Recessive Genetic Conditions:

The experiences of parents and professionals

healthwatch Sheffield

Firvale Community HUB

Acknowledgements

We would like to thank all parents and professionals who shared their views and experiences for this piece of work.

Contents

Executive Summary

Background: Families from all communities can be affected by genetic conditions (e.g. Thalassemia, Cystic Fibrosis). The Sheffield Genetics Literacy Project works with families and professionals to raise awareness about recessive genetic conditions. The aim of this report was to understand the experience of families of accessing support and information and the confidence of professionals in supporting families.

What did we do? Healthwatch Sheffield and Firvale Community Hub carried out a survey and had conversations with parents, most of whom identified as Asian / Asian British: Pakistani. We also carried out a survey with professionals.

What we found:

- Getting a diagnosis could be a challenging process. The news might be a shock to some parents and may affect their emotional wellbeing
- Parents had mixed experiences with support from their GP and the Regional Genetics Service
- Support provided in the community (e.g. by the Genetics Literacy Project) is an example of good practice in terms of cultural competency and language accessibility. However, professionals may have a limited awareness of its potential
- Overall, professionals felt confident providing support but highlighted some training needs in cultural awareness and sensitivity
- Overall, both parents and professionals felt well informed but information might not always reach people early on
- Parents had mixed experiences with how supportive the community was. There are some signs that views of younger generations may be changing

Recommendations: We have made recommendations about building the skills of professionals, strengthening the links between available support and improving cultural competency and the accessibility of information.

Background

What are Recessive Genetic Conditions?

We all have thousands of genes inside our cells, the codes that control our bodies. We have two copies of all our genes, one copy from our mother and one from our father. People from all communities carry several changed genes no matter where they are from or what their background is.

Some genetic disorders only occur if a person has two copies of the same changed gene, one from their mother and one from their father. These are called recessive disorders, and include conditions such as Thalassemia and Cystic Fibrosis.

Recessive genetic conditions can affect everyone, regardless of whether you are related to your partner or not. However, the chance is increased if you are related.

More information for parents/members of the public and Healthcare professionals is available here.[1]

Information on Genetic and Genomic testing is available here.[2]

The Sheffield Genetic Literacy Project

The Sheffield Genetic Literacy Project works with health professionals and community groups to raise awareness of the issue of close blood marriages and the links to recessive conditions. They work closely with families who have a child or children with a recessive genetic condition and help them access information and support and also help them share the information with extended families. More information is available here.[3]

The project was established in 2013 with the help of Genetic Disorders UK. After the first year, the project was taken over and funded by Sheffield City Council. This work is a part of their Infant Mortality Strategy and focuses on reducing infant deaths in Sheffield that are a result of recessive genetic conditions.

They have set up a referral system with GPs and heath visitors. This is in the form of letters that professionals can hand to a family whom they have identified as

[1] www.geneticsaware.org
[2] www.nhs.uk/conditions/genetic-and-genomic-testing
[3] www.firvalecommunityhub.org.uk/health-and-well-being

having a child with a recessive condition, or have referred to the regional service, or a family who may need extra information and support.

Staff have visited health visiting teams and GP practices in the Firth Park, Burngreave and Pitsmoor areas of Sheffield twice over the last 4 years. However the GPs and health visitors are not referring families to the project. Staff have reported that referrals are mostly through self-referral or via community organisations.

Healthwatch Sheffield and Firvale Community Hub worked together to understand the experiences of families affected by recessive genetic conditions. We also wanted to understand the experiences and confidence of professionals in providing support.

Objectives:

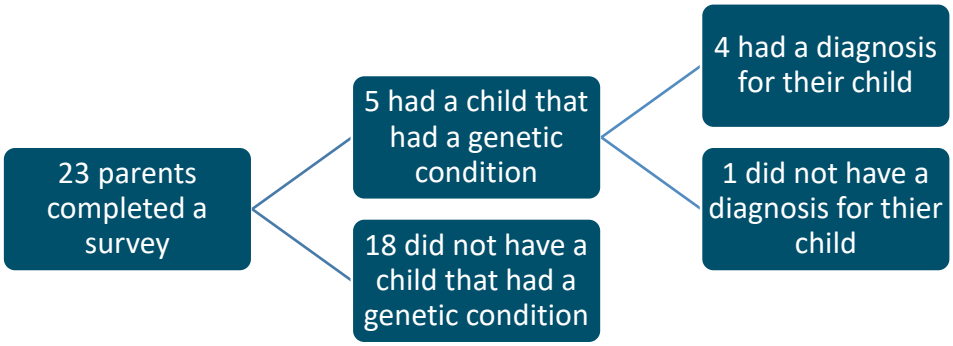
- Explore families' experiences of support services in Sheffield after a child is born or diagnosed with a recessive genetic condition
- Find out where families access information about recessive genetic conditions, and how useful this information is to them
- Explore the confidence and experiences of people about having conversations with family members and others about recessive genetic conditions and close blood marriages
- Find out what health professionals know about the Sheffield Genetics Literacy Project, how they are engaging with it and what further support they need
- Make recommendations to improve service design, commissioning and practice

What we did

We carried out two surveys – one with parents and one with professionals who support families. We asked people different questions depending on their situation. We also had more in-depth individual conversations with parents.

Surveys and conversations with parents

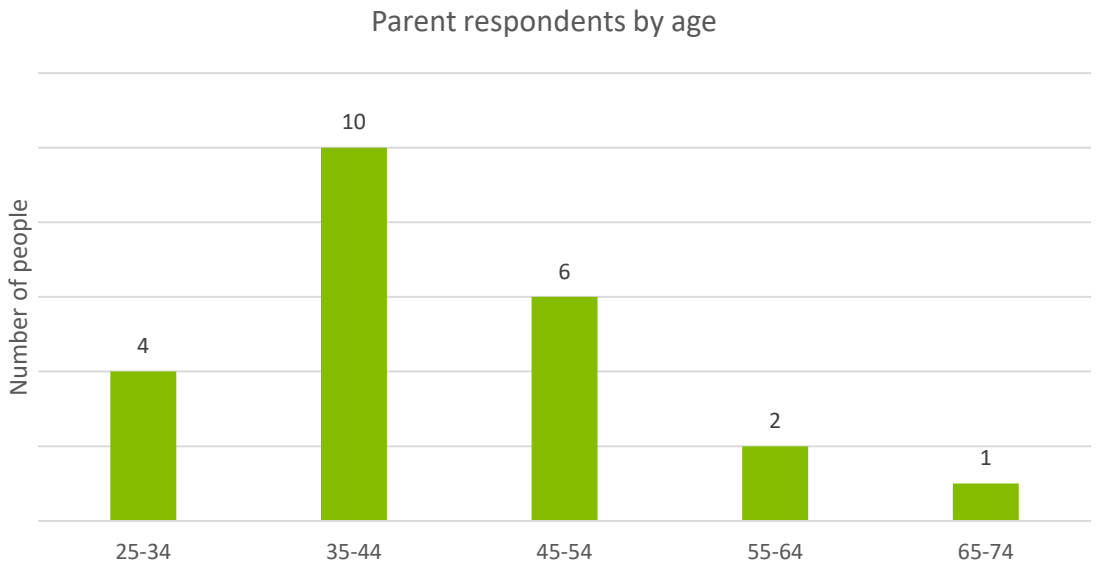
Survey with parents: 23 people completed the survey for parents. We asked people different questions depending on whether they had a child that had a genetic condition or not.



The questions for the parents who had a child that had a genetic condition focused on parents' experiences with accessing support and information, etc.

The questions for people who did not have a child with a genetic conditions focused on people's confidence in their awareness about the topic, etc.

The majority of parents were aged between 35-44 (see graph below).



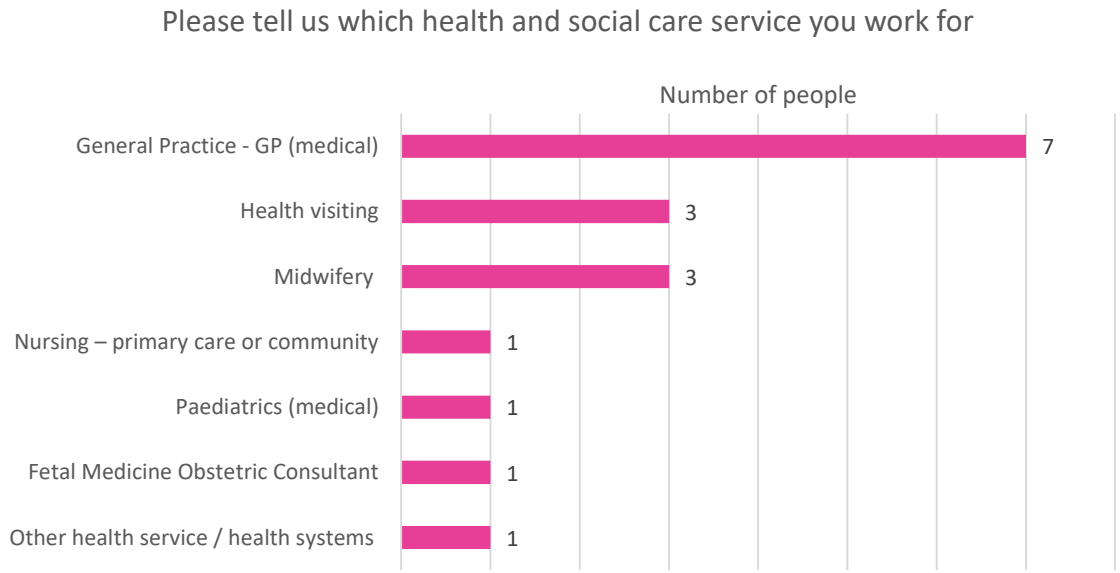
In terms of ethnicity, the majority of parents who completed our survey identified as Asian / Asian British: Pakistani (see table below).

	Number of people
Asian / Asian British: Pakistani	17
Asian / Asian British: Indian	1
Asian / Asian British: Indonesian	1
Mixed / Multiple ethnic groups: Black Caribbean and White	1
White: British / English / Northern Irish /Scottish / Welsh	3

Conversations with parents: We also had individual conversations with 5 parents (3 conversations took place in English and 2 conversations took place in Urdu). All of them had a child that had a genetic condition. These were different people from the survey respondents. The conversations covered a range of topics such as getting a diagnosis, getting information, and experience of services.

Surveys with professionals

15 people completed the survey for professionals. They worked in different geographical areas across the city. Most professionals we spoke to worked for GP practices (see graph below – some people selected more than one answer). We asked professionals about their experience and confidence in providing support.



The majority of professionals we spoke to identified as White: British / English / Northern Irish /Scottish / Welsh.

	Number of people
Asian / Asian British: Pakistani	2
Asian / Asian British: Shri Lankan	1
White: British / English / Northern Irish /Scottish / Welsh	12

What we found

Parents had mixed experiences with the support they received

2 out of the 5 survey respondents who had a child that had a genetic condition, felt well supported by health professionals.

Challenges in getting a diagnosis: 4 parents who completed our survey had a diagnosis for their child.

The time it took to get a diagnosis varied from a couple of weeks to a year:

> *My daughter was 4 when she was diagnosed with a metabolic condition and diabetes. I went to A&E when she was 4. The Consultant at the hospital diagnosed her after carrying out several tests. Took about 2 weeks to find out what was wrong with her.*

Confusing process: One person explained how getting a diagnosis was a long process which involved seeing different professionals:

> *The whole process took over 1 year. Teachers were not very helpful and direct with me. I kept seeing different consultants every time I visited. I didn't like that and it wasn't good for my son either. Eventually after maybe 1.5 years we met a nice doctor, young lady who was consistent for a while.*

Covid-19: One person explained that they had been *"Given the diagnosis, but treatment is taking time due to covid"*

Still no diagnosis: One person described the ongoing challenges in getting a diagnosis:

> *Still don't know what is wrong with my child. Doctor didn't tell me much. I have done my own research on the illness and talked to the genetic development worker. She has helped me understand that I might not ever get a diagnosis. She gave me courage to share with my family. She also gave me some good leaflets.*

Emotional wellbeing, diagnosis and treatment: When children became ill or were diagnosed, this caused distress in some families:

My son developed stress and anxiety and that affected me. He was put on medication which effected his sleep and eating habits and that was causing me anguish because his appetite dropped and he lost a lot of weight.

We were very sad and devastated when we heard the news. Even though the midwife had already given us information it was a big shock. I didn't know anything about Thalassemia and had never heard of it before.

The shock could also make it difficult to remember the experience:

I can't even remember half of what happened in the last 3 months of pregnancy because I was so stressed and I can't remember many things.

Not getting good advice from GPs:

All 5 survey respondents said they did not get good advice from their GP.

GP was always rushing and no spent time answering questions.

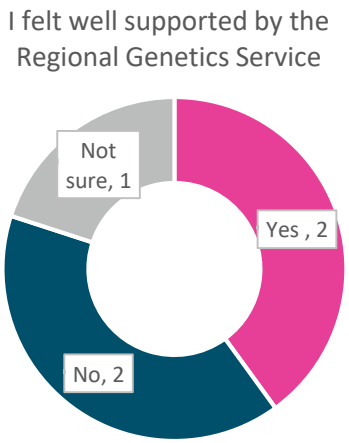
A parent told us:

Got rejected from Ryegate [House] first time around. Had many conversations with GP over the years. I think GPs also struggle with diagnosis when the problem is not visible.

Mixed experiences with Regional Genetics services:

4 parents were referred to Regional Genetics Services and they all attended their appointment. People had mixed feelings about the support they received from this service.

Genetic appointment was confusing I only went once didn't see the point after.



One person was happy with how they were treated by professionals:

Regional Genetic appointments were good, didn't feel like I was being judged or being held responsible for marrying a cousin. They talk more about the genetic side of things and explained we are both carriers rather than dwell on the fact that we are cousins. They said that this can happen to anyone - which made me feel a bit better.

Sheffield Community-Based Genetic Literacy and Support Project - an example of good practice:

Parents shared some positive feedback about this programme. Parents mentioned the following aspects of the project that they found useful:

- Connecting families whose children have genetic conditions
- Helping parents' understanding
- Providing resources in a preferred language

I think that it is very important that there is support available in the community to help families. Because hospital appointments are so quick and not very often. And sometimes you forget to ask the right questions and the I can just pick up the phone and ask [member of staff].

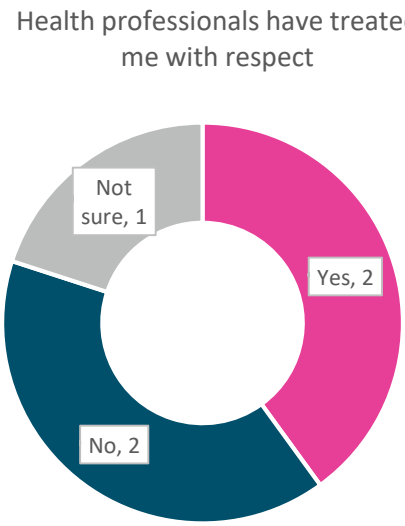
Professionals however, showed little awareness of the project:
Out of the 15 professionals who completed our survey:

- 7 said they had never heard of the project
- 4 said they had heard of the project, but were not clear what they offer
- 4 said they had heard of them and knew about the services they offer

None of the 15 professionals we spoke to had made a referral to the project. This suggests there is a lack of joined-up working.

Being treated with respect by professionals:

Parents also had mixed experiences with how they were treated by professionals.

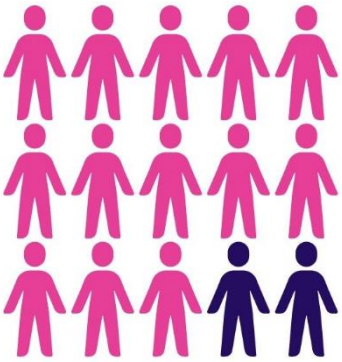


My GP said to me that I should have termination because my child would be born with an illness and that I should not have any children. I was very upset at this.

No referral to Regional Genetic Services. The consultant at Rotherham district hospital diagnosed her. Attitude was ok, one consultant was really nice but then handed to another who had a funny attitude. We got mixed messages as to what caused it from him.

Confidence of professionals to have a conversation:

13 of the 15 professionals had had a conversation about close blood marriages and recessive genetics conditions. We asked professionals who started the conversation (it was possible to select more than one answer):



- The majority (8 professionals) said they started the conversation
- 2 professionals said a family member started the conversation
- 1 professional selected both that they and the family member started the conversations
- 2 professionals were not sure who started the conversation

We asked professionals to rate how confident they felt about having a conversation about close blood marriage and recessive genetic conditions. The scale ranged from 1 (Not at all confident) to 5 (Totally confident). Most professionals rated their confidence as three or above.

We also asked professionals what makes it easier and what makes it more difficult to have a conversation with parents (see table below).

Things that make it easier for professionals to have a conversation with parents	Things that make it more difficult for professionals to have a conversation with parents
• Having an interpreter where there is a language barrier • Having a question about this on relevant forms • Asking parents how they met • Understanding the community • Understanding the patients' perspective • Professionals having prior knowledge of the patient • Knowing how to start the conversation • Trust between the patient and professional • Training for professionals • Parents already having some knowledge • Parents telling professionals that they are related	• Language barriers • Time constraints • Worrying about showing prejudice • Fear of causing offence • Parents not wanting to talk about complications • Families feeling judged • Lack of awareness • Not knowing how to start the conversation

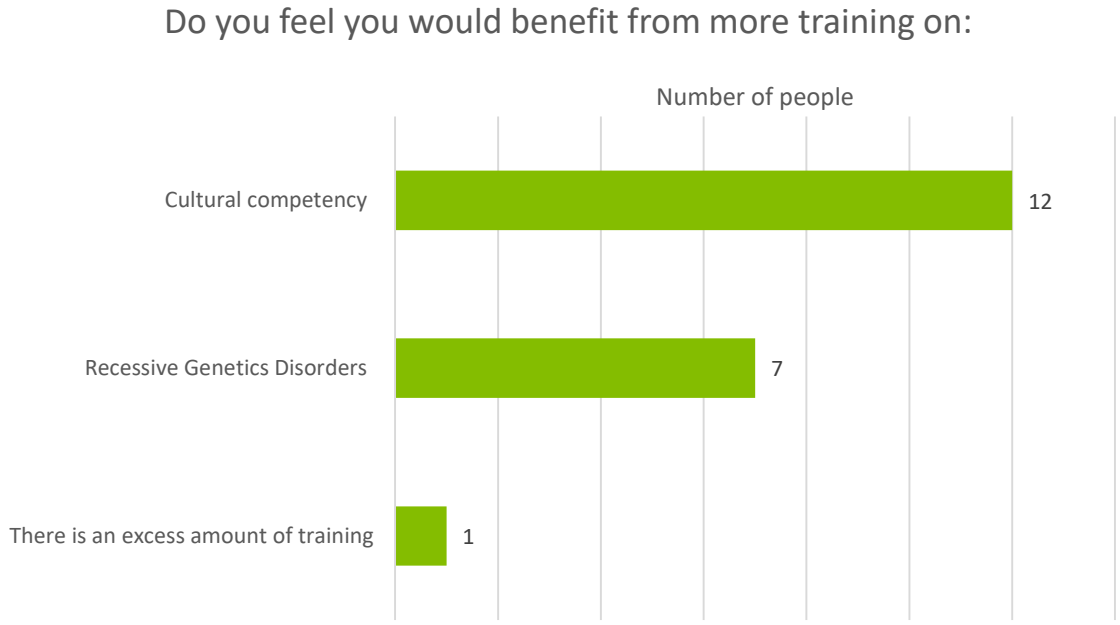
Some professionals told us about their experience in having this conversation:

Usually in the context of bad news from the scan, it is hard to ask about consanguinity at the same time, although it is important in genetic testing/follow up.

I had some training a long time ago and that made me more confident in having the conversations. Being a trusted professional that is welcomed into their home helped

Training needs for professionals:

We asked professionals about the training they felt they could benefit from. The majority wanted training on Cultural competency (some people selected more than one answer).



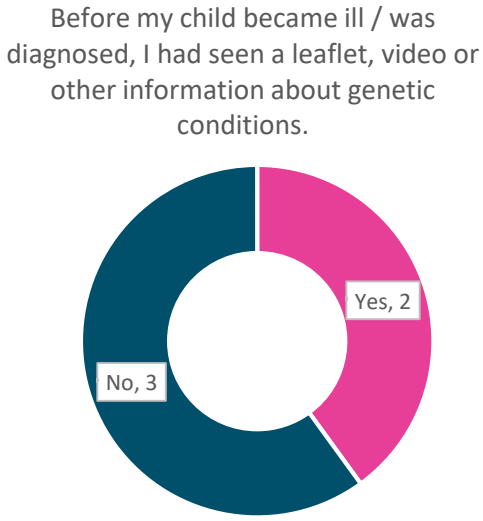
Further suggestions from professionals about training
• Brief video clips of these conversations going well and stories from the people whose lives are affected • Empowering families to be experts • How to approach the topic in a sensitive manner • Information about the common conditions and services available to couples/families • Training on the cultural aspects to pitch conversations appropriately (for GPs) • Cultural awareness

People received information but this was not always accessible and provided with cultural competency

Access to information for parents who had a child that had a genetic condition:

Having information early on

The majority of parents we spoke to had not seen information resources before their child became ill / was diagnosed.

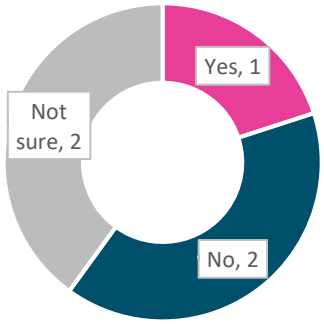


Information: Accessibility and cultural competence

The lack of information in different languages made it difficult for some family members to understand:

My partner's English is not very good and there was no material in Urdu that he could read to understand the condition. And because it was not a physical disability he found it difficult to digest. I found some YouTube videos in Urdu that I shared with my family. I googled a lot of information myself. I also asked a lot of questions to my consultants and GP's which helped me.

Overall parents had mixed feelings about the opportunities they were given to ask questions.



I had had the opportunity to ask lots of questions at every stage. The midwives and consultants were very helpful.

Parents found it useful when the information was provided in a community language. It was also helpful when the professional was culturally-competent.

The consultants at the hospital diagnosed my child. They were OK. Me and my husband don't speak good English and I felt the consultant would sometimes get frustrated with my repeated questions. The second consultant was Pakistani which made a huge difference. Helped us relax and spoke to us in Urdu. It made a huge different to the quality of information we received.

However, language was not always a barrier:

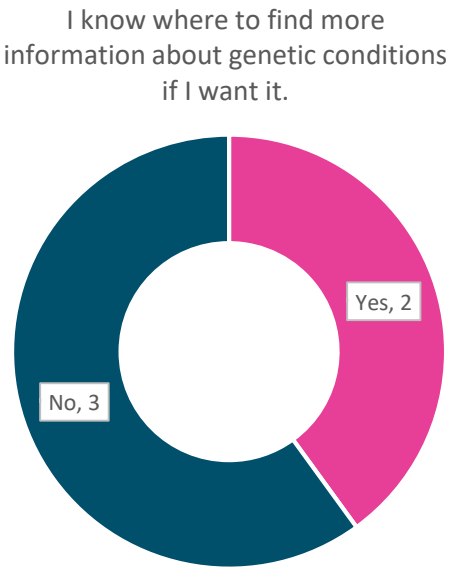
We got lots of leaflets about all the conditions we were effected by. We also got lots of information about things when I was pregnant. I got extra care, scans and tests whilst I was pregnant. All of the information was in English but we didn't have a problem with English.

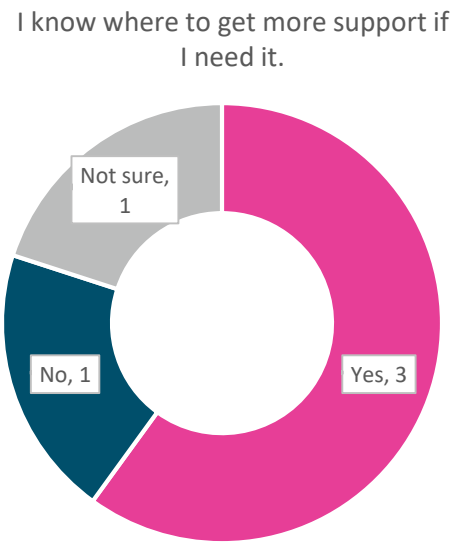
Some people explained how they got information from community-based initiatives. Staff also answered their questions in a culturally-competent way.

I have learnt a little bit about recessive conditions thanks to [member of staff from Sheffield Community-Based Genetic Literacy and Support Project] and the video workshop that she invited me to. She helped me understand what recessive conditions are and how we have been affected by them. I wouldn't have understood without the diagram and pictures she showed me. Thanks to this project my other family members avoided marrying cousins. I am lucky that I met her at the right time.

I got a lot of help and information from the SCAT [Sickle Cell and Thalassaemia Foundation] and a lady called [name]. The hospital gave me her number. She was so good in visiting us at home and giving us information in Punjabi. She helped me a lot and provided a lot of support to the entire [family]. I had multiple opportunities to ask her questions for many years and she also supported me through the blood transfusion process and through the bone marrow transplant. I asked many questions and became confident enough to do my own research as well.

Getting more information and support if needed: The majority of parents who had a child that had a genetic condition did not know where to find more information if needed. However, the majority did know where to get **more support** if necessary.



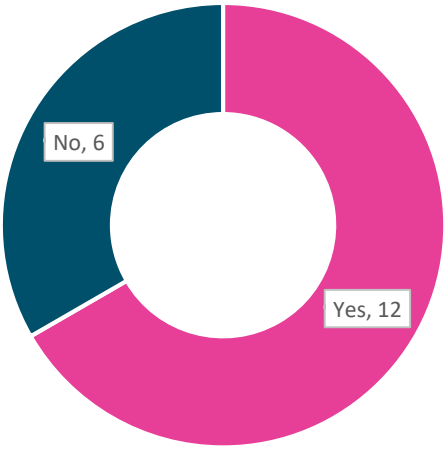


Access to information for parents who did not have a child that had a genetic condition:

Access to information: The majority of parents who did not have a child that had a genetic condition said they had seen information resources such as a leaflet or a video about genetic conditions.

I think I read/was told of the information about genetic diseases during my pregnancies

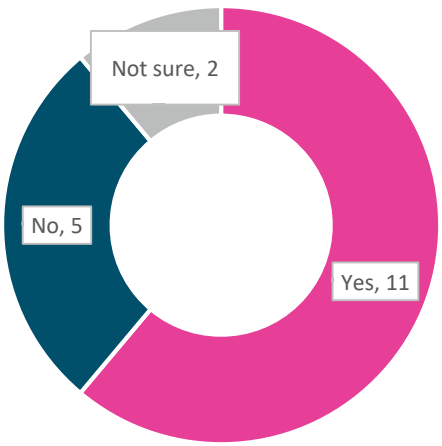
I've seen a leaflet, video or any other resource about genetic conditions.



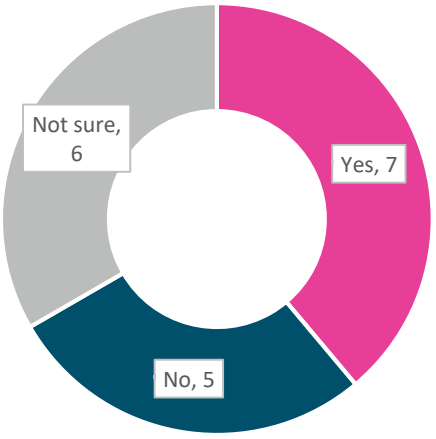
Informed decisions: The majority felt confident that they knew enough about genetic conditions to help them make decisions about marrying and having a family.

Getting more information/support: The majority didn't know or weren't sure where to get more information and support.

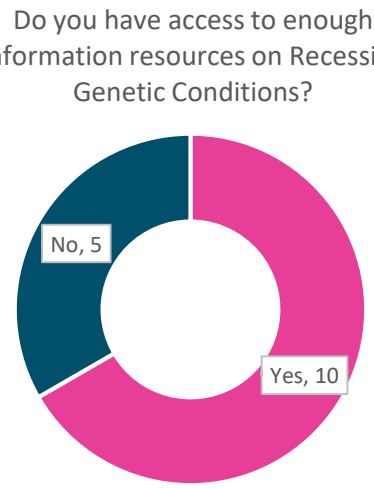
I feel confident that I know enough about genetic conditions to help me make decisions about marrying and having a family



I would know where to go if I needed to get more information or support on genetic conditions

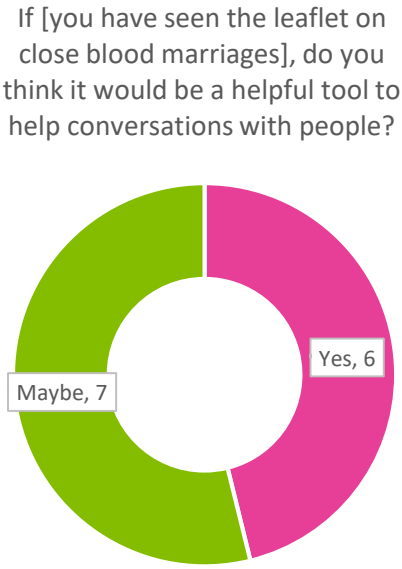


Access to information resources for professionals:



Access to information resources: The majority of professionals felt that they had enough information resources on recessive genetic conditions.

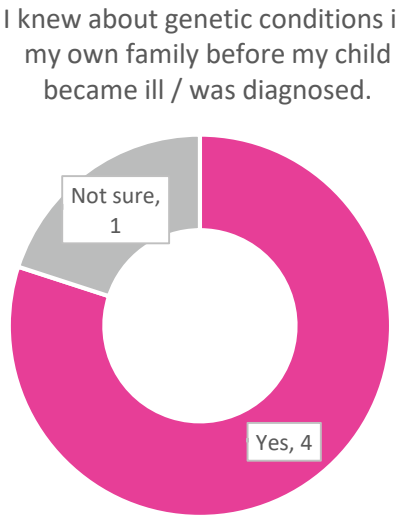
In terms of particular resources, 11 of the 15 professionals had seen the leaflet on close blood marriages.
To receive a copy of this leaflet, please email Saima Ahmed: saima@firvalecommunityhub.org.uk



Overall professionals felt that it would be a helpful tool to help conversations with people (13 people responded to this question although only 11 said they have seen the leaflet).

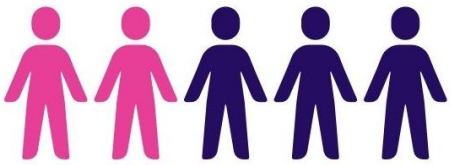
Views can vary between families and generations

Conversations in affected families:



The majority of parents knew about genetic conditions in their own family before their child became ill/was diagnosed.

Confidence having a conversation:
2 of the 5 parents who had a child that had a genetic condition said they feel confident talking to their family about genetic conditions.



One person shared their experiences with having conversations with the community:

Not much support available from the community. Families don't really share things with the community. People can be old fashioned and like to gossip. People don't understand recessive genetic disorders and how they are passed on. Very early I tried to have conversations with people but they were so negative about it and refused to believe that me and my husband carry this gene. Later, I stopped explaining things to people and just got on with my life.

At the same time close family could be a vital source of support:

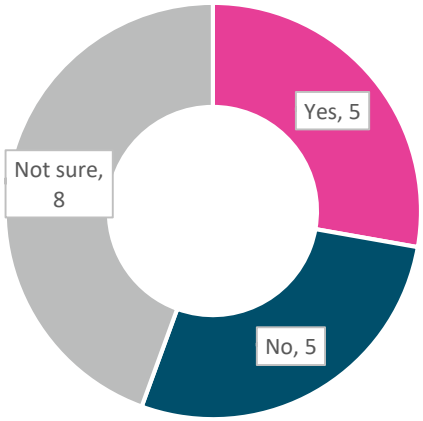
My siblings have been very supportive and we know how the other is feeling because we all have children in the position.

Conversations in families that were not affected:

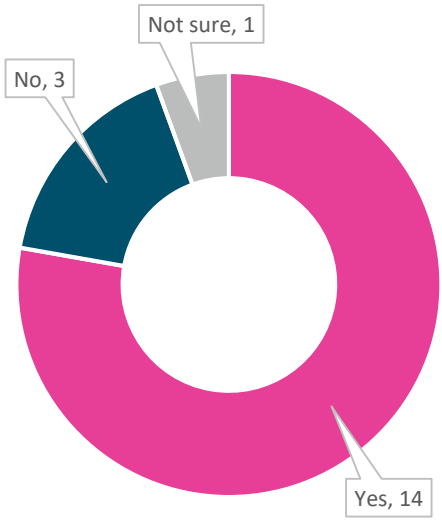
Knowing about genetic conditions in the family: Nearly half of the survey respondents who did not have a child that had a genetic condition were not sure if they had people in the family with a health condition which is genetic.

Confidence having a conversation: The majority of those who did not have a child that had a genetic condition felt confident having a conversation with their family about genetic conditions.

I am aware of people in my family with a health condition which is genetic (e.g. parents, grandparents, cousins, brothers, sisters).



I feel confident having a conversation with my parents / family about genetic conditions.



Change in attitude:

It seems that there might be an ongoing change in attitude in younger generations towards close blood marriages. All of the quotes below are from people who had a child with a genetic condition:

It's very hard to educate older parents. Young people are learning now the hard way.

Most other cousins of mine have also agreed that we shouldn't marry cousins in our family.

My younger nieces and nephews have not married into cousins and I see that our family is learning and changing their views. And I hope others can also learn.

Recommendations

- Emotional support should be made available and prioritised for parents around the time their child becomes ill / diagnosed.

- Health professionals should offer longer appointment times to allow parents to ask questions.

- Strengthen the referral pathway between health professionals and the Sheffield Genetic Literacy and Support Project to enable more families to access culturally competent and language appropriate support.

- The information provided to families should be:
    - Specific to the condition (e.g. managing the condition; inheritance patterns, etc.)
    - Culturally-appropriate
    - Provided in a preferred language
    - Followed up with additional conversations with professionals.

- Support the family with sharing the information to the wider family. For example:
    - Offering an additional appointment with family members
    - Providing a letter that explains the condition that family members can take to their GP.

- Sheffield Clinical Commissioning Group (CCG) should promote the uptake of the available training/awareness raising sessions and resources on recessive genetic conditions and close blood marriages and particularly resources that address cultural competency.

If you would like any further information about the Sheffield Genetic Literacy project, recessive genetic conditions, close blood marriages and reducing infant mortality in Black, Asian and Minority Ethnic communities please contact:

Saima Ahmed - Project Lead at
saima@firvalecommunityhub.org.uk

For information on training and resources via The Sheffield Genetic Literacy project and to obtain a copy of the leaflet on close blood marriages please also e-mail Saima Ahmed.

Healthwatch Sheffield

Healthwatch Sheffield helps adults, children and young people influence and improve how services are designed and run. We're completely independent and not part of the NHS or Sheffield City Council. If you have an experience of health or care services that you would like to share with us, or would like to get involved in our work, please get in touch:

The Circle
33 Rockingham Lane
Sheffield S1 4FW

Email: info@healthwatchsheffield.co.uk
Website: healthwatchsheffield.co.uk

Tel: (0114) 253 6688

Text: 0741 524 9657

Follow us @HWSheffield

Like HealthwatchSheffield

Firvale Community Hub

Firvale Community Hub is an award winning organisation committed to improving social equalities and inclusion for communities experiencing disenfranchisement and marginalisation in Sheffield. Firvale Community Hub empowers groups and individuals to find their voice and find leadership, to influence and inform a more inclusive society, through targeted and deeply personalised education and confidence building based projects.

A key civil society innovator in Sheffield, Firvale Community Hub works in partnership, in leadership, and alongside local agencies, leading development strategy and implementing pioneering methodology in the projects we deliver in our lively community hub.

127 Page Hall Road,
Sheffield,
S4 8GU

E-mail: info@firvalecommunityhub.org.uk
Website: https://firvalecommunityhub.org.uk/

Tel: (0114) 261 9130

Follow: @FirvaleComHub

Like: Firvale Community Hub

Subscribe: Firvale Community Hub