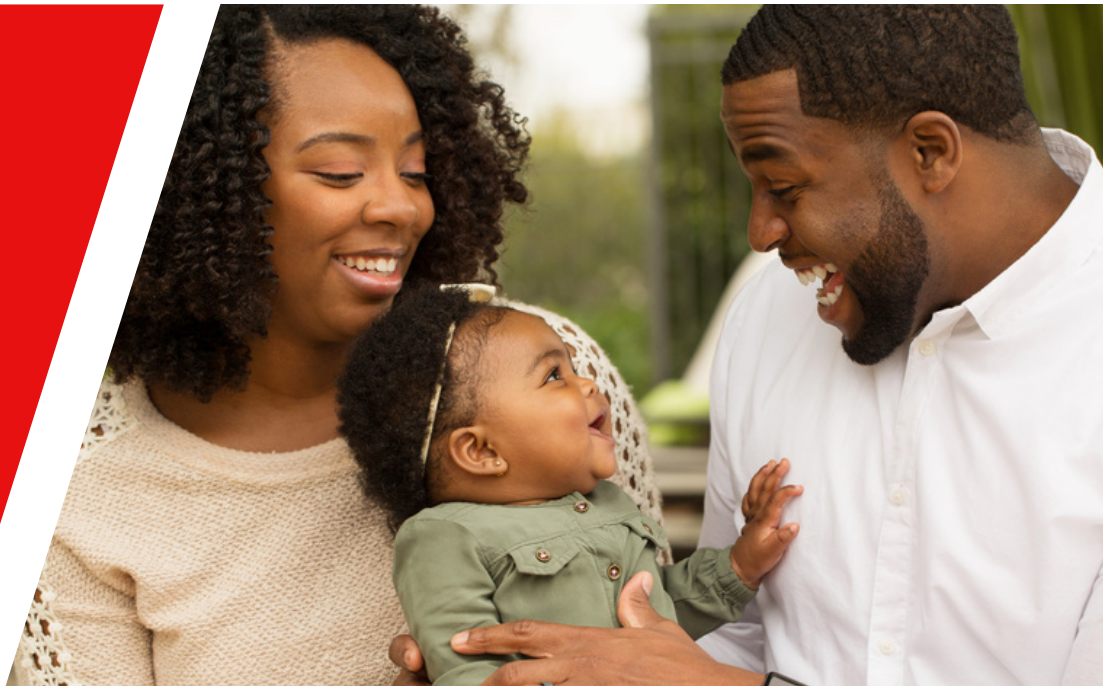
"It's in our genes"



Service User Experiences and Feedback on the Communication of Screening Results for Sickle Cell and Thalassaemia

APRIL 2023

CONFERENCE EDITION

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 ISBN: 978-1-8383098-3-1

Acknowledgements

We would like to express our sincerest gratitude to the individuals and parents who participated. Your contributions and willingness to share your thoughts, feelings, and experiences has provided us with invaluable insights of the experiences and challenges faced by families with children diagnosed with thalassaemia and sickle cell disease.
We know that discussing a child's diagnosis can be challenging, but your openness and honesty have made it possible for us to better support families like yours.

We would like to thank Professor Karl Atkin and Dr Maria Berghs for their advice on the report's content, participating in two focus groups discussions and for their critical feedback on the analysis.

We would like to thank the NHS Sickle Cell and Thalassaemia Screening programme for the funding to carry out this project and in particular we would like to thank the following individuals from the screening programme for their advice, support and contribution to this project and other streams of work

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Executive Summary

The Sickle Cell Society (SCS) and the UK Thalassaemia Society (UKTS) work in partnership with the NHS sickle cell and thalassaemia (SCT) screening programme to engage with communities less likely to access health information through usual NHS channels. The societies provide feedback from people that share the same population background or have experience of the condition and to feed into the programme updates and improvements.

To develop strategies for supporting parents with a newborn carrier or positive screening test result, the SCS and the UKTS undertook user feedback sessions to understand how health professionals communicate sickle cell and thalassaemia results to parents as part of the screening pathway and how support could be improved. The overall aim of the sessions was to share feedback on the lessons learned from the experiences of parents, so the screening programme can update its standards and guidelines and public and professional educational resources.

A total of thirty-six people were interviewed, twenty by SCS and sixteen by the UKTS. The SCS held three focus group discussions with all participants aware of their haemoglobin genotype and had acquired knowledge of sickle cell disease, as per the recruitment remit, which was for adult carriers, parents of children who were either affected by sickle cell or thalassaemia major and children identified as carriers. The UKTS used a combination of focus groups and one-to-one interviews. The UKTS also included discussions with people who did not know about thalassaemia or their haemoglobin genotype, thus giving an additional perspective to the feedback. There were commonalities within the feedback from both the sickle cell and thalassaemia groups such as, communication, signposting, and learning, these will form recommendations to the NHS Sickle Cell and Thalassaemia Screening Programme (NHS SCT Screening Programme) for further development of the programme.

NHS SCT screening programme offers all pregnant women antenatal screening for both sickle cell and thalassaemia. Newborn screening for sickle cell disease is offered to all babies in England as part of the newborn blood spot screening test. Babies with beta thalassaemia major are also identified.

From the original “Parent stories”, published in 2017, the findings show significant progress in the antenatal screening pathway for couples. Couples reported timely carrier screening and pre-natal diagnosis being offered. However, in the newborn screening pathway, some participants reported they were not given their baby’s results within the 28 days, as stated in the programme’s screening standards. Participants felt 28 days was too long to be anxiously waiting for results, furthermore some had been erroneously told “no news is good news” by their health professional. Within the preconception participants it was felt that 28 days was a long waiting time and suggested this was due to inequalities in sickle cell service provision compared to other conditions. It is therefore important for health professionals to explain to parents the technical reasons why newborn screening results might take up to 28 days to become available. Additionally, parents should be provided with the contact details of the health care providers responsible for reporting the newborn bloodspot test results.

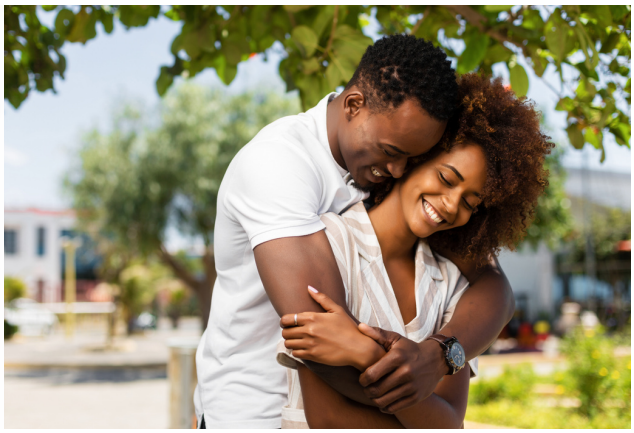
The findings showed some delay and reporting of results outside of the 28-day reporting standard, impacting on follow up care or diagnosis. This appeared to be consistent throughout the country. In the case of thalassaemia diagnosis this delay caused heightened anxiety for the parents and can have long term health implications for the child. Newborn thalassaemia screening is a not a target condition of the newborn screening programme; however, it is anticipated that the majority of cases will be detected. Babies with suspected thalassaemia major must follow the same referral pathway as those with sickle cell disease. The thalassaemia care pathway is subject to the same quality assurance processes.

The “no news is good news” was common to both SCS and the UKTS findings. This phrase being said to reduce worry when results were not received, gave false reassurance, and will form part of the new recommendations gained from this report.

The overall feeling from all the discussion groups underpins the enormous impact a child’s sickle cell or thalassaemia diagnosis can have on parents and the wider family, especially when parents knew little about the condition, or that they carried the gene. Some participants spoke of the psychological effect it had on them, after their child was diagnosed with the condition and felt support was needed in this area. Within the preconception group, some participants identified as having carrier status, felt a need for psychological support after their diagnosis, especially considering that their carrier status could influence future reproductive choices. In addition to this, both sickle cell and thalassaemia conditions tend to carry stigma, which was cited amongst participants in the sickle cell focus groups more than the thalassaemia group. These participants discussed the difficulty of having to tell wider family members that they were carriers or had a child affected with either condition or carried the gene, and that family members should also consider testing.

It is important for healthcare professionals to be aware of family dynamics and the social consequences ofstigma, which may influence decisions. This may require negotiation along the screening pathway from health care professionals to protect these relationships, whilst providing support. However, both the sickle cell and thalassaemia focus groups felt it was important for individuals to learn of their haemoglobin genotype early, suggesting in their teenage years. As such, the NHS SCT screening programme should consider raising awareness of reproductive choices, including young people having access to their newborn blood spot screening results from their mid-teens.

The need for follow-up information and support after diagnosis of the condition or carrier status, was discussed at length, a common theme between the focus groups for both sickle cell and thalassaemia identified those able to access nurse specialists felt better informed and supported.



Participants spoke of the need to develop more trustworthy resources on sickle haemoglobin C disease and resources aimed at children. Parent support groups were thought to be useful for information sharing and meeting other parents. Men from both the sickle cell and thalassaemia focus groups spoke about a need for financial support when caring for a child with sickle cell or thalassaemia. It is therefore important that information on welfare and support services is communicated to new parents who might need financial and other help. Participants all referred to a variety of methods to access information, such as the internet, social media, YouTube, apps, and podcasts, but cautioned the importance of ensuring this information came from reputable sources such as the NHS or Patient organisations. It was suggested that new parents be signposted to reliable NHS and other websites.

The focus group and one-to-one interviews have provided a rich source of service user experiences whilst accessing the screening pathway, highlighting parts that work well and parts requiring some improvement and support. If the NHS SCT screening programme implement the learning and recommendations that have come out of these successful discussions, it will go some way

Introduction

This report is an analysis from the focus group discussions held by the Sickle Cell Society (SCS) and UK Thalassaemia Society (UKTS) between June 2021 and February 2022 and forms part of the work commissioned by the NHS Sickle Cell and Thalassaemia Screening Programme. This document is a 'Conference Edition' prepared for launch on 18 April 2023, at the 'Service user experiences of Sickle cell and Thalassaemia Conference'. The plan being to publish a final report in conjunction with NHS England later.

This is a joint report; however, the feedback sections have been written as separate sections by both Iyamide Thomas (SCS) and Roanna Maharaj (UKTS) and show different styles of writing. The discussion is a combination of both reports to reflect overall service user experiences and recommendations have been formed from both reports for the ongoing development of the NHS SCT screening programme.

Within this report, it is important to note that the term 'woman' or mother' as used in this document encompasses all gender identities and is intended for anyone who is pregnant or has a baby. Similarly, where the term 'parents' is used, this encompasses anyone who has main responsibility for caring for the baby.

Background

Sickle cell disease and thalassaemia are severe genetic blood conditions that can be passed on from parents to children through altered haemoglobin genes. The NHS sickle cell and thalassaemia screening programme (NHS SCT Screening Programme) offers antenatal screening for sickle cell and thalassaemia to identify carriers of unusual haemoglobinopathies by 10 weeks gestation to facilitate early specialist counselling and offer prenatal diagnosis (PND).

The NHS Newborn screening programme offers screening on day 5 of life for 9 rare but serious conditions and includes sickle cell disease. Thalassaemia major is not part of this screening but can be identified through this process.

In 2012, the NHS SCT screening programme produced a 'Protocol for Reporting newborn screening results for sickle cell disease to parents', targeted at health professionals. This publication is due for review and as part of this work, the NHS SCT screening programme asked, the Sickle Cell Society (SCS) and UK Thalassaemia Society (UKTS) to gain service users feedback which would form part of the revised protocol.

Methodology

An evaluation method was used by both the Sickle cell society (SCS) and the UK Thalassaemia society (UKTS) to focus on specific knowledge around newborn screening and to provide knowledge on how results are given to parents. The societies recruited participants via their networks, to form focus groups. Interviews were either face to face, virtual interviews, or a hybrid of both, to utilise uptake, in accordance with COVID restrictions at the time.
Both societies took a qualitative approach using a mix semi-structured interviews and focus groups using a set of previously agreed questions. Interviews were recorded and all feedback was professionally transcribed. The subsequent transcripts were then analysed by each society, themes agreed, and both independently wrote their findings.

Recruitment criteria was set out as, mothers whose babies are up to two years old, and the baby's result are either; sickle cell or thalassaemia carrier. Sickle cell disease or thalassaemia major. Fathers with babies up to two years old, with a sickle cell / thalassaemia carrier or sickle cell disease/thalassaemia. Finally, a group set up for users with no children, thinking about their reproductive choices in the future, some knowing their genotype for sickle cell or thalassaemia and some unaware, (i.e., preconception). Both groups had difficulty in recruiting men despite designing targeted graphics. This giving three groups for both societies. Participants received £20.00 shopping voucher of their choice as a token of appreciation.

Thalassaemia focus groups

1. One focus group comprising of three mothers took place virtually.
2. Two focus groups comprising of eight individuals, all aware of their carrier status, with no children (pre-conception) took place using a hybrid model.
3. 5 Individual interviews

Sickle cell focus groups

1. Focus group comprising of ten mothers all having children with sickle cell.
2. Focus group comprising of five men (three fathers, two preconception)
3. Focus group comprising of five preconception women (three carriers and two with sickle cell)

Upon completion of the interviews, the recordings were sent to a professional company to be transcribed. All participants were then given pseudonyms in order to preserve their identity.

Feedback from Mothers and Fathers

Thalassaemia

The analysis revealed the following four main themes: disclosure of screening results, health literacy, reaction to diagnosis and lessons learned.

Disclosure of Screening Results

Service users reflected on their experiences of receiving their own carrier results and or children's thalassaemia diagnosis. Parents identified several factors which affected their overall experience. The following sub-themes emerged: timeliness of diagnosis, language/ communication, setting/format, and support.

Timeliness: Receipt of antenatal screening results

Many mothers reported receiving antenatal screening and subsequent referral to prenatal diagnostic testing within 13 weeks of their pregnancy. ***"It was between two to three months during the pregnancy when you have the full tests done, after that they came back to say you have beta thalassaemia and then they said well your husband needs to get tested for it – do you know if he has it and I said we didn't. I think it was around the 12-week scan that they did the other tests and offered us more."*** **Farzana (mother)**

Another mother, an employee in the armed forces spent part of her pregnancy overseas in Europe. Though she was aware of her carrier status, she recounted that her husband was not offered testing as part of their pregnancy journey. On return to England, the mother was subsequently offered pre-natal diagnostic testing but explained she declined, due to how late in the pregnancy it was offered. Indicating a different screening pathway within the armed forces but highlighted on return to the UK the screening pathway had been instigated.

Newborn Screening and diagnosis

Parents articulated the need for the results and their children's diagnosis to be reported to them in a timely manner. The group felt that receiving no news, was a sign that their child had not inherited a severe form of thalassaemia and as a result was **"healthy"** and unaffected by the condition.

"***When she was born, they did a heel prick test the next day and then the results didn't come through the post a month later, so I just thought I've not heard anything, so it was fine- like my older child." Saadiya (Mother)***

Many went on to explain that they needed to follow up on their baby's screening results after not hearing back from anyone from the team.

"I had to ask for the results of the heel prick test- the nurse said it was a little bit odd, she would ring. It took a while and it just said I had an appointment to see one of the doctors because my child's haemoglobin was too low… that was the first time I ever knew something was wrong". Isabella (Mother)

Others commented that the delays in their child's diagnosis also affected their ability to cope, manage and accept their children's diagnosis.

"***The length of time it took to diagnose was concerning. It changed our life. We had well over a year of uncertainty, of not knowing if it was thalassaemia… looking back at all of that knowing things could have been better for us as a family, knowing our child had thalassaemia." Virat (Father)***

One mother expressed her disappointment in the NHS services with the delays in diagnosing her child. ***"I guess with us so many mistakes have been made along the way, finding out my child had thalassaemia after such a long time in the UK was not acceptable and was a year too late for me." Aariya (Mother).***

Communication and language

Some parents reported having a "**better understanding**" of the results and feeling "**comfortable**" when health care professionals avoided medical jargon. They felt that this was an important aspect which positively affected their experience of receiving their children's diagnosis.

"She [counsellor] spoke to me in normal language and I understood how serious it was. It was scary but I got what she was saying, your [child] is going to be poorly for the rest of [their] life and need blood transfusions for the rest of [their] life. She gave me lots of leaflets and information and her number. And she did sit with me for quite some time while trying to comfort me and trying to make me understand about the condition and everything." Saadiya (mother)

On the contrary, when medical jargon was used by health care professionals delivering the news, parents reported feeling "**scared**" and "**confused**" and even likened it to sounding like another language to them, "I don't remember what they were saying.

"***They were saying a lot of serious things from their expressions. It felt like another language, I knew it was bad and scary, but I didn't understand. I left there and took the leaflets to my family, and we then looked on the internet. I couldn't make sense of what they said about what I know now is thalassaemia. I just knew it was serious and I was scared." Farzana (mother).***

One mother highlighted the importance of healthcare professionals using direct and simple language to explain the condition and treatment options. ***"We had a call from someone in genetics. She went round and round in circles, she wasn't very straightforward, she was like, oh you have options and I said to her straightaway, if you're thinking about abortion, it's not going to happen." Farzana (mother)***

A clear difference in the parents' experiences was observed depending on whether the diagnosis was communicated by health care professionals who were informed and experienced in thalassaemia than those who were not. Parents reported feeling reassured or relieved when health care professionals were perceived as thorough in their explanation and knowledgeable on the subject. Parents described these characteristics as "**informative**," "**educational**" or "**helpful**."

"They explained it all to me and was helpful answering all the questions I had. I could tell they knew a lot about the condition and that really helped me. I know it was serious, but it wasn't that scary anymore." Isabella (mother)

Conversely when the news was not reported by a confident health care professional, maybe less experienced in thalassaemia, parents recalled having a different experience. ***"But I think the issue at that point was that the midwives and doctors didn't have enough information and I don't think they knew enough to say to us that this could be a problem." Farzana (mother)***

All parents expressed the need for clearness from health care professionals who were delivering the screening or diagnostic news. They felt that if they were given all the information including the poorer outcomes and "**worst case scenarios**" it would have helped them learn more about the condition, what was needed from them and what to expect in the future.

Two fathers went on to explain, the benefits to them and their families with regards to managing their children's condition better. "***Finally, when my child was diagnosed, the doctor put us down the right road. And they didn't shy away from anything. The only way I can describe it is that the thalassaemia Society and the hospital have given us a lot of information and they've been transparent with us from day one. They have reassured us and the whole set up is right for us to deal with my child's condition. We feel we've got a contact at the hospital if we feel things aren't going right or need advice. We've got yourselves [UKTS.]" Virat (Father)***

One father also shared positive feedback with regards to the way health professionals conveyed the news to his family. "***We don't have any complaints about the nurses and doctors. The way they told us they made us understand. It was simple. They give us leaflets and supported us with everything they can. They were friendly and they talked to you normal like a family. They are open and honest and tell you the full story. They are not discouraging but always supportive. They are good people". Ali (Father)***

Relevance of information:

Within this sub-theme there was a clear difference between parental experiences in receiving a diagnosis during the newborn period and if they had received adequate information during pregnancy. With the information given during the maternity period, as opposed too, after their children were born. With regards to learning about thalassaemia and how the condition could affect the baby, some parents who received the news of their carrier status during pregnancy, in some areas, reported not having enough information or understanding the severity of the condition. ***"They told me that I had the trait and that there was a small chance that the baby could be affected. I thought that because I had the trait and was healthy and didn't know, that it would be fine. I think if I understood more about what thalassaemia was, it would have helped me grasp how challenging it was.", Farzana (mother)***

All parents felt that during the course of their pregnancy, the information on the process and risks involved in chronic villus sampling was very detailed and explained to them in a way they could understand." ***I felt I got enough information, they gave me all the pros and cons, they told me there was a risk of miscarriage after the procedure, they told me exactly what they were looking for, and how they were going to do it I felt like I was given enough information to make that decision." Aariya (Mother).***

Most parents felt that their children's newborn blood spot result and subsequent diagnosis of thalassaemia was not well explained. One mother recounted that she was not adequately prepared for unexpected changes to her baby's care and had a lack of understanding of the potential progression of the condition and resulting treatment.

At the first appointment, they said the baby won't need a transfusion for at least 6 months so we would have some time to get our head around it and to see how it was going to be as we had no family around and it was all new to us. We came back to an appointment when my baby was 2 months old and then they were like literally like, let's take the baby's blood and the next day we're going to have a blood transfusion, that's how quickly it was as the fetal haemoglobin was very low and wasn't holding as they had hoped for a few months later." Farzana (mother)

Setting/ Format

Parents reported several ways in which they received their children's diagnosis. This took the form of written documentation i.e. a letter, a telephone call, and in person. Their experiences with regards to receiving the news are described below.

Written documentation.

Most parents who received their child's diagnosis in a letter found the process to be extremely distressing and fear inducing. They explained this format made them feel unsupported and alone after receiving life changing news. ***"It wasn't really appropriate to send a letter to explain something which was life changing for a child and our family." Isabelle (Mother)***.

Another explained that receiving a letter with an appointment was concerning because it took them by surprise and provided limited information on what to expect. ***"I spoke to the midwife when she came to do a couple of visits at home. And I told her I hadn't received the heel prick test and she mentioned that she was going to find out from the hospital. I let a few weeks pass and I got a letter through the post saying that my child had been booked in to see one of the consultants at the hospital. This was the first time I knew something wasn't right. It was literally a shock." Isabella (mother)***

Over the phone

Some parents reported a preference for hearing the results over the phone because they could speak openly with informed and experienced health care professional and ask questions "**directly**".

Not receiving any communication

One mother who was told to expect a letter from the nurse who arranged her child's newborn blood spot test, however, after not receiving a letter and having to contact the nurse again was told the letter was sent only to the hospital. ***"She said she would ring to find out what's happened, or what's the reason why I didn't get anything to explain. But I never received a letter or a call. apparently, the letter was sent to the hospital; it never came to me." Farzana (mother)***

Face to face

Parents who received their child's diagnosis face to face, with experienced and trained healthcare professionals found this method of disclosure to be the most helpful. ***"I remember the door going and inviting her in. She looked really serious, and I was like do you want a cup of tea. And then she told me the news and I just remember I started crying. I just couldn't believe it. She gave me lots of leaflets and information and her number. And she did sit with me for quite some time while trying to comfort me and trying to make me understand about the condition and everything. I think this was the best way for me to find out and I will always be grateful to her." Saabiya (Mother).***

The benefits of an established relationship and continuity of care with their children's health care professionals were described as "**comforting**", "**reassuring**," or "**personable**." **"She was lovely, and she was really comforting and every time I went to the hospital for an appointment, I saw her. She would be with the consultant, and it always made me feel at ease." Saabiya (Mother).**

However, some parents also commented that there was no right or wrong way to disclose life changing news**."There's no right way of giving a result like this. But I think it would have been better if we had all the information about thalassaemia first and then told the result that this... or even when we done the screening this is a possibility, your child might have Thalassaemia and worst-case scenario, but it might not be." Virat (father).**

Whilst one received the news via a letter and subsequently offered an appointment, he felt his past experiences of having multiple children with thalassaemia influenced how he coped. **"Honestly speaking in person or a letter, it doesn't matter because the news is news, no matter how you get it. and because we had an experience before, we were optimistic, and we were praying but it didn't work out. And when we got the result, we just thanked god for blessing us with another baby and that's what we thought, like maybe god doesn't want us to go somewhere else.", Ali (father)**

Support

When health care professionals prioritised assessing the parents' level of understanding or took time to answer questions, parents reported feeling "**reassured**", "**confident**", "**comfortable**" or "**less worried**." This positive experience encouraged them to ask questions and ensured they understood not only the results but where to seek help or advice when they needed it. ***"She gave me lots of leaflets and information and her number. And she did sit with me for quite some time while trying to comfort me and trying to make me understand about the condition and everything." Saadiya (mother)***

However, some parents perceived they were not always supported by health care professionals in their quest for information during the early stages of their children's' diagnosis, ***"And that's when the doctor said he thinks it's thalassaemia and then I grabbed hold of that and made them test him. I knew nothing about thalassaemia, they didn't give me any information, no doctor or nurse gave me any information on thalassaemia whatsoever. All I knew about thalassaemia is what I read on the internet." Aariya (Mother)***

Health Literacy

Within this theme, the parent's prior knowledge about their carrier status, ability to understand their children's diagnosis and the use of information to make decisions about their child's health and manage the condition as well as seek support will be explored.

Within this theme, four sub themes have been identified: knowledge, informing family, friends, and ethnicity/ religion &culture.

Awareness, knowledge of thalassaemia

The majority of the mothers interviewed were aware of their carrier status, prior to the pregnancy. "I knew I had thalassaemia trait before because of my first pregnancy." Saadiya (Mother), conversely most of the partner's carrier status was unknown. **"I knew I was a carrier but what we didn't know that my husband had thalassaemia, especially with his background being Caribbean. Thalassaemia wasn't picked up in my first pregnancy." Isabella (Mother)**

In one instance, a father recalled advising the health care professionals during the antenatal and newborn period that he was a carrier. His wife had not been identified as a carrier during the antenatal screening pathway and therefore he felt that his status was dismissed. His child was subsequently identified as having beta thalassaemia major, due to the mother having a silent mutation. Silent mutations are not always identified through the screening programme. This is a known and recorded limitation of the screening algorithm

.

Finally, some parents explained that their knowledge of thalassaemia was skewed due to them being an asymptomatic thalassaemia carrier. **"So, I knew I had the trait but because I didn't have any issues with the trait growing up and it never affected me personally, it was never something I questioned". Farzana (Mother)**

Declining pre-natal diagnosis screening tests

Many parents reported that they had declined pre-natal diagnosis (PND) testing due to a variety of reasons. One mother explained that she decided to decline PND testing because it was offered to her very late in her pregnancy. **"Late in the pregnancy when I got to the UK after being on military posting abroad, I had a nurse ring me from the hospital to check whether I wanted to do the screening test and I said I wasn't interested in doing it at that stage. Whatever the outcome I would deal with it. We were quite informed when we got back to the UK about all the tests and procedures I needed to do." Isabella (Mother).**

Another reason given for decline was due to risk of miscarriage associated with PND testing. ***"But I didn't want to have the test [chorionic villus sampling] because before I had two miscarriages already, so I didn't want to risk it and whatever the outcome was going to be, I wasn't going to change my mind. If my child had thalassaemia, I wouldn't do anything about it. I would just accept it and deal with it." Saabiya (Mother).***

Sharing the news with family, friends, and the wider community.

On finding out that their child inherited thalassaemia, they described it as a "**challenging**" and an "**emotional**" experience. ***"It was clearly so emotional for you at the time when you find out so I couldn't talk about it for ages. Literally, I couldn't talk about it for ages. I couldn't. Even my husband and I couldn't even talk about it to each other for a little while." Aariya (Mother).***

All parents found that one of the most difficult aspects of this time was sharing the news with family and friends. Influencing factors were identified as; parent's knowledge and understanding of the condition, coping with their emotions, and coping with the emotions of others and the wider community. ***"We came to tell our families it was…they found it difficult to understand probably because we explained it badly. And trying to tell them that it's something we've passed onto our child, it's something that we've given our baby because we carry the gene and I guess when my family members found out it was difficult because I had to deal with their emotions as well as my own and that just made everything so much more difficult." Farzana (Mother).***

All parents commented that relaying the news to their parents, grandparents and to the "older society" was challenging when compared to telling their siblings and peers. **"I think that was more grandparents and my parents and my husband's parents just were very emotional about the situation and got the most upset. It took them a long time to come to terms with it all, but our siblings were all very supportive". Aariya (Mother)**

Parents explained that telling their family about their child's condition was their "duty" as it was a genetic condition that could affect their entire family. ***"I guess that for me, it was important my family knew about it as soon as possible because I have siblings and they could have been affected." Zayn (Father).*** But not all felt that their siblings, whilst supportive, did not understand the true implications and meaning of a genetic condition and the importance of screening.

"I told my sister about it and she hasn't been tested herself and her children haven't been tested either. However, you can only pass on the information. My sister said to me that obviously her children doesn't have thalassaemia and as she is not planning to have any more children, she didn't see the point in testing." Aariya (Mother).

One mother also went on to explain her experience of sharing the news with her in-laws who resided in another country. ***"My husband's family lives in Pakistan and all his siblings are married as well but I don't think they have been tested. I think they are quite ignorant. I don't think they see the importance of it, but I have explained the importance of being tested on several occasions". Saabiya (Mother).***

When sharing the news with the wider community, all parents found this process difficult. They expressed feelings of anxiety and fear in how individuals would perceive the news. ***"It was really hard to tell everybody, actually I've had a baby and the baby has actually got this disorder and nobody at all, even had a clue about it, they were all so shocked, what is it, what's going to happen and obviously telling people that actually it's a lifelong condition, it's not just one or two transfusions here and there and my baby will be fine.. I think a lot of people had that in their head that okay she must have a transfusion, and all will be okay but actually telling them no, this is how it is going to be and explain to people that's its life long and even though my baby looks well, they actually won't be." Farzana (Mother).***

The influence of coronavirus

Most parents interviewed received their news during the pandemic. This greatly inhibited their community engagement due to lockdown measures and social distancing guidelines. ***"Covid stopped us from doing a lot and we really haven't been in the mosque or in the wider community. I was quite fearful being around so many people in close proximity, so we never really told anyone or really spoke a lot about it." Farzana (Mother).***

Ethnicity, Religion and Culture

Parents identified their family origins as being from the Caribbean, India, Pakistan, and Afghanistan and identified as being Muslim, Sikh, Hindu, and Christian.

Whilst religion was not cited as a deterrent in accepting further carrier screening or prenatal diagnosis, parents mentioned leaving the fate of the baby in the hands of God. ***"So, we were optimistic, we were praying, we left it up to God, but it didn't work out. And when we see it, we just thank God for blessing us with another baby and that's what we thought, like maybe this was our God's plan. You know what I mean, to keep praying. So yeah, I mean, we are happy, you know. We have an amazing child and I love my child." Ali (Father).***

When offered the choice of carrying on with the pregnancy or termination following pre-natal diagnosis, one father expressed his and his wife's view on the choices they were given. "**And obviously, we did test to find out if the baby is healthy or not. And then we were given a choice, "You want to keep the baby, or you want to do abortion, it's up to you. So, we came to the house and – I mean, we're Muslim. We don't want to kill a human. It's against our religion. I mean, I don't want to be a part of a murder. So, I just spoke my mind to my wife and said, "Whatever comes in. I know I'm the dad, there's a lot on my shoulders already, but I can take responsibility. Now it's up to you." And then my wife said, "Well, obviously, if you are backing me up, I don't want to do abortion. Ali (Father)**

Reaction to diagnosis

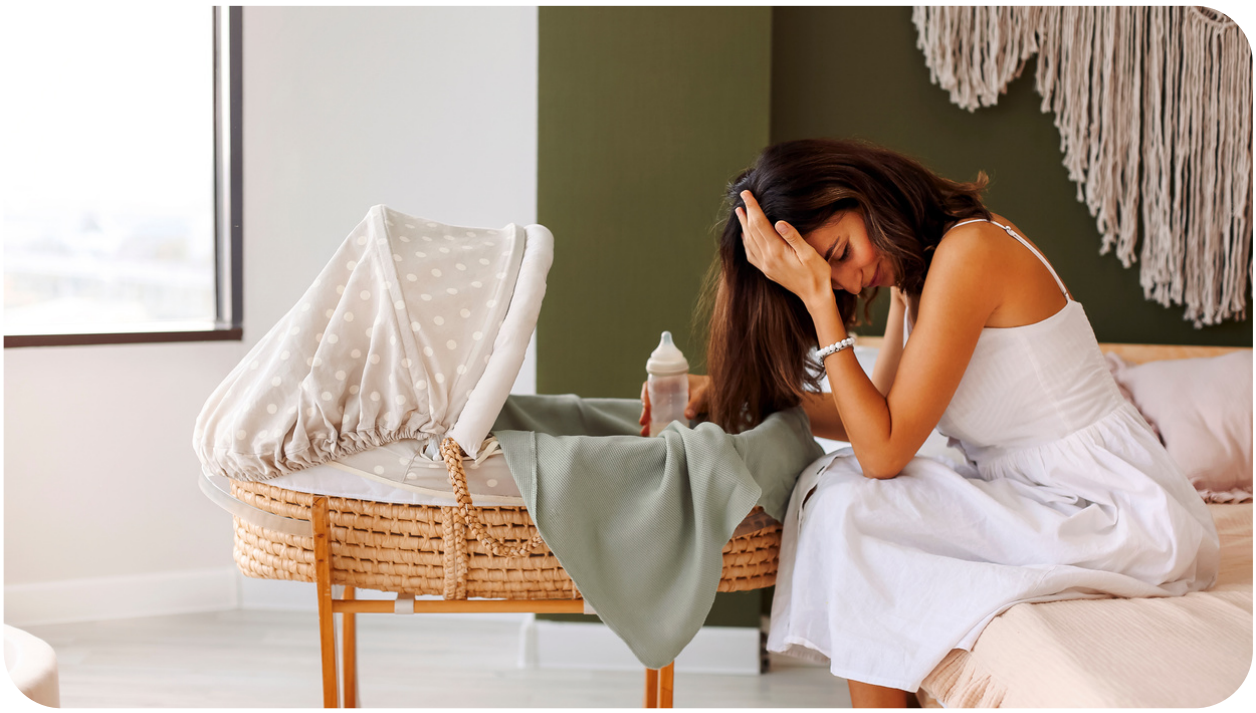
Parents received their children's diagnosis across a range of different ages with earliest diagnosis being made at two months old and the latest at two years old. Despite the differences between their children's diagnostic age, all parents expressed being overwhelmed and devastated with the news. Parental reactions ranged from shock, disbelief, denial, sadness, anger, anxiety, guilt, worry and confusion.

"I was just shocked and confused when she (health care professional) told me…I felt pretty numb to be honest. It was the most distressing news we have ever gotten in our lives." Aariya (Mother).

Many parents also felt responsible for their baby's condition and experience a great deal of guilt and blame. **"I did blame myself and my husband. I was just like oh god we've given it to our baby and we're perfectly healthy and the baby won't be.. but the baby could have been healthy. There were so many emotions". Farzana (Mother)**

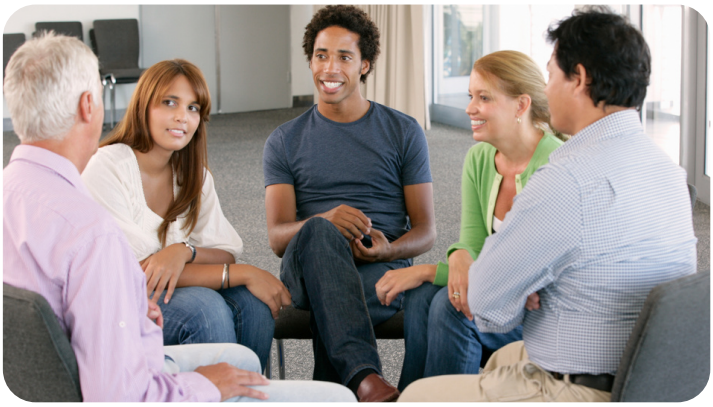
One father expressed a strong feeling of guilt for his child inheriting thalassaemia and wanted to ensure his child had the best possible chance and opportunities in life.

"We feel like we've already punished our child by passing on thalassaemia, so we want to give him the best start possible and like taking him out of school is not viable because obviously we don't want him to fall back at school." Virat (Father).



Thalassaemia treatment can be expensive, and the financial burden can be overwhelming for some families. One father shared his concern and expressed a need for support with regards to the financial burden of caring for a child with thalassaemia, as well as the emotional and physical toll it can have on the family.

"It's quite a lot for a parent to go through, but there is no support, not at all. Because now I'm self-employed, I can't even work like a fulltime job, why? Because if I'm on a fulltime job and I have appointments, my employer, he's going to say like, "Well, find another job. I can't give you like five, six times a month off so that you could do your appointments," you know what I mean, because schedule is schedule. So, self-employed is like low income, because we are family, you know what I mean. So, sometimes you work. Sometimes you sit in the house. Most of the time, you are in appointments. So, there is no support for thalassaemia patients. You know, thalassaemia patients should be considered as disabled – not disabled, I mean, that way. I mean, they should be given support, as they do support disabled people." Ali (Father).



Lessons learned.

This section explores some lessons learned and other feedback from parents who received a thalassaemia diagnosis for their child.

Education is key!

Many parents who received a thalassaemia diagnosis for their child, stressed the need for ongoing education and support from thalassaemia specialist healthcare professionals. They often feel that they lack knowledge about the condition and its management. "The key thing is when you have a diagnosis it's getting the right information and reading leaflets and everything is all good and well but absorbing that information when you're not in the right frame of mind, just been told your child has something and possibly could have something."Farzana (Mother).

Parents also mentioned that they felt the choice of educational materials used was not always helpful. "I would have preferred to have video or something. Reading leaflets is very difficult to absorb that information especially during such a hard time. Most of the time I had leaflets and to be honest I read through it briefly, but most of the time I'd go on the internet. If there's a video, I'd be more inclined to watch that video instead." Isabella (Mother).

One father also felt that his family would have benefited from attending an educational course or training to learn more about the condition and how to manage it appropriately.

"***But it's essential in my point of view that new parents should be given at least training in person, to know how to deal with this, and to give them courage, you know, to explain the whole process and what's waiting for them, what should they expect as well. Because I have seen people, that they sometimes get fed up, you know what I mean, with the transfusions and all the medication giving and things like that, you know what I mean, appointments and things. It's really a lot." Ali (Father).***

Support

Parents stressed the importance having collaborative relationships with hospital teams and the UKTS from early on in their pregnancy and child's diagnosis. Parents reported not being told about the UKTS or not being given any information on how to contact the society. Parents commented that in retrospect they wished they had been given details about the society as they felt it would have aided their diagnostic journey and offered them support during the most challenging time of their lives.

"The first time at the hospital they told us about the UKTS and gave us your details straight away. When we met the professor, he forced me, to put the website in my phone and download it so I could get in touch with you guys. And it was brilliant, and it was the best thing that he could have done for me because I got in touch with Romaine, and we spoke things through. It just made me feel a lot better about my child's condition and it was a massive relief. The hospital was brilliant at making sure we got put in touch with you." Saabiya (Mother)

One mother who received information about the UKTS from a professor at the hospital, highlighted the benefits during their diagnostic journey. ***"I think meeting the UKTS at that early stage when I was told, would have been really good. Because maybe it would have helped my depression and help me to accept this. It should have all linked up because I think this made me become closed off because I found it hard to accept and didn't know where to go." Aariya (mother).***

Additionally, parents spoke about the profound impact of speaking to other families and patients with thalassaemia, had on their perception of the condition. They felt this would have helped them prepare for their new reality. ***"And I just wished we had all the information to hand, and we were told at the beginning, this is what's wrong and this is what's going to happen, and you could plan your life. And you could have emotionally prepared yourself that this is what was going to happen. And maybe we would have come to the Society quicker, got information and looked at other people and stories and exchange. Cause there might be questions, there might be silly questions. And like with the UKTS, there's so many things that you can tell us about your experiences, which you have done on previous conversations with my wife and myself which helped put us at ease. I think this human touch would be so much better earlier in the journey than later." Virat (Father).***

One father also went on to explain that having access to other parents, patients and patient groups would have helped with preparing them on what to expect in the future and help put their mind at ease. ***"But it could also be support I think for the family if they could talk to somebody, my child's doing this, my child did do that... it would be a like for like comparison. As close as you could possibly get it rather than comparing a sick child to a child that's perfectly healthy or..." Zayn (Father)***

Advocating for your child

All parents found advocating for their child during an emotionally challenging time without sufficient knowledge, educational materials, and support, very difficult. One mother, whose child's thalassaemia major diagnosis was not identified as part of the Newborn Screening Programme, described feelings of frustration, worry, sorrow and helplessness when her motherly intuitions were dismissed by attending doctors when her child repeatedly became unwell requiring several hospital admissions without a diagnosis made.

"My child kept being unwell and having fevers that we couldn't control, and I knew something was not right. I also felt like my child wasn't growing compared to other children in that age group. My husband and I visited the GP as well as the hospital and they just said it must be a bug picked up from day care and that my child was probably a fussy eater and I needed to try more. I was made to feel that I wasn't a good mother, and I wasn't taking care of my child like I was supposed to. It left me feeling broken and I didn't know what else to do". Aariya (Mother).

Future Outlook

With time, parents began to process the diagnosis and learn more about their child's condition, they experienced a range of emotions such as determination, hope, and resilience. ***"And to be honest with you, we're living with thalassaemia now, but the thing is we've got a perfectly healthy child and the way we manage this, and this is my understanding and the way I tell myself, we need a blood transfusion and medication to give our child a good quality of life and that we are doing everything to make them a normal happy child. You know, I think it's so much easier to talk about thalassaemia face to face. I'm looking at you now, Roanna and I'm not going to say your age, I can see my child growing to where you are now and it's not as scary and I always remember the conversation we had on National Thalassaemia Day, we were online, and there was a woman there that was 60 years old plus. So that makes me think that my child is going to have a long life that could outlive me". Virat (Father)***

Preconception Group

In the pre-conception focus groups four themes were identified; disclosure of diagnosis, timeliness of result, communication and language used and support available.

Participants identified their family origins as being from Lebanon, Bangladesh, Iraq, Britain, Hong Kong, and Nigeria. Their ages ranged from 24-33 years old, all of whom did not know their carrier status and had no children.

Analysis revealed the following four main themes: disclosure of screening results, health literacy, disclosure of screening results and educational format.

Health Literacy

The group discussion revealed all those born in England, had not heard of thalassaemia or about carrier screening before they saw the advert for the focus group. ***"I had not heard much about thalassaemia before contacting you, but I did a little Google search to find out more, but it's a relatively new topic for me". Sara***

On the other hand, most of those who were born overseas; in regions where thalassaemia was highly prevalent, recalled hearing about thalassaemia at an early age and knew of carrier testing. ***"What I know about thalassaemia is that it is a blood condition that needs blood transfusions. I do not know if its treatable or not, and it is like a Mediterranean condition. I came across this in a TV series as a teen in Hong Kong." Lianda. All individuals expressed that they were not aware of the NHS sickle cell and thalassaemia screening programme. "I didn't know they had testing for women. I knew they tested babies because of my nephew but I didn't know for what." Rayhaan***

Disclosure of a positive newborn screening result

All participants expressed the need to receive the news from a health professional who was either a specialist or knowledgeable about thalassaemia. ***"Probably hearing from a specialist, I think I'd prefer face to face, just because I know I will probably have some follow up questions and also finding out how severe it is and the next steps as well." Rayhaan***

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Others added that they preferred, face to face meetings with a health professional who was knowledgeable about thalassaemia would help them tremendously by answering any questions they would have and putting them at ease. ***"Yeah, I think if you are not really aware of what it is and you have been given news you might panic or feel shocked and it would be better to have face to face interaction from someone who really knows about it, where they can give you information and reassure you about it." Awele.***

The majority of the group expressed a preference of receiving news about their child's condition in person rather than in a letter. ***"Yeah, I don't think a letter would be the best thing for me. That would worry me. I think in person would probably be the best, to find out like what next steps have to be taken and to be able to talk to someone." Hussain.***

One, however went on to explain that she would prefer having a phone call as the first point of contact before she met face to face with a health professional. ***"I think initially via a phone call to arrange an appointment, so I could have time to gather my thoughts and really think about the important questions I needed to ask." Zia***

Like with parents, all pre-conception group expressed the need for health care providers to be as clear as possible with the positives and negatives of the condition. They felt this would give them a balanced viewpoint and advise them on what to expect. ***"I would want to know everything that my child could potentially go through. I think this would help me make a better decision but if they are only saying that they may need blood or whatever, I wouldn't think it was as serious as it really is.. they need to tell the whole truth." Rayhaan***

Accessing health related information

Accessing information, participants expressed a preference for using trusted NHS and Department of Health and Social Care resources either online, or in paper form to learn about health related concerns. ***"The first place I would turn to is the NHS website or app to find out about any health condition, as I know its a trusted place. If I had to learn about thalassaemia, I know a little about what you do, and I've seen your link in the NHS website so I know I can trust the information you provide. I think that's what is important- you can get information from anywhere, but it might not be true." Sophia.***

In comparison, some of the men indicated a preference for videos as a source of educational insight of the condition. ***"I think leaflets are okay, but you can lose it, I think electronic things are better because you can always come back to it. I tend to get bored with only reading things, but videos for me would be the best method of education because it makes the condition and the challenges real and not abstract. I would watch short videos." Rayhaan***

When asked if they would ask relatives and extended family about thalassaemia, as a first port of call, the group all opted to research the condition themselves before asking family. ***"I probably would avoid talking to my family or my partner's family, because I know they can go to unreliable sources when they're looking up medical conditions. I know they're the type that going to give me the wrong information. so, first port of call would probably be some of my friends, who I know think a bit more along the same wavelength as I do, when it comes to medical stuff, or GP appointments". Olivia***

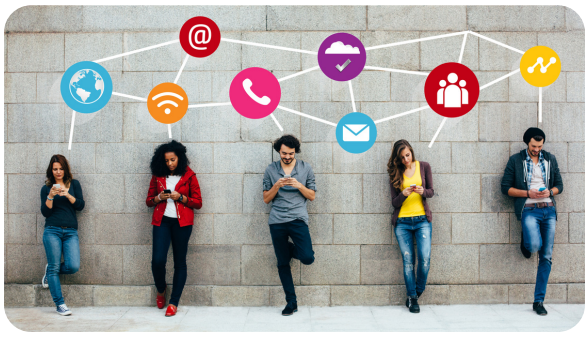
Knowing your screening test

Some participants said that they would like preconception screening or would like to have their newborn screening test result available. ***"I believe screening tests are essential to health outcomes, and knowing what I know now about thalassaemia I will definitely be screened. It is important for everyone to do so and its worrying that people my age and from my culture, don't know about it but it affects us the most.", Zia.***

They also felt strongly about their future children undergoing the newborn blood spot testing. "***Yes, I if I have children, then I would ensure they do the necessary tests to make sure they are okay and receive treatment quickly. I think its beneficial because then you'd sort of know what you're dealing with and the earlier the better." Lianda***

Educating the younger generation using various platforms and offering blood testing

All participants agreed that it would be beneficial if information about thalassaemia, was provided during sexual health education during schooling (16 and above), also an optional blood test to be offered during pastoral care. ***"Yeah, sexual education in schools for ages 15-18, maybe TV programmes or Netflix dramas that have different conditions because I know there is like several conditions that I wouldn't have heard of until I'd seen it on a TV programme. So, I think that would raise a lot of awareness to people that maybe aren't looking online, and just sort of like looking for YouTube videos and stuff like that. And maybe not looking for medical stuff." Olivia***



Social media

Everyone felt that social media played a crucial role in raising awareness of thalassaemia. ***"I think social media is a great way to learn about things, everyone uses social media and if it's made to look appealing or short videos, it captures your attention and makes you think about it." Hussain.***

Learning and Recommendations from thalassaemia focus groups

1. Timeliness of results disclosure, parents need to be given an estimate of how long new-born spot test results could take and who to contact. Parents need to be told that not hearing from the health care provider re: screening results does not mean there is no diagnosis or further test needed.

2. Results should only be given by individuals who are knowledgeable or a specialist in thalassaemia, using simple terms that are clear and concise should be used.

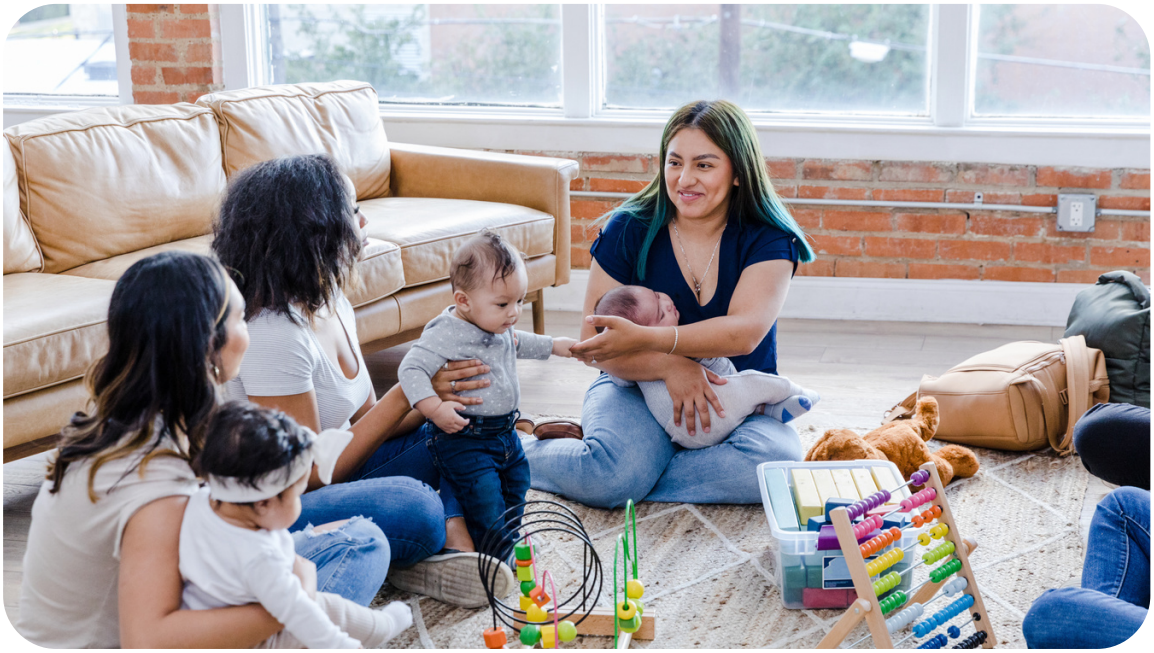
3. A choice of how results should be given with a clear method of support to reassure families and answer questions or concerns.

4. Effective communication with healthcare providers and access to information and support can help parents to develop the necessary coping strategies and to provide the best possible care for their child.

5. There should be a caveat applied to newborn results highlighting the limitations of screening.

6. Parents should be referred to UKTS as soon as possible for support and holistic care.

7. Partnerships with healthcare providers: Parents stressed the importance of developing collaborative relationships with their child's healthcare team. They want to feel like they are a part of the decision-making process and that their voices are listened to. They appreciate being involved in their child's care and management plans.



8. Psychological and emotional support: As with any serious medical condition, parents who receive a thalassaemia diagnosis for their child need access to psychological and emotional support. Parents often experience anxiety, stress and depression, and the provision of psychological and emotional support is crucial.

9.Early screening and diagnosis: Starting medication and treatment at an early stage is crucial to managing thalassaemia symptoms and prolonging the life expectancy of the child. Parents appreciated the importance of early genetic screening and detection of the disorder, as this can improve treatment and quality of life for their child.

10. Addressing financial costs and concerns: Thalassaemia treatment can be expensive, and the financial burden can be overwhelming for some families. Parents appreciate it when healthcare professionals offer guidance and support with the costs associated with thalassaemia management.

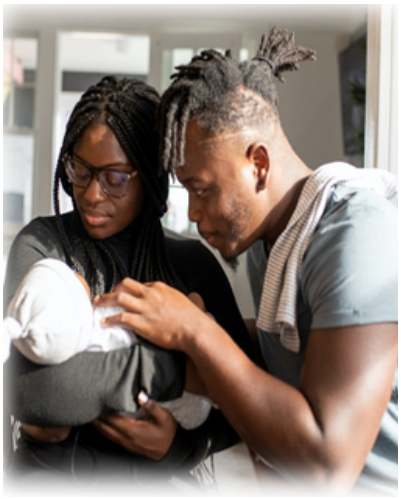
11.In order to address stigma in thalassaemia, increasing public awareness about the condition and its impact is essential, while challenging harmful beliefs and attitudes towards people with thalassaemia.

12. Community outreach: Community outreach programs can be an opportunity to raise awareness about genetic conditions. It can help inform young couples about genetic testing and counselling services they can avail of.

13. Pre-conception counselling: Lastly, pre-conception counselling can help young couples to explore their genetic risks before becoming pregnant. It provides a platform to discuss potential genetic risks, testing options, and get advice and support from medical professionals.

Feedback from Mothers, Fathers and Preconception Men

Ten women participated in the focus group, all having children with sickle cell disease and no carriers. The women identified with African and African Caribbean ethnicities and were located around the country giving a broad geographical spread and insight of screening service delivery.



One of the women had been pregnant during the pandemic and she recalled that this influenced the way her services had been provided, this echoed by another who felt the pandemic had affected her access to services after the birth of her child.
Recruiting men to the focus groups proved more difficult, despite efforts being made to target men to join the project.

Interviews comprised of a combined discussion with three fathers and two men without children. Two of the fathers had children with sickle cell, the third having sickle cell himself, and his children were carriers. The men's group covered areas, across the South East of England. Only one man had a child born in England whilst the children of the other two fathers were born abroad, bringing an interesting perspective to the discussion.

From both the sickle cell groups and the thalassaemia groups, terminology around an individual's genotype was given as either "carrier" or "trait", both are correct and "trait "appears to be used more commonly within the community. To avoid confusion, it should be noted, the NHS Sickle Cell and Thalassaemia (NHS SCT) Screening Programme uses the term 'carrier' to describe someone with just one copy of the sickle cell gene.

The sickle cell facilitator probed further asking the group if they knew what the term 'carrier' meant? ***"I would have thought that carrier meant the same as trait. I've not heard anyone use the term carrier before. I've only ever heard trait. I class trait as somebody who carries a gene for sickle cell." Charlotte.***

Knowledge of Sickle Cell Carrier Status, Diagnosis and Disclosure

This section looked at prior knowledge and understanding of haemoglobin genotype, and how participants felt when they received the diagnosis for their newborn baby.

Various subthemes were explored such as, disclosure, once these results were known, which family members or other people were told, what impact this had on family and any stigma caused.

Two fathers of children with sickle cell explained they knew very little about sickle cell before their child's diagnosis: ***"I've got sickle cell trait, and my son unfortunately has got sickle cell disease. I'd heard about sickle cell, but I didn't know nothing about it. And in 2018, I had a son. And my wife also, she had a trait as well, and she didn't know about it. So, when we had a son, obviously, because our son was born in Senegal, they missed the screening." Hassan.*** Another referring to anaemia being common in Africa: ***"My kid is recently diagnosed with sickle cell, and I want to know about it more. Yeah, we are told anaemia is pretty much common in Africa, so I'm trying to find out more about it." Joshua***

As expected, some knew about the condition: ***"Sadly, I've got sickle cell myself, and I've got two kids that are carriers, so I kind of, you know, understand what some other people are going through, yeah."*** **Olu**.

"***I have sickle cell anaemia. I'm single and I don't have any children stashed anywhere. Well, my parents told me when I was younger. I always hate the cold, still hate the cold. But my parents found out when I was born. Part of the reason why I was a sickly baby. I was born midwinter, a cold winter at that, so they found out then. The blood test – we're talking '62, so obviously the tests weren't as rapid as they are now." Courtney***

When the women were asked if they knew they were carriers, prior to antenatal screening, there were a variety of responses. For some this included perspectives on their partner's status. Some of the women had known they were carriers prior to antenatal screening, whereas others found out their status and that of their partner through antenatal screening.

The women who already knew their haemoglobin genotype cited a variety of ways they had found out. One woman said she and her partner took the test when they were engaged and although both were carriers they had decided to go ahead with the marriage. Another saying she and her husband had both been told (by their parents) they were carriers at a young age: ***"I always knew that I was sickle cell trait. My mum told me when I was quite young. And my husband also knew he had sickle cell trait quite young as well. And then when I was pregnant with my daughter, they recognised his results weren't on the system, so they called us in for him to get tested, and then that was more confirmation of what we already knew." Raissa***

This indicating some couples had accepted testing before conceiving and knew due to their carrier status they were at risk of having a child with sickle cell so made early informed choices.

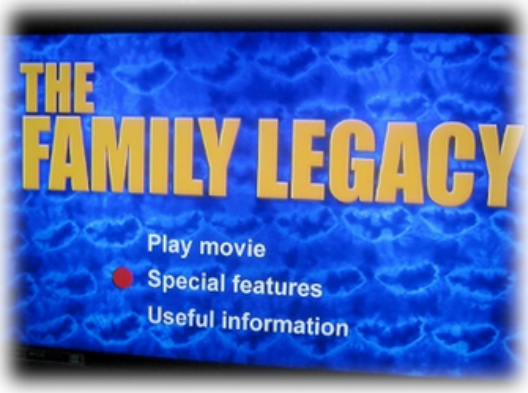
For other couples they only found out their carrier status through antenatal screening, one woman recounting that although she knew about sickle cell and had worked in a field where she dealt with children who had the condition, she never got tested and couldn't believe it when she found out she was a carrier: ***"I actually didn't know I had the trait until the screening, so it was hard for them to explain to me that I had it. I couldn't believe it. Until now I'm not believing it. I knew about it. I knew everything. I was schooled about it. I used to work with an organisation that used to deal with those children. But I myself, uh-uh. My fiancé, he got tested and, you know, he had the trait as well, but he said, "It's okay. All what is important is the life that is coming out." Florence.***

Another explained how she and her partner found out: "***Yes, I have sickle cell trait, and I didn't know my partner has the sickle cell trait until I was about – I'd say maybe about four months, when he has to do – you know, like when they ask a partner to do a blood test. And then that's when we found out that he has a trait as well, so yeah." Laura.***

One service user felt little could be done, but testing was not a problem: ***"I didn't know that I was a trait, so when I was pregnant with my daughter then I knew that I'm a sickle trait, and the dad too. But we were married already, so there was nothing that we can do. But it is so good, she is not – and the second one too is not bad. It's only the third one that has the sickle cell anaemia." Afia***

Being aware they were carriers and testing was not a problem, was sometimes still difficult to accept a diagnosis, ***"I knew I had the sickle cell trait, and I've also got a little sister with the sickle cell trait. My boyfriend said that he's sure that he has gotten it, and then it got confirmed when he had to come and get his blood test done. But I knew it was a possibility, but you just didn't think, "Oh yeah, it's going to happen." Tasha***

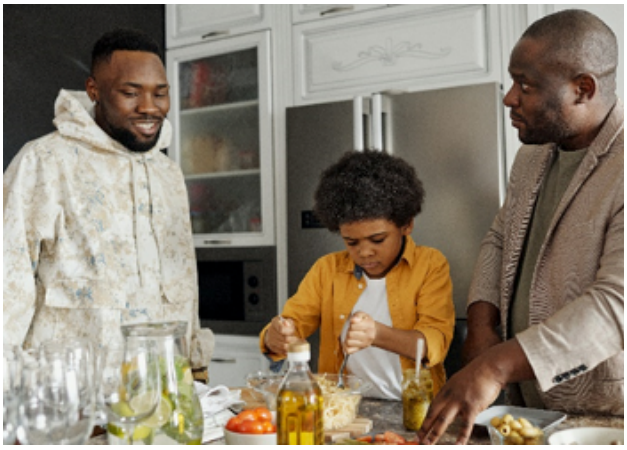
There were also partners who were reluctant to be tested or insisted they were not carriers (when in fact they were). This is a view we came across during outreach and as such was included as a scenario in 'The Family Legacy' DVD produced by the NHS SCT Screening Programme to raise awareness of sickle cell, screening and associated myths: (https://www.sicklecellsociety.org/resource/the-family-legacy/)



"I knew I was a trait through my first son. That's when I found out. That's when all my family found out if they were traits or not, through me. His father insisted that he wasn't a trait. I still insisted he does the blood test and everything, and he said he's not, blah, blah, it was a shock to me when they came to tell me, he (the child) had sickle cell anaemia." Melissa.

Partners were reluctant to be tested: ***"I always knew that I had the sickle cell trait. His dad didn't. He didn't want to go for the tests. I did speak to his mum, and she was adamant that he didn't have the sickle cell trait, so it was a shock when, you know, I found out that my son had sickle cell." Gloria***

One father described how they found out about their child's diagnosis and subsequently their own haemoglobin genotype too, after arriving in the UK.



"After three months my son felt a bit poorly, and then my wife took him to hospital in Gambia, which is the MRC (Medical Research Council). Then they did some blood tests and everything, then they told her this. And then I was like, "What's this all about? How come?" So, we were both shocked you know, to be honest. So, because I live in the UK, I went down to my GP, and I spoke to him about it. I said, "Can you take me to the hospital because I need to get my blood checked?" So, when I went down to the hospital, they did it and they were like, "Well yeah, you have the trait." So, I was like, "What?" And then my wife did the same thing as well. So, when that happened, I didn't believe it. I was like, "No." Because there's nobody in my family or my wife's family has had sickle cell." Hassan

Another father had a similar experience in that he and his partner only found out they were carriers after their daughter started feeling ill. These children were born abroad where no testing is offered, but on arrival in the United Kingdom, now had access to the screening programme. ***"Well, when we had our daughter, we went to the hospital - she was okay for something like nine, ten months. So, when she started experiencing this fever, pale, pale eyes, pain, we decided to go to the hospital. The doctor told us, "You guys should test yourself for anaemia." None of us wanted to do that. They told us, "Your daughter may have anaemia." Everyone was shocked. So, he explained to us how it's a hereditary condition, but he told us, "Don't worry. If you take medicine…" The child was diagnosed at an early age, so he told us, "It can be managed," and everyone was relieved. So, now we are in the process of treatment." Joshua***

Thus showing the benefit of having an NHS Sickle Cell and Thalassaemia antenatal and newborn Screening Programme in the UK, where new children under one year of age are offered screening on arrival into the country.

Both men and women spoke of the challenges they faced communicating results and the possibility of stigma, whilst trying to understand their feelings after receiving their child's diagnoses with the condition or carrier status. ***"At first, for me, we sat something like three hours, not talking to each other. I was thinking and she was thinking like, what's next? Will she abandon me? Will I abandon her? Rejection, yes. So, after a long period of time, there was no option but to just accept and deal with this problem together." Joshua***

The men tended to speak more about stigma and guilt. ***"You do feel a bit guilty as a parent that has got the trait, and then now you have a sickle cell child. I feel pretty guilty sometimes. Had it not been for my wife, obviously, I wouldn't have a child with the disease you know what I mean. So that's why I feel sceptical about having another kid." Hassan.***

Parents of one man knew from birth he had sickle cell disease but still did not tell him when he asked, showing the extent of stigma or guilt felt by some. ***"They found out when I was a baby about sickle cell. Sadly, we are 18, as in my father has got 18 kids, because I'm from a polygamous zone, so yeah, different wives. I'm the only one with sickle cell. That's the sad truth. I remember when I was young, asking my mum, "Why am I always taking medicine every day?" She just said I need it, but at that time I didn't understand." Olu.*** He expressed how he felt stigmatised. ***"Back in Nigeria, I'm not even sure any of my friends know that I've got sickle cell, because even if they know, obviously they don't want to associate with you, because there is this thing, I don't know how to translate it in English, like you know, someone that dies and wakes up something like that. You would want to keep it to yourself, because, you know, there's so much stigmatisation when first people know you've got sickle cell and all that." Olu.*** It must be acknowledged that stigma can be worldwide***. "Even in the western world, yeah, there's still stigma." Om***o

In one case the mother, passed responsibility of telling the child to the father: ***"So, I just knew, when I was like a teenager and I started asking questions, "Why do I feel ill?". I think there was a conversation with my mum that I said, "What's the problem? And she said, "Oh yeah, ask your dad." Omo***

Hassan felt it was lack of awareness that caused stigma. ***"For me in Gambia, I think it's a lack of awareness. Because I told a couple of friends, and even my mum and that, she knows about sickle cell, but still, it's the detailed information, that's what she's missing. I think it's a lack of awareness, to be honest, because I don't feel stigmatised at all. As a father I'd rather be stigmatised than put my son at risk of dying and that's a fact." Hassan.*** So, he made sure people knew of his son's diagnosis. "***For me, I told everybody. The fact I did was for everybody to know how to treat my son as well, not to, you know, leave him running everywhere, making sure he drinks water enough. That's why I told everybody. Sometimes I even feel he has special treatment, because they look after him so well, you know, because they know he's got a condition." Hassan.*** He also urged siblings to get tested. ***"Obviously, when I had my son that had sickle cell disease, I told my sister. I said, "Listen, I don't know whether you know about this or anything, but this is what's going on with my son. If I was you, go and have it checked whatever your status is, before you have a child." And fortunately for her, the children are alright."*** Hassan.

However, he appears to hold some disbelief, ***"My siblings also, they all had kids, but none of them had sickle cell. Well, I don't even know whether they have the trait, because if I believe the trait, I believe they should also. If I had, why didn't my brother have it, some kind of mentality like that, you know what I mean. Or if I had it, why didn't my wife's family have it, something like that."***

"Yeah, but maybe their partners don't, so that's why they haven't got no sickle cell in their kids or anything. That's why we're holding on to have a second child, you know. We just want to make sure what – so, that's another dilemma as well, you see. I don't want to be selfish or careless." Hassan

Others are very cautious to tell family about having sickle cell disease or being a carrier. ***"Yeah, I think for me, I tell the most concerned and the most caring ones, so that they can inform others in a nicer way. You just don't go and tell everyone. The whole family would be scared." They will be frightened, like, "This is a family member, he has anaemia, and the anaemia is inherited." Joshua***.

Telling an employer wasn't such an issue, as it may offer protection or support to hold down a job. ***"I think if I eventually go back to work, I will let my employer know. Because I remember back in Nigeria, I was working with an insurance firm. I didn't let them know. It made me stay out of work for a very long time and at that point I lost my job. But eventually, when I saw my boss, we had a discussion and he said to me that I was at fault, that if I had let my employers know that this was my situation, probably I would have kept that job at that time. But they just thought "Oh, what sickness will keep him out of work for about two months?" Olu***

Those born in the UK however, reported not feeling stigma living with sickle cell and were perplexed that others did. (Caribbean heritage). ***"I was born in Britain, lived in Britain and all the rest of it, but if some people say stigma, I'm missing something here, so why? To me perhaps it's the way I've been brought up. I suppose, because I've known it since I was so young, I really do not understand this whole stigma thing, because it's like, well, you've got the gene, do the best you can for yourself, do the best you can for your child, move on, nothing to see here." Courtney***

The women's group also did not share this concern around stigma especially when the group facilitator mentioned that to dispel stigma during her talks, she always mentions that the sickle cell gene was a mutation to help protect against malaria***. "Going back to what you said about the history of sickle cell, I think for the black community that's important to know, because we just know like, yes, it affects the black community or the BAME community, but I don't really know like, okay, well, why does it affect us." Sheril.*** Another felt having some protection against malaria had benefits***. "I just can't believe – I didn't know that there were any negative connotations associated with it, which I'm quite shocked by. I had no idea. I was always kind of told that it made me stronger against malaria, if you know what I mean, so I was going around thinking I had this armour." Charlotte***

Accepting the child's diagnosis had varying levels of acceptance. One service user with two children both with sickle cell already said: **"*So, 2014, and she's full-blown sickle cell, which – I didn't take it well, even though I knew it would happen. When I got the phone call, I was like, "Oh no". "We went and had another one. When I got the news, I felt like dying, because I'm like, Oh, he's the only boy, I wish that he didn't have it. But I'm looking after them okay. They get their medication regular, every day, on point, no missed appointments, stay up to scratch". Nadia,*** *indicating the cultural importance for some of having a healthy male child still exists.*

Pathway in Screening

The screening pathway itself was discussed, participants discussed thoughts about the antenatal screening invitation (where relevant), the offer of counselling, when it was determined they were an at-risk couple and whether the offer of Prenatal Diagnosis (PND) was accepted or not.

There were varying responses to the antenatal screening invitation, offer of PND and decisions made. Whilst several of the women's partners had accepted the invitation to be tested, one woman had to decline the letter and subsequent phone calls as her partner did not want to go for the test. "***So, when my son's dad didn't want to go for the test, I just had to carry on with the pregnancy, go to the antenatal, you know, classes, knowing that there could possibly be a chance. So, obviously, I received a letter, but there was nothing I could do if his dad didn't want to participate." Gloria. Adding to this Gloria's mother-in-law had insisted her son was not a carrier. This an example of possible fear of stigma by the family.***

The lack of acceptance of screening by a partner can potentially impact the screening pathway affecting the users' journey (e.g., lack of counselling or consideration of PND). This lack of acceptance may be linked to uncomfortable feelings around accepting PND due to the risk of miscarriage. ***"So, for us, I remember – like I say, I always knew that I was trait, and my husband wasn't sure, and I remember, he was abroad at one point doing his work, and they offered to do the one where they put the needle in the belly. I can't remember what it's called again. But then they said, "There's a chance of miscarrying." I just said, "No, I don't want it." And then when he got back, he was then able to do his blood test, and that's when it came back that he was AC. Till then, we still didn't think anything, and then when she was born, she had the heel prick, didn't hear anything, and then we got a letter in the post saying they'd like to come to our house and have an appointment with us. And obviously at that point I knew it was something negative, for them to want to come and have a meeting with us". Betty.***

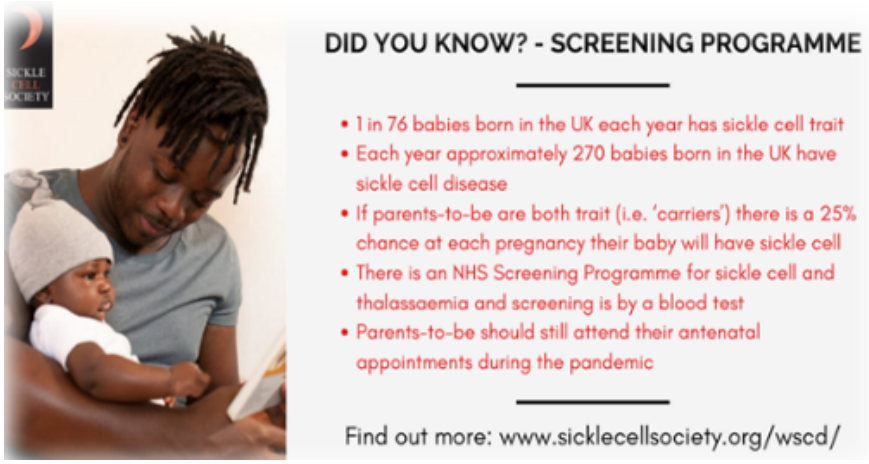
The group felt that there was no pressure around agreeing to a PND and felt able to exercise choices. ***"So, we already knew that he was sickle cell trait, and then we had a letter to say, "Come in and let's test you." So, he got tested, and at that appointment they had a sickle cell community nurse come in, explain to us what sickle cell was all about and explain what could happen, and then they took the blood. So, it wasn't at a centre. It was at another hospital, and they brought somebody in….. Yeah it was well explained. We chose to do the heel prick test and we didn't want to do it with her inside my tummy, but there was no pressure, and no shame if you wanted to do it that way. It was just "Look, these are your options. I'm not going to ask you to make a decision now. This is just for you to be informed." Yeah, and then they sent through the results to the doctors, as in my husband's blood results". Raissa.***

These accounts confirming informed choice is being given and choices supported. Partners also showed support: ***"We've both looked into that as well. But then again, if my wife gets pregnant, for example, now, and then we go for this antenatal screening and then the child in the womb has sickle cell disease, what option would that leave me? My wife to have an abortion? That's hard that would lead to trauma in my wife's head, mentally, because going for an abortion is hard. Why would I put that to my wife, can she even handle that as well? That's another problem as well I'm facing". Hassan***

One service user declined, based on information being sent in the post due to the pandemic. ***"No, I didn't (go for antenatal counselling). This was all during like the pandemic, so during Covid, so they just sent me things through the email". Tasha***

Interestingly this service user who was pregnant during the COVID pandemic, couldn't recall being offered a face-to-face antenatal appointment.

Throughout the pandemic many NHS Sickle Cell and Thalassaemia Centres still encouraged women to come for their antenatal appointments. In fact, during this time, the Sickle Cell Society circulated a message about this on social media, as shown. However, the woman did say she recalled "***speaking to someone at the hospital",*** apart from sending emails.



The men's group were asked about antenatal screening and when testing should be offered. One service user with sickle cell had ensured any potential partner was not a sickle cell carrier: ***"So, growing up, when I was ready to choose a life partner, I was always asking for their genotype. At one point my girlfriend was so sure she was AA. Then she found out she's got the trait. But she wanted us to be together, but I said, no, consider all my experience and all that." Olu. Others were fearful of being tested:***

"No, both of us did not know our traits. So, the doctors were telling us to go and get our blood screened, and we were quite scared. Yeah, there's this phobia, your blood is taken, you are afraid like what they will find, some three or four diseases in your blood. So, it's quite scary and up to now we are trying to see the results." Joshua. After receiving results, concerns grew around telling family members and urging them to go for testing: ***"They are also human beings. I don't know what they are thinking. I don't know how they'll react. So, I'm trying to figure out a nicer way to tell them. Yeah, they don't know. That comes with the story of stigmatisation and everything. I'd rather be conservative first before telling them. I'd rather have control of the situation." Joshua***

The group was asked when the best time to test was and this resulted in a variety of answers. ***"Yeah, the best time for testing is now, because everyone knows that they will have a baby someday, sometime. So, if you are still alive, you just go to the hospital and you get screened. The earlier, the better." Joshua. "Before having a baby. The earlier you know, the earlier you know what to do and what to avoid, and who to date, and who to potentially have a kid with, so the less likely you'd have the disease spreading." Hassan***

Those living with sickle cell agreed and said testing teenagers could be helpful, ***"I agree. It's just a genetic thing and people get more information about genes these days, so if you test them as a teenager, I can't see it doing any harm. For some people, it might actually be helpful." Courtney. Especially since young people are having babies earlier, "Kids having kids, yeah, I think someone should let them know." Hassan. "So, we should test, but for children, we should test them, but we should tell them when they are old enough to accommodate the message. We don't want them growing up with stress."Joshua***

Communication of Newborn Screening Results, Resources Given and Follow-up Support

One of the key purposes of these focus groups was to capture feedback on when and how newborn screening results are being communicated and the support the users received from various health professionals. Users were asked what resources they felt they needed after diagnosis to the first hospital consultation and beyond.

This would allow the parents' perspectives to be fed into the review of the Screening Programme's 'Protocol for Reporting Newborn Screening Results for Sickle Cell Disease to Parents'. Participants were also asked to say what worked well, what did not and how they thought things could be improved. Mothers recounted several examples of good practice after getting their results: ***"Yeah, a heel prick there. And we got the results before we left the hospital actually, which is quite good. However, adding to that, I got a phone call from the sickle cell centre, saying they'd book an appointment when we came home, and also again to come and inform me about sickle cell and things like that, to just say that, am I aware that he has sickle cell. And I say, "Yes, I am aware," and they just kind of went through some bits and bobs with me." Laura***

Further appreciation of the timeliness of follow up after receiving a diagnosis. "Actually, they had to call me like after two months. ***Yes, when I had the phone call from the sickle cell centre, booking me in an appointment, after letting me know the results. No consultant. I didn't see the paediatrician at that time, no. And it was a phone call. So, basically, after receiving the phone call in the morning, then in the evening the sickle cell centre called me to book me in." Florence.*** The speed of the process can help alleviate stress of waiting for a result. ***"Yeah, they came to the house to do the screening. Because of the two I had, there was nothing wrong with them, but this one, I was thinking that maybe definitely this one would be a sickle cell, so I was ready for it, and I got the results after four days". "Yeah, they phoned me. They gave the results to the hospital and the nurse called me, that, "I'm so sorry," this and this. I said, "Oh, it's alright." I knew maybe it will happen, so I'm ready for it, so I wasn't surprised. I was a bit down, but I wasn't surprised." Afia,*** expressing her upset at the result, but not the process.

Positive test results can generate considerable emotional pressures, some will require more support than others. ***"I got the results; I think it was between two to three weeks. Yeah, and like I said, it was a shock, as his father told me he didn't have anything. So, when I got a call to say that the specialist nurse would like to come to talk to me about his results, I was confused and I was thinking, "Oh, what's happened, what's wrong with him?" I even said to my mum, "Oh, maybe if he's got the trait, do they have to come and talk to us about a trait?" So, I was a bit confused, until she turned up later that day and said that he's got sickle cell anaemia, which I was in total shock about." Melissa***

There will always be a variation in delivering results, the group were asked about the timeliness and method used to deliver newborn screening results. Most women reported that results were delivered in person after they had received telephone calls, although a couple of women said their results were given over the phone.

One user reported waiting 2 months and then received the result by telephone, whilst another reported receiving theirs in just over a month. Overall, a variation of a few days to 3-4 weeks was reported. These very different experiences raise concern in relation to the 28 days standard not being met in some cases. Within the same NHS Sickle Cell and Thalassaemia Centre (SCAT) reporting of results varied, implying the service may benefit from some auditing processes or further investigation.

One area of concern was around some health professionals giving the 'no news is good news' message, which can be devastating for women who have held on to this only to be told that their baby does have the condition.

This was the case several years ago when the two Societies consulted women on antenatal screening and produced the 'Parents Stories' in 2017 and it seems this has continued in some areas.

"I think I waited about maybe a month, maybe a month and a bit, but I kept on asking my midwife, "What are the results?" And one of the things that my midwife said to me is that, if there was anything, they would have been in contact with us straight away. And if I don't hear anything, just assume that it's good news.", "I then received a phone call. My daughter was born maybe – was it three and a half weeks before we went into national lockdown, and I received a phone call and a voicemail from the hospital, saying that they would like to come and visit, and discuss the results of the test. And the specialist nurse, I phoned her back, because I was out at the time, and then I literally – I remember dropping my basket in Sainsbury's and just running out. And she was just like, "Where are you?" And I said, "I'm not far." She was willing to come to where I was to calm me down and then take me home then to tell me, but I wasn't that far away from home. I got home, I let her know – in fact, I think I met her outside of my house. She came in, and I could not have asked for a better person to tell me. She was phenomenal. She was so good. She was really good. She gave me literature. She couldn't have broken it to me in a better way. And I was devastated. But her sensitivity and knowledge, and the literature that she gave, and just her empathy, not only just for me but for my husband as well – because she was just like, "Do you want me to wait for him to come back from work? Where is he working? Do you want us to drive there"? She was just superb, absolutely superb." Raissa

This highlights the power of words spoken and reassurance the service user had taken from the health professional, indicating this type of practice needs to change. We see exemplary care when Raissa met the sickle cell nurse specialist.

Feedback suggests that some parents prefer face-to-face meetings, when being given bad news: "***So, I received my results more than a month. In fact, I was told the same as the other lady, that if you don't hear from them, that means – so, I was in shock, because I was really positive, "Oh, my baby is…" I thought, "Oh, thank god," you know. And then this phone call – in fact, I received the news by phone call from the specialist nurse, who told me, "Your baby is SC. So, I just broke down on the phone. I wish I had someone telling me face to face, not over the phone as well." Estelle***. The interviewer asked if she had been given the option of a face-to-face meeting, confirming that she was only given the result on the phone, and this was followed by a letter.

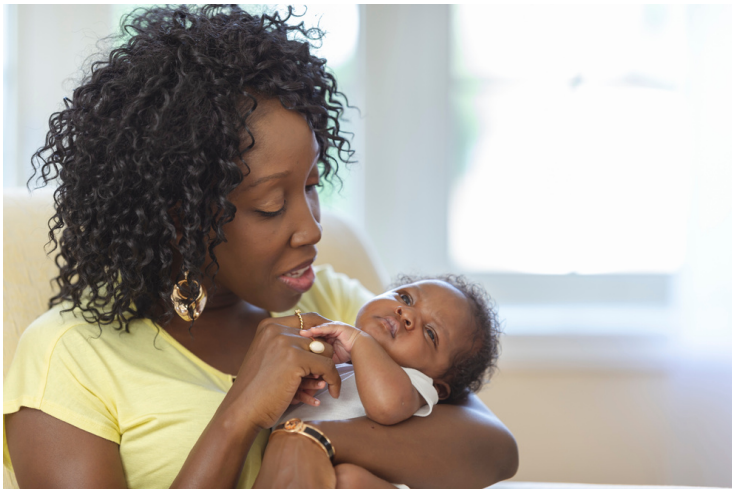
Women were specifically asked which health professionals had given their results and what they thought about communication and follow-up information.

The feedback gave greater insight into the communication of newborn results from sickle cell nurse specialists as compared with other health professionals. Specialist nurses were well informed in comparison to community healthcare providers. Though most women with access to specialist nurses reported good support, not everyone's first experience from a specialist nurse was positive, luckily things improved when a different specialist nurse took over: ***"Yes. I have to be honest; our first initial meeting wasn't very good. Just listening to Raissa there in comparison, it sounded that it went as well as it could. For us, the particular nurse that came – so, we received the letter, then she came to our house, and then she basically told us that our daughter was SC. But then she kind of proceeded to tell us, you know, life expectancies of people with that, and we were just literally, my husband and I – like he must have thought I was crazy – because by then my daughter was like just over a month old and I could tell she was strong, and I was like, "No, that's not my baby." I was just sitting there like, "No, no, she's covered by the blood of Jesus, no." Like I was literally like that, because she was basically telling us that our daughter's going to die early."***

Carrying the conversation,

"***Then she said to us, we're going to have an appointment with the actual paediatrician, that we'd be followed up at the hospital. And then it was actually when we went to the hospital that things started to get a bit more positive. Sorry, I forgot, she did give us some literature, because I said I'd like to read more about it, so she gave us an information booklet, which I started to read through. And then it was our first initial appointment, there was a different nurse from that same centre, and then it was much better, and we're still in touch with her up to now. Maybe it was just that person." Betty***

Many described more positive encounters: ***"The support was good, yeah, because she gave me her number and she told me that I could contact her whenever I needed." Tasha. "Yes. I got a lot of support. I got the nurse specialist to come round. Yeah, so she came round. She was quite good. Then when ("Althea" Nurse Specialist) took over, "Althea" is very, very supportive, you know. I do call them. Any little thing, I call them at the sickle cell centre, because nine out of ten times I can't go to the centres because, you know, I've got other kids. But I get all the support that I need. Any updates, "Althea" will call me. She would let me know. And I had that book, the first book that you just said, I don't remember the name, but I've got that one from birth. I had that one. And then I also have the other one. I have two." Nadia***



Raissa, received a lot of support when the nurse specialist delivered her newborn screening results, however all further support came from the hospital's haematology team and not the Sickle Cell and Thalassaemia Centre (SCaT Centre), possibly due to the pandemic: ***"I know that my experience is probably a very unique experience, but one of the things that has been actually very, very sad is – I know you keep on mentioning centres, and our centre is quite far from where I live, and I've had no contact with them. Because we were on the shielding list at the beginning, they couldn't do any home visits. So, I'm completely reliant on my haematology team. They are the people who have given me all the education, all the support. I think (the SCaT Centre) they've actually been in contact with me two times and that's it, and I think that's really, really sad. And I know it's probably the effects of the pandemic, but I'm like, actually, that service needs to be restored, because we're now – you know, my daughter's one and a half, and I still haven't heard anything from them, and if things were going wrong with the hospital, I wouldn't have any other way to get any other support". "It's not just because of the distance. It's because they haven't been in contact. I've been in contact with them twice and that's it. So, like in terms of my support, there is none coming from them, and all of my support comes from the specialist nurses at the Children's Hospital." Raissa***

An important issue that came up regarding follow-up which was repeated by several women was linked to penicillin. One relating to communication around its use and secondly around GP's prescribing the correct amount. ***"I think one of the things that I find very stressful when I had my son is regarding medication. I think what could have happened is that the Penicillin – when I think about it, I get emotional. My GP – the Penicillin basically – nobody told me that I have to discard it every seven days, because, you know, they give them Penicillin, folic acid. I didn't know that every seven days you need to get rid of what's left and then remake another one. I was stretching it up to a month. Yes, so nobody told me anything about that until I now called my GP and said, "Well, can I have more than one, because I'm a working mum. I work fulltime. I can't be calling every week. Can I have at least two or three?" And they basically refused to give me extra. What the pharmacist said to me, "If your child has sickle cell, you can't be using one for the whole month." So, I said, "Oh." So, nobody had that conversation with me, until I now called ("Althea" nurse specialist) and she said, "Well, that is true. It's supposed to be for seven days, and then what's left over, you throw it away and then you get another bottle."***

Adding ***"I don't know if other parents have that conversation about how the medication process – you know, how long you're supposed to keep it for before throwing the Penicillin away. I think that would have been very helpful for me as a new mum, dealing with a child with sickle cell." Laura. Further confusion and concern, "About the mum (Laura) who has just spoken, about the Penicillin, because when you go for the first appointment with the paediatrician, the Specialist Nurse is always there, and actually they explain to you about how you're going to administer the medication. And they are supposed – like they give you the prescription themselves, because you're supposed to take the baby to your GP, which states they are supposed to give you a supply of a month, letting you know that each bottle is weekly. Those are seven days, then you discard. The concern, like most of the mums, is about giving the babies Penicillin, which is cold from the fridge, like from the fridge to mouth, – what I did is I get the Penicillin, then I put it in the measure container, then I put it in hot water to warm up, because the stomach is still young. But GP is supposed to supply for a month when you pick the medication from the pharmacy, so they missed it." Florence.***

Another stated how stressful it had been liaising with her GP and how it was Specialist Nurses that had come to her rescue, ***"I had an absolutely horrendous time getting the Penicillin from my GP. The letter was sent. The communication was very, very clear, and they would not give me more than one bottle. And then after – so, my consultant said, "Look, she needs a three month supply". I phoned up over five times to explain, "This is not how it's done." After the fifth time, I said, "Forget this. This is becoming too stressful," and I had to inform the specialist nurses before it was put right." Raissa***

Feedback on Resources

Participants were asked whether they felt they had been given enough follow-up information after they had been informed of their newborn results, or if they were signposted to the Sickle Cell Society. Several confirmed they were given enough information, although one expressed there was not enough information given on SC Disease, which is what her child had. Another said she had to supplement the information she was given by going online or talking to others. However, searching online was often found to be distressing and they appreciated that not all online information was appropriate. This latter point about internet information was also expressed in the men's focus group.

"Yeah, I was given maybe three sheets, you know, photocopied, but I wasn't given enough information, so I had to do my own research. I managed to download – you know the handbook. But everything, all the reading, I had to get it for myself. I did not know where to turn to. Obviously, you speak to family and friends, and they try and console you and stuff, but you're in so much turmoil, you're in so much hurt, you're going through so much pain, that there just isn't anything. So, you go on the internet, you start reading things that maybe you shouldn't read. It was tough." Gloria.

"All we got was the booklet at first. And because my daughter is SC, there's only a small section within that booklet about SC specifically. So, maybe have a booklet for SC that's more detailed. So, I had to do a lot of internet, which sometimes is good and sometimes bad, I know, but you feel like you need to get more information. But I think maybe talking to other parents with children with SC might have helped, you know. I know you say each experience is different, but at least to kind of get a gist of what people are going through and how they manage it, and what it actually entails, would have been good." Betty

Service users with access to Specialist Nurses felt well informed and supported: ***"Yes. I received a lot of information when she came to talk to me as well, so I feel that I was very well informed. Because it just hit me, the information seemed quite a lot actually at the time, but yeah, they're very supportive. If anything, you just call them and ask them. There's been times where I was concerned, they will book straight away a blood test appointment and things like that, so they are very good." Melissa.***

Some nurses also offered support with accommodation: ***"I just wanted to say something about how (a city in the Midlands) is treating them. They are doing really well. They are very supportive. The nurse can call about twice in a month to know how the baby is doing. Even when we came home from the hospital, because I did a caesarean, they asked me how I am doing. Even when I'm leaving, they say, okay, they help me through the council and they move me to a new place, where it's warm. So, (City in the Midlands) people, they are doing really well. She called twice in a month, asked how the baby is doing, how everything is going. And they send me leaflets to read about sickle cell. So, that city, they are doing really well." Afia***

The men gave more general feedback about the sort of information they would like on sickle cell and where they would want to find out about sickle cell and screening. They were also asked what information they'd like for fathers who were identified as 'carriers. ***"I think it should be the way everybody's aware of HIV, cancer, things should be more spread out everywhere, so everybody will be aware of it. Then hopefully there might be help, because there's a lack of help as well, more money into the NHS and help people, then, you know, develop a treatment or something. Because there are certain people, especially here in the UK, they don't even know about sickle cell. I've spoken to some of my wife's colleagues– they've just heard about it, but they don't know how it even operates, because it's not their kind of sickness". Hassan.***

Omo gave innovative suggestions on where people might be targeted with health information, this included barbershops, a venue the Sickle Cell Society have conducted successful outreach with the Family Legacy DVD project on behalf of the NHS SCT Screening Programme.

Iyamide Thomas, NHS Engagement Lead, SCS conducts outreach at barbershop in South London

"***Information, it has to be inside where we interact, where there's parties, churches, mosques, those are the places. Because if you put the information out there and say, "Oh yeah, go to your GP," they wouldn't go there. But today's Saturday. Parties are going on everywhere, and those are the places that we can then talk about prostate cancer and things like that, thalassaemia, all these things, yeah, screening. A lot come to the barbershop and people are still - even though I sent them all my podcasts about prostate cancer, they're still like, "Oh what's that?" I'm like, "Are you serious?" Omo***

Joshua had different views on consulting the internet, which brought some laughter to participants, ***"I disagree with that because the internet is not a very good doctor. It will give you more thoughts, more difference, yeah. There is this phrase that says -Never Google your symptoms-"***

Other Follow-up Support

It is pleasing that most women reported seeing a consultant within 90 days, as per standards. Participants were then asked to discuss any further support they felt was needed from the time they were given their child's results (which is often when they are most anxious and contemplating the diagnosis) to seeing a paediatrician or consultant within 90 days. Parent support groups were also discussed as this is offered by some NHS SCT centres, particularly in high prevalent areas.

Some participants also spoke of family support: ***"Well, I live with my grandma, so as soon as I received the call, I told my grandma and I called her dad, my boyfriend, because we knew it was a possibility, but obviously this was just the confirmation of it. My mum was at work at the time, so I told her when she finished work. With my dad's family, my dad's daughter's got sickle cell, so they're familiar with it and they know how to like treat it, so my dad was just advising me on what he's been doing. And my little sister, so I've even spoken to her and asked her like what she does and how she feels. So, when she starts nursery, obviously, the nursery needs to be told, yeah, and I need to be assured that they will know how to like monitor and see it." Tasha***

When the men were asked who they had communicated with to understand and get support about sickle cell, partners were their choice. ***"Yeah, your partner is the best person because you are facing it together, so you encourage each other." Joshua,***

or family: ***"The family is an important source." Omo or a patient's organisation: "Yeah, Sickle Cell Society as well. I did have a look at that as well, yeah. In fact, when I went to the Sickle Cell Society, that's why I started now donating blood. So, now I'm on the blood bank as well." Hassan***

A few service users spoke of the psychological effects the diagnosis had on them, only one had been able to access a clinical psychologist: ***"When I had my eight-week appointment. I was saying, "I know I need some help to get through this. It's been too hard". I was very, very lucky the hospital got me in contact with their clinical psychologist, and I'm assuming, because I only have one daughter, that that service is available for everybody. And if it's not available for everybody with sickle cell then it's something that needs to happen. Because as well as the support groups, they let me know that, "Look, this service is available for you now and throughout your lifetime. If you need it, if your daughter needs it, if your husband needs it, they're there to support you." I feel like that was something that was communicated to me because of the way that I took it, because I specifically said, "I need counselling. It's hit me really hard." I had a hard labour and that was literally just – yeah, that was the final straw for me." Raissa.***

Another expressed possibly needing 'therapy' or counselling after the birth of her child but did not seem to have access to this. Several wanted to talk to other parents, sadly few had been signposted to a local support group: ***"I think for me, maybe some sort of therapy, because there is nobody to talk to. You're given that devastating news. From the moment you're given that news, the person you may have been before is lost, because all you're thinking about is your child's wellbeing, and it feels like at times you can't cope. I just wish that there was some sort of – I don't know, a counsellor that you can speak to, who can help you, give you ways that you can cope, because you're basically left on your own". "You know, I would have maybe been able to speak to other parents, because it is speaking to parents that you're able to empathise with everybody, and you just know that everybody is going through the same thing." Gloria.***

Others agreed speaking to other parents would have helped: "***I think maybe talking to other parents with children with SC might have helped, you know. I know you say each experience is different, but at least to kind of get a gist of what people are going through and how they manage it, and what it actually entails, would have been good." Betty***

The men's group were asked what the most important thing, policy makers need to understand. They felt the financial aspect of caring for a child with sickle cell needs to be considered:

"I think, because our kids already have the condition, I think now it's all about the financial help, because most of us lose work, and most of us – say our sons have it. We go to work, we get a call from their school, we have to leave work, and we have a responsibility in our job, and that's financially impacting on us, unfortunately." Hassan

The men's focus group ended with one participant saying something very pertinent and which will help improve advocacy, service provision and some of the inequalities of care we see with sickle cell**: *"Before I log out, if I could suggest one thing, to please try and get people with sickle cell themselves to ditch the whole stigma thing, because I think, unless people stop with the stigma, it will never improve. I think, until people break through that barrier, that itself is going to be a problem." Courtney***

Feedback from Preconception Women – Sickle Cell

This group consisted of five women, all around 30 years of age, with no children. Three were carriers and two had sickle cell disease. The women identified with African and African Caribbean ethnicities, and all lived in London.

Knowledge of Sickle Cell Carrier Status and Disclosure

Women were asked about their knowledge of sickle cell and how and when they found out they had the condition or were carriers. As two women had the condition and two others had close family with sickle cell, it was not surprising that these women knew their haemoglobin genotype quite early in their life.

"So, I've known that I had a sickle cell trait from a very young age, because my brother has the full sickle cell. So, I grew up seeing him go through his episodes." Amanda. Another knew their carrier at a young age and the implications when choosing a partner: ***"So, my dad has the disease. I think over the last 20 years, he's managed his condition outside of hospital. Growing up, as a child, I knew that I had the trait. So, I think, when I was going to have an operation as a young child, that's when I found out that I had the trait. And I guess, you know, I'm at an age now where this has implications for, you know, relationships and stuff like that." Sheril. "I've got sickle cell trait. Essentially, every single time I go to the doctor's and have a blood test, someone tries to make me aware of the fact that I have got sickle cell trait. I think my parents told me when I was a child. They'd be like, "This is your sickle cell trait card." Charlotte***

This group had no experience of the antenatal screening pathway, so were asked what they understood by the terms 'antenatal' and 'preconception' screening. One description of antenatal screening (inadvertently or unknowingly) included non-invasive prenatal diagnosis (NIPD) a screening test of the future: ***"Yeah, so, antenatal is the lady is actually pregnant and she's going to appointments, and maybe through like a routine blood test, they want to find out if their child is a carrier with the trait, and then they find out that way." Belinda.***

Preconception testing was understood: ***"So, is that when they would test you to see if you have the trait before you're thinking about having children." Sheril***

Participants were asked about the best time for testing people for the sickle cell gene. Most thought this should be when the person was young, agreeing with the men's focus group***. "I would say towards the late teens, and maybe earliest would be mid-teens, so then they have time to know their options and know sort of what they can do, and what is available to them, and maybe what they should look out for." Barbara***

Identifying schools to help: ***"I found it quite useful finding out when I was about five or seven, just because then it became a normal conversation. So then my school learnt more about it, because I would ask my teachers and they would do research and things like that to tell me things." Charlotte.*** Others agreed that carriers as young as six should be told as it can impact on other conditions***: "I agree, in a sense of the younger you know, the better, just so you have more options available for you. And although having the trait, you may not have as severe symptoms as someone with sickle cell disease, you might have other issues that, aligned with the trait, can cause some serious complications. I have a heart condition and having the sickle cell trait has actually made my condition a lot more difficult to manage. My condition being so rare, they don't necessarily know how having the trait fully affects it or having sickle cell disease could affect it. So, now I have to think about having the sickle cell trait, having the heart condition that is also a hereditary issue, and it can be very intense to think about all of those things. But that's why I think it's so important that, from a young age, children and families should be educated about it and how they can navigate with the trait." Amanda.***

Teenagers should have more education of haemoglobin genotype**: *"I think I agree with everybody else, that you should know from when you're young, because I always knew growing up, okay, yes, I'm a trait, and I just thought, okay, well, that's going to have implications, you know, if I want to have children. Very recently, I guess it's something that I've had to sort of really face, you know. I think it's important to know quite early on, and maybe there could be more education during like your teens, as a woman, like what does that mean for me. And I think also just like the psychological aspect of that as well." Sheril.***

In 2012 the Sickle Cell Society collaborated with sociologists from University of York on a research project "Living with sickle cell or beta thalassaemia trait', which explored the experiences of individuals who are 'at risk' of having /or have been identified as a sickle cell or thalassaemia carrier; the impact it has on their life-choices; and relationships and the level of support available across statutory and voluntary sectors. https://www.sicklecellsociety.org/living-with-sickle-cell-or-beta-thallasaemia-trait-leaflet/.

Service users with sickle cell disease were asked about their thoughts on relationships, including meeting someone who was a carrier. **"*So yeah, as someone with sickle cell – my sister also has sickle cell, and we have this little running joke that, whenever you meet somebody, you introduce yourself, you're like, "Hi, my name is…" And then you're like, "Do you know your genotype?" That's the second question you ask instantly. You don't go any further until you ask that question because it's not going to go anywhere, honestly. I'm 30 as well, and thinking about having children, it's interesting in the sense that I can't just date anybody. I don't have that luxury, like a lot of people do, just dating anyone, marrying anyone, just having children with anyone. I have to think a bit more in terms of, I don't want to pass this disease onto my child. So yeah, it's definitely there at the forefront of everything." Belinda.***

"***So, okay, when it comes to the whole relationships, dating thing, I'm single, never dated, all that stuff, but in the future, that is exactly what I'll say, you know, "Hi, I'm so and so, I have sickle cell. Can we go to the hospital so we can get you tested before we can start this whole thing? Don't worry, I'll pay for the taxis and stuff." Barbara.***

The reality of living with sickle cell disease is not always understood**: "*You know, people can say, "Oh, you're inspirational," all they want, but the reality is that unfortunately a lot of people are in hospital a lot of the time, and so they need to realise that "Hey, can I handle this? Am I equipped to deal with this?" And at that point it's not about love anymore." Barbara***

All the women felt preconception testing was a good thing and gave suggestions of where in the community this could take place. ***"Maybe the church or other places of worship, particularly where the congregation is predominantly black." Sheril.***

An interesting venue suggestion was given***: "I go to a lot of tech conferences, and when they can, they do testing for like bone marrow and things like that, to find out people's blood types for bone marrow, but I've never seen it where there's a stand about sickle cell. So, I think that's another space where – it's like a black tech conference, so you've got a lot of people there who are focused on technology or whatever it could be, but then also have a testing booth. I don't know if you can do it with a prick, but they do it with pricks for blood types, so that could be quite a useful space as well." Charlotte***

When asked which health professionals should be told about their carrier status Charlotte mentioned how being a carrier had given her symptoms. Most people with sickle cell trait do not have any health problems caused by sickle cell trait. However, because a certain percentage of sickling does occur, there are a few, rare health problems that may potentially be related to being a carrier. E.g. Some people may experience pain when traveling to or exercising at high altitudes. This sickle cell trait toolkit will increase your understanding of sickle cell trait and some of its associated symptoms: https://www.cdc.gov/ncbddd/sicklecell/toolkit.html

"I often tell doctors sometimes if they don't already know. So, I was having quite a lot of joint problems for a while, and I'd mentioned the fact that I was sickle cell trait to one doctor, who used to do a lot of work in Nigeria, who then said, "Oh, your sickle cell trait could actually be making that worse for you." And it was only when I'd mentioned sickle cell trait to a Nigerian doctor that they made that sort of link. So, for me, it was really useful. And then he gave me guidance that actually helped, rather than just physio, which didn't do anything for it. So, it was actually quite useful telling my GP and other people in that sort of sense for other things that you might just not be aware of. But that was only after I'd done quite a lot of research online to know that there could be a link, to then mention it to somebody who could help." Charlotte

Feedback on Communication of Newborn Screening Results, Resources Given and Follow-up Support

Although some had no experience of the antenatal screening pathway, many were aware of the NHS Sickle Cell and Thalassaemia Screening Programme, particularly those living near an NHS Sickle Cell and Thalassaemia Centre. Like the mothers' they felt that results should be given face to face, including carrier results.

"Obviously, I don't know how anything is ever communicated about anything when you have a child, but I feel like it should be communicated in the same way as any other like condition or thing that you're carrying. So, I would assume that that would be face to face, especially if there are any elements of, "You'll need to do this to make sure that your child stays healthy," that sort of thing. I'd expect that to be quite close communication, where the doctor's sitting you down and talking you through it, or the nurse or whoever. But I'd expect that to be done in a way where you can ask questions if you need to. They'll be informed of what they need to tell you as well, and they'll understand it and know what that could actually relate to. Because yes, you know, a few of us were given a card when we were young, but that was it. Then the internet came and then we could Google it. But before that, there was very limited information that we had on it. So, parents should have the information when they have a child." Charlotte.

Others questioned possible inequalities that might arise, **"***I think it should be told just after you've given birth, when they do the tests to see what could potentially be wrong with the child. If they have the trait, they should just let them know. As we know, sickle cell predominantly affects black African and Caribbean people, and I feel like if we're not given that opportunity to have conversations face to face and show empathy, it can quickly become something that's disregarded, because we're not the majority in this population. So, we should be given that opportunity to have face to face conversations. I don't think it needs to be as intensive a conversation as if your child has the sickle cell disease, but even if it's like a, "Hi, we've noticed that your child has the trait. They have iron deficiency," or whatever. So, the same way they let you know after you've given birth like what the child could potentially have, then is when they should be told whether they have the trait or not." Amanda.*

Several of the women thought parents should not have to wait that long: ***"So, I don't think it's a good thing that health professionals would allow parents to wait that long. As said, they can be worrying. But it's not just the worry. They need to know what they can do and how they can go forward with their lives and with their new child's life. And they need to know the options and know what's available to them, and how best they can help their child. Even if it's with trait or sickle cell disease, it's still something that affects the child. Even if it might not affect them to the extent of like sickle cell disease, it's still something that they need to know." Barbara.***

Those living with sickle cell agreed and thought that parents should meet the haematologist earlier than three months: ***"I was going to mention about the fact that the symptoms don't usually start showing until after six months. But also, I think 28 days, it's kind of long as well. I feel like meeting a consultant should be a bit earlier, because that consultant's going to be in the child's life for the rest of their life if they have sickle cell disease, as long as the consultant's working there their whole career, and I think it's important to get to know them, build a rapport, especially if you're going to be living in the same area for many years to come. From my experience, I've had the same haematologist since birth. So, it's really good to get to know them as early as possible, the parents especially, because until you get to like teenage years and you get moved to adult care, it's usually a parent that's always with the child in hospital and appointments and things like that. So, meeting the haematologist I think should be a bit earlier than three months." Belinda.***

Some felt possible inequalities: ***"Whether or not you have the trait, sometimes you might have to re-evaluate how you raise your child, things that you do, and you wouldn't necessarily wait 28 days to disclose a different hereditary element, so I don't understand why that for sickle cell, you know, they can wait up to 28 days. Again, maybe because it's something that doesn't affect the majority of the population and rather just a small minority. But I think when they disclose the information of whatever a child is dealing with, sickle cell trait should be high in that priority as well." Amanda.***

Overall, the group felt waiting times associated with screening was too long, this having potential to generate inequalities when compared to other conditions.

Feedback on Resources and follow-up support

The group were asked to feedback on the support and resources which should be available to new parents, all agreeing that support groups offer a good source of potential support: ***"So, it was about like what support they would need. So, I think definitely a support group. I think they should be informed of what's out there, you know, and hearing from other parents as well. So, that emotional and th*****at psychological support, and what does this mean, and how can I cope with this, etc." Sheril.**

One service user's mother had attended a support group, showing a well tried and trusted model: ***"Places to get information from, thinking about when I was younger, we had a really good sickle cell centre in our borough, so my mum used to go to meetings on Saturdays and meet other parents with children with sickle cell. And also, those that are much older – so, it's nice to see like a range – because if you're a brand new parent of a child with sickle cell, you'd have no idea what you're doing, especially if you had no experience. So, it's nice to meet parents of children who are like teenagers or even adults to give you like little hints and tips and things on how to basically raise your child or help them through crises, and ways to avoid crises and all of this." Belinda***

All participants agreed on the importance of providing psychological support and counselling to new parents, irrespective of whether their child carried the trait or had the disease: ***"Okay, so, I do think that psychological help, whether it is the trait or the disease, would be of help to parents, because – for example – okay, I'm going to use the word normal, even though we don't know what that means. But like normal healthy parents, and then you've been thrust into this situation whereby your child has this illness or has the gene or the trait for this illness, and you genuinely do not know what is going to happen, that can freak a lot of people out, I would say. And if it does come about that the child has sickle cell disease, sometimes the parents feel really guilty, genuinely guilty." Barbara***

This further highlighted the importance of developing resources for children. Being told at a young age they were a carrier they spoke about the importance of developing resources for children too: ***"I was thinking that there could be, or maybe used to be at least, some sort of resource book as well. I'm sure that when I was young – because you mentioned the 25 percent and 50 percent, I'm sure I remember seeing that in a book that I then explained to my teachers, and I explained to them what sickle cells were when I was quite young. And it must have been in a very visual way for me to understand that. So, even something for the child to understand alongside the parents having that support network, but the child understanding what it actually means. Because definitely for when you're a lot younger and in a population like the UK, you have to explain it over and over and over again to almost everybody you ever meet. So, I think having a pack that's almost for the parents and then a resource pack for the child, so they've got something to go by as well." Charlotte***

This group were then asked where they looked for health information, reporting a variety of sources including the worldwide web like those from the other focus group discussions. They did however, express reservations about information available on some websites and reflected on how people may access information online:

"I tend to use the internet, but I guess I always want it to be on an official site. So, NHS, WebMD, even Sickle Cell Society, like if there's resources there, that I know it's backed up. So, I might see something on social media or see something on WhatsApp or wherever I see it, and then I would probably then Google to make sure that it's on a legitimate site, to know that it's true. But with sickle cell, I feel like I do read articles that aren't backed up necessarily by the NHS, because they have less information. So, I probably read more random articles than I would for other types of ailments". "I think it's hard to know – with the UK, obviously, we know which sites are to do with the health service, whereas in America I don't know if it's just a random person or if it's backed up by something. So, even if there was a list of official sites that you could direct people to or something like that." Charlotte

"Yeah, I think apps are a good idea, however sometimes I find that it's not really updated as often as it needs to be. For me, I follow a lot of sickle cell pages on Instagram or Facebook, where – I feel like, as you're browsing through your general social media, you can just come across a nice article, click the link and then have a read about any new research or any developments." Belinda.

"I know there's a lot of Facebook groups. I know Facebook isn't used as much, but I think the groups are, because people connect with each other and share – it's almost like an online support group. And the same with Reddit, people use like Sub-Reddits to talk about different ailments. With reference to the app idea, the NHS app, now that everybody has it, it could be a place where you've just got like a thing that says, "Sickle cell trait," or, "If you have this, these are some resources you could go to." It could be quite an easy way to add additional health information." Charlotte

This participant had looked for sickle cell information on video platforms:***"I've looked through like Tik-Tok to see if there's anything about sickle cell, but there's nothing there. It's just more about chronic illness in general, like people with just different chronic illnesses explaining what they have and how it affects them, but nothing on sickle cell. There's stuff on YouTube about sickle cell about people's experiences and stuff. But I think it would be beneficial if – I don't know if the Sickle Cell Society have an app, but I think that could be something that could be done, and then like if you have events or something, it could alert you. I think that would be a good thing." Barbara***

Like the men's focus group discussions, the women mentioned podcasts as a possible and valuable way to access health information: ***"So, on Spotify, very recently I listened to some podcasts about sickle cell. I can't remember the names of the podcasts off the top of my head, but I have listened to some very recently." Sheril.***

Podcasts might be a good way to reach men in particular: ***"I do think, you know, there should be a little bit of a difference in the way that we approach speaking to men about sickle cell or about getting tested or whatever, just for the simple fact that they don't really get checked much for anything. So, to now ask them to just check and see, oh, you might have the trait, I don't know, it seems like a hassle to them, to be brutally honest. So, I think maybe something like podcasts could help them a little bit more, so then it's not so daunting." Barbara***

Organisations no longer provide large quantities of printed resources preferring to rely on online information, with this one suggestion: ***"So, whilst I know that people don't really use them that much anymore, I know that when I've run events before, I've been able to, from different charities, order like leaflet packs that I can then distribute at my events. So, that could be a way of distributing within the community, if people that run different networks or communities are able to then order them to distribute to other people." Charlotte***

Learning and Recommendations from sickle cell focus groups

• It is important for healthcare professionals to be aware of family dynamics and the social consequences of stigma that might influence decisions and negotiations that might have to be made along the screening pathway and also impair healthcare interactions.

• NHSSCT Screening Programme should consider raising awareness of preconception testing among young people from their mid-teens.

• Health Professionals should on no account be telling the parents 'no news is good news' whilst they are waiting for their child's newborn screening results.

• Newborn results should whenever possible be delivered before 28 days as participants felt this was too long and is an inequality in service delivery

• Health professionals should deliver positive newborn results face-to-face, with sensitivity and knowledge to answer parents' questions.

• Parents need info on sickle cell treatments such as hydroxyurea and exchange blood transfusions.
• Where possible new parents should be given access to a Clinical Psychologist and /or Counsellor after being given their child's positive newborn results.

• Specialist sickle cell nurses appeared to relate better to service users and this difference with non-specialists could be addressed with more training.

• Communication to new parents, GPs and pharmacies on the prescribing and use of Penicillin for sickle cell is needed.

• Parents should be provided with a list of reliable NHS and other websites such as the Patient Societies as sources of information.

• More trustworthy information is needed on SC Disease.

• Podcasts could be used to give health messages.

• The NHS SCT Screening Programme should signpost people to the comprehensive sickle cell information on the NHS app.

The Sickle Cell Society should look into developing a sickle cell app with information and alerts for forthcoming events etc.

• Parents should be signposted to support groups some of which are nurse-led and /or the Sickle Cell Society when their child has been diagnosed with sickle cell.

• The updated Protocol should contain information on welfare and support services for new parents who might need financial and other help when looking after their children with sickle cell.

• Need to develop resources for children with sickle cell disease or the carrier status.

• More research needed to investigate and develop resources and support for parents in the first year of their child's positive diagnosis.

• NHS SCT Screening Programme should consider a variety of ways to disseminate information such as social media, apps, and podcasts. Leaflets can still be useful especially for people who don't access the internet.

Discussion (Joint)

The findings from the focus groups underpinned the enormous impact a haemoglobinopathy diagnosis had on parents and wider family. This project undertaken by SCS and UKTS gave great insight of the experiences of service users with regards to the newborn screening pathway and the NHS sickle cell and thalassaemia screening programme. Whilst, service users recalled many aspects of service improvement and good practice, this project also undercovered the areas in which there is a need for further development.

The findings highlighted the significant progress, made to the antenatal screening care pathway since the publication of the original “Parent stories” in 2017. Screening of couples for sickle cell and thalassaemia, were well established to allow a timely offer of pre-natal diagnosis. Some users were aware of their carrier status, prior to antenatal screening, whereas others only found out their genotype or that of their partner through antenatal screening. Many women expressed disbelief around partners who had refused to be tested or insisted they were not carriers when in fact they were. Overall, women felt they were well supported and given choice during this part of their pregnancy journey.

However, it is important to note that screening tests will not detect 100% of conditions or carrier states. Screening typically acts as a sieve, classifying individuals into higher or low chance groups. Those who are classified as having a ‘screen positive’ result often require further investigation, however, there can be limitations of screening. Screening for thalassaemia may not detect gene variations which are known as silent mutations. This is where the screening results appear entirely normal, but there is a need for a genetic test to identify a carrier state. These types of gene variations are very rare. Participants in the focus groups and interviews did not always understand the limitations of screening and the differences between screening tests and diagnostic tests.

To improve the service, the programme should formalise the reporting and delivery of care for babies and families who present with a haemoglobin abnormality which is a classic marker for a thalassaemia diagnosis. All parents should be made aware of the estimated time the new-born screening results may take to be reported. Additionally, parents should be provided with the contact details of the health care providers responsible for reporting the new-born spot test results.

Some of the feedback obtained from parents took place in the amidst of the coronavirus (covid19) pandemic. The covid19 pandemic caused significant changes to healthcare systems during its entirety, the most notable being lockdown and social distancing, causing great disruption to services. These were exceptional times and some service users commented on the implications of this, highlighting the effects of how lockdown inhibited community engagement for parents with recently diagnosed children. New parents especially those who received their children’s diagnosis with thalassaemia, found it a very isolating experience. With restrictions in place, parents felt more alone as they struggled to understand and adapt to their child's diagnosis. Expectant parents were unable or chose not to attend face to face meetings with their health care professionals or interact with their family, friends, and wider community due to the risks of the virus, which in turn left them feeling unprepared and unsupported.

Though the NHS sickle cell and thalassaemia (SCaT) centres remained open during the pandemic, many of the usual services were reduced or worked differently, leading to delays or gaps in care. This in turn meant families faced longer waiting times for appointments and missed out on opportunities to connect with others, obtain information and support, and crucially establishing relationships with their healthcare professionals.

Stigma remained a prominent topic across both groups. In our experience, this can manifest in a number of ways, including discrimination, negative attitudes, and a lack of understanding. One of the most common forms of stigma is related to misconceptions and beliefs about the conditions. for instance, some people may wrongfully view thalassaemia as a being a contagious or shameful condition. Not only can this lead some individuals living with thalassaemia to feel isolated but it correlates with having low self-esteem, anxiety and depression. The lack of knowledge and understanding of thalassaemia can contribute to the stigma surrounding it and there are many misconceptions which can lead to prejudice and discrimination towards those living with the condition. In order to address stigma in thalassaemia, it is important to increase public awareness about the condition and its impact, while challenging harmful beliefs and attitudes towards people with thalassaemia. Education and awareness campaigns can help to break down stereotypes and promote understanding of the condition.

Within the sickle cell group participants discussed stigma in terms of having to tell wider family members that they or their child had the full condition or were carriers and that these family members should also get tested. It is important for healthcare professionals to be aware of family dynamics and the social consequences of stigma that might influence decisions and negotiations that might have to be made along the screening pathway and impair healthcare interactions.

This point exemplified by one woman whose mother-in-law had insisted her son was not a carrier. Her husband had ignored the invitation for testing, so she had continued with her antenatal classes and navigation of the screening pathway on her own. The men spoke a lot about stigma, also suggesting it varied depending on which country one lived or had grown up in. There are myths about sickle cell disease in some African and Caribbean countries, and this could partly explain why. However, other participants did not think there was stigma associated with sickle cell disease. Interestingly, this was voiced from participants who were born and grew up in the UK, indicating some minor shift around stigma. All participants agreed that stigma was caused by lack of awareness. Within the sickle cell disease, participants appreciated learning that the sickle cell gene arose from a mutation to help protect against malaria. However, stigma was not an issue when it came to informing educational, employment or other relevant institutions about an individual's sickle cell or thalassaemia status, to ensure necessary support or aids could be put in place if needed.

The receiving of newborn blood results was an area of concern for many parents, parents reporting waiting over a month for their results, causing anxiety and concern which had impacted on their newborn experience. Unfortunately, some healthcare professionals continue to use the 'no news is good news' message to offer reassurance, this being reported in the 2017 ***'Parents Stories'***, and requires addressing as it gives the parents false hope and can be very upsetting when they subsequently receive their child's positive diagnosis. A robust pathway, with realistic expectations and communication channels, needs to be established to ensure a smooth transition. At the time of offer for newborn screening, parents could be made aware of the timescales involved in reporting and the communication of results. Contact details could be provided giving a point of contact, more information around the complexity of testing could be provided to help understanding and ease anxiety. It is therefore important for health professionals to explain to parents the technical reasons why newborn screening results might take as long as 28 days.

It is clear from both focus groups the enormous impact a diagnosis has on parents and wider family. Parents reported feelings of disbelief, guilt and for some a sense of loss for the life they had imagined for their child and worried about the impact of the condition would have on their child's health and quality of life. Parents spoke with, strong emotional recall about being faced with the disclosure of either a sickle cell or thalassaemia diagnosis. This highlighted the importance of the first contact with health care professionals, specialist nurses, and their realisation that their world as they once knew it had been changed forever in that moment. Similar to findings in 2017, women generally reported better experiences when they had access to sickle cell nurse specialists who were well informed in comparison to community midwives or other health professionals. Specialist nurses are usually based at the NHS SCaT centres, and not everyone has direct access to these. Haemoglobinopathy services are distributed across all regions with allocated specialist input from specialist teams. The introduction of haemoglobinopathy coordinating centres has enhanced support to centres in lower prevalence areas. One user reporting that as there was a specialist nurse in another area and they now travelled attended that hospital there as it was not too far away and offered more support.

Parents felt they had a positive experience when they felt their health care professionals had considerable knowledge and confidence in delivering the information. Receiving news from a healthcare professional who was not confident in their knowledge, was found to have a negative and detrimental physical and psychosocial wellbeing on the parents
.
The use of positive and informed dialogue in non-medical jargon between parents and their health care professionals was instrumental in forming trust, successful education and developing healthy relationships. This not only alleviated some of the anxiety and worry by creating a safe, supportive space but also influenced the way parents' developed methods of coping with their future.

Within the thalassaemia group, parents felt an increased level of distress when one parent received the possible diagnosis alone. Health care professionals should emphasise to benefits of bringing someone for support or sharing this information with family and friends once they have had time to process their own emotions and thoughts and have a clear understanding of the diagnosis and treatment plan. Sharing the news with others can help build a supportive community around their child. Though this may be difficult to talk about, being open and honest can help reduce feelings of isolation and helplessness.

Within this discussion some spoke of the psychological effect it had on them, after a diagnosis was made, the sickle cell disease group, felt support was needed in this area, especially for new parents to be able to access psychological support and /or counselling, for the time between diagnosis to first consultant appointment, this applicable to both groups. This highlighted some users require more time and support than others to process the news.

The findings also highlighted the importance of the first meeting as it underpinned the need for a professional, well-designed, and structured care pathway to support parents throughout their new journey.

The use of social media to access information were mentioned, Examples included, YouTube, apps, and podcasts, the use of infographics and videos all came up as ways to disseminate information, one user commenting she had not been given enough information and searched the internet only, to find some of the information inappropriate and distressing, this feeling shared throughout both reports, with all participants agreeing that some internet websites might not be reliable, one father put it “the internet is not a very good doctor”! strengthening the need for accurate and evidence-based information to avoid spreading misinformation.

One suggestion was for service users to be signposted to reliable social media resources. This could be accessible to a vast network of individuals from both the sickle cell and thalassaemia communities, or those trying to raise awareness. It was suggested that such an approach can be particularly effective if the messages come from influential social media accounts or celebrities with substantial followings. It can also be used to create a community and increase engagement. Another suggestion was for NHS SCT screening programme to investigate adding sickle cell information onto the NHS app, like other health conditions. However, the NHS app has comprehensive information on sickle cell and thalassaemia including signposting to both the Sickle Cell Society and the UK thalassaemia society. Therefore, it is important for this app to be advertised more to stakeholders as a reliable source of information on sickle cell disease.

Suggestions around support groups was discussed, some centres facilitate new parent support groups, where they can meet the specialist nurses, other parents and learn about support available from both the statutory and voluntary sector, such as the Sickle Cell Society and the UK thalassaemia Society with representatives sometimes attending the meetings. Many parents found this engagement to be an important source of information and emotional support as they learn more about the condition and how to care for their child's ever-changing needs. These groups offering support to siblings, grandparents, and extended family.

The sickle cell feedback highlighted some areas as requiring further information for parents, it was felt that there was not enough information for children living with sickle cell disease. Another voiced concerns about understanding by GPs around penicillin use and it is therefore important for new parents and health professionals to receive accurate information on Penicillin as children with sickle cell disease are more prone to infections and will benefit from the prophylaxis it provides. Finally, it was suggested information around treatments available to those affected by sickle cell, for those who had moved in from abroad should be readily available.
Only in the father’s groups did financial issues come up, around the costs of caring for a child with sickle cell disease or thalassaemia which indicated the need for further signposting and information of help available.

The NHS SCT screening programme has provided online and face to face training sessions for health professionals involved in the screening pathway and counselling, for many years and this will continue, however, from the feedback this may benefit from being promoted to a wider audience, such as health visitors, and GP’s etc.
Many parents reported a great sense of fulfilment, purpose, and drive in providing care and advocating for their child. They gain a sense of community by connecting with other parents who share similar experiences.

These findings offer insight and great opportunity for learning and development of the NHS Sickle Cell and Thalassaemia screening programme.

Joint recommendations

Sickle cell group	Thalassaemia group	What can be developed
	Time from accepting baby testing and results	Communication
	Missed diagnosis, delayed treatment	Information Programme
Carrier or Trait	Carrier or Trait	Information Communication:
Waiting times for results - participants felt this was too long. Told, no "news is good news".	"No news is good news"	Communication Training
Experience of receiving results varied between high/low prevalence areas and within same clinic settings.	Giving of results	Communication Programme Training
	Further information sources	Signposting to resources
Feelings of guilt, disbelief, some insisting they were not carriers. Difficulty in accepting diagnosis and telling family members	Education, understanding and stigma.	Information Communication Education and Training
Participants felt testing when as a teenagers would help it become a normal conversation.	Importance of knowing your status, raising awareness	Programme Communication Information.
Young people, testing to know their status. Giving bloodspot results as an adult	Access to testing as preconception or young person	Communication Information: Programme
	Inequality not accessible screening pathway?	Programme
Results to parent protocol	Results to parent protocol	Programme
Psychological support after diagnosis of status	NHS Primary care	NHS Primary care
Parent support groups	Understanding the lived experience to help understand the query.	Communication Information signposting
GP'S and pharmacies understanding of prescribing suitable amounts of penicillin.	Timely referral to specialists to monitor baby's haemoglobin, growth etc.	Communication programme

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UKTS Related Resources

Accessible via website – www.ukts.org
1. Asian Awareness
2. For people who carry alpha zero thalassaemia
3. Guide to living with thalassaemia
4. Information for people who carry alpha thalassaemia
5. My baby has thalassaemia
6. Parent Stories
7. Standards for the clinical care of children and adults with thalassaemia
8. Teaching a child who has thalassaemia.
9. Thalassaemia . Your Life. Your Test, Your choice
10. Thalassaemia in your language
11. Videos and documentaries on our YouTube - for children, adults, clinicians and the general population
12. What is thalassaemia trait
13. What you need to know about beta thalassaemia

About UKTS

UKTS's overriding aim is to improve the lives of people living with thalassaemia. We provide invaluable support to patients with the condition across the UK. This means supporting not only the patients and families themselves, but also a broader network of medical and educational professionals involved in their care. We organise conferences for health professionals to share information on latest treatments and support groups for patients to share their experiences.

Over the years, we have funded life-saving equipment and ground-breaking research and development. We work with Public Health England and the NHS to promote screening and to develop clinical standards to ensure the best care for thalassaemia patients. We have given rise to a number of other charities around the world and continue to support them. We do not receive any government funding and are wholly dependent on private donations. It is vital that we continue fund-raising in order to support all those – patients and their families, medical and educational professionals, overseas charities and many more – that are dependent on the work we do.

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Useful Sickle Cell Resources

1. 'Parents Stories – Personal Experiences of the NHS Antenatal Sickle and Thalassaemia Screening Programme' https://www.sicklecellsociety.org/resource/parents-stories
2. Parent's Guide to Managing Sickle Cell Disease' (4th Edition) –' Parents Handbook'. https://www.sicklecellsociety.org/resource/parentsguide/
3. Sickle Cell Disease in Childhood: Standards and Recommendations for Clinical Care' (3rd Edition) which has a full document and an 'Executive Summary' - 'Paediatric Standards'https://www.sicklecellsociety.org/resource/paediatricstandardsresource/
4. Sickle Cell & Thalassaemia Counselling, Knowledge & Skills Guidelines' (2020) – 'Counselling Competences' - https://www.gov.uk/government/publications/sickle-cell-and-thalassaemia-counselling-knowledge-and-skills

About Sickle Cell Society

The Sickle Cell Society was first set up as a registered charity in 1979. It was formed by a group of patients, parents and health professionals who were all concerned about the lack of understanding and inadequacy of treatment with sickle cell disorders.

The Society's mission is to enable and assist individuals with a sickle cell disorder to realise their full economic and social potential. This is achieved by improving opportunities for sickle cell affected individuals and families by raising public awareness through education, advocacy together with the provision of direct welfare services, assisting in research and lobbying.

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