

Parents as partners

A report and guidelines on the investigation of children with developmental delay;
by parents, for professionals



This document summarises the input of participants in the patient and public involvement component of the Cambridge Genetics Knowledge Park (CGKP) *Learning Disability & the Interface with Genetics Project*. This was an interdisciplinary project by CGKP that sought to develop expertise and services for the genetic investigation of learning disability. The document portrays the parent and carer perspective and has been created through a collaborative involvement process with families affected by learning disability.

The document is divided into four sections. The first provides a background to the project and the guidelines. The second section summarises evidence collated from families and the third section contains parent/carer recommended service changes. The final section is a set of recommended guidelines for the investigation of children with developmental delay written in a collaborative manner by the parents and carers who participated in this project.

All principles and guidance contained within this document have arisen from participant input and we have tried to use participants' own words wherever possible.

Please note that the document does not necessarily represent the views of the professionals involved in the CGKP project, nor professional guidance.

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Acknowledgements:

We acknowledge with thanks the many parents and individuals for their generous and thoughtful contributions.

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Foreword

Just as parents and professionals have collaborated to create this report, so it is imperative that parents and professionals work together as mutually respected partners at all stages of investigation and diagnosis. It is refreshing to read in this document acknowledgement of families' experiences and perspectives of diagnosis of a genetic cause of learning disability. For a successful working partnership, professionals need to be sensitive to parents' needs and wishes. To this end, the report highlights parents' recommendations for service changes and presents guidelines for the investigation of children with developmental delay. I would hope that professionals will read the report with interest and adopt as many of the recommendations as possible for the benefit of parents and their learning disabled children.

Beverly Searle
Unique CEO

January 2006



The consultation process

Background

Participants

Participants were drawn from across the country with a strong representation from the East Anglian region.

Represented conditions included: Angelman's, Batten's, DiGeorge (Deletion 22q11), Downs, Fragile X, Klinefelter's, Lissencephaly, Miller-Dieker, Rett, Tuberous Sclerosis, Turners, Ring 18, Phelan McDermid Syndrome (Deletion 22q13) and Syndromes Without A Name (SWAN).

We also had representation from Contact a Family, Community Living, Mencap and The Learning Disability TaskForce.

Process

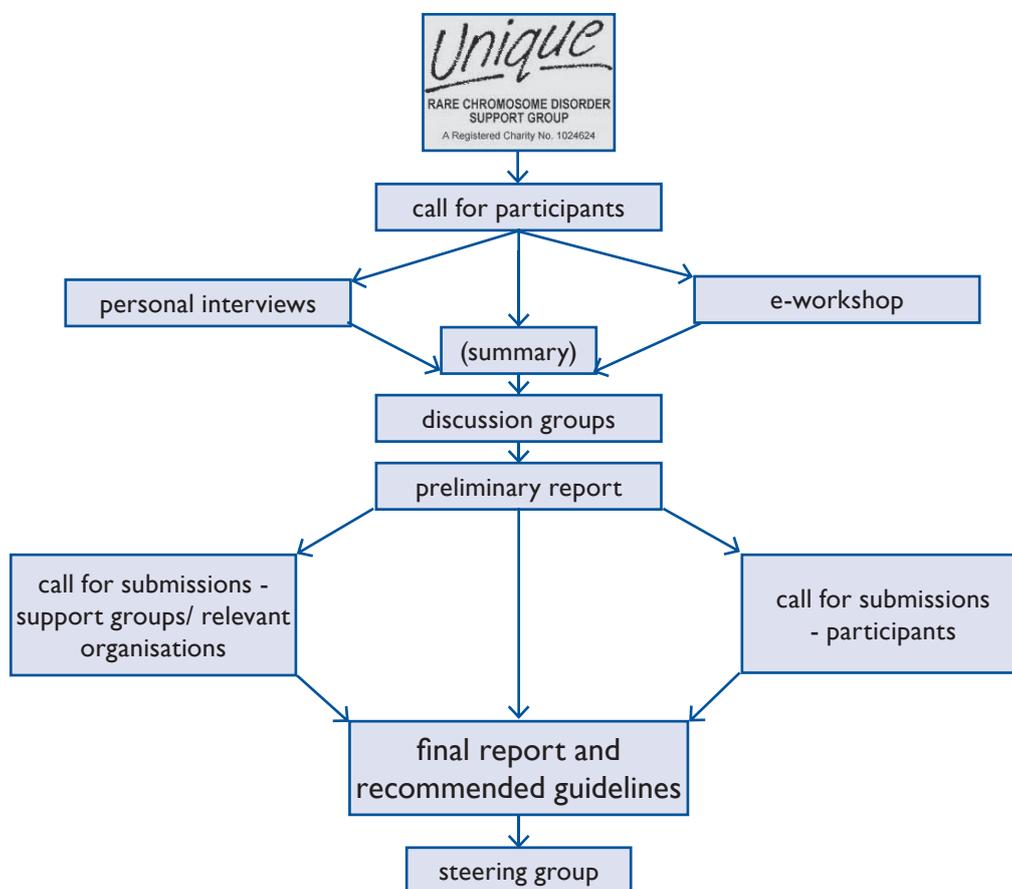
An initial survey of families with rare chromosomal disorders was undertaken by *Unique*, a rare chromosome disorder support group, examining what benefit accrued from a late genetic diagnosis. Just over 100 families responded and results suggested that in 80 per cent of cases a genetic diagnosis does not make a substantial difference to the services and support for patients and their families where a learning disability had already been identified.

As a response to the survey we established a consultative process aimed at understanding parent and carer perspectives of genetic testing for, and diagnosis of, learning disability. The process was iterative, starting with a broad set of general questions, refining or expanding the questions through consultation and deliberation, and then feeding questions and responses back to participants. The input methods for involvement are described below.

Interviews

We conducted interviews with 38 participants, two with a genetic condition, the remainder parents and carers. Of the parents, six were fathers. A total of eight interviews were conducted in person in Cambridge, the remainder by telephone. All parents were members of support groups.

The interviews were conducted in systematic semi-structured narrative style. We opened with an explanation of the purpose and scope of the project and why we were seeking parent and carer involvement. We then asked participants to tell us about their personal experience of investigation and diagnosis. There was no time limit on interviews and most lasted more than forty minutes.



Forum

To provide a platform for participant discussion, we also conducted an online moderated forum. The forum had 46 registered members. Not all members actively contributed to the forum, although from follow-up discussions it seems apparent that they did read the posts.

The forum allowed people affected by learning disability from across the country to participate in an anonymised group discussion. Posts were moderated to ensure that they conformed to our data protection and use rules.

There were four main areas in the forum: advantages/disadvantages, diagnosis delivery, services and support and other. Participants were free to discuss issues within these areas and there was no direct intervention by moderators, allowing participants to direct discussion and highlight important points. Discussions were summarised and structured and sent back to participants for review and approval.

Discussion groups

The final stage of the project was to take the feedback from face to face, telephone and online consultations to two discussion groups, one in London (15 people) and one in Cambridge (21 people). The former was primarily constituted of support group representatives and the latter

with family members. Groups were asked to review consultation work, make suggestions and 'brainstorm' possible guidance.

Feedback and review

As stated above the process was iterative, so draft documents (including this one) were sent out to all participants in each of the above-mentioned groups (interview participants, forums, discussion groups) for feedback. They were then forwarded to a number of key stakeholders, support groups and relevant experts.

Along with the organisations who participated directly in the project (Mencap, Contact a Family, Community Living, The Learning Disability Taskforce) the document was forwarded to other key stakeholder and support groups. These included: London IDEAS, Genetic Interest Group (knowledge translation group), Royal Mencap, SOFT UK and Oxford Genetics Knowledge Park. Participant input was included in subsequent drafts. We would also like to thank the Foundation for People with Learning Disabilities for the use of their electronic forums. Draft documents were posted to the forums, which have a combined membership of some 3500 subscribers (who, according to the Foundation, include "people working with people with learning disabilities, national and local policy makers, service providers and purchasers, parents, carers and people with learning disabilities") and we received a great deal of positive and helpful feedback via electronic and conventional post.

There were a few cases where edits, revisions or comments were deemed inappropriate, but where they were, contributors were informed of the reasons for that decision and provided an opportunity to respond or resubmit their comments.

The guidelines

The main document subject to this review process is the guidelines on the clinical investigation of children with developmental delay, which are attached to this report (see page 31). These guidelines began as a loose skeleton draft, collated from discussion group brainstorming. Over the course of the project they were refined into their current form. The guidelines reflect a 'gold standard' approach to genetic testing and diagnosis and can be said to have been collaboratively written by parents and carers affected by learning disability.

In talking to patients, parents and carers we have tried to reflect on both the positive and negative aspects of their experience. We asked them to highlight what they found beneficial about the process and what they disliked. Whilst we listened to criticisms, we also encouraged participants to think of ways that such events might be avoided in the future. We also asked them to highlight positive things that their health professional did which they thought other families might benefit from.



Evidence from families

Why parents seek a diagnosis

According to participants, the most important reason for a family to obtain a diagnosis is to gain knowledge about the condition. The most common reasons this knowledge is seen as important are set out below.

Just ‘to know’

The most frequently repeated basis for seeking a diagnosis was also the most non-descript. That is, parents ‘just need to know’ what is causing the anomalous behavior. Simply wanting to know should not be underestimated nor put down to mere curiosity; it is a genuine and profound emotional and intellectual need shared by almost all the parents of children with learning disability no matter what their experience or what the condition. One parent stated, “uncertainty is the worst torture of all”. There are also peripheral reasons why knowing may be important to some parents. Many want assurance that they are not “making up the condition” and that their concerns have been realistic and valid. Many parents have unfortunately found that, without a diagnosis, some people – be they family, outsiders or even health professionals – do not take them seriously. Hence, parents often seek reassurance in a diagnosis that the condition is not a result of their own behaviour or parenting. Having a name for the condition is also important to parents, particularly if the child is not obviously disabled. Parents who have a child who has no named diagnosis often state that “it would be much easier to say she has X syndrome rather than having to explain her disability to everyone I meet”.

To understand the future needs of the child

Parents often seek a diagnosis in the hope that it will allow them to predict what the child will be like in the future, by comparing them to others who have the same condition. Participants who have been through the process stress that a diagnosis never allows you to completely or adequately predict how a child will progress, how the disability will express itself or indeed all the potential problems a child will face. However, they do agree that it helps provide an insight into some of the future needs of the child. This allows parents to “anticipate potential problems and

“While not knowing in the early days allowed us a period of normality with our baby, later on we reached a point where we simply needed to know more about why he was not developing and what the future would bring. It was a very unsettling time because I was using every waking hour of the day to try and ‘reach’ my son. I was clutching at straws”

“Don’t ever let anyone think that it’s too late to get a diagnosis ... although they might never get over the grief it’s so much about closure”

“I did feel it helpful because people can sometimes think of the person as someone with mental health difficulties or badly brought up - a medical condition with lots of information around it does help focus people and make them understand and accept it and deal with it. Without the diagnosis people don’t have to face the problem and kind of find excuses for it.”

“The diagnosis was worth getting although traumatic – we have been able to re-plan our lives and drive our lives more pro-actively.”

“A lot of the guess work would be taken out of treatment, hopefully resulting in better control of symptoms and appropriate medication with minimal side effects.”

get something in place earlier rather than later” and “know what the best case scenarios are likely to be and aim to make these possible.”

To understand what the condition means for the rest of the family

For many families a learning disability automatically equates to a genetic condition, whether or not it actually is the result of one. This can cause concern about future family planning, and has meant that some members of the family have chosen not to have children. Parents may also have other children who might be affected by the condition and whom they want tested. Finally, knowing about the condition allows parents to plan for the needs of other siblings or family members, because the disability will create specific needs for them too.

To stop the uncertainty

Even though parents may recognise their child has a disability or learning difficulty, there may still be uncertainty if it will ever improve or whether the child will ‘grow out of it’. Parents can spend a long time worrying if and when the situation will improve. These parents seek a diagnosis to find some closure on the matter.

To find others affected by the condition

For many parents having a child with a learning disability can be a very isolating experience. They are unable to share the experience of parenting with their peers because their child has different needs and behaviour to others. Having others to talk to and share experience of learning disability can be extremely important. Families may seek a diagnosis, in part, to find other families to identify with and speak to.

To help the child obtain services and support

Regardless of whether or not families actually receive increased support subsequent to a genetic diagnosis (where the learning disability itself had already been assessed) – according to the *Unique* survey and consultations this may not always be the case – it is undeniable that this is one of the primary reasons parents seek a genetic diagnosis. Parents hope that the diagnosis will help them gain access to medical, social and educational services. They also see it as important to obtaining an early and accurate statement of special educational needs.

“We ... know that it has happened because of a genetic mutation... Therefore our other children should hopefully go on to have families of their own without the additional risk of this particular condition.”

“Our time and energy could be focused in the right places - and this was not always with my newly diagnosed son, but with his brothers who I realised were going to have their own needs in relation to all this.”

“I still remember the day my daughter was diagnosed at the age of 4 ¾ . Till then I had been told that she would ‘catch-up’ eventually.”

“In retrospect I think that I really wanted to know if there are other children out there with the same condition or other families who might have also been through it ... I just didn’t want to feel alone anymore ...”

“E is a beautiful articulate tall elegant looking child – people look at you and think ‘what is your problem?’ Because she is compliant, well behaved, non-disruptive behaviour [sic] she slips through the net. A diagnosis and statement brings recognition and support that don’t come naturally to a child like her.”

The advantages and disadvantages of genetic diagnosis

Do parents' hopes about the process translate into beneficial outcomes? We asked parents to talk about the advantages and disadvantages of obtaining a genetic diagnosis and what their personal experience was. The response was mixed. Whereas some families have found the process distressing, badly managed and painful others have felt supported and cared for and have a strong connection, respect and affinity with the health professionals involved. Most processes contained a mixture of good and poorer aspects. No participant stated that there was nothing positive or nothing negative about the process.

It was pleasing to note that parent's experiences during the investigation processes seem to have improved over recent years. The most negative stories generally come from more than a decade ago. Families more recently in contact with services are, on the whole, much happier with the process and the way health professionals have dealt with it. Again there are exceptions to this rule.

Families seemed to have more positive experiences from the investigation processes where these were undertaken by specialist genetics services rather than more general or local services. As an aside, we are also very happy to note that all participants who received a diagnosis in the East Anglia region in the last decade found the process positive.

Common responses to a diagnosis

Parents describe a variety of emotional responses at the time of diagnosis. These include:

Relief

For those parents who have been seeking answers for a long period of time the news can come as a great relief. They describe having an 'at last' feeling, regardless of how positive or negative the news. Receiving the diagnosis: allows them to 'move on', provides a degree of closure and lets them concern themselves with living and caring for their child.

"[W]hen a child is diagnosed at 4 or 5, following years of suspicion and doubt ... by [people or professionals] who put the [parent] down as an overanxious fussy hysterical woman, then to some extent there may be relief - at least initially ..."

Hope

The loss of hope has been a major theme that ran throughout consultations. On the one hand, a diagnosis may help restore hope to parents who thought: they would never find out what was wrong with their child, their child was the only one who experienced such symptoms or they would never receive medical recognition of the disability. On the other hand a diagnosis can serve to diminish parents hopes, because "there is nothing you can do about a genetic condition".

"This is for life its not like an infection or an injury, so you lose any hope of it getting better in the future"

Affirmation

A diagnosis can affirm parents' concerns about the child and put at rest questions of whether the child's condition was the result

of bad parenting, a bad family situation or events during the pregnancy or birth.

Shock

The news of a genetic condition can come as a shock to many families, even though they already know their child has a learning disability. They may have been hoping that the child would “grow out of it” or “mature eventually”. Furthermore, a bad prognosis, in particular where the lifespan of the child will be shortened can also cause profound shock.

Anxiety

Many parents describe feeling very anxious at the time of diagnosis. This may be because of concern for how the child will be affected, potential familial impacts of the condition, uncertainty for the future, or how they themselves will cope with the condition.

Fault

Some people – particularly from older generations – can see finding the genetic cause as tantamount to ascribing “blame” or “fault” to the person who “passed on the responsible genes”. In several cases families have chosen not to tell other family members for such reasons.

Anger

Parents who have been seeking answers about their child’s condition can often feel anger at professionals when a diagnosis is finally confirmed. They may have been turned away from services previously, told that their child did not have a condition, or previously have been refused testing on the grounds that “it would be a waste of time”.

Grief

An extremely common emotion described by participants is that of grieving or feeling as if they have ‘lost their child’ – meaning the hopes and dreams of who they thought their child might become (a lawyer, teacher, athlete) have been shattered. Similarly some participants report feeling as if they had ‘lost

“We just wanted to know that they weren’t imagining [the learning disability] ... We wanted someone to affirm that ‘we believe what you are saying’”

“I knew my daughter had Down’s Syndrome the minute she was put into my arms - the diagnosis some 12 hours later was relatively superfluous - but make no mistake - it was still a huge shock to hear it. My husband didn’t realise at the birth ... so the diagnosis came as even more of a shock to him.”

“D’s condition is manageable at 9 [years old]. I just wonder what life ahead of her is going to be like. School provides a good amount of support at the moment but I have been told that a mainstream secondary school may not be able to support her. The specialists can’t really tell me if she will cope with her life ahead.”

“Grandparents on both sides were adamant that nothing like that had ever happened on their side of the family, each thinking that the “blame” must lie with the other side.”

“Mum and dad kept blaming themselves about who passed [on] the hereditary gene’ We didn’t want [them] to be tested because blame would point to one person.”

“After spending years seeking an answer from everyone I was told by [the specialist] that I was being over-obsessive about my child and damaging her psychologically. When we got the [diagnosis] I wanted to go and throw it in [the specialist’s] face. He had no idea how much damage that did to me and my relationship with D.”

“When we were told ... it felt like we’d lost all our futures.”

control' over the care of their child or that "it felt like nothing I did would matter".

Guilt

The negative emotions outlined above can be compounded by a severe sense of guilt about actually experiencing them. No parent wants to feel grief about a child that is still living, yet such instinctive emotional responses can be unavoidable. Parents feel bad about having such responses and being unable to control them.

Overwhelmed

In all the range of emotions experienced, the amount of information to take in and the alien nature of clinical diagnosis can be very overwhelming for those involved.

The impacts of the diagnosis

Once the immediate emotions have subsided, parents and carers have a chance to consider the impacts of the diagnosis. We asked parents to describe some of the advantages and disadvantages of genetic testing for learning disability. Some of the most common responses are outlined below.

"There is no question that [many] families feel grief when they hear the diagnosis."

"I felt all these uncontrollable feelings of loss and grief. I felt so guilty about feeling them too."

"My own psychological reaction to my daughter when I found out abhorred me and it took me a long time to forgive myself."

"It was a tragic diagnosis – we were overwhelmed by it all and went through a complete withdrawal"

Advantages	Disadvantages
<ul style="list-style-type: none"> May provide a name for what is wrong. Can help put you in contact with a supportive community. May help you find out more about the disorder. Other family members can be tested to see if they are affected. Can help long term planning for your child's and your family's future. Can alleviate concerns that developmental delays are the parent's or family's fault. Can mean that others become more sympathetic/supportive. You may be able to access more/improved services. It can allow parents to find a degree of 'closure.' 	<ul style="list-style-type: none"> There might not be a diagnosis. There might not be a community to connect to if the disorder is very rare. There might not be any recognised medical treatment regime for the named condition. It may cause stress for your immediate and wider family. It could impact on relationships with the extended family. It may make the future look more bleak. Having a named condition can sometimes result in negative stereotypes and labels. It may cause problems with securing insurance cover. In a worse case scenario it may impact on medical services.

Labelling

A predominant concern voiced at all levels of consultation was the impact of a 'label' on the child, family and care pathway. Labels can potentially change the way that individuals are looked upon by members of the family, outsiders and even health professionals. This can be both positive and negative in nature. In some instances a label can help gain further support or indicate what

specialist services might be needed. A label may also assist to increase tolerance of what otherwise would have been interpreted as bad or odd behaviour. On the other hand, a label can serve to obscure the child as an individual and result in generalisations either about the child or the way care should be given. Participants highlighted that diagnosing the underlying condition may not reveal the extent to which a child will be affected by developmental disabilities or 'how severe a cognitive impairment will be if diagnosed with a genetic condition'. They were concerned that a genetic diagnosis may be seen as providing a complete description of the child and his or her future. Participants were emphatic that a genetic test should only be utilised as a tool in the care pathway of the child, to 'open doors not to close them'. They urged continued investigation and refinement of the diagnosis throughout the person with learning disability's life.

"[the diagnosis can] stop us looking at the individual and put our trust in an unreliable body of knowledge. Empirical sciences do not work as well for the diversity of the human population as they do for the laws of physics. We must never take our eyes off the individual as one condition could impact two people quite differently ..."

"We love and care for the individual, so the diagnosis helps us interpret what we experience. The diagnosis and associated actions need continual refinement in the light of REAL experience rather than theoretical extrapolation."

"It can take away the individuality of the person ... even within the confines of your knowledge about your particular syndrome or condition that they have they are still individuals and they still confound us."

Is a genetic diagnosis worth pursuing?

No family reported that in retrospect, they would have preferred not to have received a genetic diagnosis. In fact participants emphasised the need for a genetic diagnosis as early as possible. All saw it as necessary, even though not every family saw it as having actual, tangible benefits. Some saw the benefits of knowing the basis of the condition as being undermined by bad management at the time of investigation and beyond. These families do not regret that a genetic test was undertaken but rather how the genetic diagnosis was handled.

Hence, we can say that generally, genetic diagnosis is seen as beneficial for families affected by learning disability, so long as it is properly managed.

Practical examples

Parents are most happy when the health professional treats them with dignity, empathy and respect, admits their own gaps in knowledge and assures parents that they will work with them to find out about the condition and the best way to care for the child. They are least happy with professionals who provide diagnosis in a cold, detached or dismissive way.

Two examples of patient experience are set out below:

Positive	Negative
<p>“Although [The health professional] admitted that she had not heard of [the condition] she had found some information on it for me to read and had found the number for the support group. She said that she would read more about the condition and suggested that I did the same ... [the health professional] met with me about five or six times after that ...”</p>	<p>“My daughter was born floppy ... but by 9 months [the time of diagnosis] she was sitting up and looking ok ... so although I knew she had a disability I wasn’t really expecting anything drastic. I was shown some pictures and asked if my daughter looked like any of them ... they were pretty scary. I was told she had a condition ... he read off a list of things that she would be like ... she wouldn’t have any children, would be cruel to children and young animals and her teeth would probably rot ... and that was it, I never saw [the specialist] again ... I probably didn’t show much emotion but it was a huge shock – my beautiful child had been taken away from me.”</p>

Participants were realistic about what could be achieved within the health service. They understood the pressures on health professionals and recognised that no family could be given the undivided attention and dedication of a specialist service. Rather, they expect a balance between the two above extremes. Furthermore, they would like to see a normalisation of practice and certain benchmarks for clinical investigation set. General principles developed and agreed to by participants are set out below. They have been used as the basis for the attached guidelines.

General principles for investigation and diagnosis

Participants were emphatic that there could never be any steadfast rules governing the process of investigation and diagnosis because every family is different just as every condition is different. Rather, health professionals must act with flexibility and discretion, responding and altering practice to suit the patient and family’s needs, capabilities and understanding. Nevertheless there was a consensus that certain core principles should be observed in interacting with patients, carers and families.

What parents expect from the investigation

The best interests of the child

The primary principle in any care framework is that health professionals need to act in the best interests of the patient/child. In practice this means balancing the impacts of clinical investigation against its benefits. Whilst parents overwhelmingly

“One of the guidelines is the testing MUST be beneficial for the patient and the family ... the testing mustn’t be done because of any external pressures ... for my mind that has to be a headline here.”

support genetic testing, prolonged investigation can be stressful for a child, particularly if there is no immediate diagnosis. In such a scenario it may be appropriate to consider – in partnership with the parents – if the child is already receiving adequate care and support and if a genetic diagnosis will make a substantial difference to their care.

Continuity of support

Families affected by a complex condition will need to interact with a series of professionals. Being moved from specialist to specialist and from clinic to clinic can be overwhelming, tiresome and stressful. Parents don't want to have to repeat the same stories for each health professional, care worker and educationalist they encounter. They would prefer that existing knowledge was built upon, with each professional contributing to the overall profile rather than re-collecting the same data. Ultimately participants would like to have one central person who can act as the intermediary between the family and those responsible for the care of the child in question. In the absence of such a key support worker, they would like to see coordination, collaboration and communication between all professionals in the care pathway to ensure there is continuity of service and support. Participants see regional genetics centres as particularly good at achieving such a holistic and 'joined up service'.

Social as well as medical support

Although genetic diagnosis is inherently medical in nature it will have more than medical repercussions. Indeed the diagnosis might have immediate emotional and psychological repercussions but change little in the clinical management of the condition. To truly make the process of genetic diagnosis beneficial to patients and their families, those responsible for delivering it must be tuned in to these needs and be able to provide adequate support or to signpost the relevant support services. The most important of these, according to participants, is the support group. The support group gives parents a chance to talk to others who are, or who have been through the same experience as them. It is particularly important to draw upon the strength of others and to see that there is a 'light at the end of the tunnel' once the emotions surrounding the diagnosis subside and 'real life begins'. This is not to say that participants were not aware of the potential flaws of support groups. They

"Before anything else we must remember that this is a child and the child should always come first."

"I would really prefer to have had the same person from start to finish."

"having to tell your story to every person you come into contact with is a bit like being dragged back over the same hot coals over and over and over again."

"I often feel we are an anonymous number in the system ... not feeling as if anyone really knows or cares what we are going through."

"At this point in time the diagnosis is more about me than my children ... its about finding support groups and information ... I don't think that the diagnosis should actually make a difference to the services, medical or otherwise that my children receive because I believe that should be based on the social model."

"I think when he's younger its really social and educational support we need although I know it will become more medical as he grows older."

"The medical profession didn't need to be involved at diagnosis stage because nothing medical that could be done at that stage - other bodies have to be involved. Doctors only look at ways of managing the medical side of things - but there is a whole person there with their own experience and own personality and own ways of being engaged with and motivated - psychological and emotional aspects of things."

recognise the difficulty in evaluating reliability of information and support provided by such groups. Contact a Family was agreed as a good resource to help evaluate the support group information. The length of time a support group has been running, its size, website, and how well known it is, are all indicators of its quality. Participants also recognised that some parents are not willing or comfortable with contacting such groups. Hence they urge health professionals to make parents aware of the relevant support group without forcing it upon them. Some participants have also recommended putting the parents in touch with disability advocacy groups and specialist care organisations. These organisations help parents see from the disabled person's perspective, to realise that there is a community of like-minded people and serve to motivate families in a positive way.

Reliable information

There is a great deal of erroneous information about genetic conditions in the public domain, particularly on the internet. Parents can happen upon shocking and disturbing worst-case scenarios and graphic photos when searching for information about a condition. Participants highlighted the need for guidance by a trusted individual towards the right sources of information. Participants believe that most parents will go straight to the internet to research the condition following a diagnosis. Arming parents with the appropriate information about the condition, where to look and what to look for on internet sites will not only mitigate the potential impact of such information on them it will also diminish the need for the health professional to counter unfounded misconceptions at a later date.

"Gaining the knowledge and insight of people affected is vitally important."

"Parents are often well supported by other parents. The support goes on a variety of levels: information, shoulder-to-cry-on, even just having someone else who is going through it themselves and also doesn't have the answers."

"[Support groups] provide some of the friendship and continuity that people need - not to mention the understanding of what they are going through."

"Also put them in touch with advocacy groups because that helps parents see from disabled persons perspective"

"parents need to talk to all the people around the disability or with the disability to see their own value"

"the group can support and advise the family on credible, family-friendly information."

"Parents are bamboozled with information, they want one central place to go and seek that information out."

"The best person to pass reliable information to you is the person who looks you in the eye as you take hold, rather than an anonymous webmaster. We must be able to trust the data."

"Many modern families will go straight to Google with a single keyword and get some horrid stuff in return."

"The quality of supportive information available generally is patchy or inaccessible. There may be good information on the Internet, but there is also a lot of really bad stuff with a focus on what will make your child stand out as abnormal."

"If you put information in peoples hands they actually look for what is right for them."

What parents need from the professional communicating the diagnosis

Expertise and understanding of genetics

When parents are informed of a genetic diagnosis they may need to ask questions about: the meaning of the test and results, the condition, genetics or the genetics service. Those who have been through the process believe that diagnosis should be delivered by someone with expertise in genetics, and if possible the condition. Parents will want to ask questions about genetics, the condition, familial implications and the genetics service. Participants would ultimately like this person to be a geneticist or genetic counsellor. They also believe that person should be someone whom parent's respect, who has authority and seniority.

Present the child not the disability

Many of those who have been through the process of diagnosis see it as one of the most profound events in their lives. It can cause huge emotional upheaval, which can be greatly exacerbated if the diagnosis is not properly framed and delivered. If too much emphasis is put on the prognosis and too little attention given to the child, parents can end up "viewing the child through the veil of the diagnosis". This can consequently impact on the way parents bond and relate to their children.

Good management will help focus parents on the positive aspects of diagnosis, mitigate unnecessary negativity and help parents "regain a sense of control". Participants therefore see it as imperative that health professionals "present the child and not the disability".

In practice this might mean:

- reinforcing that "the bond between a child with learning disability and the rest of the family is no different than in any other family"
- reminding parents that "they still have the same beautiful child they had before diagnosis; that nothing has changed, except now there is a name for that child's condition"
- recognising parents' emotional vulnerability and "avoiding throw-away comments" about disability or prognosis.

"GPs are not the best person [to deliver the diagnosis]— We would want someone who knows more ... a specialist, preferably a genetics specialist."

"The person should be an expert in the room otherwise some very unfortunate things can be said which just aren't right."

"The nice thing about having a geneticist tell us was that she could tell us at least a little bit about this rare condition ... However, we've not been able to see her again, we see our paediatrician who we have to take our support information to."

"Medical professionals tend to see conditions as a 'group' i.e. 'they are prader willies' rather than a group of individuals."

"news [of a genetic condition] is often broken badly, and this impacts on the way that parents bond with their babies. ... So many throwaway comments by medical professionals (and those associated with them) are tactless at best and downright prejudiced at worst. [Parents] are presented with the disability rather than the baby, who happens to have an extra chromosome."

"the diagnosis can 'make the future look bleak' if not handled properly."

Empathy without negativity

Given the sensitive nature of diagnosis there is a need for health professionals to be empathetic in the way they deliver information to families. However, in doing so health professionals need to consider the above-mentioned themes, specifically the need to focus on the child and give a sense of hope to parents. Thus, whilst it is important to recognise parents' emotions, the health professional in question should endeavour to retain a positive focus. Participants described empathetic practice as being more than simply what was said to parents. Rather it is evidenced in the way that health professionals are attuned to the immediate and long term needs of individual families and family members. Factors such as body language and the environment in the consultation room are very important. Simple things like sitting next to parents rather than across a table, providing a space for parents to talk, or even giving them a cup of tea may seem a simple gesture but they are remembered and valued long after the event. They also help build a relationship of trust and respect between family and the health professional in question. Participants overwhelmingly emphasised the need for parents to feel they have the health professional's time, to ask questions and just "to take it all in", intellectually and emotionally. Some elements of the diagnosis may be overwhelming in nature or result in substantial shock to one or both the parents. At these times families may not be capable of taking in every aspect of the diagnosis and prognosis. Being empathetic may mean spreading out the delivery of the diagnosis over several, well timed, consultations.

Honesty

Whereas participants emphasised the need to avoid negativity they simultaneously expect health professionals to be honest about the challenges that will be faced by the child and the family. Parents need honesty about the prognosis and what is known and not known about the disorder. They want health professionals to be honest about their own gaps in knowledge and the gaps in scientific understanding of the condition and its aetiology. Depending on the situation, health professionals might need to encourage parents to think about practical issues such as long-term care and how this will impact on the person and the family.

"Most medical professionals are busy, their offices are busy and cluttered, and you are just one appointment in a string. Parents are aware of how annoying it is to be kept waiting long past an appointment time, so feel pressured for time, to allow the next appointment in."

"Although a closed door and confidentiality are important, the quality of the environment is important too: to sit down on equal terms with a friendly person in a relaxed environment, free from time constraints, may allow just a bit more to sink home and provide the opportunity for the parents to take away more than a label."

"If I knew beforehand I may have been able to read up on it more, instead of ... finding out most of the information when I was not in the best frame of mind to take it all in."

"Over the first few consultations then you start building up what information the parents would like to know - you don't have to take it all on board at once."

"Make people understand the range of feelings they will go through and validate that but also show them there is a different perspective."

"You need to recognise the potential for institutionalisation ... parents should start thinking about how their children will make friends at an early stage, because it becomes much harder as they get older ... but [people with learning disability] really do need friendship."

"Today I read that they have found the faulty gene. M is 30 years old this year, we were told that he probably wouldn't live past two, then five, then teenage then young adulthood. How do you plan a future when you keep being told that there won't be one?"

They will need to ensure that their child is in a suitable residential environment, whether at home, school or in a community home and that they are able to develop socially and have friends. Finally, it might be important to ensure that the family is aware of and prepared for social stigmatism, labelling and stereotyping. As noted above, this information has to be handled sensitively and empathetically. Being overly blunt or factual about the prognosis is never helpful. On the other hand it is important to be forthcoming when asked direct questions and not to withhold information when parents request it.

Avoiding stereotyping

As noted above, the potential for labelling and stereotyping is a predominant concern among participants. Those involved in the diagnosis and care of people with learning disability need to be aware of the stereotypes others might hold about specific conditions and avoid adopting them themselves. Furthermore, health professionals should endeavour to ensure that a diagnosis never becomes a barrier to further investigation, support or care.

Affirmation

No matter how well handled the diagnosis, discovering that a child has a genetic condition can still cause shock and distress. Even in cases where participants are happy with the manner of delivery they simultaneously report feeling variously: grief, loss, despair, depression and emotional detachment from the child [see above]. Often these feelings are unavoidable, yet experiencing them can in turn cause a great deal of guilt. Parents need to know that they are not 'bad people' and that many other parents have similar emotional reactions. Those telling them of the diagnosis will need to affirm that other parents also experience such emotions. It is also important that health professionals do not feel as if these reactions are their fault.

The needs of parents/families

Parents as partners

Parents are their child's foremost champion, primary carer and spend more time with the 'patient' than any other person. They have a wealth of knowledge about the child. They need to be listened to and made to feel like partners in the process of diagnosis and care. Parents of children with

"I was given a three line diagnosis about how the condition would affect A. When I look back I laugh because you can't possibly explain in that little slot all the problems that A has in real life and how they impact on us all."

"[T]he professional who says 'they will never do anything' about a baby prejudices its development by expecting it not to be able to do things. You have to expect the child to learn or you will effectively prevent it from learning some things it might otherwise cope with. You have to keep believing it will learn so as not to further handicap the child."

"The specialist neurologist nurse gave a [hour and a half] slot to talk about initial diagnosis ... She talked through the issues sensitively and supportively and knew where we [the parents] were coming from. She knew about the feelings of guilt. She validated that feeling – it's alright to feel bad that it's normal."

"Make people understand the range of feelings they will go through and validate that but also show them there is a different perspective."

"You must recognise and deal with the question of fault early on."

"If handled sensitively, the ... process [of diagnosis] can feel like beginning a partnership with the professionals, handled badly it can be very isolating."

learning disability describe “not being taken seriously” by health professionals, when they reported atypical behaviour. When it was later discovered that in fact the child had a learning disability, they became extremely angry with not only the health professional in question but also the health service in general. Therefore it is imperative that parents feel they are listened to, treated with dignity and their intuition respected. Moreover, health professionals must recognise that by the time parents have reached the clinic they are emotionally and intellectually vulnerable. They should be cautious about dismissing parental concerns out of hand. Parents feel guilty about reporting their concerns about the child.

Health professionals need to recognise that the parents are individuals too. Parents of a child with learning disability might have different views from each other, different levels of understanding of child development, different knowledge of their child and different ways of reacting to, or coping with their child's problems.

The amount of information shared

Whilst participants generally felt it was important to include parents in all aspects of the child's care, including the process of investigation, they did recognise the need for discretion on the part of the health professional overseeing the process. Where a series of tests are being undertaken it may not be appropriate to name each condition being tested for, because this could cause unnecessary stress and uncertainty for the family. Informing parents does not always mean providing every detail about the

“As a mother I need compassion, humanity and a recognition of my role as a mother. My doctor acts with humanity and that heals me ... restores my personhood ... thank god we have moved forward ... every consultant I meet nowadays talks to me like informally, like a person ... they aren't dressed in medical gowns ... they introduce themselves by name and don't use professional titles and they don't condescend to me.”

“I wanted the doctor to listen to me ... to take on board what I was seeing happening at home and in the classroom ... and not told I was being oversensitive.”

“I am suspicious of people who want to catalogue families and put them in boxes - we are all different in every which way. And there is no proper way to grieve/cope or otherwise progress through the trials of being a parent.”

“15 years ago ... most of the information was provided by researchers who came, saw and went away again. Now parents and patients are much more involved and the balance has hopefully changed.”

“The paediatrician told me that I was wasting her time ... not listening to the parent is the ultimate sin.”

“We need to give greater honour to the intuition of Mothers.”

“Caring for a child is a partnership between parents and health professionals ... so parents should have a say in everything, right from the start, including the testing.”

“the prognosis was worse than we could have imagined ... apparently [the specialist] had 'suspected as much' but wanted to be certain before he discussed it with us. I was red raw, really angry that he hadn't told us. But later I realised that if he had been wrong in his suspicions we would have spent the eight months [waiting for the results] in absolute agony ... uncertainty can be the worst torture of all ...”

process. Rather it is about being responsive to what parents need and what impact providing or withholding information will have upon them.

Familial considerations

By its very nature genetic information has familial implications. In most cases people will have a sufficient understanding of genetics to understand that the diagnosis may impact upon other family members and for future children. However, this knowledge should not be assumed and such implications will need to be discussed where appropriate. Participants emphasised that all families should be offered genetic counselling. Some thought that this might be appropriate from the outset of an investigation if the health professional is reasonably certain that a genetic condition is involved. Their experience was that, discovering they were a carrier at the time of diagnosis increased the shock and emotions of the whole diagnosis. In the absence of genetic counselling at this time it may be appropriate to provide parents with a general information sheet on genetics and what it means for families. It must also be remembered that many people will find genetic information complex and potentially confusing. Without expert support people can find it hard to make sense of the diagnosis for other family members, or to adequately translate what they were told within the clinic. Telling the rest of the family can also be challenging if they need to tell others that they may be 'at risk'. Furthermore, as was noted above, family members can feel as if they are being 'blamed' for a genetic condition. Hence, participants stressed the need to provide parents with information and support to tell other family members.

"I was really wanting a diagnosis for my son ... so when my paediatrician said we're going to give us some genetic tests I thought 'oh great this might give us the answer' but I didn't think much beyond that ... when we went to get the results of the test and I found I was the carrier the shock value was enormous ... thinking back on it now it would have been very valuable if someone had said 'do you know what this really means ... do you understand what this might mean for you as parents too?' I think it would have been helpful to have some counselling at that point ..."

"Genetic testing is a huge thing because it can have implications for the whole of the family and it can have implications for you having other children ... you're talking about the essence of somebody's life here really ... I think that you don't need to take lots of time in broad terms what a genetic test is ... and if the doctor doesn't have the time then I think that you should have access to a genetic counsellor."

"It would have been better to hear first and then tell [child]. You are not at your best to deal with things and sensitively to deal with these emotions ... by telling the child you may have created a situation which the parents can't handle ... the family are the ones who give the child the basic support - you MUST listen to them - because they are on the front line. What is 'ethical' isn't always right because it has major family consequences which that 'ethical' person will walk away from"

Recommended service changes

Moving investigation and diagnosis to genetics service

There is an extremely high level of appreciation, respect and support for the work of the genetics service. The service was seen as most competent and capable in providing the services and support parents need at the time of investigation and diagnosis. Subsequently there are strong grounds to recommend that the investigation and diagnosis of learning disability – especially at the severe end of the spectrum – should take place within the genetics service.

“There is a call then for understanding that there are certain health professionals that are not as successful [at this process] than others ... but there are others who they should be referred to, to my mind that is a geneticist.”

“there should always be a referral to the genetics service, they have the support, facilities, compassion and time to deal with these issues.”

“The person centred approach is only being taken up in a patchy way through the medical service ... but it is something that the genetics [service] does quite well ... they work with parents and patients and look at how they can do something for them, understand them and work with them.”

“I am extremely grateful to all the geneticists I have seen who have spent inordinate amounts of time telling me about what the tests might mean for me ... that the tests might throw up something like reduced life expectancy [or] whether they would continue certain types of medical treatment ... geneticists have been extremely important in delivering that sort of information sensitively in a way that can be understood and digested ... and also telling me in a way that allowed me to decide whether I wanted to pursue further testing ...”

“It would really help if there were one person who could help support the parents all the way through rather than being passed from person to person.”

“[Parents] want someone to be an advocate for them and take away the stress of finding their way around the Health Service, benefits system, pre-school education, support groups etc.”

Key support worker

As noted above, participants consistently emphasised the need for a key support worker to take them through the process of investigation, diagnosis and into the care pathway and be their main point of contact within the health service.

A key support worker would optimally:

- have some level of expertise in genetics, although this is not completely necessary if the diagnosis is to be

delivered by a genetics service or a genetics specialist

- have a good understanding of local and national health and social services
- already be known to the family or become known to the family.

Participants were strongly in favour of extending the role of the genetic nurse rather than creating a new type of professional to undertake the function of key support worker. Genetic nurses are seen by parents as having sufficient competencies in genetics as well as the ability to interact with families in a caring and supportive way over long time periods. Participants would like to see the genetic nurse brought in from the outset of the investigation.

Follow-up consultation

Participants are emphatic that parents should always be provided the opportunity to have a follow up consultation. They argue that it is rarely possible to fully grasp the implications of a diagnosis during the initial consultation and that certain elements may need to be explained in more detail once the initial shock and emotion has subsided. They also strongly support the need for the professional responsible to spread the diagnosis over more than one consultation so the most pertinent elements of the prognosis can be provided at the outset and then less immediate concerns are dealt with subsequently.

“The support person [needs to become] more like a friend ... rather than like an outsider. Sometimes it is easier to open up to someone you can trust rather than a complete stranger.”

“A genetic nurse/counsellor might be able to help the person through the process ... it may be that someone who understands genetics in more general terms might be able to help people through quite a lot.”

“Genetic nurses can explain things so much better ... they understand how to translate information ... they have a way about them and an understanding of what the family needs ... they are brilliant and they can stay with you from start to finish and they are someone by and large you can contact.”

“Genetic nurses are the last port of call but they shouldn't be ... when you are initiating diagnosis or suspecting something genetic they should probably be brought in then ...”

“If a genetic counsellor had been available at some of the appointments I could have asked her afterwards ‘what did that mean.’”

“There should always be a follow up appointment because it is too hard to absorb all that has been told.”

Bringing GPs ‘back into the loop’

It is undeniable that parents want the investigation and diagnosis to be undertaken by those with expertise and authority in genetics. This does not mean that they wish all their child’s medical care to be undertaken by such persons or in such institutions. Travelling to the specialist clinic can be time consuming and expensive and large hospitals can be isolating and intimidating.

One recommendation brought forward was that the ‘GP should be brought back into the loop’. Many parents would like to have GPs deal with minor ailments rather than having to take their children to the specialist for things as simple as common colds, cuts or abrasions. Participants report reluctance on the part of some GPs to undertake such consultations because they know nothing about the main condition or its implications.

Having the GP on board may expedite obtaining common referrals for physiotherapy and occupational therapy. The GP is also seen as a local carer, who can act as a champion for a family and provide them with a “foot in the door of the health service”. Some participants saw the GP as someone within the medical system who might be able to take the lead in ensuring continuing support throughout the care pathway as an alternative to a key support worker.

In order to achieve a greater level of involvement it would be necessary for parents to visit their GP and encourage them to learn about the condition. In such situations it would be helpful if the specialist were to call the general practice and offer to discuss the diagnosis and answer any of the GPs questions.

Whilst it may be said there was a general consensus on the need to bring GPs back into the loop, there was certainly no unanimity on the issue. Some parents have had less than pleasant experiences at this level. Alternatively their child’s needs are so severe that they can only be dealt with by specialists. Others said they simply, “already see their specialist enough”. Although these families recognise the potential benefits of GP involvement for some families they would prefer that it be at the family’s discretion.

The involvement of the GP may be more relevant in regional areas where there is some distance between the family and a specialist or paediatrician. It will also depend on the service user’s relationship with their local or family doctor.

“It would feel very beneficial to have the support of the family doctor.”

“The doctor is really reluctant to see K, because he doesn’t know much about the condition and seems to be worried about complications, even for simple things like colds ... which means I have to travel to the city to see the specialist for pretty much everything”

Other recommendations

Participants recommended giving health visitors increased recognition and competencies. They would like to see health visitors have the ability to refer children to other therapies. In some areas such referrals can only be made via the GP and paediatrician.

Participants have also highlighted the need for increased awareness of conditions among health professionals and training for all those concerned in genetics.

Parents would like to see greater use of contemporary aids to explain diagnosis, to both the immediate and extended family. For example, using video and internet.

“Where is the (e.g.) video to take away, presented by a friendly face and using contemporary styles and visual aids to communicate key issues, and give us something to show grandparents etc without us having to face a barrage of questions and get the message over that we haven’t fully grasped ourselves?”

Conclusion

Many participants have had negative experiences of genetic diagnosis, yet all seem to believe that attempts to find an underlying cause for the condition should be made. How do such responses resolve with the *Unique* survey which showed that there was little marked difference to some aspects of the lives of patients and their families, such as enhanced service delivery? The answer lies partly in how the term 'benefit' is defined.

If benefits are taken to be only tangible returns, such as medical treatment, more services or support, then genetic testing for learning disability is unlikely to be seen as highly beneficial. If and when genetic knowledge can provide therapeutic tools rather than simply diagnostic ones this may change. Until such time however, there will be few immediate medical benefits to obtaining a genetic diagnosis.

There is perhaps a greater likelihood that families will receive services and support benefits as a result of the diagnosis than health benefits. Yet, participants emphasised that this was not always the case and certainly cannot be said to be the norm. In most cases medical and social service and support remained the same following a genetic diagnosis. Ironically however, some families report that where a named diagnosis cannot be found, standards of support actually dropped below what they were prior to testing. Conversely, others reported that where a diagnosis was provided, it meant that some health professionals stopped looking deeper to discover that in fact another separate condition was present. Thus, it seems that whilst some families will receive support benefits, many others will not.

There is no doubt that parents hope to gain medical and service benefits from genetic diagnosis. However, participants generally had a much broader outlook on what they wanted from genetic testing. Much of the discussion during consultations centred on how a genetic diagnosis could address the emotional, intellectual and psychological needs of families. Hence, participants described the need to "move on", "know they weren't a bad parent", "know no-one was at fault", "stop worrying about the cause", "find others in our situation" and so on. These are emotional and psychological issues, not medical ones. Similarly when parents describe the need for a diagnostic label to alter the perceptions of others around them towards them or their child, they are describing social issues.

Health professionals need to recognise that genetic diagnosis creates and resolves as many emotional, intellectual and social dilemmas as it does medical ones. The process of diagnosis should therefore be more than just a medical procedure. It must be managed so as to take into account the wider implications that the disclosure of the information will have on the family. This is why so much of the discussion in consultations involved what happened when the diagnosis was communicated rather than what went before or after. How that information is handled is extremely important to families and is the dominant issue for those who have been through the process. As one parent put it, "the three things I will remember till the day I die are marriage, birth and diagnosis".



This is not to say that the benefits from genetic diagnosis are merely emotional or intellectual. As noted above, some parents do report receiving improved service and support. Yet, what is clear is that merely receiving raw information in the form of a diagnosis does not axiomatically lead to such improvements. Simply, a diagnosis of itself is not a solution. It needs 'value adding' and even then can only be considered a component of the overall care pathway.

Just as health professionals need to import more than medical knowledge into the process of diagnosis, so too must they be willing to consider how to add more than purely medical value to the diagnostic information. The guidelines in this document give help on how this can be achieved. A genetic diagnosis needs to be passed on to the right people in the right form. Parents need to be armed with sufficient knowledge and tools to take the diagnosis out of the consultation room and into the real world. This requires a certain degree of proactivity on the part of health professionals. It also requires health professionals to inspire parents to be proactive once they are capable of it. Utilising the information to signpost services goes part of the way, but the greatest benefits will come if health professionals can build the bridges necessary to reach them.

Guidelines

Initial considerations

Parents will most likely be aware that there is something different about their child and will have a heightened sensitivity about the wellbeing that child. It is therefore appropriate that discussion with the parents about the child is properly managed from the outset and that certain guidelines for interaction are established.

- If possible identify a **key support worker** or discuss the benefit of having one.
- Parents sometimes feel that they are not being listened to.** They spend more time than anyone else with the child and they are the best placed to detect physical or behavioural abnormalities.
- Focus parents' attentions on the child,** not the developmental delay or disability. Highlight that parents have a beautiful, wonderful child who will bring them joy and who needs their love and care. Make sure parents feel they have your support and the support of the health profession.
- Consider whether an **interpreter** is required, if so double the time you would usually expect to take.
- If a parent is a sole parent, ask if there is another family member that he or she would like to ask to attend consultations.
- Explain how the health and/or genetics services work. Understanding the process and who is involved in assessment, investigation and care helps make the system seem less daunting and intimidating.
- Ask questions about the parents' experience of caring for the child. Encourage them to keep care diaries. This provides valuable assessment information and it also affirms parents' roles in the process of investigation and care.
- Avoid delays in investigation and referral wherever possible.

If you would like further information on how to break the news of a disability to parents, see the Scope 'Right from the Start' website at <http://www.rightfromthestart.org.uk>, which provides a template on telling parents about disability generally. Scope also provides Right From The Start Training to health professionals in some areas. For more details Email rfts@scope.org.uk or telephone 01432 370860.

Contact a Family provides a support pack for health professionals with practical suggestions on how best to communicate information and offer appropriate support to parents at significant times from pregnancy to pre-school.

It is available from their website <http://www.cafamily.org.uk/packs.html> or by calling 0808 808 3555.

Investigation

Professional initiated investigation

In most cases clinical investigation will be initiated by a health professional. Parents may not have noticed that the child has learning disability or may believe that slow development is due to immaturity. They may not have thought of the possibility of a genetic component to the condition, nor of any other specific cause.

In these cases the health professionals should take into account:

- The impact on the child.** Will testing cause stress to the child? How long will it take? Is this the first time the child has been investigated? Such considerations must be balanced against the potential benefits of having a named diagnosis. However if the health professional(s) think that the process of investigation would cause stress or harm to the child, she/he should discuss concerns with the parents. The decision not to pursue testing should be taken together.
- Parents as the primary carers.** Parents as partners in the process of diagnosis and investigation. They should be provided an opportunity to understand what is happening to their child and to make informed decisions in respect of them. However, health professionals need to **apply discretion** in how much parents are told before a diagnosis is provided. Expect parents to leave the clinic and undertake independent research about any named conditions. This may cause uncertainty, fear and stress on the family while they wait for results of a test to be returned.
- In most cases it is sufficient to tell parents 'we are looking for a number of potential causes including genetic conditions'. Where parents ask for more details of tests, or whether the child is being tested for specific named conditions, they should generally be told. However, it is important not to overestimate the chance of a specific condition at this stage.
- If it is relatively certain that the child has a genetic condition it may be important to provide the parents with some general information about genetics and what it means for families

from the outset of the investigation. If a parent finds out, for example, that he or she is a carrier at the time of diagnosis it can compound shock and other negative emotions. The health professional should consider whether it is appropriate to refer the family to a genetic counsellor.

- Parents rarely choose not to pursue testing. If they do, assure them that the option to pursue testing will remain open. Provide the parents with the contact details of Contact a Family.

Contact a Family provides a fact sheet on genetics entitled 'A Genetic Condition in the Family'.

It can be downloaded at It is available from their website:

<http://www.cafamily.org.uk/genetics.html> or by calling 0808 808 3555.

The Centre for Genetics Education (Australia) provides free general fact sheets on human genetics which are presented in a simple and accessible manner. They can be downloaded (HTML or PDF) from <http://www.genetics.com.au/factsheet/default.htm>

Parent or non-professional initiated investigation

In some situations a child might be recommended for testing by a non-medical professional, by a social worker or school. Parents too might request genetic testing for a specific condition. In these scenarios a health professional should consider the following:

- It is important to make parents feel that you **recognise the importance of having a diagnosis** and their need to know as much as possible about their child's condition.
- Do not underestimate the need for resolution and closure for the family that a diagnosis might bring.
- Counsel parents** about the process and ask what they hope to achieve by it. Parents should be aware of the advantages and disadvantages of genetic testing before the process is initiated [some of these are discussed on page 6 of the attached report]. In particular they need to be made aware that a genetic test may not always lead to a positive diagnosis.
- Ask parents whether they know what learning of a diagnosis will mean for them and how they would respond if they found out they were a carrier. Consider referring them to a genetic counsellor if they are uncertain or had not thought about these issues.
- Parents will need to **consider the welfare of the child**. That is, will the diagnosis actually change the way the child is cared for? Help them consider whether the stress on the child is justified or if the child is already receiving adequate care and support.

Communicating the diagnosis

Preparing for the consultation

There are several things that can be prepared in advance that can help improve the environment and make the overall experience more positive for parents.

- Schedule the appointment early in the day** – and, if at all possible, not on a Friday afternoon – and ensure that extra time can be allocated to the consultation if necessary.
- Obtain relevant **interpretation services** if necessary.
- Parents will want to **ask questions about genetics and understand the implications of this condition**. Do you have sufficient expertise in genetics or knowledge about this condition? Is there an appropriate person within the institution who can provide this level of expertise?
- Parents may find the process daunting or forget much of what has been told to them. If possible **prepare information about the condition** for the parents to read later. It may be valuable to have a tape recorder ready, so that the consultation can be recorded and transcribed.

Contact a Family's website is the best place to find parent/patient oriented information about conditions and the contact details for support groups. The website is located at <http://www.cafamily.org.uk/> and they have a free hotline on 0808 808 3555.

The Genetic Interest Group provides condition specific fact sheets for a number of recognised learning disability conditions. These are available free to families and health professionals by contacting the Genetic Interest Group (GIG) Unit 4D, Leroy House, 436 Essex Road, London N1 3QP, Tel. (020) 7704 314; e-mail: post@gig.org.uk; web: <http://www.gig.org.uk>.

BDF Newlife (Birth Defects Children's Charity) has a nurse helpline for families and professionals. The service is free and confidential and staffed by qualified nurses experienced in supporting patients and parents regarding inborn conditions. The helpline number is : 08700 707020 or e-mail info@bdfnewlife.co.uk. BDF Newlife, BDF Centre, Hemlock Business Park, Hemlock Way, Cannock, Staffordshire, WS11 7GF.

If there is no named diagnosis

For various reasons genetic tests for learning disability may not provide a diagnosis. Health professionals need to be aware that this can prove as upsetting to parents as receiving a named diagnosis. Parents might have placed emotional reliance on knowledge of the diagnosis. Not

receiving a diagnosis may serve to compound family tensions, or heighten the uncertainty about the child or the cause of the condition. In these situations parents should be reassured:

- They are **not alone**, many other people with learning disabilities do not have a named diagnosis.

There are many learning disabilities for which there is not yet a genetic test.

- That you **recognise that a learning disability exists**, despite the absence of a diagnosis.
- The child's condition is not their fault nor the result of bad parenting.
- The lack of a diagnosis **should not be a reason to withhold services** or a referral to relevant specialists.
- Assure the parents that the child will continue to be cared for and discuss whether further investigation is a possibility.
- Consider referral to a community paediatrician. They should then automatically institute a multidisciplinary assessment.

It can still be useful to refer parents to support organisations that deal with specific symptoms or aspects of their child's condition that cause concern (e.g. support organisations for children with visual or hearing problems).

Contact a Family provides a simple fact sheet entitled 'Living Without A Diagnosis'. You can obtain the fact sheet for free by contacting the Contact a Family Helpline on 0808 808 3555 or online at <http://www.cafamily.org.uk/undiagno.html>.

Contact a Family will also be able to direct parents to support organizations that deal with symptoms specific to their child's condition.

BDF Newlife (Birth Defects Children's Charity) publish leaflets 'Awaiting Diagnosis' and 'Diagnosis Unknown' which can be downloaded free from their website at: www.bdfcharity.co.uk

Where a diagnosis should be communicated

The environment where parents are told of a diagnosis has been identified by families who have been through the process as pivotal to how they receive and take on board the news. Get it right

and they may not even notice, but get it wrong and it will greatly exacerbate the negative emotions experienced.

- Parents should feel the **environment is private**, ensure that they have agreed to the presence of everyone who is in the room.
- Ensure the door is shut and that you will not be disturbed.
- Reinforce to parents that their **privacy and confidentiality** is paramount and will be maintained.
- Make parents comfortable and, if possible, sit at an equal level to them.
- Be flexible about time.** This is a huge event in parents' lives. They will need space and time to take in the information and deal with the emotions of the event. However, be aware that telling parents that you have cancelled other appointments may make them feel guilty about taking up too much of your time or the health service's time. Be aware of when parents want to leave, or if they have had too much. If a 'time out' space is available, tell them about it, but do not pressure them into staying.

Who should be present

Parents should always be told in person, whether or not results provide a positive diagnosis. It may be appropriate to alert parents of this practice early on. This way parents will not automatically make assumptions about the results when they are asked to visit the clinic.

- Ideally **both parents should be present**. If that is not possible then another family member, such as a grandparent may provide emotional support.
- There should be a senior professional and a support person in the room.
- The health professional should have an expertise in genetics (parents would prefer a geneticist) and if possible a good understanding of the condition.
- However, do not allow too many people to be present as this can prove very uncomfortable.

How the diagnosis should be communicated

There is never one completely successful way to tell parents a diagnosis. Every family is different and will respond in a different way. What is most important is that the health professional is able to adequately read the reactions and respond to the needs of the parents during the consultation. However there are some general principles that should be observed:

- Before the consultation, make sure you know each parent's name and surname. Do not assume that they are married.

- Foremost the child is a person**, and should be referred to by name, preferably not 'she/he' or 'your son/daughter' and definitely not as 'the patient'.
- Avoid 'throwaway comments'** about the disability or the prognosis that may have an impact on the way the parent views the child.
- Don't bombard parents with information. Recognise that the provision and exchange of information is not a one off process but a continual one. Be responsive to parents' capabilities and 'information saturation level'. Do not overload them with information if they are clearly not coping, but do not withhold information if they clearly want it.
- In general **parents need plain, clear language**, particularly in the beginning. Parents will find the situation daunting and confusing enough without highly technical language and medical jargon. On the other hand, expect that parents will want to do their own research, understand the medical basis of the condition and eventually become 'experts' in their own right. Therefore, key medical terms may be used with an appropriate explanation. It may be helpful to write down key terms for parents, not only relating to the condition but also generic genetic terms (e.g. chromosome, gene, recessive). Parents will need an understanding of these terms to understand the literature they will read and other experts they will talk to.
- Encourage parents to write information down if they so choose.
- If parents request copies of reports or referrals for patient held records, consider flagging up areas of concern with a sticker to highlight what the important features of the report are.
- Expect parents to undertake independent research about the condition. They most often do this on the internet, sometimes immediately after the consultation. Unfortunately they can sometimes happen upon erroneous or shocking information. If they are not forewarned about this or provided with credible information, you may be left to remedy misconceptions at a later date. Provide parents with any fact sheets about the condition that are available from reputable sources such as the Genetic Interest Group or the relevant support group. It is also helpful to tell parents how to identify good quality medical information about the condition.

The Genetic Interest Group provides a generic guide entitled 'A Genetic Condition in the Family'. It is available free to families and health professionals by contacting the Genetic Interest Group (GIG) Unit 4D, Leroy House, 436 Essex Road London NI 3QP, Tel. (020) 7704 314; e-mail: post@gig.org.uk; web: <http://www.gig.org.uk>.

Contact a Family provides an easy to read fact sheet on accessing internet information. It is entitled 'Medical Information on the Internet: Seeking Quality' and is part of their Parents and Paediatricians Together project. You can obtain the fact sheet from <http://www.cafamily.org.uk/info.html> by contacting the Contact a Family Helpline on 0808 808 3555.

You can also point families to the Judge Project's guidelines for consumers accessing health information online.

http://www.judgehealth.org.uk/how_to_search.htm

What should be communicated

As noted above, different parents will react to news differently. What is imperative is that every parent is told in a sensitive caring manner. Consider the following in deciding what information should be provided.

- Different parents will need different amounts of information in different time frames.
- In all, **honesty is the most important feature of the diagnosis**. Be honest about what you do or don't know. Be honest about what can be expected in the future or what is unknown about the future.
- Although it is sometimes hard to provide news of a disability in a positive light, **the way that information is framed can be important**. Hence focus on what the child will be able to do rather than their likely disabilities.

Tell parents something positive about their child so that they realise they still have the same beautiful child they always had. The diagnosis helps identify the basis for the child's condition and assist everyone to improve their child's care pathway.

- Affirmation is extremely important**. Parents report feeling a wide range of feelings following diagnosis, many of which can be negative, to the extent that they may feel 'grief' for the child they expected to have. The diagnosis may shatter parental illusions of the child and the aspirations for their future life. These feelings can, in turn, cause guilt, both in the short and long term. Make parents aware that such feelings exist, affirm that other parents experience them and that it takes time to adjust. Inform them that those who have been through the experience find that the feelings do abate and are replaced by the reward and love felt by all parents.
- Consider the cultural implications of obtaining a genetic diagnosis.

- Be aware that some people associate genetics with 'blame' or fault and it may cause tension within the family as to who is 'responsible'. Emphasise that it is no-one's fault that the condition occurred and no one could have known about or been responsible for the disability.
- Emphasise that the diagnosis doesn't provide all the answers and that each child will be different. Make it clear that the diagnosis provides a profile of the condition but does not allow you to make completely certain predictions.
- Emphasise that scientific and medical knowledge and understanding develops and changes over time. Tell parents that 'this is what we know now - what we know in 10 years time may be very different'.
- Highlight any immediate benefits arising from the diagnosis such as a clearer care pathway or extra services available.
- Remind parents that 'it helps us all just to know what the condition is, because we can now move on and focus on the best way to care for her/him.'

At the close of the consultation

- Make an appointment for a **repeat consultation**, preferably no more than two weeks away.
- Consider whether parents need **time by themselves** or a space where they can sit and talk.
- Are one or both parents in shock?** Do they need immediate support? Will they be able to drive safely? If not can another family member or friend be contacted to pick them up?
- Ask if any other family members may wish to contact you or an appropriate person to talk through the diagnosis.

Depending on the situation the following may be appropriate to discuss either in the initial or follow-up consultation.

- Parents need to be told that they are entitled to **consult with a genetic counsellor** and why this might be important.
- Provide parents information on the relevant support group.
- Provide parents with information on Contact a Family.
- Do not pressure parents into contacting anyone. Rather tell parents that 'many people who have been through this process find this helpful'.

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- Tell parents about 'parent held records'.
 - Encourage parents to be proactive** – tell them that it might be helpful to make an appointment with their family doctor and explain the condition to them. If the parents take up such an option it might be also worth seeking the parents' permission to call the family doctor to explain the condition prior to such a visit.
 - Map the care pathway with parents**, let them know which specialists or support workers will be responsible for the family. Most parents would prefer not to have to explain about their child's condition, background and prognosis to every health professional, carer, public servant and special needs coordinator they come into contact with, so talk to parents about what information they would like disclosed and to whom.
 - If there is a key support worker he or she should be provided with relevant contact details of all health professionals in the patient's care pathway.



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