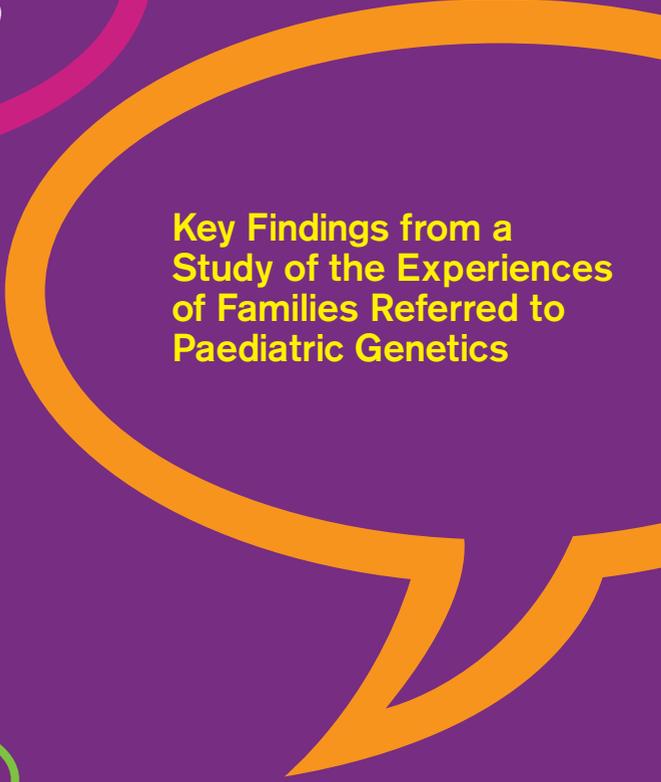


# Genetic Journeys



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Key Findings from a  
Study of the Experiences  
of Families Referred to  
Paediatric Genetics



Policy  
Ethics  
And  
Life  
Sciences



E · S · R · C  
ECONOMIC  
& SOCIAL  
RESEARCH  
COUNCIL

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*Top left to right: Dr Janice McLaughlin, Dr Emma Clavering, Professor Erica Haines and Dr Michael Wright*

# Table of Contents

1. Executive summary	4–5
2. Introduction	6–7
3. Background to the study	8
3.1. The process of genetic assessment	8
3.2. Aims of the study	10
3.3. Our approach	10
3.4. Overview of the families	11
4. Clinic experiences	12
4.1. Expectations	12–13
4.2. Consultations	14–18
4.3. Watchful waiting	18–19
4.4. The Internet	19–21
4.5. What does a diagnosis mean?	22
4.5.1. What is a finding?	22–23
4.5.2. Getting a name	23–25
5. What genetics means to people	26
5.1. Sibling fears and concerns	26–27
5.2. Questions of inheritance and responsibility	28–30
5.3. Future children/grandchildren	30–31
5.4. Genetic and developmental difference	31–32
5.5. Uncertainties	32–33
6. Concluding remarks	34–35
7. Papers and presentations	36–37
8. Resources	38

# I. Executive Summary

**The report summarises findings from a 3 year study looking at experiences of families referred to genetic services when concerns had been raised about the developmental progress of a child within the family but with no immediate or obvious cause. Previously other research has questioned whether genetics might 'medicalise' family relations by emphasising biological connections and differences over other forms of family connection and meaning. We wanted to see if such associations were made by families themselves.**

The study followed 26 families through the 'journey' they undertake when a child is referred to a genetic service. The approach taken in the study included in-depth interviews, observations of clinic consultations and informal observations in everyday life.

Genetic investigation centres around 'clues' from family history, the pregnancy and birth of the child, the child's history to date, close examination of the child and subsequent laboratory tests. This can happen over several years in a process described as 'watchful waiting'. Families in this study went to the genetic service with a number of concerns and questions. These were the search for a diagnosis; life-course potential for the child; and risk factors for possible future pregnancies (both for the parents, the child, and their wider family).

In the full report the key findings we discuss in more detail are:

- Exploring ways in which a child may have some form of genetic variation that could explain why they appear to be different to others, does not necessarily mean that families feel they are treated only as objects of medical scrutiny and curiosity.
- What geneticists and families take to be a 'finding' of significance is not always the same thing.

- Families value clear, consistent and open communication between themselves and the geneticists they come in contact with. The geneticists they remember positively are often those who respond to all their questions, maintain a relationship with them over time, acknowledge the emotional aspects of what they are experiencing, and seek to provide additional support and advice.
- Once the possibility of 'passing on' faulty genes is raised, parents can approach future reproductive decisions with fear and concern, even if they are told that their child's problems are not inherited.
- There are multiple ways in which feelings of blame and responsibility can come into play when genetics is used to explain a child's problems. People do not always believe that passing on a genetic fault should bring with it a sense of blame. Others struggle to accept that they are not to blame even though they are told a genetic fault has not been inherited. Finally, genetic explanations can be sought to challenge suspicions that parents are responsible for their children's problems because they are not raising them appropriately.
- Family members can find it difficult to make sense of the uncertainties that are often associated with genetic investigations and diagnoses.
- Families know that children develop in different ways and at different speeds. Even if a child might never be 'normal', this does not mean they are less loved and cared for, or less loving and caring.

## 2. Introduction

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**Children can be referred to genetic services for a number of reasons. In this study we describe the experiences of those families where concerns had been raised about the developmental progress of a child within the family but with no immediate or obvious cause.**

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Clinical geneticists work with other medical and laboratory staff to explore the possible explanations for the developmental pattern that is observed in a child. The underlying cause may include variation in chromosome structure involving a number of genes or changes in a single gene which can involve alteration to the genetic instructions. Some genetic differences are well known and easy to identify and much is known about what they will mean for the child and how the child can be best looked after. Other kinds of genetic variation are much harder to spot and it is less easy to predict what the future holds for the child. This report summarises findings from a project which has worked with families to explore what happens to them when a child is seen by a genetic service and the possibility of this latter form of genetic variation is investigated.

The report will begin by giving some background to the study and will then explain how the project was carried out. The rest of the report will detail some key findings, sharing what different family members, including children, said about their experiences.

We do not use the names of the families in the study and have omitted personal details to ensure anonymity of all participants.



# 3. Background to the Study

## 3.1. The process of genetic assessment

Children can now be tested for a range of single gene disorders that are well known, for example Achondroplasia and Cystic Fibrosis. Paediatric genetics also explores associations between variations within ('deletions' or 'duplications') and between ('translocations') chromosomes and problems in childhood development. Some of these associations have become established enough to be given a name (for example, Prader-Willi syndrome), others are so rare they are only referred to via the pattern of chromosomal difference found (for example, '2q37 deletions'). Many patterns of developmental variation seen in children have no currently identifiable single gene or chromosomal cause but this does not necessarily rule out identification of a diagnosis. It may be possible to do this by comparing the pattern seen in that particular child with those which have been previously recognised in other children. In at least 50% of cases it is impossible to make a specific diagnosis.

Identification of such 'genetic syndromes' in a young child occurs through a combination of techniques. A key component of this is the story of the child's development and the pattern of problems that they currently have or have had in the past. The child's family history is discussed to identify patterns of ill health which might indicate what the problem is. Family history is important because, if the problem is genetic, then it may be the result of a variation in chromosome structure or gene sequence which was passed to the child by either or both biological parents. Often, however, the family history gives no clues either because the condition has arisen as a new event in the child or because of the particular inheritance pattern in the family. In some cases it may be possible to make a specific diagnosis based on these historical descriptions alone.

This history also informs the physical examination and laboratory investigation. Examination of a child's physical features seeks to identify physical patterns that may reinforce the diagnostic thoughts identified through the history or may open new avenues for investigation. Geneticists also use many different laboratory techniques to investigate the possibility that the child may have a variation in chromosome structure or DNA sequence that may explain their presentation. This however is normally the final part of the diagnostic process that takes place only after careful assessment of the child's history and physical examination. It is not possible to 'screen' a child's DNA for all possible variations. Even if this was technically possible, interpretation of the significance of this information would rely on the other components of genetic assessment.



### 3.2. Aims of the study

Our study aimed to explore, from the perspectives of family members, what it was like to enter the world of genetics in order to see if that world could explain why a child was developing differently to other children.

When the project began we wanted to address these questions:

- How might experience of paediatric genetics affect families?
- What happens during genetic consultations?
- What do families draw upon to help them through the process?
- What are the long term consequences of living with or without a diagnosis for children and their families?

While these are the questions with which we began, they evolved through our work with the families so that our focus reflected what was of significance to them.

### 3.3. Our approach

We obtained ethical approval from the Local Research Ethics Committee (LREC) of the NHS National Research Ethics Service (NRES). Families were mainly recruited through letters of invitation sent to parent/carers via one genetic service which is located within a NHS Hospital, but undertakes clinics across a large region. We also publicised the project in newsletters in covering the same region for parents and families of disabled children. Families who were interested in participating received more detailed information about the study, and the researcher on the project

spent time talking to them before individual family members agreed to become involved.

Our approach was to follow families through the 'journey' they undertake when a child is referred to a genetic clinic. With some that meant being with them from those early stages and going along part of the journey with them, with others it meant reflecting back on previous experiences. Within each family we sought to include those people that the parents/carers defined as members of their family and who were involved in the referral process. This meant that we spoke with grandparents, aunts and uncles, close friends, siblings, and the children who had been referred themselves.

Our work with families involved carrying out a range of interviews (most of which were digitally recorded and transcribed), to capture changes in their lives. We also observed clinical consultations when geneticists first met a child and where findings from tests were discussed (observations were recorded via detailed notes which were taken during and after the consultation). To get a sense of family life, we spent time informally with families at home or on trips to, for example, the supermarket or the cinema. We also offered art and note books to the children and young people to help encourage self-expression. Material from these included stories about themselves and their families, encounters with geneticists, and photographs of themselves and with other family members.

All participants understood they could withdraw at any time, and that they did not have to talk about anything they did not want to.

### 3.4. Overview of the families

A total of 26 families participated in the study. This included:

- 44 adults (24 mothers, 14 fathers, 2 aunts, 3 grandmothers, and 1 grandfather) who undertook up to three interviews with us.
- 27 children (aged between 5 months and 18 years old) who we observed either in clinic consultations or in their everyday life.
- 9 children who agreed to be interviewed, 5 of whom had been referred to the genetics clinic, and 4 siblings. They were aged between 9 and 18 years old.

17 of the families were followed from their first referral to the paediatric genetic service, while the others had been referred to the service several years earlier and were reflecting back on their experiences.

The families live in a region of the UK that includes cities and suburbs, towns once dominated by heavy industry, and rural – and sometimes isolated – communities.

During the time of the fieldwork some family-life situations changed. For example, parents in 1 family got married, and parents in 2 families separated; 2 sets of parents had further children; 1 mother became a grandparent; and contact was lost with 2 families, due to change of address.

Most adult participants described themselves as 'working class British' though several participants said class was irrelevant to them, or of not wanting to place themselves within a specific social group. 8 participants defined themselves as 'middle class', although 5 of these talked about having middle class lifestyles, but working class values. 1 participant described themselves as 'British-Asian, class is not relevant'. In 2 families, parents disagreed about their household class identity, with one saying 'middle class', and the other 'working class'.

Study data				
Families	Interviews (adults)	Interviews (children & young people)	Observations of consultations	Informal observations
26	53	9	12	10

# 4. Clinic Experiences

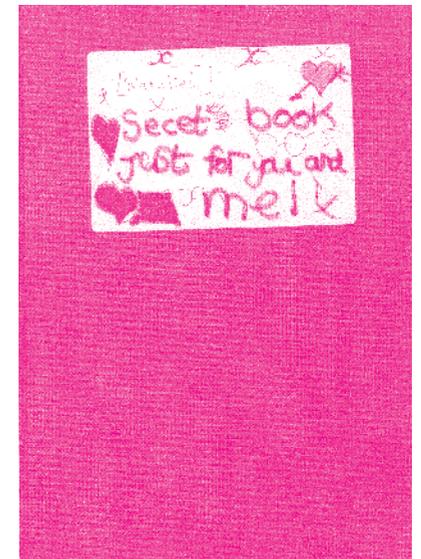
## 4.1. Expectations

For all families in the research, the genetics service was one of several services (health, education, and social) involved in their child's care. Part of their relationship with the genetic team within the clinic they attended was framed by earlier and on-going experiences with a wide range of professionals. However, genetics also offered new possibilities, and at times renewed hope that answers previously unavailable by other medical routes might now be obtained. Given the high profile media coverage that is associated with the reporting of new findings in genetic knowledge, and popular science fiction accounts of brave new worlds in which DNA can be manipulated freely, it is perhaps not surprising that parents often approached the genetic service with a high level of optimism that new and conclusive information might be found. They also did so with some trepidation because the genetics world was unknown to them.

Prior to attending a genetic clinic, each family was sent information from the service informing them about what to expect. This information was addressed to the parent/carers of the child being referred. Parents who remembered receiving this information confirmed that the language was clear, and most found it helpful to be informed about what to expect, and to be given the opportunity to prepare for the first consultation by, for example, gathering background information likely to be relevant. Attending the clinic often occurred at stressful times in family life, particularly when the child was very young, when they were dealing with a range of concerns, new information and previously unfamiliar services. In these times of increased pressure, written information such as this was not always closely read. This factor was acknowledged by geneticists and space was often given in first consultations for introducing the service, and talking through what would be covered.

Almost all the first consultations we observed began with, or quickly moved to, the geneticist asking parents what they were hoping to find out. Parents' responses tended to fit under one or more of these concerns: to be given a diagnosis; life-course potential for the child; and risk factors for possible future pregnancies (both for the parents, the child and their wider family). While it appeared that families were ready to indicate to those present in the

consultation what they wanted, they were less comfortable disclosing some of the background reasons for why this was what they wanted to achieve. In discussions with us they disclosed contexts such as long-running family issues and tensions, personal worries or battles with other services. Such contexts informed their decision to accept the referral to the genetic service, and also informed both their expectations before and response after that referral.



## 4.2. Consultations

Each consultation we observed was unique and varied due to factors such as what other medical treatments or services were involved with the child at that time, what the family's main concerns were, which types of professionals were present, and the geneticist's approach to carrying out the consultation.

Although consultations were led by a named paediatric geneticist, other professionals could be present such as junior health professionals, paediatric consultants, or other paediatric specialists (genetic counsellors were not present in the consultations we observed, but they can be). Referral letters invited parents and carers to bring their child with them to the clinic. Other family members (such as grandparents, siblings or cousins) also attended sometimes, either out of convenience or because they might be part of the assessment.

Parents often described consultations as being relatively friendly and informal, a place to explore what was most important to them:

*Mother: "Everything was answered that we wanted... That, I think, that was probably one of the most useful things because I think, if you don't answer that question, I potentially could've missed other bits of it because I was thinking, mustn't forget to ask, mustn't forget to ask, mustn't forget to ask, where as if you say that at the beginning, you get it out the way, then you can just go ahead and, and listen to everything there that's being said. I am fairly sure I would've missed what [the geneticist] said about the different chromosomes had I not known that she was gonna return to the two things that I'd asked."*

*(Family 6. First interview. Present: mother and father.)*

While being unique occasions, almost all the first consultations we observed involved four key aspects: questions around family history; the pregnancy and birth of the child; the child's history to date; and a close examination of the child. The geneticists described this process to families as 'looking for clues'. Most parents were used to telling the birth story to medical professionals. Their closeness to their child meant they had lots ready to say about their child's medical history and development. The aspects of the first consultation which tended to stand out for them were the questions around family history and the close examination of the child.

Family history was not something any of them had discussed in depth before, sometimes not even with other family members. Recounting details of relatives' medical conditions could be quite difficult emotionally too. In anticipation, some parents had researched this subject with wider family members. One mother spoke about trying to find information when there were no detailed records available. Her own mother was able to fill in some of the gaps, but it still meant they could not give the geneticist a complete picture. Parents in one family had adopted their child and had to rely on outside agencies to piece together some of the family history of the birth mother and father. Geneticists acknowledged with parents that it is never possible to fully complete a family tree with every single detail added. However, it was still difficult for some parents to be left wondering if there were potentially crucial 'clues' that were missing from the investigation. Recounting family history could also raise other issues about what family meant to participants and this is further described in section 5.2.

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**Mother: "I am fairly sure I would've missed what the geneticist said about the different chromosomes had I not known that she was gonna return to the two things that I'd asked."**

*(Family 6. First interview. Present: mother and father.)*

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The process of gathering clues means there are times in consultations, particularly the first consultation, when there is a lot of focus on the child's body – while this happens in other paediatric contexts, it stood out as important to parents. Differences in how the child looks as compared to their family members and the geneticist's perception of what would normally be expected in a child of that age, sex and ethnicity, help the geneticist narrow down what diagnosis might explain the issues for the child. The differences in the child's features may not cause any problems for them – but they help suggest possibilities to the geneticist.

GENETICS

Geneticists record any significant physical features in writing and/or by drawings in the child's medical record and often by taking one or more digital photographs. The taking of photographs was often seen by the families as one of the more unusual features of the consultation compared to other types of medical examinations they had attended. Taking each photograph entails the consultant getting very close to the child with the camera, taking photographs of fingers, feet, nails, hands, ears, eyes and other features:

*Geneticist asks the child referred: "Mind if I take a picture of you?... It's for my friends in [the service]."*

*The geneticist gets on to their knees in front of child, with the digital camera ready. I can see the geneticist, but the child has his back to me, sitting on his father's knee.*

*The father speaks to the child: "Don't pull your face." The geneticist takes one photograph of the child – a shot straight on of his face.*

*The geneticist then holds the camera out, and asks him: "Would you like to see yourself?"*

*The geneticist turns the camera round so that the child can see the picture that has been taken in the viewer: "There you go." Then, looking at the image, the geneticist turns to the mother: "They [the child's eyes] are a little wide but not anything to worry about."*

*(Family 7. Observation notes from first consultation. Present: mother, father, child referred, 2 siblings, grandmother and grandfather, and geneticist.)*

Geneticists use their skills at looking at physical features and knowledge of children with genetic syndromes in order to make sense of what they see in the child and on photographs. This can be an odd process for the children and parents. In everyday contexts photographs are taken to mark special occasions and relationships. Here however, the photographs are being used to capture what is considered to be different about a child – according to the geneticist – and to use it to see if that can help explain what may lie behind those differences. Geneticists often sought to make the child and other family members present comfortable with the process by using informal language and humour. But it is important to remember that the photographs are part of a medical process.

Families bring their own interpretation of the significance of how a child looks into their response to what the geneticist says. They can make use of their own photographs to do this. For example, at the end of a first consultation, a mother and father used a mobile phone photograph of their other child to show how – in their view – the child being seen by the service 'took after' the father, while the other child, with no known health problems, 'took after' the mother. In subsequent interviews it became clear that this view of the pattern of inheritance was an established family story, which was influencing how they thought genetics could explain the child's problems.

For the most part parents and children were comfortable with the process of examination. The ways in which geneticists spoke directly and carefully to the child often helped create a relaxed environment. For many of the children this was not their first time in a hospital consultation room and they were familiar with adults wanting to look closely at them. This familiarity meant that some children were at ease with the process, for others it meant yet another appointment, and some children expressed their dislike of having to go for the appointment even before the consultation had taken place. Occasionally parents did feel uncomfortable at certain times during the close examination:

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**Mother: "She (the child referred) was a bit of meat on the bed. The geneticist was doing their job; I don't have any resentment about that. But it just became, she became like an object. It was very, it just felt clinical and I didn't like it. The geneticist was looking at bits of her."**

(Family 16. First interview. Present: mother.)

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While this level of discomfort was unusual amongst the families we spoke to, it acts as a useful reminder of the negative responses that are possible.

After an initial consultation, some families will not need to return to the service. This might be because a diagnosis has been made and all available information has been given, or the geneticist feels that there is little indication that further genetic exploration could aid an understanding of the child's problems. The four key aspects of the first consultation (family history; the mother's pregnancy and the birth; the child's history; and close examination of the child) helped inform the next step/s. Not all of the newly referred families returned for follow up consultations during the time of the study. We were however able to observe a small number of 2nd and 3rd consultations with families.

What tended to mark these subsequent consultations out as different from the first visit was far less attention being directed to the child. There were still times when the geneticist's attention was fully on the child, for example, when further examination was undertaken, or when new questions arose that needed considering from the child's point of view. However, for the most part attention was centred on communication of any test results, questions about other family members, and discussions about whether further tests were required. This led one child to speak about feeling almost totally invisible in the follow up consultation she attended with her mother and sibling. She said she did not mind this too much, but was not very sure why she had to be there if no one asked her if she had any questions. She remembered the only time when she felt acknowledged was as they were about to leave and the geneticist stroked her head. We return to how parents and other families responded to discussions of what geneticists had or had not been able to find out in greater depth in the sections 4.5.1. and 5.5.

### 4.3. Watchful waiting

Once a geneticist gets a clear idea about what might be the particular diagnosis affecting a child's development then they may move straight on to laboratory tests to establish whether or not they are correct. However in some cases this is not possible. This can be because the child is too young for their physical features and development to give a clear 'clue' to what may be the problem, or it can be that they have a very rare, perhaps never seen before, variation in their genetic makeup which is hard to spot by looking at their features or hearing their family history. For a significant number of children there are no laboratory tests currently available to provide confirmation of a diagnosis made on the basis of history and examination. In all these situations families can be invited back for a follow up several years after the last consultation. This could be because the child's development provides new or more specific clues, or because new medical knowledge has come to light or a new test has become available. It could also be as part of a systematic check-up to see if anything had changed for the family over time. Geneticists sometimes refer to this period of time as 'watchful waiting'.

Parents in the study were often surprised by watchful waiting; they rarely seemed prepared that this might be the case, or that significant time might occur between appointments. However, the opportunity to come back to review new possibilities was important to some families.

The questions parents brought to these later consultations could be very different from when they were first seen. For example, one mother told us how she no longer sought a diagnosis, and just wanted the geneticist to keep her and her child in mind should new resources become available. For other parents, a return to the service meant re-visiting that earlier optimism. One mother spoke about the renewed sense of hope she had that the geneticist would be able to identify the reason why one of her children was developmentally delayed and another had behavioural issues.

### 4.4. The Internet

Almost all the participants interviewed, across the generations, had searched the Internet for further information either before or after consultations. This was particularly the case if a specific syndrome was mentioned, even if this was in passing. Their aim was often to find out further information, in particular what kind of future their child could expect. Internet searches for syndromes often produce photographs of children with that and other diagnoses. Given that many syndromes vary in the physical effect they have on individual children, there is a high probability that families will come across images and descriptions that suggest possibilities which are not useful guides for their particular child and situation. It can be hard for families to make sense of the different images and they can be shocked and unsettled by what they see. One mother, for example, went on the Internet when a particular syndrome was being considered:

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**Mother: “So we came away from the hospital just really. And I had, silly really, went on the Internet, started having a look, and as I say it's, it's absolutely terrifying what you find. So I was just distraught, absolutely distraught.”**

(Family 16. First interview. Present: mother)

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In consultations geneticists did spend time warning families of what they might find on the Internet and the likelihood that they could find things that were not relevant. One parent had been told not to go on the Internet before the consultation, but both her and the geneticist agreed it was near-impossible to ignore it. One approach geneticists took was to indicate particular Internet sources which they thought could be useful, supportive and relevant.

It is understandable that parents use the Internet in these and other ways. Indeed some families used it a great deal to reach out to other families with a child with a particular syndrome or to those with broader known or unknown genetic differences. In addition, images they found on websites detailing different problems associated with certain syndromes were not always distressing. They could also be reassuring. One family discussed the range of images they found, stressing the feelings of unease and then reassurance they had experienced. The first sites they found contained images of children older than their child, with a range of very marked physical differences; the site also listed learning disabilities associated with the condition. The family had been clearly troubled by how physically different the children looked. Eventually the maternal grandmother found an alternative site. On this site she found photographs of an older child, which also contained pictures of her at a similar age to her granddaughter. The grandmother quickly shared them with the mother and father:

*Father: "There is also, when you look into it, some recent case histories with other people, of adults, where this is one, there's one little girl, when they show you a picture of her when she was first born, and she is identical to [child referred]."*

*Maternal grandmother: "That's what I saw, I showed [the mother], I says, I got quite a shock when I saw her."*

*Father: "And then you see a picture of her when she's four and,"*

*Maternal grandmother: "She's beautiful isn't she?"*

*Father: "and it was completely different"*

*Mother: "Yeah she's lovely, she is a normal little girl, she's just little."*

*(Family 11. First Interview. Present: mother, father, and maternal grandmother.)*

One family were informed via telephone prior to a second consultation that the analysis of their child's blood tests appeared to show that they had a particular deletion on a numbered chromosome. Soon after the parents received this information, they were on the Internet and found images of other children who had what they believed to be the same deletion. They were immediately struck by how similar their child looked to the other children, confirming for them that their child must have that deletion.

When discussing this with the researcher before the consultation, the father said that when he looked at the images he thought he was looking at his own child. It was therefore a considerable surprise to be told in the consultation that what the geneticists had found was a specific form of deletion pattern on the numbered chromosome that they would not have seen on the Internet. When the father reflected later on this he argued that it didn't matter that his child's deletion was different to those children whose images he had originally seen:

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**Father: "We could see how all these children look the same. Just having something different about your genes brings them all together, whatever the deletion-point-this-that-or-the-other is."**

*(Family 19. Observation notes, discussion following second consultation. Present: mother, father and child referred.)*

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While the analysis of this child's blood was able to produce a level of detail about the specific pattern of deletion, this did not mean very much to the father. Instead what was meaningful to him, and also to the mother, was seeing other children whose specific pattern of chromosomal variation may be different, but who looked similar.



#### 4.5. What does a diagnosis mean?

We observed several consultations where findings from assessments and tests were fed back to families. The main issues for families being given this sort of information are highlighted below.

##### 4.5.1. What is a finding?

During the research, one issue that emerged was what 'findings' meant to the different people involved. For geneticists a finding is being able to use the different laboratory techniques available to identify an alteration in gene or chromosome structure. Parents were asked to come to the clinic to have such findings explained to them. Several families in the study, in particular those who were given a diagnosis of a very rare chromosomal condition, spoke of going into such consultations with high expectations of hearing about what this 'finding' meant for their child. They hoped it would give some indication of what the future held, and what level of developmental problems the child might experience, for example whether they would be likely to go to mainstream school, live independently, have a normal life expectancy etc. Often what they found instead was great detail about the specifics of how their child's chromosomes were different, but little about what that meant for the child:

*The geneticist returns to the hand written diagrams made during the consultation:*

*"So, if we look here [pointing to the picture on the page] you can see [numbered chromosome] is in the right place, where the red blob is, [numbered chromosome] is also in the right place, but one has ended up on chromosome [number]. So we had a close look at that. Chromosome [number] had a normal look to it, but here there is a bit of chromosome [number] on [numbered chromosome]."*

*(Family 16. Observation notes from second consultation. Present: mother, father, child referred, and geneticist.)*

The families, while interested in the detailed information about the chromosomes, were disappointed when it could not tell them what they needed to know:

*Mother: "Cos we still don't know what the future holds for [child referred]... we want to know, now that they've found this, what the effect is. And it's like, 'well we don't know, it's such a new thing.' And I think, well fair enough but it doesn't really help us, it's just a label.*

*... And then [the geneticist] explained [the chromosome test] saying, it's all about you know, 'we dye them, and we blow them up.' And we were like, oh how clever and what have you, you know, but, hm... It's great and, it's good we've got these brainy people around but you know this is about [child referred]."*

*(Family 2. Third interview. Present: mother.)*

The parents response was influenced by what made something a finding to them, which was that it was something that could be useful for looking after the child.

##### 4.5.2. Getting a name

Even though there were limitations to what a diagnosis could actually say about the future, obtaining a diagnosis was something most families were positive about. All families went to the genetic service seeking an explanation, a name they could use to tell others this was why their child was different from other children:

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**Maternal grandmother: "Before we had a name we had nothing to tell us what this syndrome was, and I think that was the worrying bit. You know, well what does it entail, what does it entail, like you say, for them and her herself?"**

*(Family 11. First interview. Present: mother, father, and maternal grandmother.)*

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Children, even if very young and with significant developmental problems, could also become invested in finding out what could explain something about who they were. One child's diagnosis had led to the diagnosis of the mother, which in turn led to a re-evaluation of all that the mother and her parents and her siblings had experienced in their lives. The child's diagnosis was welcomed by the whole family including the child:

*Researcher: "Did your mam say how you have the same [genetic syndrome] as her?"*

*Child referred: "Yes"*

*Researcher: "What do you think about that?"*

*Child referred: "Hm [long pause, looks up and smiles] Happy."*

*Researcher: "Happy, in what way?"*

*Child referred: "We're the same."*

*(Family 22. First interview. Present: child referred.)*

While some families primarily obtained a 'name', others did receive practical help and/or treatment options for the child based on their diagnosis. For example, one family were directed to alternative feeding options after the diagnosis, which reduced the level of vomiting their child was experiencing. Some families were forwarded to other specialists in rare syndromes who could give a clearer sense of what to look out for in the future and advise on practical ways of looking after the child that were making a difference. Indeed, there could be times when the geneticist seemed to be a linchpin in the wider care around a child as they were able to provide highly specialised knowledge about the child's condition and their family context. In some of the consultations we saw the geneticist take time out to ask about wider services involved, and offer both to put a family in touch with new resources and to prompt existing ones to come back on board. This active role, however, could not always be maintained indefinitely. In one family, for example, gaining a clear diagnosis and then being put in touch with other services had been a major benefit of going to see a geneticist. Their relationship with the service had lasted over 5 years, and they had seen many changes during that time. The parents talked about their concern that the geneticists' roles were changing as the service's workload demands grew. They had noticed recently that the geneticists took less and less of an active role in connecting the family to wider resources and services.

Part of the usefulness of getting a genetic diagnosis is social in character. For example, several parents used the genetic diagnosis to rebut criticism and assumptions of irresponsibility. In one family, the maternal grandmother had suggested that personal difficulties the mother had experienced during pregnancy may have contributed to the child's problems. In response the mother pointed out to her mother that the geneticists did not believe that anything had happened during the pregnancy that contributed to the outcome. In several situations we saw parents use the notion of genetic 'bad luck' and the authority of the geneticists' medical expertise to challenge other explanations that tried to place responsibility in their character or behaviour.

Parents living in contexts of economic deprivation and patterns of family living that are different from the norm of nuclear family (married couple looking after their own children) can find that people are particularly quick to judge them for being at fault. For example, women raising children on their own, if they are also reliant on welfare benefits and are young, are often accused by others of 'living off' benefits and not looking after their children appropriately. Some families in our study had experienced such negative attitudes and this influenced why they wanted genetics to help explain the problems their children had. One lone mother had to challenge various assumptions about her mothering skills and her character in order to gain acknowledgement that there was possibly something medically wrong with some of her children:

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**Mother: "I had a three year battle, going through a Learning Disabilities Team saying I was 'molly coddling' them. There was nothing wrong with them... At first they said, 'oh you just want her to be disabled.' All sorts of accusations came out. They said 'you are just making it up,' or you get people in the street saying, 'you're doing it for the DLA benefit.'"**

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*(Family 14. First interview. Present: mother.)*

Another young lone mother found herself under considerable scrutiny by medical professionals when her baby did not gain weight:

*Mother: "I kept on saying he's not growing, he's not putting on any weight. This was probably at about a year old, about eleven months, and they just kept on saying, 'Ah, just give him more calories.' They weren't doing anything to start with, and then it was, when he was like really, really, really tiny, they were like, 'We'd better have another look.' When he was about three that was. Yeah, great that!"*

*(Family 5. First interview. Present: mother.)*

Getting a named diagnosis, then, was generally seen as adding a new piece of the jigsaw, or tool in the box of care and resources around the child. However, it was not the be all and end all – the family focus always returned to questions around what this meant for the child and their future.



# 5. What Genetics Means to People

**Sibling:** “I think genetics means, things in your blood that make you you, which you inherit from your parents.”

(Family 4. First interview. Present: sibling.)

Family members spent time making sense of what genetics appeared to say (or not say) about them, the child referred and their family past, present and future. When doing so they often drew on existing understandings they had of inheritance, responsibility and blame. They also had to work through how the uncertainties embedded in genetics affected what they knew and didn't know about the future.

## 5.1. Sibling fears and concerns

Siblings told us about a range of concerns they had. Siblings, like the adults in the family, were interested in finding out why their brother or sister was different:

*Sibling:* “I'm also curious, because all my life I thought my brother had autism. But I have been told that he has a type of autism. I, I want to know what it is, and why they are different. [Pause] What is the difference?”

(Family 3. First interview. Present: sibling.)

Seeing their brother or sister undergoing genetic investigation also threw up questions for siblings' own health. We observed times in consultations when a brother or sister of the child originally referred showed concern over potentially being drawn into the search for a genetic diagnosis. For example, during one family's third consultation, while the geneticist and parents compared head sizes, body sizes and behaviours between siblings and the rest of the family, we observed a look of worry crossing the older sibling's face, eyes moving rapidly between geneticist and mother, as if watching a verbal tennis match as they spoke in turn.

Although this sibling was apparently not too keen about having attention turned towards them, there could also be times when a brother or sister initiated an interest in their own genetic makeup and whether they might share the problems their sibling faced. For example, one young person had questioned their own health for several years. They spoke to us about their worries, and how they were starting to experience problems throughout their body that were stopping them from doing the sorts of activities they had previously enjoyed:

*Sibling:* “Like it could be just in general everything why [things are] bad [for me], or it could actually be some of [child referred]'s problems that I've got.”

(Family 24. First interview. Present: sibling.)

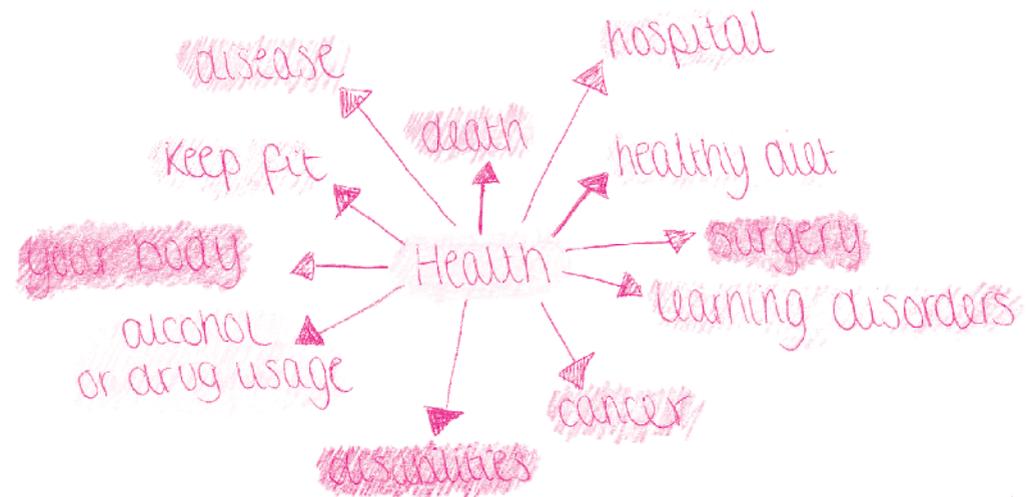
At the time we spoke to this young person, they were thinking of following up these questions with both parents and the family doctor. They were not considering going to the genetic service as, rather than obtaining confirmation of their genetic status, for the moment they were concentrating on getting support on how best to manage the problems they had.

One point raised by all the siblings was that having a brother or sister with a health condition, particularly one that required long term assessment and uncertainty increased their awareness of health issues in general.

*Sibling:* “I think about health, I think about hospital wards and stuff, and erm, to have a good health you've got to have a healthy diet, and you've got to keep fit [points to each word on the diagram they have drawn in the art book] and it's about your body as well, and also you can have surgery as well. Erm, learning disorders, like what [child referred] has, and erm, I think about cancer because it's really bad at the minute, and my grandma died of it.”

(Family 15. First interview. Present: sibling.)

Siblings, then, are very much involved in the range of questions being asked in families although, for them, the implications may seem particularly confusing at times.



## 5.2. Questions of inheritance and responsibility

Inheritance is an important component in what people think makes them a family. We can see the importance people give to inheritance in the way people often comment on which child looks like which parent, or other family members, such as grandparents, aunts or other siblings. As we have highlighted, geneticists draw from family history in considering what may be affecting a child. This can often bring into the open existing views about patterns of inheritance within the family. For one maternal grandmother this meant that the source of her granddaughter's problems had to lie with the father's side of the family:

*Maternal grandmother: "The only thing I can see is that [child referred] looks like her dad. She's got my daughter's small features, but other than that I don't see anything. Now in my other granddaughter, certain mannerisms, she's me mother, believe it or not, [laughs] I think oh, she's me mother, born long after me mam died... But I can't see anything at all in my granddaughter with learning problems, apart from that she looks like her dad and she's got, but she's got her mam's tiny little features, that's all I can say about her... 'cos she is him, which takes us back to, well it must be his side."*

*(Family 17. First interview.  
Present: maternal grandmother.)*

Geneticists are understandably focused on the biological family connections that can help to explain a child's issues. However, this means they spend less time acknowledging or considering other biological or social connections, which are important to people's sense of who they are within a family.

*Father: "They did like a little family tree of like how many brothers and sisters [the mother] had got, and [the maternal grandmother] had got, and ages and all that... I said to you [the mother] I was getting annoyed, 'cos they only did you two [mother and maternal grandmother], they didn't actually ask any questions about my family and my brother's and sister's heights and that. But you said it is just because it was your genes..."*

*Mother: "Because it comes from me, so they wanted to know my family history and things like that."*

*Researcher [to father]: "When you say annoyed, why did you feel annoyed, if you don't mind me asking?"*

*Father: "Well they was doing their family tree, I wasn't even on it."*

*(Family 11. First interview. Present: mother, father, and maternal grandmother.)*

The drawing of the tree without the father's inclusion implied that his biological relationship to his child was of less significance than that of the mother and her side of the family. He was aware of the medical basis for this emphasis, but that was not necessarily enough to reassure him.

Ties can develop because of shared characteristics between family members which are not based in genetics. For example in one family both a grandfather and grandson had hearing impairments. The geneticists looked to see if there might be a genetic explanation for why they both had hearing loss; the conclusion was that there were different reasons behind it, but to the grandfather and grandson this did not matter, they were in a 'club' together anyway because they shared the experience of hearing loss.

14 families in the research had received a diagnosis for their child before the end of the study. Laboratory diagnosis for the child may be followed up with questions exploring the history of family members, partially to establish whether there is insight to be gained from the genetic testing of one or both parents. In 6 families in the study blood samples were taken from parent(s). Results tended to be given either on the telephone, or in a letter, along with an offer to discuss in person through a future consultation or further telephone call. The significant majority (11) of the families given a diagnosis for the child were told this was likely to be the result of a new change in the chromosome or gene structure (de novo) that was not inherited. On receipt of this information, parents reacted in very different ways. Some were happy to be absolved of any blame. For others it produced anxiety and a need for an explanation for why such a new change would have occurred. One mother spoke in her second interview about her concerns that she was still somehow to blame, even after being told by the geneticist that there was nothing she had done wrong during her pregnancy.

While families often experienced relief when told that their child's problems were not inherited, this did not mean that all families where inheritance links were made felt guilt and responsibility. People were able to distinguish between what may have been in their genes and who they were and what they should feel responsible for. It was not something they could control; instead it was, as discussed above in section 4.5.2., simply bad luck.

Families often took great comfort from the ways in which geneticists explained genetic inheritance to them as something which was indeed not their fault. It was a great help to get this reassurance, particularly in contexts where a parent or parents were being blamed by others for the problems their child faced:

*Mother: "There was always something wrong with [child referred], other people were looking at me, I felt they were judging me thinking I'd done something wrong to him. And when I looked up on the Internet to find out information about the syndrome, it said in black and white, mam's pass, mothers pass it to sons, and fathers pass to daughters. That just indicated to me that it was my fault again! But then going to the Genetics and actually having Doctor [geneticist] turn around and say, 'No, it's not your fault, it's the genes.' And telling me that the genes are the main thing that make a person and illnesses can take years to come, but they can also skip two, three, four generations, and just hit on the wrong person at the wrong time. And that made me feel a lot better, having that, a doctor actually turn round and say that it was not your fault. So that took a lot of pressure off me."*

*(Family 21. First interview. Present: mother and father.)*

For the three families involved in the project who were told that the child's condition had been inherited from one of the parents, questions were then raised about possible testing of aunts, uncles, cousins, grandparents, and so on. This could be a difficult issue to broach with wider family members. In their third consultation one mother explained to the geneticist that she had little contact with her relatives, and it would be near-impossible to arrange the testing of them all. After the consultation she told us she did not think this effort would be worth it as, like all the families in this study, the main concern always returned to questions around the child's future and she did not think anything new could be added by looking back.

### 5.3. Future children/grandchildren

As well as help to explain why their child appeared to be developing differently and what the child's future may hold, parents also went to the genetic clinic to help make decisions about whether to try for another child. For those families who were told that the problem had not been inherited they were often reassured that the chances of something similar happening again were low. However, for other families, being told that they did not pass on 'faulty genes' to their child did not mean that they approached the possibility of future pregnancies without fear and hesitation. Once a doubt had been raised about what they could pass on to their child, it was hard for that doubt to go away.

While a new genetic variation means that at least some parents can go ahead with future pregnancies without specific worries about genetic inheritance this will not be the case for the child with that variation. Parents often reflected on the issues their child would potentially face as an adult thinking about having their own children:

*Mother: "But as a woman, to think that, well at some point I'm going to have to tell [child referred] that it's not going to be straightforward for her, and when she wants to have children she's going to have to be involved with the genetics [service]. And it may be that she can't have children. If she can have children she might not be able to have perfect children, for want of a better word. For a mother to have to tell that to her daughter, it's horrible, really horrible."*

*(Family 16. First interview. Present: mother.)*

One young person who had been told they had an inheritable condition several years earlier was very concerned about whether they would be able to have children of their own. According to their reading of current medical science, there was a high possibility of passing on the genetic fault, and for this young person this was something to avoid if at all possible. Their hopes lay in what might come from future genetic discoveries and technological innovation such as gene therapies. They had raised these concerns with a geneticist, and had been told that these possibilities were probably a long way off – not quite the answer they wanted.

Questions around who can have children, and what the risks might be are inevitably affected by raised awareness of potential chromosomal differences. These questions arise with or without establishing that a condition has been, or can be passed on. Geneticists recognised these issues in consultations and further correspondence. However, these issues can cut very deeply, as assurances are often balanced with an acknowledgement of lack of certainty, and the limitations of current scientific knowledge.

### 5.4. Genetic and developmental difference

Geneticists spent a great deal of time in consultations characterising the observed differences in a child's development as being within the boundaries of what could be thought of as normal. This helped reassure families that even if a child might look different from them or others, or might not develop in the same way as others, this did not mean that the child should be thought of differently to other children inside or outside the family. This mainly occurred in two ways.

First, geneticists at times questioned whether a child really was that different in appearance or development, often by focusing on how similar they looked to their other family members:

*About half way through the consultation the geneticist examines the child by looking at him as he stands in front of his mother just in his underpants.*

*Geneticist: "Actually there's nothing about him to make me suspect an underlying genetic cause [to his height]. He looks very like you!"*

*Mother laughs.*

*(Family 5. Observation notes from first consultation. Present: mother, child referred, maternal grandfather, and geneticist.)*

In this consultation (and others) genetic difference was rejected by interpreting the child as displaying characteristics associated with his family. Both geneticists and family members talked of familial similarity in order to reject that there was anything significantly different about how a child looked, and therefore their genetic makeup.



The second route through which geneticists refuted difference was to suggest that a child's physical characteristics were within the realm of normal developmental variation:

*Geneticist [wheeling chair up closer to the child]: "It's a chart that plots two lines – this one [points to one running along the top] shows the average height measurement for your age, and this line, with all these dots [hand-written, underneath the first line] is where you were at. So there are the two lines, and we can follow where the dots go along compared to the line of averages. Your line is not too far away from the average, but we can see times when your height slows down for a while. We can also look at your bone age, which is a bit more difficult to explain. This is when we do an x-ray to see if we can predict when you might stop growing. Some children stop growing at twelve or thirteen, others keep on growing until they are sixteen. If that's the case for you, then you may catch up." All the time the geneticist talks directly to the child.*

*(Family 12. Observation notes from first consultation. Present: father, child referred, geneticist, endocrinologist, and nurse.)*

In interactions such as these geneticists reminded family members that their child was a child, whatever genetic variation she or he may or may not have:

*Maternal grandmother: "One thing I will say, just the, the last parting shot the geneticist who we saw, I thought it was a really nice thing to say, and very true. They said, 'just take her home and enjoy her. She's a little girl.' Didn't they?"*

*Mother: "Yeah."*

*Maternal grandmother: "'Like any other mum... just forget about the syndrome, take her home and enjoy her for what she is.'"*

*(Family 11. First interview. Present: mother, father, and maternal grandmother.)*

Geneticists walked a very fine line between highlighting a child's distinctiveness, while bringing them back into a picture of normal family experience.

### 5.5. Uncertainties

There are many uncertainties in genetics. We will highlight two here. The first aspect of uncertainty is that a specific syndrome or genetic variation will not be identified in every child or family. That a clear explanation remained unavailable, even after at times lengthy investigations, could lead to high anxiety and stress for families.

Being confronted with a lack of conclusive evidence, and with it a clear set of options to inform and support future decision-making around the child, their siblings, and future reproductive choices, was very difficult for parents and wider family members. By the time of her third interview one mother, for example, was left highly frustrated and disillusioned with

both the service, and scientific knowledge. She had gone to the service expecting a diagnosis that could help inform health care treatments. Frustration was made worse if parents felt they had been shut out of any future return to the service should new information come to light. This had happened to this mother, and was also the case for another mother who spoke about how angrily she had reacted when the geneticist sent her a letter following a blood test taken from her child informing her that the results were inconclusive, but that there were no further avenues left for the service to explore. This mother was left with more questions than answers about why her child had a number of health problems, and why all her other children also had health problems, but the referral to the genetics service had apparently led to a dead end.

In other families where no diagnosis was forthcoming, they remained open to the possibility that at some point more may be known. However, what often changed was the family's focus in obtaining more knowledge. One mother, after years of doing test after test with no new answers, spoke of how it was no longer one of her priorities as a parent. She remained open to the geneticist's pledge to keep on searching, and to look out for medical breakthroughs, but she felt very strongly that her focus was now just on how her child was doing day-to-day. In her eyes, her child was healthy, so she was concentrating on the here-and-now, rather than continuing to spend a great deal of time and resources on the search for a genetic explanation.

A second aspect of uncertainty was that associated with a diagnosis. Because the kinds of syndromes and variations being sought are complex and sometimes newly found, there is a great deal of uncertainty about what they are able to say about a child's future development:

*Mother: "Genetics have told me as much as they know. So it's a case of like it or lump it, and I just put it at the back of my mind and don't think about it. I have accepted it. And, at the end of the day, [child referred] will still have a good life – it will just be different to the one we had expected."*

*(Family 16. Second interview. Present: mother and aunt.)*

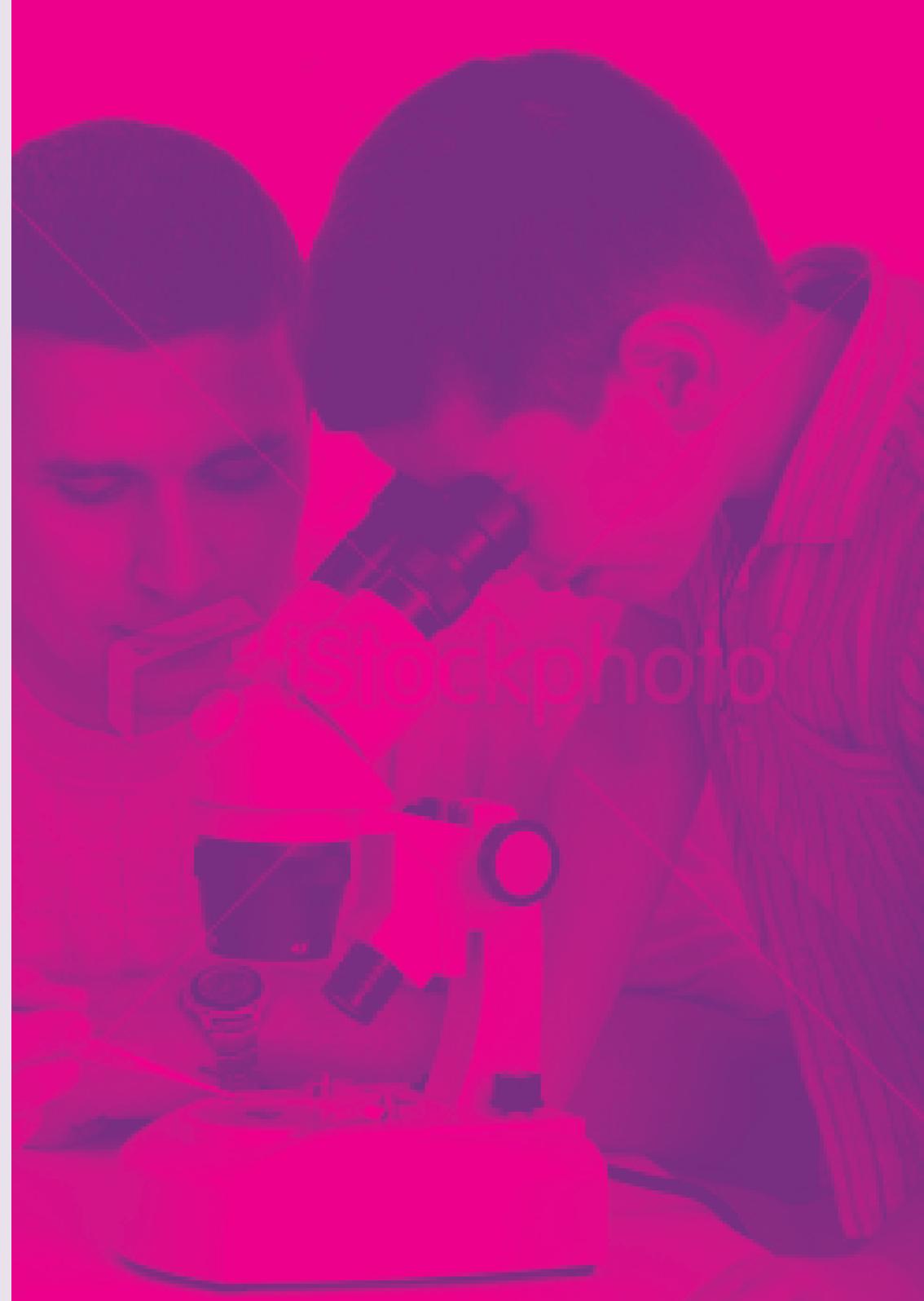
In such contexts families take up their own form of 'watchful waiting', which often ends up being open to the unpredictability of any child's future. Watchful waiting, then, becomes 'let's wait and see':

*Mother: "It has, has actually been kind of one of those big eye-opening things where the realisation that I had him in that picket fenced house and that I was looking to the last page, I'm gonna mix several metaphors here but, had him in the picket fence house and now open to the last page of the book, and missing the rest of the journey. And what is the point in buying a novel if you just only read the last page... I'm missing the journey, I'm missing the book, I'm not reading the book, I've decided how it's ending and that's it... you can just enjoy him so much better... he is different. But then who's not? Who's the same?... that's brilliant you know. And I strongly suspect we're going to actually learn more from him than he ever learns from us."*

*(Family 6. First Interview. Present: mother and father.)*

## 6. Concluding Remarks

**This report has not covered every issue and perspective that different family members have shared with us. Nor does the end of our project, and its summary here, mark the end of the families' 'journey' within genetics. There is more we could learn by continuing to follow the children and those in the family and in the world around them who care and raise them. Future advances in genetic technologies will lead to new opportunities for genetics to help explain why we all develop in different ways. As these advances grow and the scope of what genetics seeks to explain widens, it seems sensible to continue to ask what this means for those affected and how family relations in particular can be altered by such approaches to define who we are.**



# 7. Papers and Presentations

## Publications

McLaughlin, J. (forthcoming)  
*'Understanding disabled families: replacing tales of burden with ties of interdependency.'*  
In: Nick Watson, Carol Thomas and Alan Roulstone (eds.) Routledge Companion to Disability Studies, London: Routledge.

McLaughlin, J. and Clavering, E.K. (forthcoming)  
*'Questions of inheritance in pediatric genetics: substance and responsibility.'*  
New Genetics & Society.

McLaughlin, J. and Clavering, E.K. (2012)  
*'Visualising difference, similarity and belonging in paediatric genetics.'*  
Sociology of Health and Illness, 34(4).

Clavering, E.K. and McLaughlin, J. (2010)  
*'Children's participation in health research: from objects to agents?'*  
Child: Care, Health and Development, 36(5): 603-611.

## Presentations

McLaughlin, J. (2011)  
*'Potential familial and patient responses to gene therapy: lessons from current genetics provision in healthcare.'*  
TREAT-NMD 2011 International Conference, November, Geneva, Switzerland.

McLaughlin, J. and Clavering, E.K. (2011)  
*'Visualising genetics: modes of diagnosis and interpretation in the troubling body within paediatric genetics.'*  
European Sociological Association Conference, September, Geneva, Switzerland.

McLaughlin, J. (2011)  
*'Families experiences of paediatric genetics: Findings from an ethnographic study.'*  
British Society for Human Genetics Conference, September, Warwick University, UK.

McLaughlin, J. (2011)  
*'Modes of care and mothering: how does citizenship and care intersect in the lives of mothers of disabled children?'*  
Does Every Child Matter, post-Blair? The Interconnections of Disabled Childhoods Conference, April, Manchester Metropolitan University, Manchester, UK.

McLaughlin, J. and Clavering, E.K. (2010)  
*'Narratives of othering and recognition in paediatric genetics.'*  
Narratives of Health and Illness Conference, September, Uppsala University, Sweden.

McLaughlin, J. and Clavering, E.K. (2010)  
*'Visualising genetics: modes of diagnosis and interpretation in the troubling body within paediatric genetics.'*  
EASST, September, Trento, Italy.

Clavering, E.K. and McLaughlin, J. (2010)  
*'A crisis in the genes? Children and young people's responses to processes of medical labelling through paediatric genetics.'*  
European Association of Social Anthropology (EASA): Crisis and Imagination Conference, August, Maynooth University, Ireland.

Clavering, E.K. and McLaughlin, J. (2010)  
*'Engagement spaces: ethics as process.'*  
PEALS 2010 Symposium, Good Practice in Public Engagement: Expertise by Experience, May, Newcastle, UK.

McLaughlin, J. and Clavering, E.K. (2010)  
*'Othering and recognition in paediatric genetics.'*  
Child, Family and Disability Conference, April, Manchester Metropolitan University, UK.

McLaughlin, J. and Clavering, E.K. (2009)  
*'Ethnographies of genetic and kinship imaginaries.'*  
Medical Anthropology at the Intersections Celebrating 50 Years of Interdisciplinarity, Society of Medical Anthropology, September, Yale University, Connecticut, USA.

Clavering, E.K. and McLaughlin, J. (2009)  
*'Ethnographies of genetic and kinship imaginaries: kinned bodies.'*  
Vital Politics III, September, LSE, London.

McLaughlin, J. (2009)  
*'Modes of care and mothering: how does citizenship and care intersect in the lives of mothers of disabled children?'*  
ESRC Seminar Series 'Parenting Cultures', January, University of Kent, Canterbury, UK.

# 8. Resources

Further information can be found below:

**Study website:**

[www.ncl.ac.uk/peals/research/project/2745](http://www.ncl.ac.uk/peals/research/project/2745)

**Policy, Ethics and Life Sciences (PEALS) Research Centre, Newcastle University**

Website: [www.ncl.ac.uk/peals](http://www.ncl.ac.uk/peals)

**Contact a Family (CAF) For families with disabled children**

Website: [www.cafamily.org.uk](http://www.cafamily.org.uk)

National Freephone Helpline: **0808 808 3555 (UK only)**

*Open 9.30 am – 5pm Monday to Friday*

Email: [helpline@cafamily.org.uk](mailto:helpline@cafamily.org.uk)

General email: [info@cafamily.org.uk](mailto:info@cafamily.org.uk)

**Genetic Alliance UK/Syndromes without a Name (SWAN)**

Email: [SWAN@geneticalliance.org.uk](mailto:SWAN@geneticalliance.org.uk)

Website: [www.geneticalliance.org.uk](http://www.geneticalliance.org.uk)

Twitter: [@SWAN\\_UK](https://twitter.com/SWAN_UK)

Facebook: **SWAN UK (Syndromes Without A Name)**

YouTube: [www.youtube.com/user/SWANchildrenUK](http://www.youtube.com/user/SWANchildrenUK)

**Unique – Rare Chromosome Disorder Support Group**

Website: [www.rarechromo.org](http://www.rarechromo.org)

A large print version of this report is available from the project website:  
[www.ncl.ac.uk/peals/research/project/2745](http://www.ncl.ac.uk/peals/research/project/2745)

