National Genetics Education and Development Centre

The experiences and preferences of people receiving genetic information from healthcare professionals

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The experiences and preferences of people receiving genetic information from healthcare professionals

Contents

SUMMARY

The aim of the project

The aim of this project was to find out about people's experiences of receiving genetic information from healthcare professionals and their views on how genetic information should ideally be provided. For the purposes of this project, "genetic information" was defined as information about the genetic basis of the condition.

The project focused on the provision of information by healthcare professionals outside specialist genetics services. Knowing which professional groups patients turn to for genetic information and patients' preferences regarding the provision of such information are important steps in ensuring that relevant healthcare professionals are equipped to address patients' needs and concerns.

Main findings

The main findings from the study are that the people interviewed for this project:

- believed that there is a need for greater awareness of genetic aspects of conditions amongst healthcare professionals, who should be more willing to consider the possibility of a genetic condition, refer patients appropriately and take their concerns seriously.
- wanted healthcare professionals who provide genetic information to do so without judgement; to be mindful of their use of terminology; to tailor the information to the preferences of individuals; and to inform people where they can access further information. They felt that healthcare professionals should be aware that genetic information can have an emotional impact on individuals and may affect the wider family.
- saw the GP's role as referring patients appropriately and providing ongoing support and co-ordination. They felt consultants in different medical specialties could play a greater role in providing genetic information. More support from healthcare professionals in gaining access to appropriate genetic information would be welcomed.

These and the other findings are discussed in greater detail in the body of the report and are illustrated with direct quotations from participants.

How the experiences & views were gathered

People with or at risk of genetic conditions and parents of children affected by a genetic condition were interviewed by telephone to explore their views and experiences. This is an exploratory study, exploring people's accounts and recollections of receiving genetic information from their personal perspectives.

Potential participants were contacted through: the Patient Consultative Panel of the Human Genetics Commission; the Patient Interest Group of the North West Genetics Knowledge Park; and the Chairpersons of all organisations who are members of the Genetic Interest Group. Telephone interviews were conducted between September and December 2006.

Range of perspectives

The people who took part in the interviews represented a range of perspectives both in terms of the type of condition and who in their family was affected.

Receiving genetic information from healthcare professionals

Nineteen people had at some point received genetic information without actively seeking it, most commonly at the point of diagnosis. They had received such information from a range of healthcare professionals.

Twelve people had actively sought genetic information from healthcare professionals, most commonly from a general practitioner (GP) or genetics centre, although a range of other healthcare professionals were approached. The most common reasons for seeking genetic information were to know more about the condition following diagnosis or following the death of their child.

From which healthcare professionals would patients have preferred to receive genetic information?

People were asked to consider, with the benefit of hindsight, from which healthcare professional they would have preferred to have received genetic information.

- Fifteen people said they would prefer to receive genetic information from the specialist with whom they had regular contact: their consultant neurologist, paediatrician or cardiologist. The main reason given was that they had established a rapport with this specialist.

- Thirteen people said they would prefer to receive genetic information from someone at the regional genetics service, most commonly a genetics consultant. They felt a genetics consultant would have detailed and up-to-date information.

- Six people said they would prefer to receive information from an expert in the particular condition that affected them, because they would have the most up-to-date and accurate information in an area that is constantly developing.

- Four people said they would prefer to receive genetic information from their GP, who was their first point of contact, but felt GPs were currently not well enough informed to do so. Nine people felt that the GPs' role was not to provide genetic information themselves but to direct patients to appropriate specialists for advice.

- Two people said they would prefer to receive information from a nurse specialist, who can develop close links with families.

- Two people suggested that the personality and communication skills of the person providing genetic information were more important than which healthcare profession they worked in.

When would patients prefer to receive genetic information?

People felt there was a need for genetic information at particular life stages, such as when considering having a family or during pregnancy. They acknowledged that genetic information often needed to be given at a difficult or emotional time.

Participants suggested that the amount of information desired at diagnosis differs according to the individual person involved.

Participants wanted healthcare professionals to tailor the information provided to the needs of each individual.

Access to further genetic information at a later point in time was considered important. Provision of written information which could be re-read later was considered very useful.

What do patients receiving genetic information expect of a healthcare professional?

The interviewees perceived a need for greater awareness about genetic conditions amongst healthcare professionals to ensure early diagnosis and appropriate treatment. People suggested that healthcare professionals should be more willing to consider the possibility of a genetic condition, refer patients for investigations and take parents' concerns seriously.

There was general acknowledgement that healthcare professionals cannot be expected to know everything, particularly about rare genetic conditions. However, participants felt they should be willing to acknowledge their lack of expertise, seek advice from other specialists and refer patients on for further investigations or information. Some felt that healthcare professionals should know where to access information if they needed it.

The people interviewed felt that genetic information should be given by healthcare professionals in a non-judgemental and unbiased way.

The need for holistic care

Twelve people commented on the need for a holistic approach to care. Management of genetic conditions may involve a number of different specialties, resulting in a perception of fragmented care and lack of communication.

Some felt that one healthcare professional, either the GP or geneticist, should take overall responsibility for the patient's care, co-ordinating care and information from different specialties.

Other issues in providing and receiving genetic information

One of the acknowledged difficulties of providing genetic information was that the information desired was not always available. Some people had relied on the advance of knowledge to provide answers to their questions.

A number of interviewees said that they often have more knowledge about the particular condition that affects them or their family than many of the healthcare professionals they encounter.

Two people stressed the importance of genetic information being presented simply, using layman's terms. Some terminology such as "mutants" and "risk" was considered unhelpful.

Genetic information can have an emotional effect on individuals and on the wider family. People described how genetic information can lead to feelings of guilt or blame within families, or may raise issues that have not previously been openly discussed.

What other sources of genetic information do patients access?

All 27 interviewees mentioned sources of genetic information outside the information they had received from healthcare professionals. The most commonly cited sources of such information were patient support groups and the Internet.

Some interviewees suggested that healthcare professionals should direct patients to relevant support groups and reliable websites.

INTRODUCTION

1.1 Background

Increasing numbers of people and their families are asking whether their medical condition may have a genetic cause, and may approach a healthcare professional for more information. Patients have repeatedly stressed the importance of the quality of information they receive from healthcare professionals (Bristol Royal Infirmary Inquiry, 2001; Butow et al, 1997; Beisecker and Beisecker, 1990; Cassileth et al, 1980). For people with or at risk of genetic conditions the information they may receive from healthcare professionals falls into two types: information about the specific condition and information about the genetic basis of the condition, including how the condition is inherited and whether other family members could be affected. This project is about the latter: information about the genetic basis of the condition, hereafter referred to as "genetic information".

Before this project was carried out, little was known about how and from where patients initially receive genetic information, or about patient views and preferences regarding the provision of such information. Several reports have identified that it is important for a range of healthcare professional groups to have an understanding of genetics (Department of Health, 2003; Kirk et al, 2003; Burton, 2003). A pilot study conducted by the Genetic Interest Group (1999) explored experiences of services and support in families with four genetic conditions, but it focused on pathways of care experienced by individuals rather than specifically on the provision of information. Knowing which professional groups patients turn to for genetic information and patients' preferences regarding the provision of such information is an important step in ensuring that relevant healthcare professionals are equipped to address patients' needs and concerns.

1.2 Project aims

The overall aim of this project was to find out about people's experiences of receiving genetic information from healthcare professionals and their views on how genetic information should ideally be provided. The views of people with or at risk of genetic conditions and parents of children affected by a genetic condition were explored. The two main project aims and related areas of inquiry are outlined below.

1. **To explore people's experiences of receiving genetic information from non-genetics healthcare professionals (that is, healthcare professionals outside specialist genetics services).**

1.1 How and when did the person become aware that the condition was a genetic condition?

1.2 Did the person seek information about the genetics of the condition from non-genetics healthcare professionals? Which healthcare professionals did they approach? Were those healthcare professionals able to provide the information sought?

1.3 From which healthcare professionals did the person receive information about the genetics of the condition, and at what stage of diagnosis and care?

2. **To explore people's preferences with regard to receiving genetic information from healthcare professionals.**

2.1 With hindsight, from which healthcare professional(s) would the person have preferred to receive information about the genetics of the condition?

2.2 What genetic information would the person have liked to have received at different stages of diagnosis and care?

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1.3 Methods

A qualitative methodology was chosen to address these project aims. The approach adopted for this project was telephone interviews with people with or at risk of genetic conditions and parents of children affected by a genetic condition, to explore their experiences and views. This strategy itself was informed by patient views: at a focus group of patients with genetic disorders held by the NHS National Genetics Education and Development Centre in September 2005 patients highlighted the value to education and service provision of patients telling their stories and sharing their experiences. The project was approved by the South Birmingham Research Ethics Committee (ref 06/Q2707/221) and the Research and Development Department at Birmingham Women's Healthcare NHS Trust.

This is an exploratory study, designed to achieve greater understanding of peoples' experiences and views rather than to test a particular theory or hypothesis. It explored people's personal accounts and recollections of receiving genetic information, in many cases several years after the event. As a result, those accounts were not always linear and the sequence of events was not always clear. In addition, accounts were given solely from the interviewees' perspective: the healthcare professionals involved may have different perspectives on the events described. The purpose of this study is thus not to provide a definitive account of people's experiences of receiving genetic information but to increase understanding by exploring the interviewees' recollections of their own experiences and identifying common themes.

Information and invitations to take part in the project were disseminated to potential participants through: the Patient Consultative Panel of the Human Genetics Commission; the Patient Interest Group of the North West Genetics Knowledge Park; and the Chairpersons of all the organisations who are members of the Genetic Interest Group (who were asked to disseminate the information to their members, where appropriate).

People expressing an interest in participating in the project were sent a more detailed information pack and, if they decided to take part, were asked to return a signed consent form. All participants were informed that they could withdraw from the study at any time and were under no obligation to disclose personal or upsetting information. In total 48 people requested further information before December 2006 and 28 returned consent forms, 27 of whom were interviewed[1]. At the start of the interview participants were told that they could stop the interview at any time and could decline to answer any question, although no interviewee did so.

Between September and December 2006 27 telephone interviews were conducted by two members of the research team (SB and JB). Most interviews lasted approximately 30 minutes (the shortest was 12 minutes, the longest 1 hour 14 minutes). All interviews were audio-recorded and transcribed verbatim by a member of the research team (SB) and the accuracy of a sample of transcriptions (30%) was checked against the recording by a second member of the research team (CB). The key themes were agreed at a meeting of all members of the research team after they had each read a selection of transcripts. The transcripts were then analysed according to those key themes using NVivo qualitative analysis software. In order to maintain anonymity, conditions and participant identifiers have not been linked to quotations.

[1] Two consent forms were returned by members of the same family and only one was interviewed, having decided themselves which person would take part in the project.

RESULTS

2.1 Range of perspectives

The people who took part in the interviews represented a range of perspectives in terms of the types of conditions, who in the family was affected and when genetic information was first received.

The range of conditions represented included:

- autosomal dominant conditions (including: osteogenesis imperfecta; Gorlin syndrome; hereditary multiple exostoses; Marfan syndrome; hypertrophic cardiomyopathy and arrhythmogenic right ventricular cardiomyopathy);
- autosomal recessive conditions (including: Gaucher disease; sickle cell disorder; retinitis pigmentosa and primary ciliary dyskinaesia);
- X-linked conditions (including Fragile X; ectodermal dysplasia; adrenoleukodystrophy; and adrenomyeloneuropathy);
- chromosomal conditions (including translocation involving chromosome 22; cri du chat; and Trisomy 21); and
- a multifactorial condition (autism).

In addition, people spoke about: familial spastic paraplegia; familial dilated cardiomyopathy; sudden death they believed was a result of Long QT; and receiving genetic counselling regarding family history of cancer. Some families were affected by more than one genetic condition and were asked about their experiences for each one. This means that individuals may have given different answers, for example about when genetic information was first received, for different conditions (thus, numbers in the following Tables do not sum 27).

People spoke from a range of personal perspectives. Some individuals spoke from more than one personal perspective, for example, being personally affected and being parents of children affected by the condition. Twelve of the people interviewed were personally affected by a genetic condition and in addition four described themselves as carriers. Twenty people had a child or children affected by a genetic condition and, of those, four children had died as a result of the condition. In addition, two people had a child who had died suddenly, thought to be as a result of a genetic condition (Long QT), although the diagnosis was not definitive. Two people had spouses affected by a genetic condition and one of those spouses had died as a result of that condition. Two people had personally received genetic counselling regarding family history of cancer.

Most people described the process of gaining information about the genetics of the condition as occurring over a number of years. An indication of the number of years ago they recalled first receiving genetic information is shown in Table 1. The most recent example was someone who had received genetic information one month before the interview but the majority of people said they had first received genetic information a number of years before the interview took place.

Table 1 When participants recalled first receiving genetic information

Time Period	Responses from 27 interviewees[2]
Less than 1 year	2
1-5 years	6
6-10 years	7
Over 10 years	10
Over 20 years	3

[2] Some participants spoke of more than one condition.

2.2 Receiving genetic information from healthcare professionals

Of the 27 people interviewed, 19 said they had received genetic information without having sought it and 12 said they had actively sought genetic information from healthcare professionals[3].

The 12 people who spoke about actively seeking genetic information mentioned five main reasons for seeking that information:

- four wanted to know more about the condition following diagnosis;
- four wanted to know more about the condition following the death of their child;
- two wanted to know about the chance of having a child affected by a genetic condition;
- one wanted to know if their condition was linked to their father's condition; and
- one wanted to know why the doctor had asked whether she was related to her husband.

They had sought information from a range of healthcare professionals, as shown in Table 2.

[3] Some participants spoke of more than one condition
[4] Some participants spoke of more than one condition; some had both sought information and had it provided by healthcare professionals at different points in time

Table 2 Healthcare professionals approached by patients

Healthcare professional	Responses from 12 interviewees[4]
General practitioner	6
Genetics consultant (approached genetics centre direct)	4
Cardiology consultant	2
Paediatrician	1
Paediatric respiratory consultant	1
Brittle bone specialist	1
Accident and Emergency consultant	1
Dermatologist	1
Thyroid specialist	1
Long QT specialist in America	1
Consultant metabolic physician	1
Cancer specialist	1

Most commonly patients had either sought information from their general practitioner (GP) or had contacted a genetics centre directly. Some had raised questions with healthcare professionals they had previous contact with, for example asking questions during a routine appointment with their paediatrician or contacting the accident and emergency consultant who had treated their child. This prior contact was not necessarily linked to the genetic condition: for example, one person asked their thyroid specialist about the genetic basis of their child's autism.

In other cases, patients sought information from an expert in the clinical area, such as a Brittle Bone specialist, Long QT specialist or consultant metabolic physician, having identified such experts themselves, usually by searching the Internet. Of the 12 people who sought genetic information, eight had at some point spoken to a geneticist or genetic counsellor and four had never spoken to a geneticist or genetic counsellor (of those four, one had been offered a referral to the local genetics service and had turned it down).

The 19 people who had received genetic information without specifically seeking it had received such information from a range of healthcare professionals, as shown in Table 3. The majority (12) had been given this information at the point of diagnosis and two had been given the information during a routine clinic appointment (post-diagnosis). Two people had received the information during pregnancy: one had previously been diagnosed with the condition and had a child affected by it, and the other had carrier testing during pregnancy which revealed that she and her husband were carriers.

One person received information when her young child was diagnosed with the same condition as her, one person had been given the information when a family member was affected by cancer, another during a period of medical investigations before diagnosis had been made. Of the 19 people, 13 had at some point spoken to a genetics consultant or genetic counsellor and six had not.

Table 3 **Healthcare professionals who provided genetic information**

Healthcare professional	Responses from 19 interviewees[5]
Cardiologist	4
Paediatrician	3
Consultant neurologist	2
Obstetrician	2
Paediatric neurology registrar	1
Consultant at eye hospital	1
Dentist	1
Orthopaedic surgeon	1
Cancer specialist	1
Consultant from renal department	1
Neonatologist	1
Gaucher specialist	1

The amount of information initially given, both to those who sought information and those who did not, varied widely. For some, the initial information consisted of the name of the condition, the fact that it was genetic and that they would be referred to a genetics service. Others received detailed information about the genetics of the condition at the initial consultation.

[5] Some participants sought information from more than one healthcare professional

2.3 From which healthcare professionals would patients have preferred to receive genetic information?

People were asked to consider, with the benefit of hindsight, from which healthcare professional they would have preferred to have received genetic information.

The main answers, explored in more depth below, were: the specialist they had regular contact with; someone at the regional genetics centre; a specialist in the particular condition; and their general practitioner (GP).

2.3.1 Speciality consultant

Fifteen people said that they would have preferred to have received genetic information from the specialist they had regular contact with: their consultant neurologist, paediatrician or cardiologist. Of these 15, eight had previously received some genetic information from their specialist and seven had not. The main reason given for this preference was that they had been in regular contact with this specialist and had built up a rapport and personal relationship with them. One person explained:

"I would have preferred to have got the information from someone I knew, and the only one I knew was the paediatrician."

Two people affected by cardiomyopathy particularly emphasised the degree of trust they had built up with their cardiology consultant, one saying:

"If I had a choice in an ideal world it would be the cardiologist, because you build up a very personal relationship with the cardiologist, because … you're putting the life of your child in that person's hands, and the trust is immense … I'm not taking anything away from the genetics people that we did speak to, they were wonderful, but in an ideal world I think because of that personal trust element it would be the cardiologist."

Conversely, one person felt that this personal relationship may mean that some people feel uncomfortable discussing their family history and would prefer to talk to a geneticist or someone more distanced from their care.

There was a general belief that a specialty consultant would have more knowledge about the genetics of conditions in that specialty than, for example, a GP or nurse. However, some expressed concern that the specialty consultant might not be fully up to date with the latest genetic developments, as one interviewee explained:

"It would have been nice to get it from the paediatrician, because we had a nice relationship with her and we knew her. But I think that's probably a bit unreasonable in terms of her training and everything. I mean to be fair it has to be a geneticist who knows that kind of information."

Another person felt that the limited time specialty consultants have to spend with each patient could be problematic, saying:

"if you're in there for more than three minutes you're lucky, that's what you feel like."

2.3.2 Regional genetics service

Twelve people said they would prefer to receive genetic information from someone at the regional genetics service. Of these, eight people specified that information would ideally be given by a "geneticist" or "genetics consultant", two said from a "genetic counsellor", one a "genetic advisor" and one said from "the genetics team". One person explained that she would prefer to see a genetics consultant rather than a genetic counsellor because of her previous experience: about three years ago she had seen a genetic counsellor who was unable to answer her questions about what form of Down syndrome her son had. Following another referral she had seen a genetic consultant, who had been able to answer her questions, leading her to conclude:

"If someone's going to ask detailed questions the only person who can answer those kind of questions is someone who is a geneticist, I can't see that you'd expect any other healthcare professionals to have that level of knowledge."

Of the twelve people who would prefer to receive information from the regional genetics service, eight had had previous contact with the service and four had not. Two people said explicitly that at the time they had initially received genetic information they were not aware that a regional genetics service existed and, with hindsight, they would have liked to have been referred to the service for more information.

The main reasons given for wanting information from the regional genetics service were that they would have detailed and up-to-date information and be able to answer questions. One person explained:

"If you're going to get information like that I would feel more confident getting it from the person who is most highly trained I think, who would be likely to know most about it."

One person commented that appointment times with a genetic consultant were longer than appointments with a GP or specialty consultant, allowing more time to receive information and ask questions.

2.3.3 Expert specialist

Six people said they would prefer to receive genetic information from an expert in the particular condition that affected them. All six named a particular individual or Centre with expertise in that particular condition, one describing the person as a "super-expert". They reported that they had made contact with these expert specialists through Patient Support Groups or by searching on the Internet rather than being directed to them by other healthcare specialists. Once they had made contact they said the expert specialists were very willing to provide information, both about the genetics of the condition and about the condition itself.

The reason for preferring to speak to an expert in the particular condition was that they would have the most up-to-date and accurate information of an area that is constantly developing. One person said:

"A specialist in the actual condition, because things change from year to year and the documents that the doctors and the geneticists probably read last year won't be relevant to this year, because research and everything carries on all over the world, doesn't it? So I only believe to take notice of specialists."

2.3.4 General practitioner

Four people said that they would prefer to receive genetic information from their GP. All four said that the GP was the first point of contact for most patients, one saying:

"I do think that initially it's got to come from the GP because the GP is your first port of call."

However, all four said that they had approached their GP for information but their GP had not been able to give the information they wanted. One said:

"The GPs don't seem to know a lot about my condition…so it's usually me telling them."

Nine interviewees felt that GPs should not provide genetic information, because they know little about genetic conditions, cannot be expected to know about genetics and would not be able to answer questions that patients might have.

These interviewees felt that the GP's role was to direct patients to appropriate specialists for advice rather than providing it themselves. One said:

"They're your first sort of contact and I always think you go to see them and then they sort of know all the little avenues that they can refer you to."

Another stressed that referral was appropriate because GPs cannot be expected to have such specialist knowledge:

"I think it's very difficult for the GP, because you can't possibly expect them to have this level of particular specialist knowledge … The important thing is that within secondary or tertiary care there are referral centres… and then GPs, and I don't wish to, you know, relegate them to a referral service, but it may be the most appropriate."

2.3.5 Nurse specialist

Two people said they would prefer to receive genetic information from a nurse specialist, both of whom had had contact with a nurse specialist in the past. One said that a sickle cell nurse specialist was the best person to provide information to families affected by sickle cell, as a nurse specialist is able to make closer links with families than a doctor does:

"They actually come to the home, you know, so they're able to listen to you more, and take on board other factors like family life and stuff like that, to be able to give you the right information, and to also know how you're actually feeling."

The other person said they would be happy to receive information from the cardiomyopathy nurse with whom they had regular contact and had established a rapport. However, a third person said that, although specialist nurses and health visitors are supposed to know about genetics, in his experience they had known very little about the genetics of the condition that affected his family, namely Fragile X.

2.3.6 The importance of personality and support

Two people suggested that the personality and communication skills of the person providing genetic information were more important than which healthcare profession they worked in. One described the variation between different consultants in the same specialty:

"One person in particular has been absolutely useless and has made me feel worse and I've actually refused to see him again... I didn't like getting any information from him because I didn't feel he cared ... I don't know that it's necessarily somebody that you've built up a relationship with, it's just somebody who's actually prepared to listen and take your questions seriously and answer them."

The other said that, as long as they are well informed, the individual is more important than their specialty, saying:

"It comes down to that individual's personal attributes".

Four people stressed that they had had to find out a lot of information themselves as they had not received adequate information or support from healthcare professionals. They felt that the provision of information from any source would be welcome, one saying:

"They can do it any way they like as far as I'm concerned, but just make it a bit easier."

2.4 When would patients prefer to receive genetic information?

The people interviewed were asked whether they had required different types of genetic information at different stages of diagnosis and care.

In their responses they talked about different factors that had affected their desire and need for information.

2.4.1 Life stages

Eleven people said that they felt there was a need for genetic information at different stages in their lives and their children's lives. Particular points at which they felt information was important were when they were considering having children or during pregnancy.
One person with cardiomyopathy said:

"I think genetics should be raised at the very outset, because I, when I was first diagnosed we hadn't started a family, and although I was sort of vaguely aware that it could be familial we weren't offered any genetic counselling or anything then, which, you know, I think on hindsight would have been important."

Other key points at which they felt information was important were when their children grew up and wanted to start a family or were pregnant. At this point they felt information was important for their children, one saying:

"Now because of my daughter's situation we thought, well wait a minute, is she worried about bringing children up? Can it pass through to her, to her own children?"

Others explained that information at this point was also important for them personally, having previously lost a child to a genetic condition:

"Why I started it all off again this year was because I suddenly thought, you know [daughter's name]'s getting older … if she has children and this is genetically linked, will this sort of nightmare begin again? … I've lost a child and the thought of losing a grandchild and my child going through the same as I'm going through."

Others felt that it was important that their children have access to genetic information when they become older and start to ask questions of their own. One person with osteogenesis imperfecta felt that people with a genetic condition should be offered the opportunity to get information from their GP or a geneticist at the age of sixteen:

"The opportunity to go and discuss it with somebody, at a younger age, rather than being left with it and thinking 'oh my god, I'm going to have disabled children', which you do think when you're a teenager because you don't know what to think … everybody else is normal you're left with this condition. And I think at some sort of transition age, a person, a young person should be offered the information or offered an appointment to attend somewhere to learn more or ask questions about the condition that they've got."

2.4.2 Adapting to individual needs

In terms of the genetic information given at the point of diagnosis, the people interviewed suggested that the amount of information that people desire differs according to the individual person involved. Several people interviewed said that they had wanted as much information as possible at the point of diagnosis but recognised that others might not feel the same way, one saying:

"Some people want to know a lot and others don't want to know anything … I would want to know as much as they are prepared to tell me."

Another person recalled the different responses of herself and her husband at the point of their child's diagnosis:

"It's very difficult, for one to be told that, you know, your [child]'s got a hereditary condition, and, whereas, you know, I wanted to learn everything and my husband was quite upset, I mean obviously I was as well, but he didn't, he needed, yes, he needed time to digest it all … sometimes it needs to be done in stages and sometimes, it all just depends on the person really."

The need to tailor the amount of information provided to the needs of the individual person was therefore considered very important.

2.4.3 Receiving genetic information at a difficult time

Several people commented that the point at which they had received genetic information had been a very difficult period for them. Some were coping with strong emotions brought about by, for example: the death of their child; the fact that their brother was dying; their child experiencing numerous health problems; and insensitive reactions of other family members.

Many talked about the conflicting priorities they had experienced at the time. For some, learning to cope with the day-to-day care of their child was a more pressing immediate concern than genetic information. One person explained:

"The questions were there, but because we were busy getting on with looking after the baby they weren't an imminent thing that we wanted, that we were going to go and sort out ourselves then because we just didn't have the time or energy."

One respondent said her need for information had first focused on how to care for her child day-to-day, then on behaviour and education, and the desire for genetic information had come later. Another said:

"In a way the genetics were just something, you know, it's happened. And there was nothing that you could do or worry about at that stage, I was just more concerned with what was physically going to happen to [child's name] over the next few months … [genetics] just didn't seen terribly relevant when I was faced with [child's name] suddenly being physically handicapped."

The shock of the diagnosis meant that some people felt they could initially take in only a limited amount of information. Many people found it difficult to recall details of the information provided at this point, describing it as "a bit of a fog at the time". One described how the provision of detailed genetic information would have been inappropriate at the point of diagnosis:

"[At diagnosis] he didn't explain the genetics, and I don't think it would have been appropriate. He was, I think at that time he was just conveying two things to us, two very important things. And the first was, yes, we were right this is genetic. And the second, what is more, you now need to be tested because in a number of cases this is inherited … I think if anybody'd tried to give me more detail then I'd have, you know, I simply wouldn't have taken it in."

However others felt that they did need detailed genetic information at an early stage, despite it being a difficult time. For example, one person whose child had cri du chat felt that, despite the conflicting priorities of dealing with numerous health problems, he wanted to have genetic information at the point of diagnosis:

"I need to know why or what's going wrong so that I can understand it, and I find it much easier to accept, so if I can picture what went wrong and where it went wrong, not to put blame on anyone, at least I bloody understood it, you know. And if it was a genetic thing, then we could warn [siblings] … if there was congenital abnormality that was familial then of course they would need to know as well because they were producing at the time too."

Some people recognised that being in hospital affected the way that they engaged with the information given, one saying that, although they were given the opportunity to ask questions, they did not do so because of the influence of the hospital setting:

"I think I had the time, I could ask what I like, but I just didn't, it was given out, the literature, and I think they were just doing the tests on me, and yes, I think you're in hospital and you're sort of in another world, aren't you?"

2.4.4 Access to further genetic information

Given the different responses of individuals at the point of diagnosis, and the fact that initial information may be given at a very difficult time, several people said it was important for people to be told where they could access further genetic information at a later point in time. This would mean that people who later wanted more detailed information could access it easily. Provision of written information which could be re-read later was considered very useful, both to increase their own understanding and also to help them explain the basic genetic concepts to others.

It was also considered very helpful if healthcare professionals invited patients to contact them later if they had questions or required further information.
One person said:

"I feel very strongly that clinical genetics departments should always leave the door open, there should always be a 'phone number which families can use to make contact as and when new questions arise, because they do arise."

2.4.5 Genetic testing

Five people talked specifically about genetic testing. Genetic testing can be a technologically demanding procedure and it may take many months before the final result is available. Four interviewees spoke of the long wait they had experienced between giving blood and receiving the test results. One person felt that the results should have come back much quicker; another two said that, while the wait was expected, it was difficult, one describing it as an "anxious time" and the other saying it was "almost like holding your breath". However, a fourth person felt that the wait had enabled her to adjust to the implications of the test results, saying:

"It was three months before we got the results, and I don't at any stage remember wishing that the time had been shorter. I think we needed a lot of time to adjust."

Three people spoke about genetic testing for their children. Genetic testing is not usually performed in children for adult onset diseases or for carrier status until the child is able to make the decision for him or herself, unless the result is required for diagnosis or management.

However, one interviewee said that they wanted to have their child tested and did not wish to wait until their child was older:

"I would just like to know one way or the other … You know children do get pregnant by mistake and I just feel that my request should really be taken more seriously."

Two other interviewees said they had had their children tested for carrier status before the age of sixteen, despite being informed of the ethical questions involved, because they wanted to know the implications for their family. One felt that it was less traumatic for children to be gradually introduced to the concept of being a genetic carrier than to wait until they were eighteen.

2.5 What do patients receiving genetic information expect of a healthcare professional?

During the course of the interviews the people involved talked about their expectations regarding what different healthcare professionals should know about genetics and about genetic conditions. The main themes raised are explored below.

2.5.1 The need for greater awareness of genetic conditions

A key theme was the perceived need for healthcare professionals to have greater awareness about genetic conditions and be more willing to consider the possibility of a genetic condition. Eleven people gave examples where they felt that greater awareness amongst healthcare professionals would have led to earlier diagnosis or more appropriate treatment. These included: a basal cell carcinoma that a GP treated as eczema for many years before referral by a dentist for a jaw cyst led to the diagnosis of Gorlin syndrome; and a child having a lump for several years before the GP referred to a specialist resulting in the diagnosis of hereditary multiple exostosis. One person stressed the importance of healthcare professionals having greater awareness of cardiomyopathy and the need to monitor and treat it differently to coronary heart disease:

"GPs, local nurses, nurses on cardiac units in District General Hospitals, doctors in District General Hospitals, they're brilliant with coronary heart disease and they make assumptions about people with cardiomyopathies that aren't appropriate because their experience is in coronary heart disease … It's not the same as coronary heart disease, the symptoms aren't the same, the outcome isn't the same, and it needs to be treated differently."

Some people were angry that diagnosis had taken so long, and felt that it could have been achieved earlier if healthcare professionals were more willing to consider the possibility of a genetic condition and to refer patients for investigations. In some cases, people had repeatedly asked healthcare professionals whether there was any significance in the fact that other members of their family had experienced similar symptoms and were incorrectly told that there was no link.

Three people described how they had found it difficult to get their concerns about their child taken seriously by healthcare professionals. One said:

"My health visitor was telling me that I was a neurotic, overprotective mother … And it wasn't until I started finding out more information that, you know, it was a case of well the health visitor's wrong."

Another, whose son was diagnosed as having Gorlin syndrome at age 12, said she had raised concerns when he was a toddler that he was not meeting the normal milestones such as sitting up and crawling and felt that the doctor and health visitor had labelled her as an anxious parent and had not listened to her concerns. The third had been advised to see a psychiatrist because her concerns about her child were imagined rather than real: the child was later diagnosed as having dilated cardiomyopathy.

2.5.2 Acknowledging lack of expertise

There was general acknowledgement that healthcare professionals cannot be expected to know everything, particularly about rare genetic conditions. However there was strong agreement that healthcare professionals should be willing to acknowledge that they are not experts in all areas. One person said:

"The first port of call is the GP, and if the GPs can be big enough and hold their hand up and say they don't know it all, it makes them appear to be more human."

People felt it was important that GPs in particular should be willing to ask other specialists for their opinion or refer patients on for further investigations or to get more information. One commented:

"It's about honesty, and you know just saying 'well I'm not sure what it is, okay let's just point you in the right direction and, you know, can you get an x-ray and see what happens, see it from there'."

Doctors who gave misinformation, for example that a relative was not at risk when they actually were, were viewed very negatively. One example highlights how the patient would prefer to be told that the doctor does not know the answer rather than given incorrect information:

"The registrar we had issues with because he was just completely uninformed about the whole condition, and gave us misinformation … I feel incredibly angry about it actually, I mean I don't mind somebody saying 'look this is very rare and I just don't know the answer but I will look it up for you', but to just make up an answer is absolutely unforgivable I think."

One person also said that doctors should not withhold information from patients or parents, for example about the suspected diagnosis, as they had a right to know this information.

Five people felt that healthcare professionals should show a willingness to get informed if they knew little about a particular condition. One had taken written information about her condition to the GP who responded by saying that he did not need to know about the condition. She said: "he seemed to want to wash his hands of it". Four people noted the importance of healthcare professionals knowing where to access relevant information when they needed it. One suggested that GPs could look up relevant information on the Internet before a consultation:

"Because they are now mostly netted up they could gen up on it in the five minutes before you walk in the room. So they have that possibility, but only if they have the nouse to look for it."

Another person suggested that computers in dental surgeries could flag up relevant issues, for example where a patient's medication might affect their dental treatment. Another suggestion was that healthcare professionals on maternity wards should know where to access information leaflets about different conditions that they could then pass on to relevant parents. In general people wanted more support from healthcare professionals in seeking information, including informing patients where they could access further information if they wanted it. One person suggested that healthcare professionals could show people where to access reliable information on the Internet, ideally letting them look up information on a computer then and there.

2.5.3 Providing information in a non-judgemental way

Another theme that emerged from the interviews was the expectation that healthcare professionals should give information in a non-judgemental way. One person said:

"There should be no judgements on it at all … I mean people don't like being judged by people they don't know."

This theme particularly emerged in interviews with parents of children affected by a genetic condition. One person, a mother with a child affected by a genetic condition, said she had felt judged by a number of healthcare professionals:

"People kept saying 'didn't you have any antenatal testing?' as if to say 'what's he doing here?' You know, either you shouldn't be shocked that he's here or he shouldn't be here anyway because you should have had a test and got rid of him. I came across that a few times from medics. And I found that very difficult to deal with because I had to keep justifying my son's existence."

She felt that healthcare professionals had a negative perception of the condition and had given information in a biased way:
"I think generally disability has a very negative view, and I think instead of accepting people have disabilities, the information's coming across really to aid terminations, rather than to aid people to think about, well, what's this going to mean for us now and the fact we're going to have to change our lives."

Similarly, a mother with children affected by a genetic condition felt that information at the time of her antenatal scan was biased towards abortion:

"They were trying to help me get rid of it. Whereas I just wanted help, to be told what they saw on that scan, so I could be ready for it when it did come out."

She felt that more training was needed for people who give information at antenatal scans, so that the information is given in an unbiased way and the person is supported to make a decision:

"I don't think they're ready for if they do find something that's not quite right or they do find the genetic condition's been passed on. They should have someone there to support them, the parents, through making the decision, whereas it was just a scanner and a doctor asking me did I want a termination. I don't think they'd ever heard of the condition before … I felt like I was all by myself. "

A number of people suggested training in counselling skills for healthcare professionals to prepare them for providing information in a supportive and non-judgemental way.

2.6 The need for holistic care

Another key issue, raised by twelve of the people interviewed, was the need for a holistic approach to care. They noted that treatment of genetic conditions often involves a number of different specialties as the condition affects more than one part of the body. This sometimes resulted in people feeling that their care was fragmented, with nobody addressing the condition as a whole or taking a holistic approach to the overall care. One person said:

"There certainly didn't seem to be any communication between the various specialists who were treating, and it was as if, 'well I'll deal with this bit', and 'I'll deal with this, he'll deal with such', and as such I think the psychological aspect of having a long-term genetic condition that needs to be managed by surgery and whatever was overlooked, there was no holistic approach."

Lack of communication between different specialties was viewed as a particular problem. Some people noted that different specialists appeared to work in isolation from each other rather than as one multi-disciplinary team. One person said:

"Most health professionals, they seem to be blinkered in the sense that they, they're experienced in their field and that's all they bother with, they don't actually talk to others."

Another person described the approach as "bitty", with different specialists ordering scans for different parts of the body but no-one taking an overview or co-ordinating those investigations:

"It could be a spectrum of problems, like genito-urinary, it could be cardiovascular, it could be gastro-intestinal. And it was bitty in the way that you couldn't get one person to do one scan of everything and tell all three of them in one go … It's like digging up the road. You've got BT digging up one day, the gas digging up the next, and the water the day after. And why couldn't they have done it in one go?"

Six people interviewed suggested that one healthcare professional should take overall responsibility for the patient's care, co-ordinating care and information from different specialties. Two people felt that this co-ordination role should be undertaken by the genetics service, one saying:

"The geneticists are very good, and they do I think explain things to families quite well. But in some respects they tend to sort of segregate themselves as just geneticists … They're basically doing their job, but then they're not looking at the family and thinking "well you're going to need more information from different people and these are the people you need to go and see".

Four people felt that the co-ordination of care was the responsibility of the GP, one noting that the GP should already receive information from each of the specialties involved in the patient's care and was therefore in the best position to have an overview of the care as a whole. One person described how their GP had supported them when their child was diagnosed with a rare chromosome disorder by searching for and collecting information about the condition. She felt it had probably not taken the GP much time to collect that information but described the feeling of support as "transformative".

In contrast, other participants felt unsupported in their searches for genetic information. One said she had not received any genetic information from healthcare professionals at the point of diagnosis of her child:

"We were basically just left to get on with looking after him in hospital really. There was nobody to talk to about the genetics of the condition or anything."

She felt that more could be done to support people who want more information, to make them aware of the information and services that are available and facilitating access to those services and information for those people who wish to learn more:

"They could actually do something about it for you rather than leave it to you to have to actually make the first steps yourself towards it."

Another participant also said she would have welcomed more information from healthcare professionals, as she felt she had to find the information she wanted herself, commenting:

"…trying to get information was like trying to get blood out of a stone."

Other people noted the lack of support they felt after they had been given genetic information. One described this lack of support as a "complete hole", another said "we were just left to it really, to deal with it". Follow-up contact or access to further information was considered very important.

2.7 Other issues in providing genetic information

A number of issues were raised during the interviews that were specific to genetic information and information about genetic conditions. These are outlined below.

2.7.1 Information not available

Twelve people interviewed noted that one of the difficulties of genetic information was that the information desired was not always available. Genetics is a rapidly developing area and scientific advances continue to provide more information, but there are currently many questions for which even the experts in the field do not have the answers. One person explained:
"I don't have all the information I need, but I do feel I've got all the information there is."

She added that her condition was so rare that the specialists were learning a lot from her own symptoms and treatment. Another person described how she had to wait until reliable tests were developed before her children could be diagnosed with Fragile X: the consultant had recommended that she contact a genetics department every five years as the gene would eventually be discovered.

Two people said that the advance of knowledge meant that they had initially been given information which later turned out to be incorrect, but which had been considered correct at the time. For example, one person was initially told that her children would only inherit mild osteogenesis imperfecta, of the same Type as her own. She said that this was considered to be correct at the time, but has since been found to be incorrect.

One person noted the difficulty healthcare professionals faced in keeping up to date in such a rapidly advancing field:
"My niece has just started medicine and what she'll be needing to know in five years time when she finishes will be completely different to what doctors that started five years ago need to know now, the way things are moving."

2.7.2 Expert patients

Given that genetics is a rapidly advancing field and that many genetic conditions are very rare, a number of interviewees noted that they often have more knowledge about the particular condition that affects them or their family than many healthcare professionals. Comments included,
"we normally know more about it than they do"
and,
"Doctors don't really know much about this condition as well, and we find that we actually teach them more than they teach us."

One person described feeling disconcerted that she knew more about her condition than her GP:

"Like I say, every GP I've had in the past I've sort of had to tell them about me, which sort of worries me in a way. You think 'oh dear, you're a doctor and you don't know'. So yes, I think, if they had more knowledge of these things it would make you feel more confident."

2.7.3 Terminology and genetic concepts

Another issue specific to genetic information is the related terminology and concepts. Two people stressed the importance of genetic information being presented simply, using layman's terms rather than scientific terminology, one person advising:

"Explain it in simple terms for people who sort of don't understand medical terms … Treat you as if you, you know absolutely nothing about this."

One person described how she felt the term "genetic mutation" could lead to the scary and unhelpful term "mutant" which she had heard a number of healthcare professionals use in the past five years since her children were diagnosed. Her advice was:

"I think principally, don't ever call anybody a "mutant" although it might be the technical scientific term, it's not a very nice thing to call anybody."

Another person said that in the past three years since her child's diagnosis a lot of healthcare professionals had talked of "risks" which she found unhelpful terminology, suggesting that "chances" was a better term to use:

"I mean to talk about my son's life as something which is regarded as a risk is not a very nice way of regarding him as a person."

One interview highlighted that, as well as terminology, genetic concepts may be difficult to understand. The interviewee explained that she had not understood that the 1 in 4 chance of a child being affected by the condition applied to each pregnancy and as a result had a child affected by the condition. Comments by other people demonstrated that the term "genetics" can be interpreted in different ways. For example, there were differing conceptualisations of what constituted genetic counselling.

2.7.4 Impact of genetic information on the family

People talked about the impact genetic information could have for the whole family. Some genetic conditions are inherited and therefore there may be implications for other members of the family. Six people spoke about the feeling of guilt that a genetic condition can cause within families. Some said that they or their spouse had felt guilty when they discovered that they had passed the condition to their children. One gave this advice to healthcare professionals:

"Just be a bit conscious that having a child ... with a genetic condition does make people feel guilty, whether that's right or wrong and I know there's absolutely nothing you can do about it, but all the same it makes you feel guilty. And it also makes your parents feel guilty, so grandparents feel guilty as well if they've passed on a genetic condition."

The implications for grandparents was also raised by another interviewee who said that her mother had been very anxious when waiting for test results: "she immediately blamed herself and thought it was all her fault". The interviewee went on to talk about the difficulties genetic information can cause between couples if one spouse apportions blame to the other, and although she had not experienced this with her husband, she had seen it in other families. She felt that genetic information should be given sensitively and that healthcare professionals should be aware of these potential family dynamics:

"I think it's sort of important to get over the fact that it really isn't anyone's fault, nobody can help it ... So the person that gives the information, sort of, to be aware that there may be sensitive issues between the couple really. That one's going to feel guilt and the other might feel rather hard done by."

The way that healthcare professionals provide genetic information can have a strong impact on people's emotional reaction to that information. One participant described the positive emotional impact of a consultation with a genetics registrar:

"What he said to us that was unbelievably helpful was, 'There is only one difference between you and everybody else in the outside world there, and that is that you know about your chromosomes, everybody else could be carrying it, just as you were, but they just don't know'. And that was for me an unbelievably reassuring message, because you feel like a pariah, you feel as though you've been cast out into social outer darkness ... I felt very singled out, and that was probably for me potentially the worst time in that I felt that I'd carried something that had killed my son ... And he helped very significantly to alleviate that sense of being a pariah."

Genetic information also had implications for the family in other ways. For some, exploration of the family history of the condition raised issues with family members that had not previously been openly discussed. For example, one person described how the death of her sister had not previously been talked about in her family:

"My mum was of a generation where you didn't talk about these things ... it was painful to talk about."

Getting detailed information about the medical history of the family was not always possible, because people were not in close contact with some branches of the family or because some family members had died. People also stressed that the diagnosis of a genetic condition may have implications for other family members, who may need to be informed, and again this could be a difficult process. One person described the mental strain such information had placed on her son, who was struggling to cope with the implications of his sister's diagnosis. Others explained how they had used humour within their own families as a coping mechanism to deal with the implications of a genetic condition in the family.

2.8 What other sources of genetic information do patients access?

Although it was not an explicit question in the interview schedule, all 27 interviewees mentioned sources of genetic information in addition to the information they had received from healthcare professionals.

The main sources of information they discussed are outlined below.

2.8.1 Patient support groups

Membership of or contact with patient support groups was cited by 24 interviewees as an additional source of information. This is perhaps not surprising given that recruitment for the project was largely through patient support groups.

Of the six people who stated how they had initially made contact with the support group, three had used the Contact a Family directory (available at www.cafamily.org.uk) and the others were directed to the group by healthcare professionals: a geneticist, a medical specialist and a neo-natal nurse. Seven interviewees were now involved in the running of the support group, for example as founder members or on the Committee. They felt they had gained a lot of genetic information about the condition through this involvement, either through seeking information as part of their role or through discussion with specialists on the Committee or on the Medical Advisory Board.

Six people recalled receiving an information leaflet from the patient support group which included genetic information, which they had found useful. One person explained the value of having written information that they could revisit at a later date:

"If you've got something in your hands, I've kept the leaflets and I can go back to them if I want to remind myself of something. I have found those very useful."

Another noted that, whilst the basic genetic information had been given to them by the genetics service, they had not received further information from healthcare professionals and the information from the patient support group was therefore very valuable:

"I mean everything we know about the genetics really subsequently has been through the [patient support group]."

Six people mentioned that they received regular newsletters from the patient support group, which included information and updates on the latest research findings, for example. Three had accessed information from the group's website.

Five people said that they had gained information from contact with other patients and families which had been facilitated by the support group. One person said:

"To meet other families with children like mine has been fantastic. And we all learn off each other."

People described sharing information about education, treatments and access to benefits with other families as being valuable, but genetic information was also shared: for example, one person believed that the condition only affected males, as it had only affected males in his family, until he spoke to someone with an affected daughter.

People had also gained information from annual conferences organised by patient support groups. For some, these provided a forum where they could make contact with healthcare professionals who specialised in their particular condition, speak to those expert specialists and ask questions.

For example, one person approached a specialist doctor at a patient support group conference and asked what the chances were of her passing the condition to her children: she had not received this information from any other healthcare professional.

Patient support groups were generally seen as very valuable sources of information and support. One person said that such societies:

"…are doing a wonderful job in helping to publicise the existence of these conditions and the need for proper advice".

Another praised the level of genetic information she got from the patient support group:

"I got the answers, I have to say excellently, through [the patient support group]. I was staggered at the professionalism and the level of detail that I received, actually it was much better than the service I'd received from the NHS, and as you can see from what I've said the NHS service I received was really excellent."

2.8.2 Genetic information on the internet

Eighteen interviewees mentioned using the Internet to find genetic information. Some found it a very valuable tool, giving them access to information from the latest research in other countries, such as the USA, and providing them with much more information than they received from healthcare professionals. One person said:

"I just literally looked at anything with [condition name] in and read it all, I just couldn't get enough information about the condition."

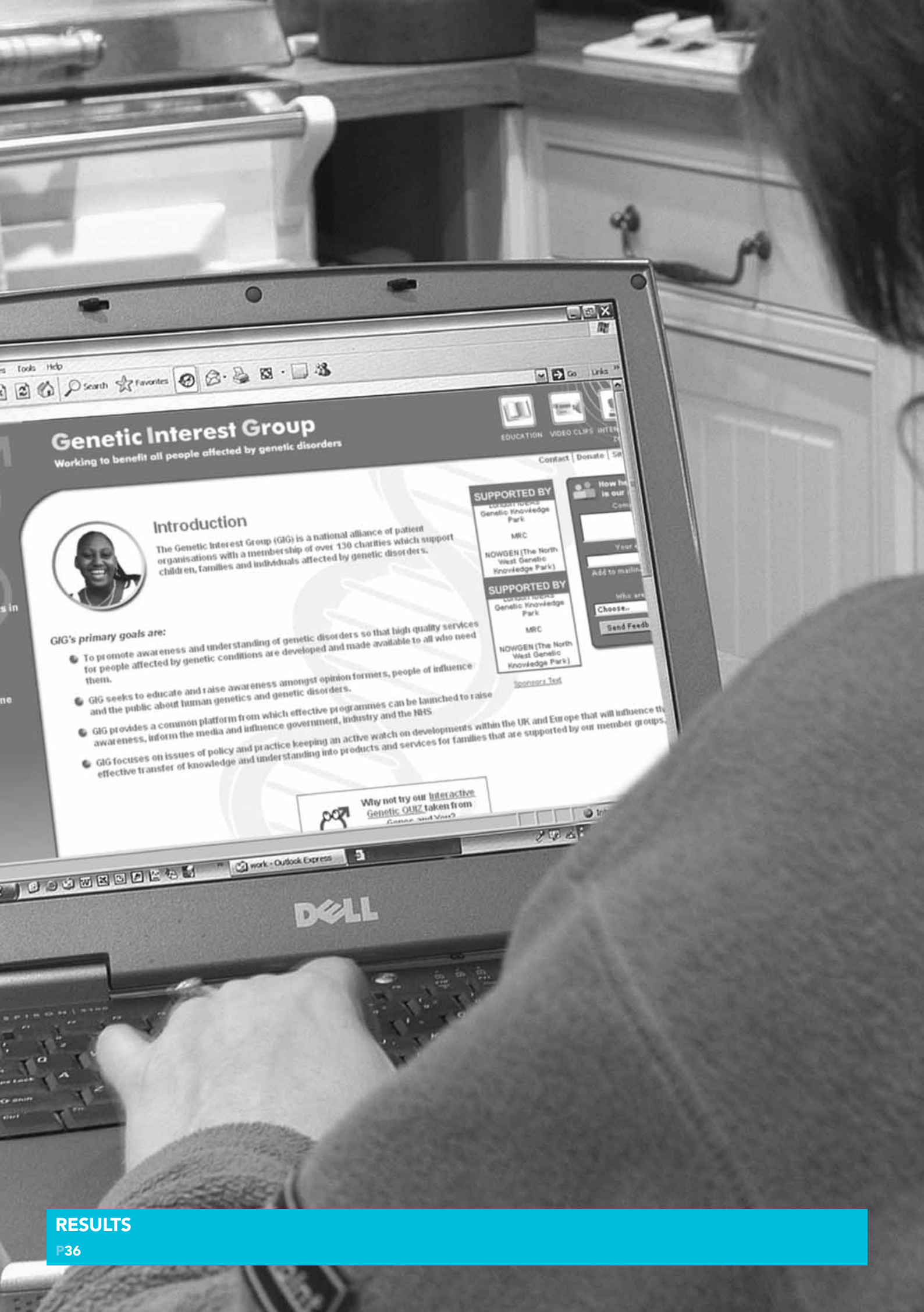
Another person felt that information on the Internet was potentially less biased than information from healthcare professionals: he had recently read an article on the Internet about the patient's right to refuse treatment, a topic which he felt a doctor was less likely to discuss.

One person had found the Internet to be a very valuable tool as it enabled them to identify and make contact with an expert specialist in their condition. They had searched medical journals on the Internet to see who had published on the topic, found contact details for one of the key expert specialists and contacted him directly by telephone.

They described the expert as being "desperate to see us" because the condition is very rare. They now have regular appointments with this specialist, who explained the genetics of the condition in the first consultation.

However, other people had found the Internet a less useful source of information. Some people had been confused as information on the Internet conflicted with what their healthcare professional had told them. Another person had initially incorrectly self-diagnosed himself as his symptoms appeared to match those listed on the Internet. Some people felt that some websites were more trustworthy than others and that some websites tended to over-dramatise the condition. One person said:

"I looked it up on the Internet as well, but to be honest early on I decided it was a bad idea because all I heard was probably the worst case scenarios which wasn't what I was wanting to hear at the time."


Genetic Interest Group
Working to benefit all people affected by genetic disorders
Introduction
The Genetic Interest Group (GIG) is a national alliance of patient organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders.
GIG's primary goals are:
To promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them.
GIG seeks to educate and raise awareness amongst opinion formers, people of influence and the public about human genetics and genetic disorders.
GIG provides a common platform from which effective programmes can be launched to raise awareness, inform the media and influence government, industry and the NHS
GIG focuses on issues of policy and practice keeping an active watch on developments within the UK and Europe that will influence th
effective transfer of knowledge and understanding into products and services for families that are supported by our member groups.
SUPPORTED BY
Genetic Knowledge Park
MRC
NOWGEN (The North West Genetic Knowledge Park)
SUPPORTED BY
Genetic Knowledge Park
MRC
NOWGEN (The North West Genetic Knowledge Park)
Why not try our Interactive Genetic QUIZ taken from
EDUCATION
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work - Outlook Express
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2.8.3 Family, friends and other information

Other sources of information included family and friends. Three people said that they were aware that the condition was in the family before they were diagnosed. However, the information they had received from other family members was not always accurate, one person describing it as "just word of mouth… which wasn't always correct". Other people knew that family members had experienced similar symptoms as they were experiencing but did not know further details about the particular condition.

Three people said that they had approached friends who were healthcare professionals and asked them for further information. In one case the friend approached informally was a genetics consultant who referred the family to one of his colleagues.

Seven interviewees said that they had drawn on their own prior knowledge in genetics. Two had covered genetics in their undergraduate University courses. The other five were familiar with genetic concepts from their previous professional training in physiotherapy (2 interviewees), nursing, general practice and osteopathy. Whilst none had detailed prior knowledge of the genetics of the particular condition, they felt that familiarity with genetic terms and concepts had helped them to understand the information given by healthcare professionals.

Nine people said they had used books or journals to get genetic information, usually medical textbooks or journals. For most, these were a supplementary source of information, but one interviewee said that her knowledge of the genetics of her condition, beyond the basic fact that it might affect her daughters, had come solely from looking up the condition in books.

Finally, two people said they had gained genetic information from watching television programmes about the condition. Both people said they had then personally contacted the expert featured in the programme.

CONCLUSIONS

Conclusions

The previous section outlined the key issues arising from interviews with 27 people with or at risk of genetic conditions and parents of children affected by a genetic condition. Whilst the people interviewed spoke from range of perspectives a number of common themes emerged in terms of their experiences and their preferences of receiving genetic information.

From these results we suggest areas for consideration in the future provision of genetic information by healthcare professionals and implications for the education of healthcare professional groups.

3.1 Healthcare professionals need greater awareness of genetic conditions

One clear message from this project is the need for greater awareness of genetic conditions amongst healthcare professionals. The people interviewed did not expect all healthcare professionals to know about their condition: they acknowledged that some genetic conditions are rare and that healthcare professionals cannot be expected to have detailed knowledge in all fields. However, they did feel that healthcare professionals should be more willing to consider the possibility of a genetic condition, to refer patients for investigations and to take their concerns seriously.

The interviewees also stressed that healthcare professionals should be willing to acknowledge their own lack of expertise and seek further information or advice accordingly. A healthcare professional who admitted that they did not know the answer to a question was considered far preferential to one who gave misinformation. Some people raised the possibility of "just-in-time" information for healthcare professionals immediately prior to a consultation, for example information flagged up on the computer system or accessed on the Internet.

3.2 Patients prefer a range of information providers

This project has also raised issues around the professional roles of different healthcare professionals.

The role of the GP was considered very important as the GP is often the first point of contact for patients. Whilst some participants would ideally wish to receive genetic information from their GP, most acknowledged that GPs could not be expected to know detailed information about all conditions and saw the GPs' role as to refer patients appropriately. Management of genetic conditions often involves a number of different specialties and some participants felt that a lack of communication resulted in care being fragmented, with no-one addressing the condition as a whole. GPs could play an important role in co-ordinating information from different specialties. The importance of co-ordinating patient care has been previously identified (Genetic Interest Group, 1999; Genetic Interest Group, 2000) and the findings of this study suggest that some patients would like GPs to take on this role.

Consultants and specialist registrars in different medical specialties also play an important role in the provision of genetic information. The majority of people interviewed (15) said that, with hindsight, they would have preferred to have received genetic information from the specialist they had regular contact with, most commonly the consultant neurologist, paediatrician or cardiologist. The main reason for this preference was the rapport and personal relationship they had established with this person.

The important role that a wide range of healthcare professionals undertake in directing people towards appropriate sources of information is highlighted by the range of professionals who were approached by interviewees. In some cases people raised questions about the genetic condition with a healthcare professional who was not linked to the care of that condition, such as the parent who asked their thyroid specialist about the genetic basis of their son's autism. This highlights the importance of healthcare professionals knowing how to direct people towards appropriate sources of information. The knowledge, skills and attitudes required for appropriate referral to the regional genetics service, for example, may be needed in a broad spectrum of healthcare professional groups. Another important source of information for some interviewees was making contact with an expert specialist in their particular condition. People described making such contact by researching the condition on the Internet, through Patient Support Groups or in two cases by contacting expert specialists they had seen on a television programme. More support from healthcare professionals in finding and facilitating access to such experts would be welcomed.

3.3 Recommendations for healthcare professionals providing genetic information

For those healthcare professionals who provide genetic information there were some clear recommendations from the interviewees. Genetic information should be given without bias or judgement. Healthcare professionals should be mindful and sensitive of their use of genetic terminology, in particular recognising the impact of terms such as "risk" and "mutant". They should also be aware that some genetic concepts are difficult for people to understand.

Healthcare professionals should also be aware that individuals have differing preferences with regard to the amount of genetic information they receive at different stages of diagnosis and care. Some people wanted to have detailed information at the point of diagnosis, but acknowledged that their spouses had not wanted such in-depth information at that time. The need to tailor the amount of information provided to the needs and preferences of the individual was considered very important.

The experiences of the people interviewed highlighted that genetic information was often received at a difficult and emotional time, when people were dealing with other concerns and priorities. Some described how the desire for detailed genetic information occurred later. Many acknowledged that the need for genetic information was influenced by particular stages in their life and their children's lives, such as when planning a family or when pregnant. Given the differing needs for information at different times, it is very important that people are informed where they can access reliable genetic information in the future. This might include: providing written materials that patients can re-read; inviting patients to come back if they have further questions; and informing patients and families of relevant patient support groups and reliable websites.

Healthcare professionals should be aware that genetic information can have an emotional impact and can have implications for the wider family. Painful memories, such as the death of a family member, may be reawakened. Feelings of guilt or blame may affect parents and grandparents. Information may have implications for other family members who need to be informed and this can be a difficult process. Healthcare professionals can influence how families deal with these issues if genetic information is given sensitively.

The Genetic Interest Group (2000) has published a checklist for those providing clinical genetics services at regional genetic centres in order that they can meet the needs of patients and their families. Some of the suggestions of good practice echo those identified by interviewees in this study, such as the need for co-ordination of care, consideration of the familial issues that may be generated and provision of information about relevant support groups. This study has highlighted that a wide range of healthcare professionals outside specialist genetics services are involved in the provision of genetic information. The development of a checklist aimed at such healthcare professionals may help to raise awareness both of the need to consider genetic conditions and of best practice in the provision of genetic information, so that patient expectations and needs are met in the future.

This study has identified that patients expect to receive genetic information or referral from a wide range of healthcare professionals and has highlighted ways in which this information can be provided to benefit patients. These findings will inform the work of the NHS National Genetics Education and Development Centre and the development of genetics education for healthcare professionals.

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