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HAL Authorization

**Parents' and children's communication about genetic risk:
qualitative study learning from families' experiences.**

Running title: Genetic risk communication within families

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Parents' and children's communication about genetic risk: qualitative study learning from families' experiences.

ABSTRACT

Little is known about how parents explain to their children about their risk of inheriting a gene that may cause disease, in the child or the child's future progeny. This study explored how genetic risk information is shared between family members and the factors affecting it, to ascertain the implications for children, young people and their parents to inform future service development and provision. A volunteer sample of parents, children (8-11years) and young people (12+ years) in families affected by or at risk from one of six inherited genetic conditions were interviewed. The semi-structured interviews explored family members' roles, language used and the self-reported psychological outcomes in discussion of genetic risk information. The findings were analysed using grounded theory.

Thirty-three families participated, which included 79 individuals. Parents often found discussing genetic risk information very difficult and emotionally painful. Discussions were not usually planned and often a major event prompted parents to finally explain genetic risks to their children, whereas children usually preferred to learn about the genetic condition gradually throughout childhood. Parents identified a number of challenges they faced related to talking to children, and many thought health professionals should provide more advice to assist them in providing developmentally appropriate information. We therefore conclude greater emphasis is required in supporting parents and children in discussing genetic risk information throughout their child's development. Open communication about genetic risks throughout childhood appeared to help children and parents cope better and come to terms with the implications of the genetic condition.

Parents' and children's communication about genetic risk: qualitative study learning from families' experiences.

Running title: Genetic risk communication within families

INTRODUCTION

In families affected by genetic conditions, parents not only have the challenge of explaining the illness, its treatment and outcomes but they also have the concomitant stressor of discussing the inherited nature of the condition with their children. Many parents struggle to know when, how and what to tell their children about the genetic risks because they fear causing distress and so feel a need to protect their children from this information⁴⁻⁸. Many parents also describe a large burden of guilt, which can contribute to their reticence in discussing genetic conditions with their family⁷. Previous studies however, have shown that withholding genetic risk information from children until they are adult can affect their coping, self esteem^{9,10} reproductive decision-making^{7,10} and family cohesion^{11,12}.

Studies examining families' communication of genetic risk information between children and their parents, which involve all members' perspectives including affected and unaffected children, have to-date been limited. With regard to children and genetic risk, the focus of debate has centred on genetic testing and the best time to carry it out rather than how to help the families cope and come to terms with the condition and its risk. Many health professionals report being asked for advice on talking to children (personal communications). However little is known about children's understanding and experiences of learning about inherited genetic risk within their families. Studies to date are small¹³, limited to a single condition and have rarely included all family members; affected and

unaffected individuals and those at risk of carrying the affected gene. Differences related to patterns of inheritance, age of onset, morbidity and mortality, where parents have to discuss the genetic condition with their children at contrasting stages of their development have not been examined.

To address these gaps, this study explored communication about genetic risk and the factors that affect it with all family members. Six different genetic conditions were chosen which encompassed different morbidity and mortality patterns in relation to the more common, mendelian inheritance patterns. The aim was to draw on a breadth of families' experiences to ascertain what information children require at different stages of their childhood, how to provide it and their likely reactions to genetic risk information according to morbidity and mortality outcomes of the genetic condition.

METHOD

Theoretical Framework

Family communication about genetic risk was investigated qualitatively from the perspective of three inter-related models. These focused on the family members' roles and influential factors (family system theory¹⁴), what was communicated and the language used (drawing on symbolic interaction theory¹⁵) and the behavioural outcomes¹⁶ of children and parents. Each model informed the design of the interview schedule and was used as part of the grounded theory development for the data analysis. Prior to commencing the study a psycho-social definition of family¹³ was established.

An advisory group of six parents and five young people from diverse cultural and social backgrounds were consulted about the conduct of the study and later, the credibility of the

findings. The group discussed the findings in context and contributed to the development of subsequent recommendations. The study was approved by the Liverpool Children's Research Ethics Committee (REC No:07/Q1502/16).

Participant families

Potential participant families were volunteers identified via charity and NHS support groups in England and Wales. Parents and their children (irrespective of their risk status; affected, at risk or unaffected) from families affected by one of six genetic conditions were invited to participate according to the inclusion criteria. It was anticipated that 6-7 families would be recruited for each condition (table 1). The genetic conditions were chosen because not only do they have a distinct pattern of inheritance and vary in the age of onset but also because genetic testing may take place at different ages, some diseases caused are life-limiting and there are different impacts on the individual in terms of illness duration.

Data collection

Semi-structured interviews were conducted with parents, children (8-12years), young people (13-17years) and adult children (18+years). Children were interviewed independently of their parents or siblings. Consent was obtained to audio-record interviews in all cases except one.

Interview schedules were developed from the literature review and discussion with the advisory and steering groups. The interviews used neutral, open ended questions and interviewers were responsive in adapting their questions, language style and question order according to the responses received. The questions focused on family members' roles in discussing the genetic condition and its risk, the language used, and the behavioural

outcomes in terms of emotional response, coping and adaptability. For children, art based portfolios were developed for use prior to interviews¹⁷ to provide a sense of control in the activity and assist with establishing a rapport and an environment conducive to the discussion of personal and sensitive information^{18,19}.

Data Analysis

Interviews were transcribed verbatim and analysed using social constructivist grounded theory²⁰. Due to the large quantity of data collected two analysis frameworks, one for children and the other for adults were developed using interviews from five families affected by different genetic conditions. The two frameworks kept the children's perspective clearly separate from the adults at this stage.

Transcripts and field notes were read for each interview. Two researchers developed each framework, initially working independently to identify phenomena within the interview and develop concepts and a list of categories before agreeing an overall list for the framework with the second researcher. As part of developing the framework the concepts were also considered in terms of family communication models; for example family members' roles in communication, and children and adults' self reporting of behavioural outcomes. Each category had clearly defined properties and dimensions for the concepts that informed its development. Once developed, each framework was rechecked for reliability and applicability against the first five interviews plus an additional two families' interviews chosen randomly.

The framework underwent modification to incorporate new phenomena and concepts as they emerged and interviews previously analysed were revisited to ascertain whether

similar phenomena had been overlooked. Data was coded into the framework using the software package NVIVO-7. Each family's transcripts and field notes were grouped together to explore the level of agreement between parent(s) and children including; how children learned about the genetic condition and genetic risk, how much they knew and understood, the level of openness and the shared language used to describe the genetic condition, genetic risk and associated morbidity and mortality.

Throughout all stages of the analysis from fieldwork to coding, observations were used to develop a series of grounded theory questions (related to the research questions and to inductive ideas) that were used to examine the data. Each researcher examined data specific to two genetic conditions on the basis of grounded theory questions, and noted any other additional observations including any negative cases. A second researcher randomly reviewed the analyses for consistency and negative cases. All three researchers discussed any disputes until consensus was reached. Finally, the findings from the family groups for each of the six conditions were compared for consensus and difference using a constant comparative analysis²⁰ to identify the overall outcomes. The findings are reported in overarching thematic headings, which reflect the grounded theory questions that tested the data²⁰.

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FINDINGS

A total of 33 families were interviewed. The demographic profile of participants and their families is provided (table 2). Families were recruited from across England and Wales but specific regional and other information that may make families identifiable to each other or to health professionals has been omitted to protect confidentiality.

Across all of the genetic conditions, many similarities were observed and exceptions between conditions are indicated in the findings by denoting which conditions the comment is referring to.

When the transcripts of all members of a family were reviewed together there was usually a strong level of consistency between reports about what and when children and young people had been told about a genetic condition and its risks. There was often a shared narrative about the condition and its effects on the family, and parents' understanding and views were often reflected in the explanations and descriptions given by their children. In families where children had better understanding and more insight, parents had usually discussed the condition throughout childhood, often as part of the family story.

Parents, children and most young people thought parents should be the main people to provide genetic risk information because they understood their children best. Young people however thought health professionals should provide guidance and advice to parents. Over half of the young people also wanted information directly from health professionals, preferably ones who understood the contemporary issues faced at school, college and the transitions they faced at this developmental stage.

Roles in communication

Mothers were usually the key person in the family who shared genetic risk information. Some fathers took on this role as key informant or shared it with the mother, but children often described fathers as too angry or too upset to talk. Whilst some parents took shared decisions about talking to the children about the genetic condition, there were also equal

numbers who did not discuss it with each other, or if they did, they disagreed on the best course of action.

“...absolutely not. My dad went “La la la under the sea.” Anything to do with HD that was it, switched off...he couldn’t cope with it I don’t think...”

Young person 15-17 years at risk from HD

Siblings rarely discussed with each other, the genetic condition or its risk. Most communication took place between an individual child and their parent(s), whom they trusted above any other information source until they reached 15-17 years. Parents often recalled talking to their older children for the first time but many overlooked talking to their younger, unaffected children. Parents assumed that because the younger children witnessed the effects of the condition in the family everyday, they automatically knew what was happening. Children did not talk to parents’ new partners about the condition but saw this person as source of support for their parent (Figure 1).

“well he (younger sibling) doesn’t hasn’t said that he knows ...we haven’t spoken to him as such, have we?...he’s just aware”

Parent - Older child affected by DMD

Making decisions about the sharing of genetic risk information

Across families, parents differed in deciding when and how they talked to their children about a genetic condition. Decisions were not usually related to their child’s cognitive development but were more dependent on the genetic condition’s morbidity and whether it was life limiting. Only a small number of parents expressed a view that their child(ren) had a moral and ethical right to know about a genetic condition affecting their family.

Delaying or choosing the right time to talk to children about a genetic condition was often very distressing and parents worried they were making the right decision. Most delayed discussions for as long as possible until a particular event either internal or external to the

family initiated the need to discuss the condition. In contrast, most children and young people said they had been aware at some level of the condition being in their family from a young age (usually by around 8 years) and wanted to be able to discuss it and understand what was happening in their family from then.

“I might be a bit angry if they didn’t tell me because I would just want to know, so I don’t think there is anything to hide really when you have got a disease, you have got a disease really, so but no I don’t think there should be any secrets” Young person 15 – 17 years CF - affected

A number of factors often compromised parents’ capability of discussing genetic risk. These were related to one or more of the following, depending on the genetic condition and stage of disease progression; shock at diagnosis, which prevented parental discussion; guilt and fears pre and post genetic testing; increased emotional and physical care giving; and experiences of grief and bereavement. A second article, currently in preparation, will expand on these factors in detail.

How and what children are told

All children whether affected, at risk or unaffected showed a high level of insight into how they learned. Children said they wanted small amounts of information at a time and to be able to discuss what was happening in their families with their parents. Children and young people reflecting retrospectively on their experience of learning about a genetic condition and its risks said that they came to learn and understand about the condition and its implications quite slowly. It took time for them to become aware of the risks either to themselves or to other family members and most children and young people said they did not spend time repeatedly thinking about it once they had initially learnt about the risks.

Information given in the gradual way children preferred, increased their understanding over time, and gave them insight without the shock of learning about the condition and its implications all at once. Parents and young people said that receiving information as younger children was less shocking, and because they learnt gradually as they grew up, it made new information and its implications easier to cope with. This lack of shock for younger children probably occurred because they had less insight and fewer experiences to draw on to recognise the fuller implications. Unlike the young people who pieced information together very quickly to ask very challenging questions when they were not always prepared for the answers.

It was important to go at the child's pace, as information that was too graphic or went beyond what the child asked about, could stop them wanting to discuss the condition further. However, this was not achieved simply by waiting for children to ask questions, which parents often did. The lack of acknowledgement of the genetic condition by parents inhibited children aged 8-11years onwards from asking questions about it because they thought it would upset their parents. Parents therefore, had to make it clear to their child that they were happy for them to ask questions. They also reported needing to check what their child knew already and correct misperceptions that frequently occurred, especially from overheard conversations.

“She (Mum) talked to me about why I should tell people if they ask me, erm sometimes I realise that it helps, erm but if she didn't ask me if anything's wrong then I wouldn't have, I wouldn't have mentioned it to her” Sibling (8-11y) of a child with NF.

Children and young people thought less formal discussions were best, and they liked to discuss the genetic condition in situations where they were also doing other things

alongside their parent(s) such as preparing a meal, gardening or riding in the car. In some families there was a ‘story’ about the condition and parents told their children how they had learnt about the condition and had learned to cope with its effects. Parents thought this taught children about coping with the condition, offered opportunities to talk and allowed different family members insight into each others’ experiences and feelings. For some it was also an opportunity for children to look to parents or older siblings as role models.

“...me and my mum would erm, in the mornings me and my mum we’re sort of people who get up first and so we would sort of just sit down by the sofa, like one of us would be on the computer and just sort of talking and, you know, it (HD) would come into the conversation” Young person 15-17 years at risk from HD

Many parents said they were completely open to discussion with their children but further discussion revealed that this often meant answering questions as they arose. Many also talked at a different time in the interview about protecting children from certain aspects of information about the condition or risk until they needed to know. Most children and young people recognised these limitations and when talking about the level of their parents’ openness qualified this with adjectives such as ‘quite’ or ‘fairly’.

The affected child was usually given more information than siblings (CF, FAP, HbO, NF). However, if the condition was life-limiting and the child likely to die during their adolescence, affected children were given limited information but their older siblings were told more about the outcomes, which they were expected to keep secret (DMD). Overall parents’ and children’s discussions of genetic risk intertwined with the communication about the management of the genetic condition. This may explain why many children and young people often struggled to explain what they knew and understood about the genetic

risks separately from knowledge and discussion about the genetic condition and its day-to-day management.

Children's understanding of genetic risk and inheritance patterns

Based on self-reports of children and observations made by parents, patterns emerged in children and young people's understanding and reactions during different stages of their childhood. This included those affected, those at risk and unaffected children (figure 2). However, some differences in knowledge were identified across conditions and between inheritance patterns.

The dominant pattern of inheritance was easier for children to understand than the recessive or X-linked patterns but it was also accompanied by most misconceptions. Children often thought that 50 % risk meant that one of two children would be affected. Even adult children who knew this was not the case said it was difficult not to think in this way. Both parents and young people looked for phenotypic traits of a genetic condition in family members and sometimes thought that children who were most similar to an affected parent in personality or looks were more likely to inherit the condition.

In families with dominant conditions (HD, FAP, NF) children from the age of 10 – 11 years upwards understood that the condition was 'passed down' through their families with some using the word 'inherited'. Most knew it had something to do with genes and sometimes talked about chromosomes and DNA. Where the condition did not manifest until adulthood or adolescence (HD and FAP) it could be more difficult for children to conceptualise risk in terms of how the condition was going to affect their future lives.

“ born with it, something to do with my DNA when I was being made” Child 8–11y HbO

Young people usually understood the risks for themselves during teenage years (around 12-15 years), but were usually 15-17 years old before they realised the reproductive implications. At this age some were also vaguely aware of possible interventions, such as *in vitro* fertilisation that would be available to them to prevent their own future children being affected. Across the age range children and young people talked about '50:50' chance of inheriting the affected gene but only a few young people aged 12-15 years and those aged 16 or older used the word 'dominant' to describe the genetic risk appropriately in the relevant conditions. With FAP, affected young people had a more detailed knowledge of dominant inheritance than unaffected siblings did, usually because parents chose not to discuss risk information until after children had received a positive gene test.

"Erm I think the FAP comes from like other people's DNA...and it goes, and goes through the family sometimes." Young person 12 – 14 years FAP - affected

Very few young people could explain recessive inheritance patterns accurately. Children and young people affected by recessive conditions (CF or HbO) usually knew there was a risk to their potential children, although they did not understand the details of this. Some affected (CF or HbO) girls knew that pregnancy would present some risks for their own health, and this was more meaningful to them than concerns about the genetic risk for their future children. Siblings at risk of carrier status who were younger than the affected child had been tested at birth but did not necessarily understand what this meant. Siblings who had not been tested did not understand risk until they were about 16 years old and had been told prior to testing or when parents thought they might begin sexual relationships. When young people knew they were or could be carriers of a recessive condition they had little idea of population risk, and thus the likelihood of them having a child with another carrier.

In families affected by DMD, girls up to the age of 16 did not understand the risks to their potential children. Children affected by DMD generally knew little about the condition, and heredity and genetic risk had not been discussed with them.

Children's emotional response to genetic risk

In families where parents treated communication about the genetic condition and its risks as an ongoing process throughout childhood, these children and young people had greater insight and appeared to cope well with the condition and their increasing knowledge of associated risk. By contrast, families who had initially tried to keep risk information secret later viewed a lack of knowledge as having led to increased stress and negative emotional experiences for children, as well as for parents.

“It wasn't ooh boo hoo Mummy's poorly, it was a relief cause she understood why Mummy was the way, she had been treating her the way she was...”

Parent of child < 8 years old – Partner had HD

Where communication was or had been less open, children struggled to understand what was happening and parents said they had observed signs of stress, such as children and young people becoming withdrawn and easily upset. Many young people said they had felt better when they knew about and understood the implications of the condition and its risks. Young people often said they had felt scared about the risk of inheriting something they did not fully understand and they had experienced problems with self-esteem. Some individuals thought this may have been a contributory factor to them being bullied or self harming.

In some families, parents encouraged and supported young people to make their own decisions about treatments, genetic testing and care provision. This appeared to encourage young people to discuss information and their thoughts about the genetic condition with their parents and usually to follow their advice. In contrast, we observed that young people

did not discuss the genetic condition and their concerns with parents who made decisions and took a more authoritative role.

DISCUSSION

This is the largest study to date that explores the sensitive issues surrounding communication about genetic risk between parents, children and siblings. It has taken into consideration the views and experiences of families from six different genetic conditions that vary in inheritance pattern, age of onset, morbidity and life expectancy. Due to differences in these factors, the detailed content of family discussions varied slightly between the genetic conditions but how children learnt, their level of insight and understanding and the emotional impact of the knowledge gained was similar across each age range for each of the six conditions.

All children and young people valued being able to discuss genetic conditions affecting their family. Gradually learning about the condition as they grew up helped them cope and come to terms with the risk to either themselves or other family members, including their own future offspring, irrespective of the disease outcomes. Demonstrating that assimilation of genetic risk information is a process rather than a single act²¹.

Where children had not always received explanations from their parents about a genetic condition, this had caused considerable strain on the individuals, parents and children, and on their relationships with each other but also their wider social networks. Once family communication became more open, this helped re-establish understanding and a shared reality that enabled parents and children to support each other and cope better.

Most parents found deciding how and when to talk to their children about the genetic condition, a distressing and major issue in family life. Despite being encouraged to talk to their children about the genetic condition by health professionals, most parents or children were unable to access appropriate advice and support at different developmental stages. However this reflected the limited support available generally for these families, who often experienced other problems and issues related to their experiences of the genetic condition. A future manuscript will explore this in more detail.

Children and young people, irrespective of their risk status also highlighted their need for opportunities to meet with a health or social care professional with whom they could discuss the implications of genetic risk. Many young people were only just beginning to realise and understand the implications of genetic risk and the possibility of a carrier status when they were having genetic tests carried out. This raises serious ethical questions about how much the young people involved really understand when and after they have consented to the tests, and may indicate a need for further advice and guidance on the genetic testing of children²².

Young people often had difficulty understanding genetic risk information, in terms of risk statistics and probability, and often relied on subjective judgments such as shared family phenotype features. Often this reflected parents' views but also characterises how lay people often view hereditary²³ and the well-documented difficulties many people have with understanding risk probabilities²⁴. All age groups of children and young people have difficulty in understanding probability terms, and younger adolescents (10 to 15 years) particularly have difficulty distinguishing between 'probably' and 'possibly'²⁵. Probability

information or risk percentages presented as a pie chart²⁶ are the most effective method to assist children's understanding. Therefore, based on this evidence and the findings presented, more effective genetic risk communication tools are required, to aid parents and health professionals in providing better explanations.

Open family communication underpins the cohesion of families, and improves family members' commitment to long term care of a parent or sibling^{8,9}. Strong and supportive families have the ability to transform both children's and parents' lives, and help them cope with illness, the risks involved and the care provision that may be needed. Therefore, the care of the family needs to go beyond preparing for genetic testing or carrying out treatments for the individual affected by the genetic condition. There is a need for greater family centred care to support parents in advising and helping them to manage the condition whilst maintaining pivotal family relationships.

Supporting better family communication is likely to lead to better health outcomes for all family members. Greater family-centred care from health professionals will help family members cope and adjust to genetic risk information. This may be a role for a family support worker within existing professional groups²⁷. Such a worker could provide advice and guidance on family communication about genetic risk communication to parents, give young people the further information they require and generally support the family dynamic in managing and coping with the genetic condition.

The study does not allow us to know the findings' generalisability to the wider population of families affected by these and other genetic conditions. Future survey studies are required to find out the generalisability and test our theories further. In addition, the

recruitment of these families via support groups is a potential bias. However, others have suggested the majority of parents in families affected by genetic conditions, share the experiences we have outlined²⁸. Further studies are therefore required to establish the prevalence of the problems the parents and children identified, and to develop and evaluate interventions and resources to assist parents in talking to their children about genetic risk information.

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Table 1: Explanation of the genetic condition and inclusion criteria used to recruit participants

GENETIC CONDITION and summary of inheritance pattern	INCLUSION CRITERIA
<ul style="list-style-type: none"> • Cystic fibrosis (CF) (autosomal recessive – onset from birth) • Neurofibromatosis (NF) (autosomal dominant – onset from birth, wide variation in signs and symptoms) • Duchenne Muscular Dystrophy (DMD) (X-linked recessive – onset of symptoms in early childhood, progressive deterioration until death in late adolescence early adulthood) • Haemoglobinopathies (HbO) (autosomal recessive – acute episodes throughout child and adulthood, vary in frequency) • Familial Adenomatous Polyposis(FAP) (autosomal dominant – adolescence onset) • Huntingtons Disease (HD) (autosomal dominant - usually adult onset, often middle age) 	<ul style="list-style-type: none"> • Participant families had been diagnosed with or at risk from the genetic condition for a minimum of 12 months, and at least one member (either child or adult) was affected or at risk from the condition, including the possibility of being a carrier. • The family had access to a peer support group or a health professional following the interview so that any personal or family issues unintentionally triggered by the interviewers could be discussed with experienced people capable of giving appropriate support and advice. • The research ethics committee directed that the children to be interviewed should be aged 8 years and above, except for families affected by HD were only young people 16 years and above could be interviewed. • Children were only included in the interviews where parents confirmed they had personally explained to the child about the genetic condition and its risks, and they were willing to answer any questions the interview may raise for the child. • Definition of a family member according to Metcalfe et al., (2008).

Table 2 - Demographic Profile of Participants

Individuals interviewed	Family backgrounds	Age Range of Parents Years (N and Gender)	Age range of Children Years (Gender)	Ethnicity	Educational background of Parents	Genetic condition affecting families (N)	Status of children in relation to genetic condition
52 Parents = 34 Women and 18 Men	Children living with parents in 29 families	30-34 (4♀0♂)	8-11 (5♀4♂)	5 Afro-Caribbean or Black British	21 left school at 16 years	Cystic Fibrosis (CF) (4)	21 affected or positively tested for genetic condition
33 Children (aged <18years)	In 21 of these, children lived with both parents	35-39 (6♀2♂)	12-14 (3♀7♂)	3 Asian or British Asian or Middle East origins	15 left school at 18 years following advanced level qualifications or vocational course	Duchenne Muscular Dystrophy (DMD) (6)	11 were at risk of being affected or carriers
11 Adult children (aged 18+ years)	In five, children lived with single mother	40-44 (9♀9♂)	15-17 (7♀7♂)	25 White and from the UK	16 had University level education	Familial Adenomatous Polyposis (FAP) (6)	12 were unaffected siblings
16 Parents interviewed as a couples	In three, children lived with mother and new partner	45-49 (8♀5♂)	18+ (9♀2♂)			Haemoglobinopathies (HbO) (6)	
17 Women interviewed alone including 6 single mothers		50-54 (2♀1♂)				Huntington's Disease (HD) (7)	
4 Men interviewed alone (2 separated fathers)		55-59 (3♀0♂)				Neurofibromatosis (NF) (4)	
		65+ (4♀0♂)					

The families represented a wide spectrum of religious beliefs and included: Atheism, Christianity, Church of England, Hindu, Humanist, Methodist, Muslim, None, Pentecostal, Roman Catholic and Sikh.

Note: The information in the rows bear no relation to each other, the findings are to be read in their columns only to give an indication of the different types of family that took part in the study.

Figure 1: Family roles in the communication of genetic risk information

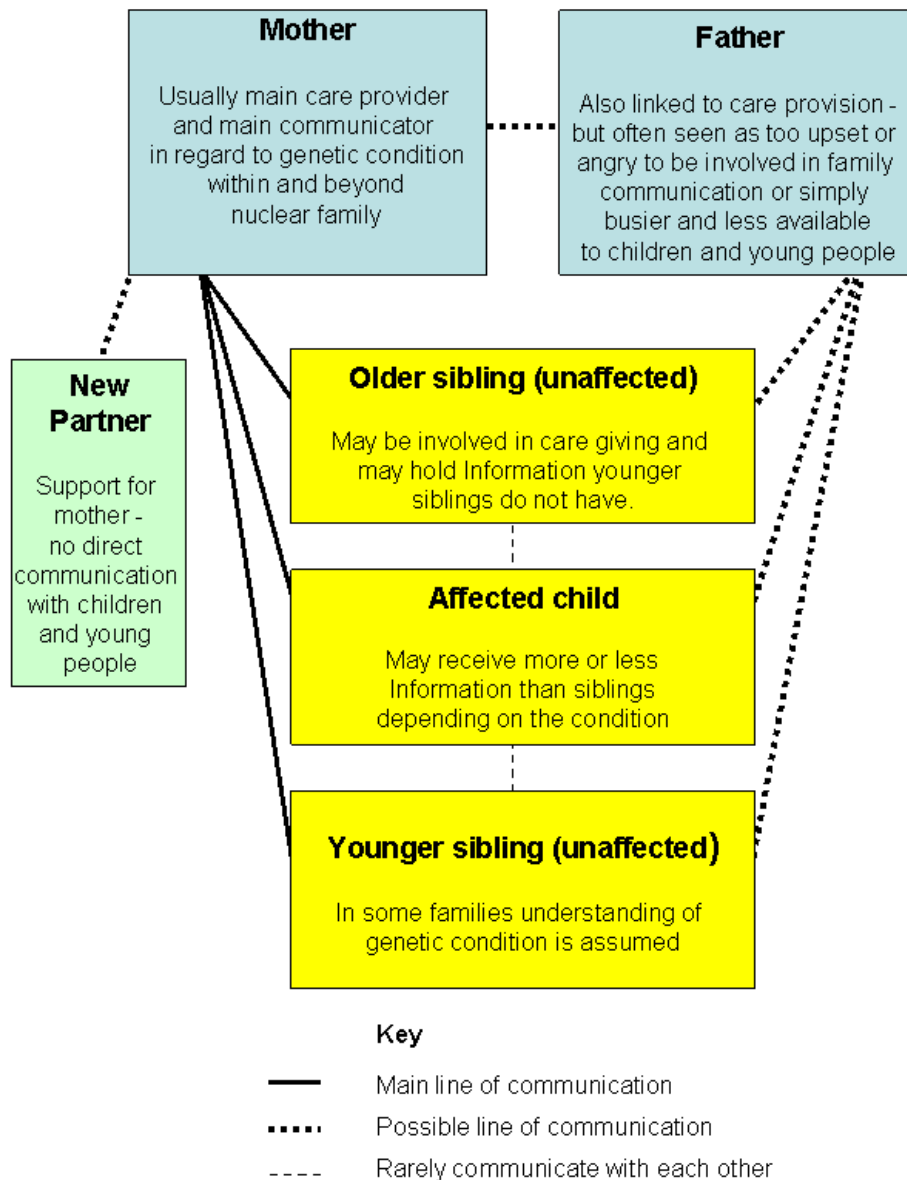




Figure 2. Children’s developing understanding and response to genetic risk.

Understanding of genetic condition and risks 				
Children began to notice and question visible difference and some asked if they were likely to be affected in the same way as what they observed. Some understood that the condition was a result of the biological relationship between parents and child in the same way as eye colour.	Children understood the condition in terms of what they could see and how it currently affects their daily lives. Most understood the notion of hereditary in terms of the condition being passed down through the family but had no idea of hereditary patterns.	Children began to understand more about the condition in terms of expected future symptoms and about mortality. Most understood hereditary in terms of their relationship to their parent(s) but not to their future children. Most use genetic language but without an underlying understanding, and they could not describe hereditary patterns accurately or quantify risk.	Young people understood that they may carry a gene that could or would affect their health. They had a clearer understanding of mortality although still not always the stages of progression. They were more able to describe hereditary patterns and began to realise the implications for their own future children.	Young adults began to understand the wider implications of the condition and relate it more to themselves. They realised how it might influence their career choices and personal relationships and how their decisions relating to genetic testing had impact on other family members.
Up to 7 years	8 - 11 years	12 - 14 years	15 - 17 years	18 years plus
Children seemed to accept information at a relatively superficial level. There were some behavioural problems or signs of stress when they were not given information in difficult family situations.	Children continued to accept information at a relatively superficial level and looked for positives and did not let the condition become the central focus of their lives.	A period of rebellion for some children who became angry and questioned ‘why me?’ Some children challenged treatments and routines.	Young people reported upset and shock as they better understood the mortality, responded to test results or faced surgery. However they looked for positives and did not let the condition monopolise their life.	The most difficult time emotionally. Young adults were coming to terms with the implications of the condition at a time when so many other important life decisions and choices are being made.
Emotional reaction 				

Timeline showing the observed changes with age of children and young peoples’ understanding of the genetic condition and genetic risk, and their emotional responses to information if they learn new information in this age category.