></td>
<td></td>
</tr>
<tr>
<td>You improve by 25% all adverse health outcomes associated with the genetic mutation</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

If you were able to choose, would you choose to remain a patient of this specific genetics centre?  

Would you choose to use this service if you had to pay £400?

The advantages of best-worst scaling over traditional DCEs is that it allows for the estimation of the importance of entire attributes, rather than just the levels of these attributes, and also that the task of choosing the best and worst attribute from one scenario appears to be a less cognitively demanding task than the traditional forms of DCE (Coast et al., 2006). This alternative form of DCE is chosen for the second choice set incorporating the psychosocial outcomes.
identified in the research. The reason for this, are the complexities inherent in a task asking respondents to process and make choices over a set of intangible, psychosocial outcomes, presented in alternative scenarios and described according to their different levels. This best-worst scenario presents the non-health outcomes of CGSs, along with a health outcome and cost attribute. This will allow investigation of the respondent’s trade-offs between health and non-health (psychosocial) outcomes. Health outcomes have so far been considered as irrelevant to the service. However, Phase Three of this study has suggested that health may be of importance to those using the service; yet defining a health attribute is especially difficult. Genetic conditions have both within and between variability in disease expression or phenotype, thus defining an overall health outcome for CGSs is almost impossible. Even if one chooses to use disease-specific scenarios for health improvements, the question remains of what do users think of when they consider health improvements in the context of CGSs. In addition, the characteristics presented here need further refinement for ensuring that they are clear enough for respondents to understand and that levels are accurately defined and are different enough to make it possible for respondents to make a choice of one over the other (Lancsar and Louviere, 2008; Coast et al., 2011). As raised previously, the perceptions of users of CGSs of what they understand as a health outcome and whether they implicitly expect improvements in health outcomes and that is why they want to access CGSs needs further exploration. Research so far has identified a number of psychosocial factors such as the need for information as main reasons for
accessing CGSs (Brain et al., 2000b). It might be the case however, that a Perceived Familial Control (PFC), emerging as the overall benefit people enjoy from being part of the CGS, may act as a means towards health benefits i.e. families are better able to deal with the negative tangible e.g. clinical symptoms, and intangible e.g. uncertainty and lack of information, implications of the genetic mutation thus better able to deal with the health implications, indirectly leading to improved health outcomes.

In order to understand the decision-making processes and preferences of users and the way people trade one attribute over another thus understanding what benefits offer most utility and at what levels, a DCE is undoubtedly the next step. This however presents a number of difficulties including making trade-offs between psychological and process attributes and the role of health within this context. Only one published study has been identified which has investigated the trade-off between health-related quality of life, a psychological outcome measure (self-efficacy), a social outcome and a process attribute (Richardson et al., 2009). This study concluded that self-efficacy is an important outcome to respondents who are willing to trade decrements in health-related quality of life for improvements in self-efficacy (Richardson et al., 2009). At the same time however, the authors point to certain limitations of incorporating a psychological outcome measure in a DCE, including firstly, the possible overlap between health-related quality of life and self-efficacy; and secondly, possible over-estimations of self-efficacy due to problems in assessing the real opportunity cost when making decisions- the DCE task “may not force respondents to focus on
the real opportunity cost sacrifice to health by presenting a direct trade-off between health outcome and self-efficacy” (p. 337).

Considering the complexity of a DCE where the trade off between health and psychosocial outcomes are examined, it might be more appropriate to conduct a methodological study comparing between the utility values obtained through a traditional DCE and a best-worst scaling study. Methodological analytical frameworks for DCE designs in healthcare are constantly shifting with more elaborate designs more suitable for handling attributes from different groups, such as the Generalised Linear Latent and Mixed Models (GLLMM) and the best-worst scaling being used (e.g. Coast et al., 2006; Richardson et al., 2009).
8.4. *Findings and the psychosocial literature*

Chapter 3 provided an overview of the psychosocial literature and the outcome measures used to evaluate CGSs. Psychosocial consequences of these services were found to fall under a number of dimensions and these included emotional, cognitive, behavioural and social and familial consequences. Research and outcome measures falling within each dimension have been independently criticised as being unable to capture the overall impact of CGSs and the subjective user experience.

Parallel to this research has been research adopting multi-dimensional outcome measures focusing on the concept of coping and adaptation with the genetic circumstances. This research has commonly used the outcome measure of Perceived Personal Control (PPC) to encapsulate the subjective experience and needs of service users. PPC has been described to encapsulate three popular dimensions of CGSs, namely cognitive control, decisional control and behavioural control (Berkenstadt et al., 1999). Its most important limitation however in the context of CGSs might be its individualistic focus. McAllister and colleagues (2007a; 2008a) have already pointed out the family nature of CGSs and have proposed a related concept to that of PPC which they have called “empowerment” and which incorporates empowerment to family members and future generations.

Current findings have proposed the concept of Perceived Familial Control to be a more appropriate outcome to consider for evaluating CGSs. This concept relates
to a process of empowerment taking place longitudinally throughout the family’s contact with the service rather than an end point resulting following contact with CGSs. This outcome has been defined as the belief of family members that: they and their family members have the tools or know how to obtain the tools to be able to deal with the tangible and intangible consequences resulting from having a genetic condition in the family. It is proposed to encapsulate four dimensions, namely knowledge and understanding; informed and shared decision-making; enablement; and reassurance.

Current findings contribute to research investigating suitable outcomes which are broad enough to encapsulate the overall psychosocial impact of the service, but also adds to the literature by proposing important attributes of the service which contribute towards appropriate psychosocial outcomes. In this sense, it illustrates important elements of service provision which add to the utility of service users and thus should be considered when assessing for suitable models of service provision. Only one research study has so far been found to describe process attributes in addition to psychosocial outcomes considered as important by service users (McAllister et al., 2008b). The relationship between process attributes and psychosocial outcomes is important to understand because of the improvements to service provision which could be made in order to achieve a more positive impact on service users.

Cognitive outcomes constitute one of the main objectives of CGSs, namely the provision of information in order to support decision-making. Despite the popularity of cognitive outcomes such as genetic knowledge and risk perceptions
(Michie and Marteau, 1996; Michie et al., 1997; Payne et al., 2008; Smerecnik et al., 2009), a number of authors point out the problems involved in using these outcomes (Clarke, Parsons and Williams, 1996; Michie et al., 1998; Biesecker and Peters, 2001). The issue raised is whether the information assessed by genetic service providers as important is the same as the information needs of patients and families. Qualitative research has illustrated that service users attribute value on more subjective aspects of the process and specifically on information that is relevant and useful to them and promotes abilities for coping with the situation (Collins et al., 2001; Skirton, 2001; Barr and Millar, 2003; Lim et al., 2004; McAllister et al., 2008b; McAllister et al., 2008a).

Current research has shown that information was a theme running throughout narratives while the need for information was a driving force behind participants’ access to the service. Current findings have shown that even though information is actively sought by participants, the service is valued because it offers more than simply information. These findings add to past literature pointing to the understanding of access to CGSs as part of a coping mechanism to deal with the perceived risk and uncertainty involved. Finding answers as to why a genetic condition has occurred has been found to be integral to subsequent adaptation following the genetic diagnosis (Barr and Millar, 2003; Lipinski et al., 2006). The search for information therefore appears to be a coping mechanism facilitating positive effects (Pain, 1999) on emotional outcomes such as adjustment.

Access to other NHS services and utilisation of risk management options has strongly emerged in participant narratives. Current findings have illustrated
elements of service provision not typically considered by the CGS literature but which should be incorporated in future assessments of effectiveness. For example, “Interdisciplinary problem-based care” has emerged as an important element of service provision, involving referrals to other specialists, establishing care pathways, enabling access to services, links to social and educational services and tangible support to enable access to needed benefits, and information about support organisations. The ability of the CGS to transcend the boundaries of the service and make links with health but also with non-health services such as social and educational support services was valued by participants.

Studies investigating the impact of CGSs on behaviours have focused on conditions where appropriate interventions can be designed in order to improve the health outcomes of conditions, such as cancer and coronary heart condition, investigating changes in behaviour following access to the service (Pijl et al., 2009; Quach et al., 2009; Zlot et al., 2010). PPC has incorporated a behavioural component in the encapsulation of perceived personal control, defining behavioural control as the availability of an instrumental response able to directly influence or change the physical characteristics of a stressful event (Berkenstadt et al., 1999). The dimension of “enablement” was presented in the current research to represent perceptions of being able to actively deal with the tangible consequences of the condition, linked to elements of service provision such as inter-disciplinary problem based care, and accessibility over time. Focusing on perceptions rather than actual behaviours has been presented as an advantage
of PPC where perceived rather that actual control is what is important (Davey et al., 2005). A focus on what users perceive as being able to do, such as being able to access needed clinical support or other actions which allow one to deal with the symptoms or other aspects of the disease, arguably tackles some of the limitations involved in assessing behaviour change. Evaluating behaviour change might be applicable only to multi-factorial conditions where changes in behaviour patterns could potentially in the long term affect health outcomes.

Assessing the role of CGSs in clinical support and perceptions of being able to actively deal with the tangible aspects of the condition is arguably important because of suggestions that health outcomes are an important outcome of the service for service users. Based on ratings provided in Phase Three as part of the pilot study aspect of this phase, “clinical outcomes” appeared among the top nine (dis)benefits preferred by attendees, a category not emerging to be widely relevant during the second phase. Health outcomes are also controversial within research since health outcomes are not a stated objective of the service (Clarke, Parsons and Williams, 1996). It appears however that an interest in improving their health outcomes might be what is motivating families to access CGSs.

The need for multidisciplinary service provision has also been raised by research reporting that women referred to a cancer genetic centre regard access to other specialists as very important (Brain et al., 2000b; Holloway et al., 2004). Access to screening services for individuals with a cancer family history is a motivating factor for accessing CGSs (Brain et al., 2000b), while access to required healthcare services is one of many needs of families affected by a genetic
condition (McAllister et al., 2008a). This might suggest that improved health outcomes might be among the expectations or hopes of service users as a result of them dealing with the situation rather than a perceived direct outcome of the service.

As a conceptual outcome, PFC encapsulates cognitive, emotional, behavioural and family related aspects, satisfying calls for multi-dimensional outcomes in the evaluation of CGSs. It also fits with the two existing frameworks proposed as outcome measures of CGSs, namely the concept of Perceived Personal Control by Berkenstadt (Berkenstadt et al., 1999; Smets et al., 2006) and the concept of empowerment by McAllister et al (McAllister et al., 2008a; McAllister et al., 2008b; McAllister, Dunn and Todd, 2011). This concept has been described and compared with the two previously mentioned frameworks, e.g. PPC and empowerment, in Section 6.6. PFC is a conceptual outcome which applies both to the overall CGSs rather than to distinct components such as genetic counselling, a limitation affecting the existing literature, but also able to capture the long-term effect of the service rather than focus on short-term implications.

**8.5. Methodological evaluation**

Current approaches in economic evaluation present difficulties for the identification, cataloguing and incorporation of all the relevant consequences, and particularly of the intangible consequences (Hall, Viney and Haas, 1998). There are a number of available methodological reviews and commentaries that discuss the limitations of existing methods for the evaluation of complex health
care interventions like CGSs, public health interventions and social care interventions (Byford and Sefton, 2003; Coast, 2004; Coast, Smith and Lorgelly, 2008a; Coast, Smith and Lorgelly, 2008b) and new approaches to evaluation have started to emerge (Grewal et al., 2006; Coast, Smith and Lorgelly, 2008a).

A new feature is the use of qualitative methods as a tool for health service evaluation. The use of qualitative research has now been included in the 5th version of the Cochrane handbook for systematic reviews of interventions (Noyes et al., 2011) in which it is recognised that more outcome studies in systematic reviews are likely to be qualitative studies while a number of economists have promoted their use (Coast and Horrocks, 2007; Smith, Mitton and Peacock, 2008; Coast et al., 2011).

This research illustrated how qualitative methodologies can support the design of economic evaluations by furthering the understanding of previously unknown variables such as psychosocial consequences, process attributes and patient pathways. Interviews with geneticists and genetic counsellors led to mapping out patient pathways through the service and the identification of variables which influence the patient load and models used by the service. Variables such as local expertise and interests, commissioning, as well as the particular characteristics of the condition and individual needs of the patients are important determinants of what is offered by the service. The way patient pathways may differ depending on these variables is illustrated in the second column of figure 8.2 p.328. Focus groups and interviews with users of the CGS helped identify several process attributes, psychosocial consequences and an emotional
concept to describe these consequences, namely “Perceived Familial Control”. This in turn can help the cataloguing of intangible consequences and subsequent design of economic evaluations. It can also assist the development of more suitable outcome measures which are better able to capture the overall impact of the service.

Methodological challenges to economic evaluation are not confined to the domain of CGSs, but have been a frequent subject of debate within healthcare interventions or services exhibiting similar complexities in their characteristics and organisation. Public health and social care interventions are two examples for which economists have had to rethink the way evaluations are approached (Byford and Sefton, 2003) and several challenges have been described (Weatherly et al., 2009). A first challenge for these types of interventions is the inherent problem when coming to attribute intervention effects, mainly because the intervention is directed towards the community rather than the individual. This requires the use of new methods of synthesising evidence from experimental and non-experimental studies which can be used in economic evaluations.
Figure 8.2: Framework of (dis)benefits of clinical genetic services to guide in the design of an economic evaluation

Factors impacting on service delivery models:
- Onset: Adult Onset, In childhood, In uterus
- Penetrance: High, Low
- Expression: Variable, Consistent

Inheritance patterns:
- Dominant
- Recessive
- X-linked
- Single-gene
- Multifactorial

Management:
- Effective management possibilities: Little can be done
- Uncommon

Prevalence:
- Common

Molecular genetics (easy versus complex)

Availability and use of genetic testing – prenatal and neonatal diagnosis)

Widespread knowledge of the condition among the public and healthcare professionals (implications for diagnosis and referrals to CGSs)

Local interest and expertise (level of support available and perceptions of what the aims of the service is)

Commissioning and funding

Elements of service provision:
- Clinical and Genetic diagnosis (genetic testing)
- Genetic counseling
- Proactive versus reactive service (level of contact with CGSs)
- Interdisciplinary working and collaboration e.g.: Obstetrics and gynecology, Oncology services, Breast screening services, Neonatal screening

Benefits and disbenefits:
- Coordination of surveillance and access to screening programs
- Referrals to other healthcare services
- Support with accessing services e.g. social, educational
- Information and contact with NGOs

Knowledge and understanding

Reassurance: Informed & shared decision-making, Enablement

Process Attributes:
- Expertise
- Family centeredness
- Individualized approach to service provision
- Interdisciplinary problem-based care
- Accessibility over time

Perceived familial control:
- Expertise
- Family centeredness
- Individualized approach to service provision
- Interdisciplinary problem-based care
- Accessibility over time
A second challenge is that given the broad nature of costs and benefits in complex interventions, standard approaches to outcome measurement and valuation may not be adequate. Consequences might impact on individuals other than the one targeted, and might involve benefits such as reassurance which are currently not specifically incorporated in outcome measures such as QALYs. These outcome measures have also been questioned for what exactly they are measuring and they have been challenged for not capturing what is actually valued by individuals (Grewal et al., 2006). Thirdly, these interventions often have implications for sectors other than health, and this means that the broad nature of the costs and benefits might require an intersectoral approach for identifying them. Adopting a wider perspective than simply the perspective of the provider which has been the norm so far has been understood to be particularly useful (Drummond, Weatherly and Ferguson, 2008) and approaches have been suggested which allow considerations of the consequences across all sectors of the economy and one such way is through a cost-consequence analysis (Weatherly et al., 2009). Finally, the issue of equity has been raised (Weatherly et al., 2009). Concerns of equity have not featured prominently in economic evaluations. Both CEA and CUA have been criticised for not taking into consideration equity (Coast, 2004) even though research has shown that individuals seem to prefer an egalitarian healthcare perspective rather than a focus on health benefits (Nord et al., 1995). Weatherly and colleagues call for more research into public and stakeholder views on equity weighting in order to
increase understanding of how much sacrifice on efficiency is warranted in order to pursue particular equity goals (Weatherly et al., 2009).

Qualitative methods could be and have been used to tackle some of the above challenges, while the methodological advances in qualitative methodologies mean that qualitative research can now be more actively incorporated in healthcare research. The design and evaluation of complex interventions by using qualitative methods has been proposed as a technique to assist in the problems faced when defining the components of complex interventions (Campbell et al., 2000). Qualitative research is already being used in new methodological approaches to evaluation which are able to take equity into consideration e.g. the capabilities approach which is now gaining some momentum in healthcare evaluation (Anand and Dolan, 2005; Grewal et al., 2006; Coast, Smith and Lorgelly, 2008a; Coast, Smith and Lorgelly, 2008b).

The use of focus groups, preliminary surveys or case studies can help define relevant components of interventions which are not well known or understood. Qualitative research can also be used to show how the intervention works and to identify potential barriers to change. Defining and justifying the level of analysis based on the aims of the study is necessary to ensure methodological rigour and the validity of the conclusions drawn from the data (Coast and Horrocks, 2007; Coast et al., 2011). In some circumstances research objectives might not call for an interpretative view of the data that requires time-consuming and exhaustive analysis. Then descriptive analysis of the data might be most appropriate rather
than in-depth analysis suitable for when research requires the exploration of relationships or the development of theoretical hypotheses.

The following figure illustrates the three stages to the analysis of qualitative data adopted in the current study, based on the research objectives and the nature of the data aimed to be obtained.

**Figure 8.3: A framework of analysis of qualitative data for economic evaluation**

8.6. **Research limitations and directions for further research**

There were a number of limitations and methodological challenges to this study. These refer to the overall design although the most significant phase limitations are associated with the final phase of data collection. First, study limitations will be presented followed by directions for further research.

8.6.1. **Study limitations**

This study adopted a case study approach by using one UK CGS as the focus for data collection. The case study approach is useful for in-depth understanding of the workings of a specific service where such understanding is limited and is
therefore often used within evaluation studies (Keene, 2006). This however, remains the study of a single genetic centre and therefore limits the transferability of findings to other genetic centres.

A second limitation associated with the research design is the decision to focus data collection on specific groups of genetic conditions rather than adopt broader or different strategies of participant recruitment. Other studies such as McAllister and colleagues (McAllister et al., 2007b) have conducted focus groups with stakeholders of CGSs recruiting participants from all conditions seen by a CGS. Focusing on a small number of specific conditions created a number of problems for both overall recruitment and recruitment to individual focus groups. The most obvious problem was the difficulty in organising the focus groups while for some conditions it was not possible to recruit enough individuals and interviews were conducted instead. Recruitment was also time-consuming due to the focus on a small number of conditions and for the majority of conditions recruitment packs had to be sent twice. The recruitment framework was designed in collaboration with geneticists from the chosen CGS based on a number of criteria listed in Chapter 4. It was attempted to cover implications from the broader spectrum of genetic conditions seen by a CGS but it might be possible that some implications associated with rarer conditions might have been missed.

Within the first phase of data collection, the recruitment of service providers who were already aware of the objectives of the research and who had been consulted about the recruitment strategy of the service users presented another limitation. The main purpose of this phase however was the understanding of
user pathways for the specific genetic conditions and of the aims of service provision. Even though participants in this phase might have been motivated to present the chosen CGS in a positive light, overall there were no major differences between what providers and users perceived as a benefit of the service. It would have been useful if differences between provider and user perceptions of the quality of the service were explored by analysing in more depth the narratives of service providers. The fact that service providers were aware of the research objective and were encouraged to participate by a senior member of staff who was acting as advisor to the project however would arguably introduce bias into the analysis. This is because service providers would have an additional incentive to present their services in a positive light if they were told that their interviews would be used for in-depth analysis. When deciding to use such a small number of conditions for data collection anonymity is also an issue since it is difficult for participants to remain anonymous when such a small number of service providers are involved in service provision raising more issues of validity if narratives are used for in-depth analysis.

Again, the qualitative characteristics and motivation of participants to present the positive aspects of the genetics service, compared to the wider group of service users is also a limitation for the second phase. This group of individuals appeared to be highly motivated and actively seeking opportunities to be involved in the CGS’s research or other activities. In this sense they might be more positive towards the service than individuals who refused to take part. It is acknowledged that the positive attitudes of participants might have affected the
analysis of the transcripts despite the initial impartiality of the researcher. This is a common issue however within qualitative research recruitment and this point has been discussed in the context of the findings from discussions with participants from each condition in Chapter 6.

Phase Three of data collection presented the biggest limitations, in particular the small number of individuals attending. Even though attempts were made to recruit a larger number of individuals from all groups of the CGS’s users, only 14 actually attended the event. A number of explanations are possible to explain the low attendance. Firstly, the research was not known to service users or to the majority of service providers. Closer individual contact with stakeholders might have been required to encourage individuals to attend such an event because of the effort required on the part of participants. Advertisements on genetic-related websites proved to be inadequate as the sole means of recruitment. Even though recruitment packs were sent to all those who had been invited to attend the second phase of data collection regardless of whether or not they actually participated in that phase, only a few who had not already participated in the second phase also attended the third. This suggests that those who had participated in focus groups and interviews were highly motivated to be involved in research relevant to the genetics centre. It is indeed the case that the majority of participants in these discussions presented the CGS in a mainly positive manner and some were particularly enthusiastic for giving something back to the service.
8.6.2. Directions for further study

Based on the limitations related to transferability of findings to both other genetics centres and service users from other conditions, further research is required to explore whether (dis)benefits emerging in the present study are relevant to the consumers or stakeholders of other UK genetic centres. Further research is also required to investigate the transferability of emerging (dis)benefits to all or at least most conditions. This was attempted during the third phase of data collection but due to recruitment limitations it was not possible to conclude transferability to other conditions. Related to this is the fact that (dis)benefits presented here apply to the specific models of service provision which in the long-term might change both within the chosen CGS and on a UK scale which again limit their transferability. Despite these limitations however, the proposed (dis)benefits provide a platform upon which to base future investigations and between- as well as within-centre comparisons.

A review of the literature has illustrated that research investigating provider perspectives on the aims and outcomes of CGSs is sparse, yet what providers perceive as the aims of the service is arguably important for the quality of the interaction between service provider and service user. This research has illustrated that service users describe CGSs to be different from other NHS services and the attitudes of service providers appear to contribute towards these positive perceptions. Research into provider attitudes and service provision in the context of prenatal care has been useful in informing service delivery (Bernhardt et al., 1998; Abramsky et al., 2001). In the same way future research
exploring in more detail how perceptions of genetic providers and users differ might also be useful in assessing and improving service delivery.

The exercise undertaken in Phase Three has illustrated some points relevant to the organisation of such future events and may be used in a future study investigating consumer preferences in the context of a DCE or other preference elicitation technique. Data collection using ARSs in order to investigate preferences might be more successful if data collection events were organised to take place within other conferences or meetings. One such example might be conferences organised by patient support groups, NGOs or condition specific organisations such as the Cystic Fibrosis Trust. In the case of service providers, data collection could take place in the context of conferences or genetic centre yearly meetings. In this way data is collected from a large group of stakeholders simultaneously without going into time-consuming processes of organising events where the participation rate would be uncertain. The ability of ARSs to automatically store and analyse data is a benefit which would enable comparisons both within the same data collection sessions as well as between sessions. This can facilitate within group comparisons i.e. differences between genetic centres, between individuals associated with the same condition or stakeholder groups such as genetic counsellors and geneticists, as well as between group comparisons i.e. differences between individuals associated with different conditions or stakeholders groups. This data collection approach would enable collection of data from a larger number of individuals than facilitated using
a Delphi study which again requires effort and dedication from the part of participants and from a wider spectrum of stakeholder groups.

Validation of the four dimensions enclosed in the concept of Perceived Familial Control will enable this concept to be taken forward and developed into an outcome measure suitable for use in the evaluation of CGSs. Section 6.6.1 discussed the similarities of the PFC concept with that proposed by McAllister and colleagues termed as “empowerment” (McAllister et al., 2008a; McAllister, Dunn and Todd, 2011), and the implications of these two studies for the adaptation of the popular concept of PPC (Berkenstadt et al., 1999) to better represent the benefits of a genetic service. Even though the concept of “empowerment” does present considerable similarities with PFC, it is argued that the concept proposed in the present research might be more suitable because of, among others, the variability in definitions existing in the literature for the concept of empowerment and the inconsistency in its use among researchers. In terms of adapting PPC, an alternative of PFC might better reflect the similarities in these two concepts but also the differences which distinguish CGSs from other services.

An attempt to develop the concept of PFC, as in any outcome representing familial implications, incorporates certain complexities. Before item development and validation of the measure, some basic decisions need to be taken concerning the outlook of the measure. For example, does one measure one individual’s perceptions of PFC i.e. asking one individual about their perceptions of benefit to their family resulting by that individual’s access to CGSs, or does
one measure one family’s perceptions of benefit. Intuitively, one might argue that the outcome measure should assess each individual’s perceptions of familial benefit, especially since service users have been found to be motivated by expectations of benefit to their family, regardless of whether the family is benefited or not. However, it would be interesting to find out whether and how perceptions of familial benefit might differ between family members, e.g. between a mother accessing the service expecting her daughter to benefit, and the daughter’s perceptions. Further exploring such possible differences might provide such guidance as to a possible “composite” measure of PFC which captures family rather than individual perceptions of familial control.

A further complexity would be the process of defining a “family” which could be the immediate or extended family members to whom this measure might be addressed. The chosen CGS has been reported to use the family unit as the currency unit for commissioning and this definition incorporates referrals for extended family members (Hughes et al., 1998). Arguably, if the outcome measure used to evaluate the service is to represent the unit of currency, then a definition of the family unit should incorporate the extended family members as well. Conceptualisations of the family unit however are arguably subjective, and further debate is needed in order to operationalise these concepts.

8.7. **Implications for future economic evaluations**

There appears to be some confusion within the literature about what can be used to evaluate CGSs. Both outcomes and characteristics of service provision have
been proposed indiscriminately and all have been presented as outcomes of the service. Donaldson and Shackley acknowledge that it is difficult to distinguish between what is process and what is outcome (Donaldson and Shackley, 1997), however it is important to make a distinction between the two since each type may be used differently within an economic evaluation. This is especially true for process characteristics and psychosocial outcomes, since when coming to investigate consumer preferences and what service users consider as important, this research has suggested that process characteristics are more meaningful as attributes within a DCE than psychosocial outcomes. This was especially apparent during Phase Three where stakeholders were asked to state how important each (dis)benefit was in the context of CGSs service provision. All (dis)benefits emerging as important were process related e.g. continuity of care, and had tangible implications i.e. empowerment in making decisions. All these characteristics of service provision however appeared to lead to an overall perception of familial control.

Bernhardt et al (Bernhardt, Biesecker and Mastromarino, 2000) in their presentation of outcomes and benefits of genetic counselling propose nine outcomes related both to the process and goals of genetic counselling including provision of psychosocial support in both the short and the long-term, provision of anticipatory guidance and facilitation of family communication. Skirton and colleagues (Skirton, Parsons and Ewings, 2005) in their design of an audit tool for CGSs incorporate both process-related outcomes including “respect for autonomy” and psychosocial outcomes such as “positive psychological change”
and “adaptation”. McAllister et al have conducted research specifically aimed to propose an outcome which can be used to evaluate CGSs (McAllister et al., 2008a; McAllister et al., 2008b; McAllister, Dunn and Todd, 2011) and have since developed their construct into an outcome measure directed towards evaluating genetic counselling (McAllister et al., 2011). These researchers appear to be the only ones who have distinguished between attributes of service provision, outcomes of the process or dimensions incorporated in an overall conceptual outcome.

Health-related outcomes have been the outcome of choice for economists interested in the evaluation of healthcare services in general and specifically of CGSs, but remain a controversial outcome for evaluating such complex healthcare services. Provision of information remains the most prevalent responsibility of CGSs and there is no direct involvement in medical care, even though co-ordinating health surveillance is a stated role of clinical geneticists in an EU context (Godard et al., 2003). The impact of the CGS on health outcomes was perceived as important by participants belonging to specific conditions, in this case VHL and to a different degree to HBOC, but not to the majority of groups, something that shows that outcomes for evaluation should be chosen based on a clear understanding of both the specific patient pathway followed by patients, as well as the actual impact the service is able to have on their lives. The findings from this thesis suggest that even though health improvements are not what service users expect directly from CGSs, they do expect the service to direct them to, set in place or co-ordinate symptom screening and management.
interventions which could potentially impact on health outcomes. This is an aspect of the service which has not received attention within evaluations of CGSs. This is an aspect which should be considered however in the context of assessing possible models of service provision.

Another impact of CGSs discussed in the economic literature has been that of insurance and mortgage problems (Harper, 1997; O'Neill, 1997; Hoel et al., 2006). The actual impact of CGSs on insurance might require further consideration, and insurance concerns were not a prominent theme in the present research since only a few participants raised these concerns. In addition other studies have found that only five out of 156 participants attending a US Cancer Genetics Programme experienced some form of insurance discrimination (Kausmeyer et al., 2006b), although another study found that 40% of carriers of the BRCA1/2 genes reported having some sort of insurance problems 3 years after having the genetic test (Foster et al., 2007). Similarly other investigators report that the most frequently reported aspect of discrimination in individuals at risk for Huntington’s disease has been that of insurance (Bombard et al., 2009). It might be that insurance problems are condition specific and clearly can also vary between countries depending on the extent to which their health care systems are insurance based and these might explain why they were not a prominent theme in the current study. It might also be the case that insurance problems might not be considered as a disbenefit relevant to genetic services or that it might not be as important as other benefits of the service. Even though insurance problems were presented among the (dis)benefits of the presentation
during Phase Three, they did not feature among the most important (dis)benefits as were ranked by contributors, suggesting that indeed this issue might not be as important as others.

Numerous health economists have to date commented on the absence of appropriate outcomes with which to evaluate a CGS and more importantly, lack of knowledge of psychosocial outcomes. Methodological discussions have pointed to the inappropriateness of equating health outcomes to utility within health economic evaluations and proposed that other (dis)benefits such as process attributes and consumption attributes need to be incorporated in a utility function (Donaldson and Shackley, 1997). The current research has identified two categories of (dis)benefits which may feature in a utility function in addition to health, namely: service attributes, and psychosocial dimensions represented by the concept of Perceived Familial Control.

Family-oriented care incorporates considerable external effects, something that might be the most obvious difference between CGSs and other healthcare services, and a characteristic which is at odds with the individualistic concept of utility within microeconomic theory. Externalities in the case of CGSs are considerable and should not be ignored. CGSs affect both current and future or potential service users and the utilities of all family members appear to be considerably interdependent. The present research illustrates why the family is the unit of currency for commissioners of CGSs and the need to consider the implications for the economic evaluation of this unique aspect of CGSs.
Externalities have been described as a central concept of welfare economics and as one of the reasons for government involvement (Labelle and Hurley, 1992). Externalities or spill-over effects refer to cases where some of the costs or benefits resulting from the consumption of goods or services of one consumer spill-over or affect the utility of third parties (Jacobsson, Carstensen and Borgquist, 2005). Labelle and Hurley (1992) refer to four sources of utility: 1) Patient utility from treatment; 2) non-patient utility derived from the treatment of patients (interdependent utility); 3) patient utility derived from the treatment of other patients (interdependent utility); and 4) non-patient utility derived from option value (option demand) i.e. from knowing that a service will be available at a later time, something that potential consumers are willing to pay for. Three of the four sources of utility refer to the presence of external benefits. The two types of externalities described, namely interdependent utility and option demand, have both emerged within current findings. Service users derive utility when other members of their family also receive services; parents derive utility when their children who carry a hereditary disease receive services, while stakeholders in the form of family members not yet accessing the service or even service users derive utility from knowing the service is available to them or their family members when needed.

Other research has also reported the presence of externalities (Jacobsson, Carstensen and Borgquist, 2005; Borghi and Jan, 2008) while models incorporating spillover effects have illustrated the important implications when these sources of utility are omitted from economic evaluations (Labelle and
Basu and Meltzer (2005) have gone so far as to use a model based on a family utility function to illustrate the welfare implications on all family members resulting from healthcare choices. They conclude that “cost-effectiveness analyses may better reflect the full costs and benefits of medical interventions in some circumstances if they incorporate family effects” (Basu and Meltzer, 2005: p.771). Labelle and Hurley (1992) conclude that if it is too difficult to adapt current cost-utility methodology to incorporate externalities, then health economists should consider the use of cost-benefit analysis, which is able to incorporate the full range of effects of healthcare services. Despite the limited attempts to incorporate externalities in CUA and CEA methodology, there appears to be a lack of consideration of spillover effects within economic evaluations (Labelle and Hurley, 1992; Basu and Meltzer, 2005; Jacobsson, Carstensen and Borgquist, 2005).

According to Gafni and Birch (Gafni and Birch, 1995) for an outcome measure to be appropriate for use in an economic evaluation it must (a) be consistent with a theory of utility; (b) incorporate individuals’ attitudes to risk in order to reflect individual preferences; (c) attach different weights for different groups of the population in order for equity considerations to be an intrinsic part of the outcome measure and take into account externalities; and (d) be abiding to the underlying economic theory.

During this early stage, the outcome proposed within the broader framework of this research appears to fulfil two of the requirements, namely it adopts a welfarist view of utility and takes into account externalities. Further work needs
to be done however for this outcome to be able to be considered as a suitable outcome measure for the evaluation of CGSs and possibly as an alternative to the traditional QALY.

As a first stage towards the design of a CBA, the identified attributes are particularly suitable for the design of a discrete choice experiment (DCE) or other preference-eliciting technique. DCEs can be used to inform the design of CBAs since by incorporating a willingness-to-pay (WTP) element in this approach it is possible to attach values to intangible outcomes as well as non-health outcomes (Drummond et al., 2005). Two examples of DCE choice sets using a traditional DCE to present the identified attributes compared to an opt-out option and a best-worst choice data set to present the psychosocial outcomes along with a health outcome were presented in Section 8.3.6.2.

Existing DCE studies have been criticised for not having methodological clarity i.e. being clear on the methods used to extract the attributes used in the design of the studies (Coast and Horrocks, 2007). The methodology used in the current study however provides both methodological clarity and a conceptual framework behind the attributes proposed for the design of DCE studies.

8.8. Overall Contribution of the research

This thesis was stimulated by a realisation that existing outcomes for the evaluation of CGSs were not broad enough to incorporate the overall impact of the service on its users which was believed to be mainly psychosocial in nature. This was especially the case in the context of economic evaluation, where
outcome measures of CGSs were focused on health or health-related quality of life measures such as the QALY (Hall, Viney and Haas, 1998; Cohen, Barton and Brain, 2004; Griffith, Edwards and Gray, 2004). Health economists have pointed out the need to better understand the impact of CGSs and particularly their psychosocial outcomes which were so far largely unknown in order for comprehensive economic evaluations to take place that can inform future resource allocation.

The thesis has presented a comprehensive framework incorporating knowledge necessary for the design of a welfarist economic evaluation: 1) variables influencing service delivery models such as genetics of the condition, commissioning and funding, and local interest and expertise; 2) service elements and characteristics such as genetic testing and genetic counselling, level of patient contact and interdisciplinary working; and finally 3) the perceived benefits of the service including process attributes, and a conceptual outcome of the service incorporating 4 dimensions.

The proposed framework has emerged through the use of qualitative methods rather than the use of qualitative techniques i.e. the use of interviews and focus groups removed from the overall framework of qualitative research, as in the majority of economic research (Coast and Horrocks, 2007). Qualitative methods utilising qualitative data analysis of varying depth to understand: 1) the range of consequences of CGS, i.e. the extent to which the service has implications for individuals other than the one referred to the service; 2) the psychosocial (dis)benefits which encompass the range of implications resulting from the use of
CGSs; and 3) for exploring the extent to which access to CGS has resource implications for other parts of the public sector or NGOs. In this sense, the present thesis has illustrated how qualitative methods can be used to further the design of economic evaluations, in this context at the initial stages of mapping out the workings of the specific service and identifying its benefits and disbenefits. Different levels of qualitative analysis might be necessary depending on the research objectives and not all studies would call for in-depth analysis of their data. Being clear of the research design and level of analysis however is important in order to ensure for research validity.

Previous to this research it was hypothesised that psychosocial outcomes were the main (dis)benefits present in the utility function of genetic service users, but as it emerged through the present thesis, process-related attributes are also utility bearing. Insurance problems did not emerge as an important disbenefit for participants. Perceived Familial Control (PFC) was proposed as a more suitable outcome for the evaluation of CGSs illustrating the gaps in other popular outcome measures which appear to be individualistic and exclude the familial element which is a distinguishing characteristic of these services.

A second contribution of the research has been the investigation of possible differences in (dis)benefits between the different conditions. Even though evaluations have tended to focus on evaluations of services for individual conditions, e.g. cancer genetics (Appleton et al., 2000; Cohen, Barton and Brain, 2004; Breheny et al., 2006), none of the previous studies have explored the possibility that outcomes and especially psychosocial outcomes might differ
between conditions. This study has illustrated that even though the patient experience differs between conditions based on genetic condition characteristics and individual needs, the (dis)benefits raised by all participants are primarily the same, with differences being present in the preferences of each condition for specific (dis)benefits. This is an important realisation in the case of economic evaluations where economists might not be called for incorporating different (dis)benefits for the economic evaluation of different conditions, but research is needed to understand in more detail how preferences might differ.

Even though the proposed dimensions of PFC, namely “knowledge and understanding”, “informed and shared decision-making”; “reassurance” and “enablement” as well as some of the attributes of the service such as “accessibility over time” and “individualised approach to service provision” are shared characteristics of quality of care with other NHS services, especially primary care (Campbell, Roland and Buetow, 2000; Sans-Corrales et al., 2006), the familial element is what distinguishes clinical genetics from the rest of the NHS and this has not been present in evaluations of the service, economic or otherwise. This is an important recognition if evaluations are to accurately reflect the impact of CGSs on its users.

Previous research has also explored process attributes and psychosocial consequences of CGSs, but none have explicitly mapped out service delivery influencing variables to specific patient pathways and benefits/disbenefits of the service. The identification of psychosocial outcomes is useful for the evaluation of CGSs within the context of service delivery and improvement and
commissioning, but in order to design a comprehensive CBA it is essential to understand the overall context of the service. This also enables comparisons between clinical genetic centres in order to further understanding into which aspects of service provision are essential for ensuring good user outcomes.
REFERENCES


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Appendices


Manne, S, Audrain, J, Schwartz, M, Main, D, Finch, C and Lerman, C (2004). "Associations between relationship support and psychological reactions of participants and partners to


Appendices


Appendices


van Helvoort-Postulart, D, van der Weijden, T, Dellaert, B, de Kok, M, von Meyenfeldt, M and Dirksen, C (2009b). "Investigating the complementary value of discrete choice


APPENDIX 1

CONFERENCE PAPERS AND CONFERENCE PRESENTATIONS
Conference submitted papers


Conference presentations


Pithara, C. What are the benefits and disbenefits of genetic services? Themes from 2 focus groups. *Institute of Medical Genetics*. 8/4/2008

Pithara, C. What are the benefits and disbenefits of genetic services? A PhD Odyssey. *University of Glamorgan Postgraduate Research Seminars*. 15/5/2008

Pithara, C. Consequences of Clinical Genetic Services: Results from user focus groups. *Institute of Medical Genetics*. 10/9/2009

APPENDIX 2

LIST OF LITERATURE REVIEW SEARCH TERMS
**Literature search strategy**

Search terms (single search terms and combinations):
- Genetic*
- Genetic services
- Genetic service provision
- Genetic disease*
- Genetic condition*
- Genetic syndromes
- Genetically inherited conditions
- *cancer
- Familial conditions
- Familial syndromes
- Familial diseases
- Inherited conditions
- Breast cancer
- Ovarian cancer
- Ovarian neoplasms
- BRCA1
- BRCA 2
- Tuberous sclerosis – neurocutaneous syndrome*
- Von Hippel-Lindau *
- Cystic fibrosis
- Duchenne muscular dystrophy
- Becker muscular dystrophy
- Muscular dystrophies
- Genetic testing
- Genetic diagnosis
- Prenatal diagnosis
- Prenatal testing
- Predictive testing
- Carrier testing
- Experiences
- Users
- Health policy
- Methodology
- Benefits
- Advantages
- Impact
- Implications
- Dimensions
- Consequences
- Demand
- Use
- Attitudes

Willingness to pay
- Uptake
- Economic evaluation
- Cost benefit analysis
- Cost effectiveness analysis
- Cost utility analysis
- Cost consequences analysis
- Discrete choice
- Qualitative methods
- Interviews
- Focus groups
- Psychosocial outcomes
- Psychosocial consequences
- Psychological*
- Psychological outcomes
- Psychological consequences
- Psychological impact
- *psychology
- Emotional impact
- Emotional outcomes
- Distress
- Adaptation – adaptive behaviour
- Adjustment
- Locus of control
- Perceived control – perceived personal control
- *control
- Stress, psychological
- Uncertainty
- Family
- Siblings
- Attitude*
- Patient education
- Genetic counselling
- Quality of life
- Coping
- Outcomes research
- Anxiety
- Depression
- False negative reactions
- Program* evaluation
- Depressive disorder
- Insurance selection bias
- Patient satisfaction
- Informed consent
APPENDIX 3

LIST OF PARTICIPANT INCLUSION AND EXCLUSION CRITERIA
Appendices

Inclusion and Exclusion Criteria

Common Exclusion criteria for the 5 conditions:
- Terminally ill patients
- Taking part in any concurrent study
- Not being able to communicate in the English language
- Extreme levels of anxiety as judged by the consultants.

HBOC:

Inclusion criteria:
- Users over 18
- High risk - breast and/or ovarian cancer
- Living
- Unaffected
- Referred between 2000 and 2005
- SE Wales only
- Attended clinic

Cystic Fibrosis:

Inclusion Criteria (provisional):
- Over 16 years of age
- Being referred to the service between September and December 2005
- Parents of a patient with CF
- Carrying a common mutation

Von Hippel-Lindau Disorder

Inclusion Criteria
- Over 16 years of age

Duchenne and Becker muscular dystrophy

Inclusion Criteria
- Over 16 years of age
- Carers or parents of people with DMD
- Carriers and people at risk of being a carrier of a germ-line mosaicism
- Patients with Becker muscular dystrophy
- Spouses of men with BMD
- Daughters of men with BMD

Tuberous Sclerosis

Inclusion Criteria
- Over 16 yrs of age
- Carers or parents of people with TS
- Carriers of the TSC1 or TSC2 genes and who are able to take part in a focus group
APPENDIX 4

INTERVIEW GUIDE: SERVICE PROVIDERS
(1) Structure of service and patient referral pathways to the CGS,
   (a) patient pathway
   (b) adherence to formal guidelines / protocols
   (c) perceived aims and objectives of the service
   (d) perceived role of genetic services

(2) Links and collaborations with other services / organisations
   (a) links between the CGS and other NHS services (referrals)
   (b) links to other non-NHS services / organisations (referrals or enabling access)
   (c) formal links to other services / agencies
   (d) are these links successful
   (e) would the patients have access to these services without the intervention of
       the genetic service

(3) Positive or negative ways in which the service impacts the lives of service users
   (a) if the service altered the impact of the condition on patients and family
   (b) positive and negative consequences of genetic services on the service users,
       whether these were psychological, social or health-related

(4) Identification of the preferred outcome on which the service provided should be
    evaluated (outcome measure).
APPENDIX 5

PERCEIVED AIMS, ROLE AND (DIS)BENEFITS OF THE CGS
(SERVICE PROVIDERS)
### AIMS OF CGSs

<table>
<thead>
<tr>
<th>Process-related</th>
<th>HBOC</th>
<th>CF</th>
<th>TS</th>
<th>VHL</th>
<th>DMD/BMD</th>
<th>PREN</th>
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</thead>
<tbody>
<tr>
<td><strong>Answer questions</strong></td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td><strong>Provide accurate information</strong></td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td><strong>Co-ordinate management of condition/co-ordinate care/access to other agencies</strong></td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td><strong>Identify people at high risk/carriers</strong></td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Provide reproductive options</strong></td>
<td></td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Outcome-related</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td><strong>Limit the impact of the condition on the patients</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

### PERCEIVED CHARACTERISTICS (ATTRIBUTES) OF CLINICAL GENETIC SERVICES

<table>
<thead>
<tr>
<th>PROCESS</th>
<th>HBOC</th>
<th>CF</th>
<th>TS</th>
<th>VHL</th>
<th>DMD/BMD</th>
<th>PREN</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>giving opportunity to have healthy children</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td><strong>Educating other professionals</strong></td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>time given to patients</strong></td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
</tr>
<tr>
<td><strong>open access policy</strong></td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td><strong>Patient-centred care</strong></td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td><strong>long-term relationships with families / follow-up</strong></td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
<td></td>
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</tr>
<tr>
<td>-----------------------------------------------------</td>
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<td>---</td>
<td>---</td>
<td>---</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Preparing patients about prognosis</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Establish links and care pathways to other</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>services/specialists/support groups/referral to</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>agencies for symptoms management</strong></td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
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<tr>
<td>Provide co-ordinated screening and management of the</td>
<td></td>
<td></td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
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<tr>
<td>condition</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Offers overview information on the condition and their</td>
<td></td>
<td></td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>care/holistic management of symptoms</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>offers choices for prenatal diagnosis/reproductive</td>
<td></td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>options</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Provision and understanding of information given</strong></td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>accurate information on reproductive options,</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>genetic risk status, diagnosis + genetic condition</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>(genetic testing)/decrease number of false positives</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>cascade screening</td>
<td></td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Concerns taken seriously by health professionals</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Psychological/practical support in coming to terms with</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>family history (adjustment)</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
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## PSYCHOSOCIAL CONSEQUENCES

<table>
<thead>
<tr>
<th>Reassurance</th>
<th>X</th>
<th></th>
<th>X</th>
<th>X</th>
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<tbody>
<tr>
<td>Psychological harm from going through multiple prenatal testings</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>Altered self/body image</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Psychological issues (e.g. depression) resulting from diagnosis</td>
<td>X</td>
<td></td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Faced with mortality early on in life</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>Worry over future of family/children</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>Psychological trauma related to the extended medical interventions</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>Certainty</td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Guilt/blame</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>Raised anxiety from unexpected pregnancy outcomes</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>Conflict between expectations and reality</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Enable sense of control in families over their lives and genetic risk status</td>
<td>X</td>
<td>X</td>
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</table>
### Sense of normality (adjustment)

<table>
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<th>Sense of normality (adjustment)</th>
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<th>VHL</th>
<th>DMD/BMD</th>
<th>PREN</th>
</tr>
</thead>
<tbody>
<tr>
<td>Empower families to take control and make decisions</td>
<td>X</td>
<td></td>
<td></td>
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<td></td>
</tr>
<tr>
<td>Disturbed family relations/communication issues</td>
<td>X</td>
<td></td>
<td>X</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Burden/impact on the family members</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
<td>X</td>
<td></td>
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<tr>
<td>Issues of identify with family unit</td>
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<td></td>
<td></td>
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<td>X</td>
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### MOST IMPORTANT OUTCOME MEASURE

<table>
<thead>
<tr>
<th>MOST IMPORTANT OUTCOME MEASURE</th>
<th>HBOC</th>
<th>CF</th>
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<th>VHL</th>
<th>DMD/BMD</th>
<th>PREN</th>
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</thead>
<tbody>
<tr>
<td>Facilitate understanding of families/patients of accurate and relevant information for informed decision-making</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>Satisfaction with the genetic service</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>sense of control</td>
<td></td>
<td></td>
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<td></td>
<td>X</td>
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<tr>
<td>sense of normality (adjustment)</td>
<td></td>
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<td></td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>long term meeting of expectations</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>X</td>
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</tr>
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</table>

387
APPENDIX 6

COMPARISONS OF SERVICE PROVISION MODELS FOR THE
FIVE GROUPS OF CONDITIONS
## Appendix: Comparison of Service Models

<table>
<thead>
<tr>
<th></th>
<th>HBOC</th>
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<th>MD</th>
<th>PREN</th>
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<tbody>
<tr>
<td><strong>Genetic Counselling</strong></td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
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<tr>
<td><strong>Genetic Testing</strong></td>
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</tr>
<tr>
<td>Diagnostic</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
</tr>
<tr>
<td>Pre-symptomatic</td>
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<td>YES</td>
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<tr>
<td>Carrier testing</td>
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<tr>
<td><strong>Reproductive Options</strong></td>
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<tr>
<td>Prenatal</td>
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<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
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<tr>
<td>Pre-implantation diagnosis</td>
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<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
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<tr>
<td><strong>Clinical Support to Users</strong></td>
<td></td>
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<td></td>
<td></td>
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</tr>
<tr>
<td>Access to screening/prophylactic surgery</td>
<td>YES</td>
<td></td>
<td></td>
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</tr>
<tr>
<td><strong>Referrals/Links to Other Services</strong></td>
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<td></td>
<td></td>
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<td>YES</td>
</tr>
<tr>
<td>Referrals/links to other services</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
</tr>
<tr>
<td>Clinical Symptom monitoring</td>
<td></td>
<td></td>
<td></td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
</tr>
<tr>
<td>Patient clinics</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>YES</td>
<td>YES</td>
</tr>
<tr>
<td>Establish care pathways to secondary care specialists for symptom management</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td></td>
</tr>
<tr>
<td><strong>Coordination Role (management of the condition)</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>YES</td>
</tr>
<tr>
<td>Co-ordinating role (management of the condition)</td>
<td>YES (needs-based)</td>
<td>YES</td>
<td></td>
<td></td>
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<tr>
<td><strong>Education</strong></td>
<td></td>
<td></td>
<td></td>
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<td></td>
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<tr>
<td>Educating other (health) professionals</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
<td>YES</td>
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389
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<tr>
<th>Proactive (recall of families)</th>
<th>NO</th>
<th>NO</th>
<th>NO</th>
<th>YES</th>
<th>YES for female carriers</th>
<th>NO</th>
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<tr>
<td><strong>Newborn screening programme</strong></td>
<td></td>
<td></td>
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<tr>
<td>geneticist involvement</td>
<td>extensive</td>
<td>minimal</td>
<td>extensive</td>
<td>extensive</td>
<td>extensive</td>
<td>minimal</td>
</tr>
<tr>
<td>genetic nurse/counsellor involvement</td>
<td>extensive</td>
<td>counsellor run service</td>
<td>extensive</td>
<td>minimal</td>
<td>minimal</td>
<td>counsellor run service</td>
</tr>
<tr>
<td>part of multidisciplinary team</td>
<td></td>
<td></td>
<td></td>
<td>informal</td>
<td></td>
<td>YES</td>
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</tbody>
</table>
APPENDIX 7

DISCUSSION GUIDE WITH SERVICE USERS
• **Introduction**
  - Introduce myself and the research

• **Opening circle**
  - Could you please tell us your name and briefly share with us how you came into contact with the medical genetic services and your journey through the service so far?
  - As we are here to discuss genetic services I’d like to know what comes to mind when you think of the genetic service?

• **Introductory questions**
  - When you were referred to the genetic service, did you have any expectations (what were they?)
  - When you think of the services you have received from genetic services do you think these expectations were met? If not, why is that?

• **Key questions**
  - What were the most important aspects of the service for you and your family?
    
    *Knowledge and understanding of the condition, the relationships in your family, any impact on your emotional state, your medical treatment or your behaviour? (prompt for these during the discussion if not mentioned) (write these on a piece of paper)*

    *Write down on board the benefits and disadvantages*
  
  - Do you all agree with these consequences and if not why?
  - We are interested to find out what you think is important in the outcomes of genetic services, either positive or negative. Thinking back to the services and the experiences you’ve had with the service, could you think of the useful things and not so useful things that resulted to you and your family from being in contact with the genetic service?
  - Could you think how your life and your family’s lives would be different if the genetic services provided by the IMG were not available to you?

    *Write down the ways the lives would be different*

• **Ending questions**
  - From all the consequences we had discussed today resulting from your use of genetic services, could you think what the most important benefit was and which the most important negative consequence?

• **Closing circle**
  - Thinking back on the discussion we’ve had today, I’d like to close our discussion by asking all of you to say again what the most important aim of genetic service should be if that has changed from the beginning of our discussion?
APPENDIX 8

PARTICIPANT CONSENT FORM
Study Number:
Patient Identification Code:

Consent Form

Title of Project: Identifying the benefits and Disbenefits of Genetic Services: A Framework for Economic Evaluation

Please initial box

1. I confirm that I have read and understand the information sheet dated ..................... (version4) for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily.

2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.

3. I understand that relevant sections of any of my medical notes and data collected during the study, may be looked at by responsible individuals from the University of Glamorgan / Institute of Medical Genetics, from regulatory authorities or from the NHS Trust, where it is relevant to my taking part in this research. I give permission for these individuals to have access to my records.

4. I agree to take part in the above study

_______________________                 ______________              __________________  
Name of Participant            Date                                   Signature

_______________________                 ______________              __________________  
Name of Person taking consent            Date                                   Signature

_______________________                 ______________              __________________  
Researcher            Date                                   Signature

When completed, 1 for participant; 1 (original) to be kept for researcher site file.
APPENDIX 9

INFORMATION LETTER TO CLINICAL GENETICISTS
Dear Mr/Mrs

**Re: Participant Recruitment for research project looking into the outcomes of genetic services**

In January of last year I have started a PhD with the Health Economics and Policy Research Unit (University of Glamorgan) in collaboration with the Institute of Medical Genetics. The title of the research is:

*Identifying the benefits and disbenefits of genetics services: A Framework for economic evaluation*

The aim of the study is to identify what the positive and negative impacts of genetic services might be on users. One part of the methodology chosen to achieve this aim is qualitative interviews with users of genetic services. Research focuses on five conditions which have been selected from the total number covered by the Institute of Medical Genetics based on their clinical and genetic characteristics. [ ] is included in these 5 conditions.

The following is a list of the inclusion/exclusion criteria which have been chosen to guide recruitment for [ ]. The aim is to recruit 10 people to take part in a focus group.

I am attaching a patient recruitment letter and information sheet along with a consent form for your information and [ ] recruitment packages consisting of the above ready to be sent out to patients. If any of your patients is interested in taking part in our study, a reply slip is included with the information sheet which they can post to the given address.

My supervisors and I greatly appreciate your support. Please feel free to contact us at any time for any comments you might have.

Kind Regards

Christalla Pithara

Supervisors:
Prof David Cohen: dcohen@ glam.ac.uk
Dr Kate Brain: kate.brain@ cardiffandvale.wales.nhs.uk
Dr Christine Shaw: cshaw@ glam.ac.uk
Dear Sir/Madam,

**Re: Invitation to take part in focus group discussion sessions**

The University of Glamorgan together with the Institute of Medical Genetics, Wales is conducting a study aiming to identify the positive and negative consequences resulting from the use of genetic services.

We are inviting people who have used the service to take part in group discussions. The researchers are interested in learning about your experiences of the Institute of Medical Genetics and during these discussion individuals like yourself will be able to discuss their experiences. The discussions will be confidential.

An information sheet which provides more information about the research is included with this letter. If after reading this information you decide to take part please complete the consent form attached and return it using the included free-post envelop to the named individual. This means that your participation is voluntary and will NOT be made known to me or other health professionals involved in your care unless you choose to discuss your participation with a member of staff.

If you would like more information regarding this research please feel free to contact the named researcher at the contact number provided in the information sheet.

Kind Regards,

[NAME OF CONSULTANT]
APPENDIX 11

PARTICIPANT INFORMATION SHEET
What are the effects of being referred to the Institute of Medical Genetics (Wales)?

This information sheet is an invitation to take part in a research study which aims to identify the positive and/or negative ways in which Genetic Services influence the lives of the people who use the services.

- Part 1 of this information sheet tells you the purpose of this study and what you will have to do if you take part.
- Part 2 gives you more detailed information about the study.

Before you decide whether to take part, it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully. Talk to others about the study if you wish. Ask me if there is anything that is not clear or if you would like more information. Contact details are provided below.

Please take time to decide whether or not you wish to take part.

Part One
What is the purpose of this study?
My name is Christalla Pithara and I am undertaking a PhD studentship which aims to identify the advantages and disadvantages of being referred to genetic services.

This studentship is a collaboration between the University of Glamorgan and the Institute of Medical Genetics in Wales and is funded by the University of Glamorgan.

Why have I been chosen?
Because we are looking for people who have been referred to the Institute of Genetic Services, Cardiff and who are willing to share their experiences.

Do I have to take part?
No. It is up to you to decide whether or not to take part. Even if you do sign the consent form, you are still free to withdraw at any time without giving a reason.

Any decision you make will not affect the services you receive from the Institute of Medical Genetics in any way.

What will I have to do if I take part?
Taking part in this research project involves attending a group discussion with other people who have experience of the same genetic condition as you, or the person you care for.

During this discussion you will be asked to discuss your experience with the Institute of Medical Genetics and in what ways it has changed your life.

If you decide to take part you can return the consent form attached to this letter using the free-post envelope included. I will then contact you to discuss the details for this group discussion and answer any questions.

A Discussion Guide including the issues to be discussed during the group discussion and also further details regarding the research study will be send to you in advance of the meeting.

With the participants’ consent interviews will be tape-recorded.

Expenses and payments
You will be offered compensation for any travelling or other expenses made in order to take part in this project. All people who participate in the discussion groups will also be entered in a draw for a £50 M&S voucher.

**What are the possible benefits of taking part?**

There are no direct benefits of taking part, other than taking part in a group discussion about your experiences of being referred and receiving services from the Institute of Medical Genetics. The benefit of this study will be in its recommendations for evaluations of genetic services that take into account emotional as well as financial benefits.

**What if there is a problem?**

Any complaint about the way you have been dealt with during the study or any possible harm you might suffer will be addressed. The detailed information on this is given in Part 2.

**Will your participation be kept confidential?**

Yes. All the information about your participation in this study will be kept confidential. The details are included in Part 2.

**Contact Details**

If there is anything that is not clear or you would simply like more information please contact:

Christalla Pithara  
HEPRU  
Faculty of Health, Sports and Science  
Glyntaff Campus  
Treforest  
CF37 1DL  
Tel: 07769774600  
cpithara@ glam.ac.uk.

If you would like more information regarding the services provided by the Institute of Medical Genetics or simply want to discuss your potential participation with a genetic counsellor please contact:

[contact details of named genetics professional]

This completes Part 1 of the Information Sheet  
If the information in Part 1 has interested you and you are considering participation, please continue to read the additional information in Part 2 before making a decision.

**Part 2**

**What will happen if I don’t want to carry on with the study?**

You can withdraw from the study before, during or after the group discussions and any information you have provided thus far will be destroyed. This will not affect any of the services you receive from the Institute of Medical Genetics.

**What if there is a problem?**

If you have a concern about any aspect of the study, you can contact me at any time (see contact details below).

During the discussion it is highly unlikely that anything should go wrong, but it is possible for different questions to arise. The contact details of your trained genetic counsellor or
geneticist are included at the end of this information sheet should you decide you would like to talk about these issues.

If you are concerned about something and after talking to me or your named health professional you remain unhappy and wish to complain formally, you can do this through the NHS Complaints Procedure. Details can be obtained from the hospital.

In the event that something does go wrong and you are harmed during the research study because of someone’s negligence then you may have grounds for a legal action for compensation against the University of Glamorgan but you may have to pay your legal costs. The normal National Health Service mechanisms will still be available to you (if appropriate).

Will my taking part in this study be kept confidential?

Yes. Any data collected during the study will be kept in a safe place within the University of Glamorgan and only I will have access to this information. Unless you choose to inform them, no member of your health care team will know whether you are taking part in this study. Further information regarding confidentiality will be included in the Discussion Guide which will be sent to you closer to the date.

When reporting the findings to third parties no names or other personal information of the people taking part in the research will be included.

Findings of this research project will be available to everyone at the library of the University of Glamorgan at the end of the study (December 2008). Findings will also be disseminated to staff and users of the Institute of Medical Genetics in the form of a presentation where you may also attend (more information will be send out later in the future).

This is the end of the information sheet.

What happens now?
If you do decide to take part please fill in and post the consent form included in the Recruitment Package. If you have any questions regarding the study please contact the researcher at the e-mail or postal address reported above.

At this point we would like to thank you for taking the time to read this information sheet and for considering taking part.
APPENDIX 12

RECRUITMENT POSTER FOR PHASE THREE
HAVE YOU OR ANY MEMBER OF YOUR FAMILY BEEN REFERRED TO THE ALL WALES MEDICAL GENETICS SERVICE?

ARE YOU PART OF A SUPPORT TEAM HELPING FAMILIES AFFECTED BY GENETIC DISEASE?

THEN WE ARE INTERESTED TO KNOW WHAT YOU THINK ARE THE MOST IMPORTANT CONSEQUENCES OF USING GENETIC SERVICES.

YOU ARE INVITED TO ATTEND A FREE EVENT ON
Friday, 21 November 2008
from 12:30-4pm
AT THE
HENRY WELLCOME BUILDING,
UNIVERSITY HOSPITAL OF WALES, HEATH PARK

DURING THE EVENT YOU CAN VOTE AND GIVE FEEDBACK ON THE FINDINGS OF AN ON-GOING RESEARCH PROJECT, LOOKING INTO THE CONSEQUENCES OF BEING REFERRED TO THE ALL WALES MEDICAL GENETICS SERVICE.

YOU WILL BE GIVEN HANDHELD VOTING SETS WHICH MAKE VOTING FUN, ANONYMOUS AND OPTIONAL.

A FREE LUNCH WILL BE PROVIDED

For more information or to register via phone please contact:
Christalla Pithara
Email: epithara@glam.ac.uk
Telephone number: 07769774600
APPENDIX 13

PHASE THREE INVITATION LEAFLET
HALF-DAY CONSULTATION EVENT

This is a FREE event

Friday, 23rd of September 2008
From 12.30-4pm
Venue:
Henry Welcome Building
University Hospital of Wales,
Heath Park

A free lunch will be provided
during the day

Participants to this event will be given the opportunity to provide comments on the consequences identified through the group discussions and also take part in a voting session. During the voting session participants will be able to vote on the relevance and importance of the identified consequences to them and their family. Voting will be optional and anonymous.

The input of participants during this event will provide the data needed to come up with a list of consequences that are important to users of genetic services, and which should be kept in mind when researchers and decision-makers are thinking of an evaluation of genetic services.

IF YOU WOULD LIKE TO TAKE PART IN THIS EVENT, PLEASE CUT AND RETURN THIS REPLY SLIP

First Name ..................................................
Surname ..................................................
Address ..................................................
..................................................
..................................................

Friends & family are more than welcome to attend but as places are limited please tell us how many people will be coming with you

For more information or to register directly please contact:
Christalla Pithara
Email: cpithara@glam.ac.uk
IDENTIFYING THE CONSEQUENCES OF CLINICAL GENETIC SERVICES

This leaflet is an invitation to attend a consultation event on the consequences of the All Wales Medical Genetics Service. This one-off event is part of ongoing research aiming to identify the positive and/or negative ways in which genetic services influence the lives of those using the service.

You might have already received information for this study, and might have already taken part in the group discussions which formed the second phase of this research.

HOWEVER, this is a one-off event and you may attend regardless of participation in other stages of this research.

WE ARE INTERESTED IN WHAT YOU HAVE TO SAY ABOUT THE CONSEQUENCES OF BEING REFERRED TO THE ALL WALES MEDICAL GENETICS SERVICE

A free lunch will be provided during the day

This event is for:
- Patients and families who have had experience with the All Wales Medical Genetics Service,
- Members of patient support groups providing support to patients and families affected by genetic disease, and
- Health professionals involved in the delivery of the All Wales Medical Genetics Service.

IF YOU DO NOT WANT TO BE CONTACTED AGAIN FOR THIS RESEARCH PLEASE TICK THIS BOX

If you would like to register to attend this half-day event, please complete and return the attached slip to:

Christalla Pithara
HEPRU, Anzani House, Glyntaff Campus, Treforest, University of Glamorgan, CF37 1DL
Or directly on: cpithara@glam.ac.uk

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APPENDIX 14

PHASE THREE PRESENTATION TALK
I’d like to welcome you all for coming to this event, especially those who’ve travelled from outside Cardiff in order to attend. This is a half day event and if all goes according to plan we will have finished by 4 o’clock. You can view this presentation as a brief interactive storytelling event, describing the progress of this research so far and asking you to give your feedback. This event is separated into several parts. The first part is a practice session to give you the opportunity to have a go at using these sets before moving on to the questions that count. The second part is aiming to set the background as to who is interacting with us, therefore asking questions like are you male or female, and what your experience is with genetic services. The third part is asking questions about whether you agree or not with a number of consequences identified through analysing the group discussions we’ve had in the previous phase. The final part will ask you to state your preferences for a number of these consequences.

As you have already noticed when you came into the room and registered, you were given a folder with the University of Glamorgan logo on it and also a small machine that looks like a calculator but without the screen. This machine is essential for your participation in today’s presentation and is also very expensive so please return it after the end of the presentation. In order to help provide some structure in the way participants offer their feedback, we’re going to use this handheld voting set which enables the individual holder to press a button from 1 to 9. These numbers represent choices which will be given throughout this presentation in the form of a question or statement followed by a number of choices. Before moving on with the story we can have a small practice session using the voting sets.

PRACTICE QUESTIONS

Now, moving on to the second part of this presentation, you’ll be asked to answer a few demographic questions, starting with whether you are male or female.

DEMOGRAPHIC QUESTIONS

Along with this voting set you were also given a folder. This folder includes a programme for the day, an information sheet which explains some background information about this PhD research project, and two copies of today’s presentation. This copy serves two purposes. One, it gives you the opportunity to look at the statements or questions you will be presented with in written form, so that it can make it easier for you to think which options best represents your opinions. This is most important for some questions where the statements are presented in a separate slide than the one the voting will take place. In that situation you might find it easier to circle your choice before the voting slide comes up so you press the right button. A second purpose of having this copy, is that space has been left after questions where we think some people might have further comments to add. Also some people might feel that some important consequences of genetic services were left out and they might want to add these consequences. They can do this on this paper and then hand it over along with the voting set. We thought that some of you might want a copy of the presentation to take home with them so you will find two copies of the presentation.

There are few things I’d like to say about the background to this event. I have organised this event as the final phase of data collection of my PhD research which is looking into the benefits and disadvantages of using clinical genetic services. I want to stress at this point that we are not interested in evaluating the All Wales Medical Genetics Service. We are interested in the consequences of genetic services in general. The funding to this research
was given by the University of Glamorgan to the Health Economics and Policy Research Unit which is also part of the University of Glamorgan in order to explore the emotional and social consequences of clinical genetics service which up to that point were largely unknown to health economics researchers. To those who are not familiar with health economics, a few words of introduction. Health economics deals with issues of scarcity, and the fact that even though the needs of individuals have no end, the resources available to satisfy these needs are limited. People therefore are faced with choices of which needs to satisfy based on the resources available to them. Health economists ask questions like, are the costs of providing a specific service more or less than the benefits that are incurred to those who receive the service. The same questions are asked about genetic services. Over the years however, researchers raised a number of important questions about the way the genetic service was being evaluated. Firstly, an important question was whether the nature of genetic services was the same as of other healthcare services.

ARE GENETIC SERVICES DIFFERENT IN NATURE FROM THE REST OF THE NHS?

Genetic services are mainly known to provide information to individuals and families about their genetic risk status and offer genetic testing. The rest of the NHS aims to improve health status while genetic services have no direct impact on health. A second distinguishing characteristic of genetic services is the importance of the family unit, genetic disease affects the whole family as opposed to only affecting the individual like other medical conditions. This leads to a second question, that of possible unknown consequences of providing genetic services.

DO YOU THINK GENETIC SERVICES HAVE A SIGNIFICANT IMPACT ON THE EMOTIONAL WELLBEING OF THOSE WHO USE THE SERVICE?

ARE THE EFFECTS ON THE INDIVIDUAL DISTINCT FROM THE EFFECTS ON THE FAMILY UNIT WHEN EVALUATING GENETIC SERVICES?

In order to explore what the unknown consequences of providing genetic services were, we organised a number of discussions with users of the genetic service and we asked them what they gained from accessing the service and whether the result was a positive or a negative one. There was one limitation to these discussions however. Only a very small number of individuals could attend because of practical reasons and from only a small number of conditions. In order to see if what these people shared with us were experiences and beliefs shared by a wider number of individuals we decided to organise this event in order firstly to present what we have found to those who have the most interest in the service and secondly to get their feedback as to the truthfulness of the story we will be telling at the end of this research project.

Your feedback today is important because regardless of how much research one does, nobody understands genetic services more than those who have experienced the service either directly or indirectly by supporting and working with individuals who are themselves users of the service.

Through our discussions we realised that the consequences of genetic services exist on a number of levels. They can be emotional, social, related to the health of the individual, or finally related to the functions and practical aspects of the service. During your registration today you were given a folder with a number of handouts in it. Included in these handouts is a list of all the questions you will be asked today to vote on, and also a list of all the
consequences identified up until this point in the research along with a brief explanation of what we mean by these terms.

A number of practical consequences relating to the functions of a genetic service were discussed by those who took part in our group discussions and these will be presented first. You will be asked to state the degree of your agreement as to the importance of these consequences when in contact with the service. I want to point out again that we are not here to evaluate the All Wales Medical Genetic Service. We are interested to identify important consequences of genetic services that should be taken into consideration by any stakeholder interested in evaluating any genetic service. We are interested to hear whether the following consequences that will be presented to you are important enough to be considered when assessing a genetic service. So when faced with these consequences please consider whether based on your experience the particular outcome is indeed a consequence of the genetic service whether you agree that it is an important one. After the end of this presentation there will be a 15 minute break and then the most important consequences will be again presented to you and you will be asked to state your opinion as to which of these are most important when evaluating the service.

1. ACCESS TO GENETICS EXPERTISE

Being able to access experts on genetics was described as a benefit of the service by participants, as they found knowledge on the genetic disease affecting their family which they could not find elsewhere. Do you agree that this is an important benefit of the genetics service?

2. CERTAINTY ABOUT ONE’S GENETIC RISK STATUS IS AN IMPORTANT CONSEQUENCE OF GENETIC SERVICES

Information about one’s genetic risk status, in other words whether one in high, medium, or population risk, whether one is a carrier of a genetic condition or whether one has or will develop a genetic condition, was an important consequence of accessing genetic services as was described by participants.

3. PROVISION OF INFORMATION IN ORDER TO INCREASE LEVELS OF KNOWLEDGE AND EDUCATE FAMILIES ABOUT THEIR GENETIC DISEASE

Moving on from the certainty obtained through the genetic service, users also described the increased levels of knowledge regarding patterns of inheritance and the nature of the genetic disease.

4. OFFER CHOICES TO INDIVIDUALS ABOUT OPTIONS REGARDING THEIR SITUATION

An other consequence of genetic services was the provision of information regarding different options that people are faced with in order to deal with their situation.

5. MAKE DECISIONS WHICH PEOPLE ARE COMFORTABLE WITH

Users also described the importance of making decisions which they are comfortable with, without being influenced by the attitudes of the healthcare professional.
6. OFFER CLINICAL SUPPORT TO FAMILIES AFFECTED BY GENETIC DISEASE

Genetic services may also provide clinical support to patients through either the co-ordination of medical treatment by scheduling and make referrals for needed tests, through offering an overview of all medical interventions, seeing which tests have been done or need to be done and make referrals, or through educating families about all the medical interventions that are needed and explanations of the medical test results.

7. FACILITATE ACCESS TO OTHER HEALTHCARE SERVICES

If you think that facilitating access to other healthcare services like screening services, paediatricians, neurologists or surgeons is or needs to be a consequence of genetic services please state your level of agreement. What is your level of agreement that facilitation of access to other healthcare services is or needs to be a consequence of genetic services?

8. IMPROVED CLINICAL OUTCOMES CAN BE A BENEFIT FROM RECEIVING GENETIC SERVICES

Several individuals believed receiving genetic services which resulted in either knowing their genetic status for some individuals or having a genetic diagnosis for others, could help improve their clinical outcomes through the clinical support they would receive from the service. If you agree with this consequence please state the degree of your agreement.

9. INFORMATION ABOUT RESEARCH FINDINGS AND NEW RESEARCH TAKING PLACE

Being informed about research findings in the treatment or causes of genetic disease and being familiar with new research was described as a benefit of genetic services. Do you agree that this is an important consequences of genetic services?

10. CONTINUITY OF CARE

Continuity of care refers to receiving care over time from a particular service. People benefited from being able to access genetics professionals after the initial contact with the service, who were aware of their particular circumstances and receiving support over time. If you think that being able to receive services from genetic services over time is an important benefit of the service please state the degree of your agreement.

11. BEING AFFECTED BY GENETIC DISEASE MAY HAVE AN IMPACT ON FAMILY RELATIONSHIPS

Relationships between an individual who carries a genetic risk or disease and the rest of their family may be influenced by the implications resulting from the identification of a genetic disease in the family. If you agree with this consequence please state the degree of your agreement.

12. SUPPORT TO INDIVIDUALS WHEN FACED WITH COMMUNICATING RISK STATUS RESULTS TO OTHER FAMILY MEMBERS
Some participants faced difficulties when having to communicate genetic status results to other family members. A benefit of the genetic service was providing support to individuals in such circumstances either through helping them in deciding what and how to communicate risk to family members, or enabling access of other family members to the genetic service. Again, please state your level of agreement with this benefit.

The above benefits described during the group discussions with users of the service relate to elements of care or functions of the genetic service which can be provided. Participants also referred to emotional and social consequences which resulted from receiving particular elements of care. You are again asked to state your agreement with these consequences of the genetic service.

13. INSURANCE PROBLEMS ARE A CONSEQUENCES OF BEING AFFECTED BY GENETIC DISEASE

Some participants described how having a history of genetic disease affected their ability of getting certain types of insurance like health and travel insurance. If you agree that this is the case, please state the degree of your agreement

14. INFORM FAMILIES OF POSSIBLE INSURANCE PROBLEMS WHICH MIGHT ARISE

If you think that it benefits users of the service to be informed in advance of the consequences on their ability to obtain insurance after being diagnosed with a genetic disease, being tested for a genetic disease or having a history of genetic disease please state the degree of your agreement

15. SUPPORT FAMILIES IN DEALING WITH THE SOCIAL EFFECTS OF THE GENETIC DISEASE

Having a genetic disease was found to result in a number of difficulties in the everyday life of individuals and their families. Problems with dealing with social services or education authorities were described. A benefit of genetic services was the support offered to families in dealing with these effects like linking families with support groups. If you agree that this is a benefit of genetic services please state the degree of your agreement.

16. ADJUSTMENT TO THE GENETIC CONDITION IS AN IMPORTANT CONSEQUENCE OF GENETIC SERVICES

Supporting families in adapting to their genetic situation has been described as a benefit of genetic services. Adjusting to their situation resulted in decreased feelings of anxiety and feeling more positive. Do you agree that this is an important consequence of genetic services?

17. FEELING IN CONTROL OF THE SITUATION IS AN IMPORTANT CONSEQUENCE OF GENETIC SERVICES

People taking part in our group discussions explained how gaining an understanding of the genetic disease and the options available to them helped them feel in control of the situation
and able to deal with the consequences. Do you agree that this is an important consequence of the service?

18. EMPOWERMENT IN MAKING DECISIONS

Imparting knowledge and educating individuals was found to lead to feelings of empowerment and the ability to make decisions when faced with choices relevant to dealing with the genetic status. Do you agree that this is an important consequence of the service?

19. REASSURANCE AND RELIEF FROM ANXIETY

Coming into contact with the genetic service can result in feelings of reassurance and relief from anxiety. Do you agree that this is an important consequence of the genetic service?

PART TWO

You were asked to state your agreement with a number of benefits and consequences of the genetics service today. An important question however is which of these consequences you perceive as being more important when coming to evaluate the service. As I’ve mentioned before presenting these consequences, they can be separated into benefits relating to elements of service provision, social, health and psychological consequences. When coming to evaluate the impact of the genetic service on its users, it is important to identify outcomes which represent what the genetic service actually does.

Adjustment and perceived personal control are two such consequences which have been discussed in the literature as outcomes appropriate for evaluating genetic services. These emotional outcomes may result after receiving certain aspects of care from genetic services. Through our interviews we have identified a number of elements of care which were found to be of value to our participants and today you were asked to state the level of your agreement as to their importance. As a final step to our research we would like to know which of these aspects of care you think are most important for promoting adjustment to the condition and personal control. You will be presented with a number of slides representing each of the 12 elements of care identified by participants as being of value and you will be asked to state how important you think each of these items is in promoting each of the two outcomes from 1 to 10, 1 representing “least important” and 10 representing “most important”. At the end of this process your answers will be presented in the form of a graph.
APPENDIX 15

PHASE THREE PROGRAMME
RESEARCH INTO IDENTIFYING THE CONSEQUENCES
OF CLINICAL GENETIC SERVICES:

USERS’ PERSPECTIVES OF IMPORTANT CONSEQUENCES FOR ASSESSING
THE SERVICE

User participation event

VENUE
Henry Wellcome Building, UG16
University Hospital of Wales, Heath Park

Friday, 21st of November
12.30-4pm

PROGRAMME

<table>
<thead>
<tr>
<th>Time</th>
<th>Activity</th>
</tr>
</thead>
<tbody>
<tr>
<td>12.30 pm</td>
<td>Welcome buffet lunch</td>
</tr>
<tr>
<td>1.15 pm</td>
<td><strong>Brief introduction by Professor Julian Sampson</strong> (clinical advisor to research project)</td>
</tr>
<tr>
<td>1.30 pm</td>
<td>Background to the research and introductory questions</td>
</tr>
<tr>
<td>1.45 pm</td>
<td>Demographic questions</td>
</tr>
<tr>
<td>2.00 pm</td>
<td>Consequences of clinical genetic services</td>
</tr>
<tr>
<td>3.00 pm</td>
<td>Brief break for tea or coffee</td>
</tr>
<tr>
<td>3.15 pm</td>
<td>Ranking of most important consequences</td>
</tr>
<tr>
<td>3.45</td>
<td>Overview</td>
</tr>
</tbody>
</table>
APPENDIX 16

PHASE THREE PRESENTATION QUESTIONS
Consequences of clinical genetic services

Half day interactive event

Practice Questions
- Who is the new president of the United States?
  - Homer Simpson
  - Tony Blair
  - Barak Obama
  - Pamela Anderson
- Have you taken part in other phases of this research?
  - Yes
  - No
- The government should provide all new available treatments on the NHS
  - Strongly Agree
  - Agree
  - Somewhat Agree
  - Neutral
  - Somewhat Disagree
  - Disagree
  - Strongly Disagree

Demographics
- Sex
  - Female
  - Male
- Age
  - 0-18
  - 19-30
  - 31-45
  - 46-65
  - Over 65
- My first contact with the clinical genetics service was...
  - Less than a year ago
  - 1-5 years ago
  - More than 5 years ago
  - I can’t remember/I don’t know
  - Not relevant
- I have...
  - A family history
  - No family history
  - I don’t know/can’t remember
  - Not relevant
- I am …
  - User of clinical genetic services
  - Geneticist/genetic counsellor
  - Working with voluntary groups
  - Researcher
  - Other
- Which statement best applies to you?
A. I have not been offered a genetic test and I would not be interested to have one
B. I have not been offered a genetic test and I would be interested to have one
C. I have been offered a genetic test but I have chosen not to have one
D. I have been offered a genetic test and I have chosen to be tested
E. Not applicable

- Please make your selection...
  - A
  - B
  - C
  - D
  - E

- Why were you referred to the clinical genetics service?
  A. Because of a family history of cancer
  B. Because I was diagnosed with a genetic condition
  C. Because one of my children was diagnosed with a genetic condition
  D. Because prenatal tests showed that my unborn child might have a genetic condition
  E. Concerned about reproductive options because of a family history of a genetic condition
  F. Because my sibling/family member was diagnosed with a genetic condition
  G. Other
  H. Not applicable

- Please make your selection...
  - A
  - B
  - C
  - D
  - E
  - F
  - G
  - H

(DIS)BENEFITS OF CGSS

- Are genetic services different in nature from the rest of the NHS?
  - Yes
  - No
  - I don’t know

- Do genetic services have an impact on the emotional wellbeing of those using the service?
  - Yes
  - No
  - I don’t know

- Are the effects on the individual distinct from the effects on the family unit when evaluating genetic services?
  - Yes
  - No
  - I don’t know

- What are the consequences of genetic services?

ACCESS TO GENETICS EXPERTS

- Strongly Agree
- Agree
CERTAINTY ABOUT GENETIC RISK STATUS
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

LEVELS OF KNOWLEDGE AND EDUCATION
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

OFFER CHOICES
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

DECISION-MAKING
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

CLINICAL SUPPORT
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

ACCESS TO OTHER NHS SERVICES
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

- CLINICAL OUTCOMES
  - Strongly Agree
  - Agree
  - Somewhat Agree
  - Neutral
  - Somewhat Disagree
  - Disagree
  - Strongly Disagree

- RESEARCH UPDATES
  - Strongly Agree
  - Agree
  - Somewhat Agree
  - Neutral
  - Somewhat Disagree
  - Disagree
  - Strongly Disagree

- CONTINUITY OF CARE
  - Strongly Agree
  - Agree
  - Somewhat Agree
  - Neutral
  - Somewhat Disagree
  - Disagree
  - Strongly Disagree

- CHANGES IN FAMILY RELATIONSHIPS
  - Strongly Agree
  - Agree
  - Somewhat Agree
  - Neutral
  - Somewhat Disagree
  - Disagree
  - Strongly Disagree

- SUPPORT IN COMMUNICATING GENETIC RISK
  - Strongly Agree
  - Agree
  - Somewhat Agree
  - Neutral
  - Somewhat Disagree
  - Disagree
  - Strongly Disagree

- INSURANCE PROBLEMS ARE A CONSEQUENCE OF GENETIC DISEASE
  - Strongly Agree
  - Agree
  - Somewhat Agree
  - Neutral
  - Somewhat Disagree
  - Disagree
- Strongly Disagree

**INFORM FAMILIES OF POSSIBLE INSURANCE PROBLEMS**
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

**SUPPORT IN DEALING WITH SOCIAL EFFECTS OF GENETIC DISEASE**
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

**ADJUSTMENT TO THE GENETIC DISEASE**
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

**FEELINGS OF PERSONAL CONTROL OVER THE SITUATION**
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

**EMPOWERMENT (MAKING DECISIONS)**
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

**REASSURANCE AND RELIEF FROM ANXIETY**
- Strongly Agree
- Agree
- Somewhat Agree
- Neutral
- Somewhat Disagree
- Disagree
- Strongly Disagree

**Did thoughts of your family have a role to play in your decision to access the service?**
RANKING

1. adjustment
genetics expertise
   i. Least Important
   ii. ...
   iii. ...
   iv. ...
   v. ...
   vi. ...
   vii. ...
   viii. ...
   ix. ...
   x. Most Important

2. adjustment
certainty
   i. Least Important
   ii. ...
   iii. ...
   iv. ...
   v. ...
   vi. ...
   vii. ...
   viii. ...
   ix. ...
   x. Most Important

3. adjustment
knowledge and education
   i. Least Important
   ii. ...
   iii. ...
   iv. ...
   v. ...
   vi. ...
   vii. ...
   viii. ...
   ix. ...
   x. Most Important

4. adjustment
choices
   i. Least Important
   ii. ...
   iii. ...
   iv. ...
   v. ...
   vi. ...
vii. ...
viii. ...
ix. ...
x. Most Important

5. adjustment
decision-making
i. Least Important
ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most Important

6. adjustment
clinical support
i. Least Important
ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most Important

7. adjustment
access to healthcare services
i. Least Important
ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most Important

8. adjustment
clinical outcomes
i. Least Important
ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...

x. Most Important

9. adjustment
   research updates
   i. Least Important
   ii. ...
   iii. ...
   iv. ...
   v. ...
   vi. ...
   vii. ...
   viii. ...
   ix. ...
   x. Most Important

10. adjustment
    continuity of care
    i. Least Important
    ii. ...
    iii. ...
    iv. ...
    v. ...
    vi. ...
    vii. ...
    viii. ...
    ix. ...
    x. Most Important

11. adjustment
    support with family communication
    i. Least Important
    ii. ...
    iii. ...
    iv. ...
    v. ...
    vi. ...
    vii. ...
    viii. ...
    ix. ...
    x. Most Important

12. adjustment
    dealing with social aspects of the disease
    i. Least Important
    ii. ...
    iii. ...
    iv. ...
    v. ...
    vi. ...
    vii. ...
    viii. ...
    ix. ...
    x. Most Important
13. personal control
genetics expertise
i. Least Important
ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most Important

14. personal control
certainty
i. Least Important
ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most Important

15. personal control
knowledge and education
i. Least important
ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most important

16. personal control
choices
i. Least important
ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most important

17. personal control
decision-making
i. Least important
ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most important

18. personal control
   clinical support
   i. Least important
   ii. ...
   iii. ...
   iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most important

19. personal control
   access to healthcare services
   i. Least important
   ii. ...
   iii. ...
   iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most important

20. personal control
   clinical outcomes
   i. Least important
   ii. ...
   iii. ...
   iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most important

21. personal control
   research updates
   i. Least important
   ii. ...
iii. ...
iv. ...
v. ...
vi. ...
vii. ...
viii. ...
ix. ...
x. Most important

22. personal control
   continuity of care
   i. Least important
   ii. ...
   iii. ...
   iv. ...
   v. ...
   vi. ...
   vii. ...
   viii. ...
   ix. ...
   x. Most important

23. personal control
   support with family communication
   i. Least important
   ii. ...
   iii. ...
   iv. ...
   v. ...
   vi. ...
   vii. ...
   viii. ...
   ix. ...
   x. Most important

24. personal control
   dealing with social aspects of the disease
   i. Least important
   ii. ...
   iii. ...
   iv. ...
   v. ...
   vi. ...
   vii. ...
   viii. ...
   ix. ...
   x. Most important
APPENDIX 17

PHASE THREE TERM DEFINITIONS
Definitions of terms used:

Elements of service provision

ACCESS TO GENETICS EXPERTISE
Being able to access experts on genetics was described as a benefit of the service by participants, as they accessed knowledge on the genetic disease affecting their family which they could not find elsewhere.

CERTAINTY ABOUT ONE’S GENETIC RISK STATUS
Information about one’s genetic risk status, in other words whether one is in high, medium, or population risk, whether one is a carrier of a genetic condition or whether one has or will develop a genetic condition, was an important consequence of accessing genetic services as was described by participants.

PROVISION OF INFORMATION IN ORDER TO INCREASE LEVELS OF KNOWLEDGE AND EDUCATE FAMILIES ABOUT THEIR GENETIC DISEASE
Users described the increased levels of knowledge regarding patterns of inheritance, the nature of the genetic disease and how it would affect them or their offspring in the future. The importance of knowledge is that participants did not feel overwhelmed by information but felt they understood the information given and felt educated.

OFFER CHOICES TO INDIVIDUALS ABOUT OPTIONS REGARDING THEIR SITUATION
Another consequence of genetic services was the provision of information regarding different options that people are faced with in order to deal with their situation, like for example treatment or prevention options, and reproductive options.

MAKE DECISIONS WHICH PEOPLE ARE COMFORTABLE WITH
Users also described the importance of making decisions which they are comfortable with, without being influenced by the attitudes of the healthcare professional. People did not feel pressured.

OFFER CLINICAL SUPPORT TO FAMILIES AFFECTED BY GENETIC DISEASE
Genetic services may also provide clinical support to patients through educating families about the clinical course of the disease and all the medical interventions that are needed and explanations of the medical test results, the co-ordination of medical treatment by scheduling and make referrals for needed tests, through offering an overview of all medical interventions, or seeing which tests have been done or need to be done and make referrals.

FACILITATE ACCESS TO OTHER HEALTHCARE SERVICES
The service could facilitate access to other healthcare services like screening services, paediatricians, neurologists or surgeons.

IMPROVED CLINICAL OUTCOMES
Several individuals believed receiving genetic services which resulted in either knowing their genetic status or having a genetic diagnosis, could help improve their health status prospects through accessing needed medical care.
INFORMATION ABOUT RESEARCH FINDINGS AND NEW RESEARCH TAKING PLACE
Being informed about research findings in the treatment or causes of genetic disease and being familiar with new research was described as a benefit of genetic services.

CONTINUITY OF CARE
Continuity of care refers to receiving care over time from a particular service. People benefited from being able to access genetics professionals after the initial contact with the service, who were aware of their particular circumstances and receiving support over time. Continuity of care was described to be important not only for the individual originally accessing the service but also for any other family member that required genetics support.

BEING AFFECTED BY GENETIC DISEASE MAY HAVE AN IMPACT ON FAMILY RELATIONSHIPS
Relationships between an individual who carries a genetic risk or disease and the rest of their family may be influenced by the implications resulting from the identification of a genetic disease in the family.

SUPPORT TO INDIVIDUALS WHEN FACED WITH COMMUNICATING RISK STATUS RESULTS TO OTHER FAMILY MEMBERS
Some participants faced difficulties when having to communicate genetic status results to other family members. A benefit of the service was providing support to individuals in such circumstances either through helping them in deciding what and how to communicate risk to family members, or enabling access of other family members to the genetic service.

Social

INSURANCE PROBLEMS ARE A CONSEQUENCE OF BEING AFFECTED BY GENETIC DISEASE
Some participants described how having a history of genetic disease affected their ability of getting certain types of insurance like health and travel insurance.

SUPPORT FAMILIES IN DEALING WITH THE EFFECTS OF THE GENETIC DISEASE
Having a genetic disease was found to result in a number of difficulties in the everyday life of individuals and their families. Problems with dealing with social services or education authorities were described. A benefit of genetic services was the support offered to families in dealing with these effects like linking families with support groups.

Emotional

ADJUSTMENT TO THE GENETIC CONDITION
Supporting families in adapting to their genetic situation has been described as a benefit of genetic services. Adjusting to their situation resulted in decreased feelings of anxiety, feelings of reassurance and relief and generally feeling more positive.

FEELING IN CONTROL OF THE SITUATION
People taking part in our group discussions explained how gaining an understanding of the genetic disease and the options available to them helped them feel in control of the situation and able to deal with the consequences.
EMPOWERMENT IN MAKING DECISIONS
Imparting knowledge and educating individuals was found to lead to feelings of empowerment and the ability to make decisions when faced with choices relevant to dealing with the genetic status.

REAASSURANCE AND RELIEF FROM ANXIETY
Coming into contact with the genetic service can result in feelings of reassurance and relief from anxiety.
APPENDIX 18

ETHICS APPROVAL
04th October, 2006

PRIVATE & CONFIDENTIAL

Ms Christalla Pithara
HEPRU
HeSAS
University of Glamorgan
Glyntaf
CF37 1DL

Dear Ms Pithara,

I am writing to confirm the details of your Honorary Contract with the Cardiff and Vale NHS Trust.

I have enclosed two copies of your contract, and I should be grateful if you would sign both copies. Retain one for your own record, and return the other to the Recruitment Office, Human Resources Directorate.

I would like to take this opportunity to thank you for offering your services to this Trust and hope that the experience you gain will be invaluable to you in your future career.

Yours sincerely

KATHRYN COOK
Recruitment Assistant
Human Resources Directorate

Enc
12 May 2006-05-12

Christalla Pithara
C/O School of Care Sciences

Dear Miss Pithara

Registration for MPhil/PhD at the University of Glamorgan – Title of Research Project ‘Benefits and disbenefits of genetic services: A framework for economic evaluation’

I am writing to confirm that DRPC Chairs Action has been taken to approve your registration for the award of MPhil/PhD.

Please note that this registration takes effect from the 1 January 2006.

Your supervision team was approved as:

Prof D Cohen, Director of Studies
Dr S Myles, Second Supervisor

If you have any queries about the Committee’s decision, please do not hesitate to contact me.

Yours sincerely,

Karen Roberts
Research Administrator & DRPC Secretary

cc: Prof D Cohen, Director of Studies
Dr S Myles, Second Supervisor
PEMBROKESHIRE & DERWEN NHS TRUST
HONORARY CONTRACT FOR RESEARCH & DEVELOPMENT

Name of researcher: Christalla Pithara

Outline
I am instructed by Pembrokeshire & Derwen NHS Trust to offer you an Honorary Contract as a Researcher from October 2007 until October 2008.

This post allows you to undertake the duties outlined in your attached contract of employment with University of Glamorgan on premises and using facilities of Pembrokeshire & Derwen Trust. If your duties involve clinical or administrative duties connected with patient care you are granted access to the associated records.

Conditions
The following conditions apply to your contract:

1. You are required to observe the policies and procedures of Pembrokeshire & Derwen NHS Trust (including Moving & Handling, Health & Safety, Fire Training etc.) in so far as they apply to this appointment and to observe all NHS policies and procedures in respect of clinical and research activities.

2. You must at all times act in accordance with the Trust’s Policies, Procedures and Guidance, copies of which are available in the Human Resources Department. The Trust reserves the right to terminate this Honorary Contract if your conduct is inconsistent with the high standards of work and behaviour expected in your placement with the Trust.

3. You agree to make yourself familiar with the Research Governance Framework¹ and agree to accept the responsibilities associated with your role that are outlined within it. Pembrokeshire & Derwen NHS Trust manages all research in accordance with the

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¹ Pembroke and Derwen NHS Trust
Honorary Contract for non-Trust employees to undertake research & development within the Trust
requirements of the Research Governance Framework. As a contract holder of Pembrokeshire & Derwen NHS Trust you agree to comply with all reporting requirements, systems and duties of action put in place by the Trust to deliver research governance.

4. You and your employer recognise the Trust’s right to benefit from intellectual property (IP) arising from work undertaken under this contract in accordance with the Health and Social Care Act 2001. In circumstances where there is a potential IP you are required to notify the Trust R&D Manager. Specific intellectual property agreements will be negotiated on an individual case by case basis.

5. You will report, on a day to day basis to Shan Owen Genetic Counsellor. If you wish to raise concerns or complaints about your commitments under this Honorary Contract you should first raise the matter with the Trust R&D Manager. The agreed procedure for settling differences between you and Pembrokeshire & Derwen NHS Trust will be in accordance with the Trust’s Grievance Procedure. This information will be fed back to your employing body as appropriate.

6. The Trust accepts liability in respect of your acts and omissions to the degree that those acts and omissions were carried out whilst working on behalf of the Trust and in accordance with your appointment under this honorary contract. You must however observe the same standards of care and propriety in dealing with patients, staff, visitors, equipment and premises as is expected of any other contract holder. You must also act appropriately and responsibly at all times.

7. Whilst undertaking officially sanctioned NHS duties, you are covered by the NHS indemnity against claims for negligence. In other circumstances (e.g. when providing services for which you receive a separate fee, or if undertaking research which has not received Trust approval) you are not covered by the indemnity. If you intend to treat private patients on Trust premises you must have a valid indemnity. Medical practitioners are advised to maintain membership of a medical defence organisation and submit a copy of current membership to the Human Resources Department.

Pembrokeshire & Derwen NHS Trust
Honorary Contract for non-Trust employees to undertake research & development within the Trust
8. You are required to be registered with the appropriate professional body and remain so for the tenure of your appointment with the Trust (applicable for all persons employed in registerable professions, i.e. doctors, nurses, AHPs, medical scientists). You should forward a copy of your current registration certificate to the Human Resources Department.

9. You are required to ensure the security and confidentiality of all information regarding patients or staff at all times. You should not release any such information to anyone other than an approved person in the course of your duties. If as an honorary contract holder you handle patient or staff-related information stored on computers, you must ensure that it remains on Trust-owned computers and is not transferred to computers owned by other organisations including those of your substantive employer without appropriate authorisation. This authorisation might be in the form of a formal agreement between Pembrokeshire & Derwen NHS Trust and your substantive employer with regard to specific types of information or a specific agreement between yourself and the trust with regard to storage of such information. You should be aware of your responsibilities under the Data Protection Act and only use such information for a registered purpose, not disclosing it to any unauthorised person. You should also make yourself familiar with the relevant Trust policies.

10. In the event of sickness or unavoidable absence, you must notify Sian Owen Genetic Counsellor immediately. You must report any accident or injury, however trivial, arising out of or in the course of your activities in the Trust to the Trust R&D Manager and complete the appropriate records and statements as required.

11. Please ensure that you wear your ID badge so that you are able to prove your identity if challenged.
Agreement

If you agree to accept this Honorary Contract on the terms specified above, please sign the form of acceptance at the foot of this page and return the whole contract to the Human Resources Department. A second copy is attached, which you should also sign and keep for further reference.

Signature: ..............................................................

Designation: PERSONNEL OFFICER
Date: 6th November 2007

CONFIRMATION OF RECEIPT AND ACCEPTANCE

I have read and agree to the above conditions & I enclose a copy of my current professional registration certificate and contract of employment.

Signed: .............................................................. Date: 21/11/07


Please sign and return to: Human Resources Department
Pembrokeshire & Derwen NHS Trust
Withybush General Hospital
Fishguard Road
HAVERFORDWEST
Pembrokeshire
SA61 2PZ
Miss Christalla Pithara
Research Student
SoCS
University of Glamorgan
Treforest
CF37 1DL

Dear Miss Pithara,

Project Title: Identifying the benefits and disbenefits of Genetic Services: A Framework for economic evaluation
REC: 06/WSE
R&D reference: PDTR/07/016

Thank you for submitting your proposal to us for approval for the project to be carried out in this Trust. I have received the comments from the Trust review panel and have not received any objections to the project going ahead.

Please accept this letter as approval for the project to proceed here according to the protocol submitted to us. If your project includes participants or resources from other health and social care organisations it is your responsibility to contact the relevant R&D office(s) in order to gain approval to commence there.

This Trust approval is given on the condition that you comply with your responsibilities under Research Governance with respect to us as a participating centre, and that you agree to provide us with information relating to your project as needed to meet the responsibilities and objectives of this Trust. Please remember that any amendments to the study should be agreed by this office, and all serious adverse events as well as those adverse events resulting in completion of an incident form relating to the work of the project in this Trust, should be reported to the R&D department. We also request that you send a copy of your final project report to our R&D office for our reference.

If you have any queries, please do not hesitate to get in touch with me and we wish you every success with the work.

Yours sincerely,

Mr Chris Tattersall
R&D Manager

Wednesday, 7th November 2007
South East Wales Local Research Ethics Committee Panel B

22 November 2006

Miss Christella Pithara
Research Student
Health Economics and Policy Research Unit
Faculty of Health, Sports and Science,
University of Glamorgan
Treforest
CF37 1DL

Dear Miss Pithara

Full title of study: Identifying the Benefits and Disbenefits of Genetic Services: A framework for Economic Evaluation

REC reference number: 06/WSE02/95

Thank you for your letter of 06 November 2006, responding to the Committee’s request for further information on the above research and submitting revised documentation.

The further information has been considered on behalf of the Committee by the Chair.

Confirmation of ethical opinion

On behalf of the Committee, I am pleased to confirm a favourable ethical opinion for the above research on the basis described in the application form, protocol and supporting documentation as revised.

Ethical review of research sites

The Committee has designated this study as exempt from site-specific assessment (SSA). There is no requirement for [other] Local Research Ethics Committees to be informed or for site-specific assessment to be carried out at each site.

Conditions of approval

The favourable opinion is given provided that you comply with the conditions set out in the attached document. You are advised to study the conditions carefully.

Approved documents
The final list of documents reviewed and approved by the Committee is as follows:

<table>
<thead>
<tr>
<th>Document</th>
<th>Version</th>
<th>Date</th>
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<td>Participant Information Sheet: Flow up letter</td>
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<td>Participant Consent Form</td>
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<td>Response to Request for Further Information</td>
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<td>Peer Review</td>
<td>31 October 2006</td>
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<td>Flow Chart</td>
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Research governance approval

You should arrange for the R&D department at all relevant NHS care organisations to be notified that the research will be taking place, and provide a copy of the REC application, the protocol and this letter.

All researchers and research collaborators who will be participating in the research must obtain final research governance approval before commencing any research procedures. Where a substantive contract is not held with the care organisation, it may be necessary for an honorary contract to be issued before approval for the research can be given.

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

06/WSE02/95 Please quote this number on all correspondence
With the Committee's best wishes for the success of this project

Yours sincerely

[Signature]

Mrs A Dowden
Chairman, Panel B
South East Wales Research Ethics Committee
(Dictated but not signed)

Email: jagit.sidhu@bsc.wales.nhs.uk

Enclosures: Standard approval conditions – Non CTIMP

Copy to: University of Glamorgan

Trsforest

R&D Department for Cardiff & Vale NHS Trust
Ref: CT/jth/pdtr/07/016

Mr Chris Tattersall
01437 773823

Chris.tattersall@pdt-tr.wales.nhs.uk

Wednesday, 7th November 2007

Miss Christalla Pithara
Research Student
SoCS
University of Glamorgan
Treforest
CF37 1DL

Dear Miss Pithara

Project Title: Identifying the benefits and disbenefits of Genetic Services: A Framework for economic evaluation
REC: 06/WSE
R&D reference: PDTR/07/016

Thank you for submitting your proposal to us for approval for the project to be carried out in this Trust. I have received the comments from the Trust review panel and have not received any objections to the project going ahead.

Please accept this letter as approval for the project to proceed here according to the protocol submitted to us. If your project includes participants or resources from other health and social care organisations it is your responsibility to contact the relevant R&D office(s) in order to gain approval to commence there.

This Trust approval is given on the condition that you comply with your responsibilities under Research Governance with respect to us as a participating centre, and that you agree to provide us with information relating to your project as needed to meet the responsibilities and objectives of this Trust. Please remember that any amendments to the study should be agreed by this office, and all serious adverse events as well as those adverse events resulting in completion of an incident form relating to the work of the project in this Trust, should be reported to the R&D department. We also request that you send a copy of your final project report to our R&D office for our reference.

If you have any queries, please do not hesitate to get in touch with me and we wish you every success with the work.

Yours sincerely,

Mr Chris Tattersall
R&D Manager

Withybush General Hospital
Pigwidd Road, Haverfordwest, Pembrokeshire SA61 2PZ
Telephone: 01437 764545
WHYN: 1720

NHS CYMRU WALES

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• Inform the Trust R&D Office of any amendments relating to the protocol, 
  including personnel changes and amendments to the actual or anticipated 
  start/end dates.
• Complete any documentation sent to you by the Trust R & D Office or 
  University Research & Commercial Division regarding this project.
• Comply fully with the Research Governance Framework for Health and Social 
  Care¹, and co-operate with any audit inspection of the project files.
• Undertake the project in accordance with ICH-GCP and the Trust’s Good 
  Research Practice guidelines.
• Adhere to the protocol as approved by the Research Ethics Committee.
• Ensure the research complies with the Data Protection Act 1998.

Yours sincerely,

Professor MF Scanlon
Chair of the Joint Trust/University Peer & Risk Review Committee

CC Dr Ian M Frayling, Institute of Medical Genetics, Cardiff University, UHW

¹http://www.wcard.wales.gov.uk/content/governance/governance-framework-e.pdf

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