



Public Health
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Parents' Stories



Personal experiences of the NHS Antenatal Sickle Cell and Thalassaemia Screening Programme

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Parents' Stories

Personal experiences of the NHS Antenatal Sickle Cell and Thalassaemia Screening Programme in women and couples at risk of having a baby affected by one of the conditions

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Executive summary

Thirteen women and couples who had recent experience of the NHS Sickle Cell and Thalassaemia Screening Programme were interviewed.

Timeliness data at significant points in the screening pathway were collected on 16 pregnancies; women also shared stories from earlier pregnancies.

Responses and the experience of women and couples were diverse and people did not always act according to how they were assumed to act.

The belief that late testing is due to late presentation was not supported, all women first presented in pregnancy at less than 10 weeks gestation. The majority of women already knew they carried the gene for sickle cell disease or thalassaemia. Carrier status was also already known in the majority of men that carried the gene for thalassaemia or for sickle cell disease.

A common theme in the interviews was an assumption on the part of the service users that the healthcare professionals would recognise the risks and refer to counselling and offer of prenatal diagnosis (PND) promptly when carrier status was known. This was not the case even in families that already had a child or children affected by the condition. Some healthcare professionals did not assess the information given to them or were not trained to ask and evaluate genetic information. There was an assumption that 'these issues' would be picked up down the line by someone else that is more knowledgeable.

There was an expectation that key information told to one healthcare professional would be automatically recorded and shared with others.

In contrast, some women did not pass on information about carrier status or affected children possibly due to denial or fear of raising a difficult subject; as if by not mentioning it they were trying to treat their pregnancy as normal.

Several healthcare professionals lacked knowledge of the conditions and the screening pathway and did not recognise the need for prompt referral to counselling and PND.

All women and couples who were counselled by specialist haemoglobinopathy nurses and midwives reported positive experiences. Direct access to specialist sickle cell and thalassaemia (SCT) centres was really valued. They felt that only these specialist healthcare professionals understood the issues of an at-risk pregnancy and the need to expedite the screening process. Too often referral processes blocked rather than facilitated access to specialists and for some the offer of PND came too late in the pregnancy to allow reproductive choice.

Nine babies were born with one of the conditions, including one set of twins. Of these 5 women declined and 3 accepted PND. Although 7 out of 9 PND tests were performed by the 12 weeks and 6 days standard, unacceptable delays were encountered.

Three of the families interviewed said that they would opt for PND and termination of an affected baby, but only if it could be done within 120 days in accordance with the belief held by some Muslims of "ensoulment" at 120 days' gestation. This was achieved in only one of the families and 2 families gave birth to affected babies.

Healthcare professionals should be aware that beliefs regarding termination of pregnancy vary and should not harbour any preconceptions about the choices a couple will make. It should also be borne in mind that women or couples may not make the same choice in every pregnancy.

Background

In March 2015 the Sickle Cell and Thalassaemia Screening Programme Advisory Group set up a subgroup to focus on timeliness of antenatal screening and PND. The subgroup included parents of children with sickle cell disease and thalassaemia and representatives from midwifery, obstetric and genetic professional organisations.

At the first meeting we held a structured brainstorming session and used a fishbone diagram to identify all possible causes for why women are tested late. This helped sort ideas into the following useful categories:

- people (split by users and Health Care Professionals (HCPs))
- methods/processes/tasks
- equipment/IT
- measurement and control

This work focusses on the 'people' category and forms part of a larger piece of work that includes clinical audit and service evaluation. The Sickle Cell Society and UK Thalassaemia Society were commissioned to do this work on behalf of the programme. We present the personal stories of women and couples who have recent experience of the NHS Sickle Cell and Thalassaemia Screening Programme.

Understanding the user perspective is essential if we are to build a service that meets user needs and meets the programme objectives. This work will be used to inform standards and guidelines and public and professional educational resources.

Methodology

Two experienced staff members from the UKTS and SCS recruited participants and held in-depth face to face interviews with volunteer service users using a survey questionnaire (appendix 1).

Women who were pregnant within the last 5 years and fulfilled the categories below were invited to participate:

- those who have accepted or declined PND
- those who knew that they (and the baby's father) both carried a gene for sickle cell or thalassaemia
- those who knew that only they carried a gene for sickle cell or thalassaemia

Elaine Miller (UKTS) interviewed 4 couples and 2 women, 1 woman and 2 couples each from the North and South of England. Iyamide Thomas interviewed 7 women: 4 from the north and 3 from the south; only 5 stories are reported due to similarities. Data from all 7 women are included in the gestation timeline. Several women gave accounts of more than one pregnancy.

Pseudonyms are used to maintain anonymity.

Parents' stories: UK Thalassaemia Society

Interview 1 - Amina's story

My name is Amina. My husband and I both come from families which originated in Pakistan; although my husband was born in the UK. I was married in 2007 and already over 3 months' pregnant when I came to the UK. We live in the North of England with my husband's family. We were screened and found out we were both thalassaemia carriers when I was 25 weeks pregnant. We declined PND because our religious beliefs forbid termination of pregnancy unless the mother's life is in danger; we prayed for a healthy child. After the baby was born a nurse came to tell us that he has thalassaemia. We were grief stricken. Nobody in our families knew anything about thalassaemia and my husband's family blamed me completely for the baby's illness. I have suffered from depression for years because of the stress of living with the hostility from my in-laws.

I was scared to get pregnant again because of what happened with my son. Five years later I found myself pregnant again. I went to the GP straight away and saw a midwife at 5 weeks pregnant. I told the midwife at the first appointment that we are both thalassaemia carriers. She did not seem concerned and throughout all my antenatal appointments nobody mentioned the fact that we were at risk again. My husband was not tested again in my second pregnancy. I was terrified of what would happen but didn't say anything in front of the family. I was too scared to sleep or eat.

When my baby was born in 2012 I did not dare to leave the house. I sat by the phone and was terrified every time it rang. I cried constantly and could not enjoy having my little girl for a long time. Weeks went by and I heard nothing; the baby seemed well and I started to hope that no news is good news. My daughter is four now and to this day nobody has told me her test result. I never dared to ask because I was afraid of hearing bad news. My youngest is well but I still feel scared when she is ill and worry that it could be signs of thalassaemia. My husband and I wanted a big family but I am afraid to go through the trauma of pregnancy again. What would happen to my family if I had another sick child?

Interview 2 - Iqbal and Khadijah's story

My name is Iqbal and I was born and raised in the north of England, although my family is from Pakistan. I have been married to Khadijah for over 20 years; she came to the UK from Pakistan when we were married in our late teens. Khadijah is not confident in speaking English although she understands quite a bit. We had never heard of thalassaemia when we got married – the first we knew of it was when our first child, a daughter, was born in the 1990s and diagnosed with thalassaemia major. We were still in shock when we found Khadijah was pregnant again and the following year our son was born. We were devastated to find out that he too had thalassaemia.

The next few years were very difficult raising 2 children with thalassaemia; it was very stressful dealing with the hospital appointments and all the worry. The hospital explained to us about PND and Khadijah and I agreed that we could not cope with any more children with thalassaemia. We decided that if any more children came along, we would have PND and terminate the pregnancy of an affected child. As Muslims we believe that termination of pregnancy can only be done if it is carried out within 120 days of conception; so early PND is crucial for us. In our third pregnancy we had PND at 12 weeks. The haematologists treating

our older children suggested that we also find out whether the baby was a human leukocyte antigen (HLA) match for stem cell transplant. Fortunately, the baby was healthy and proved to be an HLA match for his brother.

As years went by Khadijah and I did not think we would be having any more children so we were surprised to find that Khadijah was pregnant again. Khadijah went to our GP (an Urdu speaker) to report the pregnancy when she was 6 weeks pregnant. I was pleased that she got an appointment with the midwife very quickly at 7 weeks and I thought the GP had arranged it so quickly because we are at risk. Our whole family, including our 2 children with thalassaemia major have had the same GP for many years. I assumed that the next step would be that they would arrange PND for us. When Khadijah was 9 weeks pregnant I was very surprised to get a phone call from a midwife saying "your wife has been diagnosed as a thalassaemia carrier so we need you to be tested." I said "we already know we are both carriers, we have 2 children with thalassaemia!" The midwife was completely unaware of our family history and she insisted that I would have to come in and be tested because they could find no result in my medical records. I was re-tested and received the result quickly at 10 weeks. Khadijah was booked in for a scan at 11 weeks and I went with her to the hospital thinking they would talk to us about PND during the appointment. During the appointment, I realised nobody had registered that there was any risk and they were treating it as a normal pregnancy. I started asking what had been done about arranging PND but nobody seemed to know what I was talking about. I insisted on seeing a doctor that same day but even when I started talking to the doctor she didn't get it that PND had to be done as soon as possible. It was only when I said straight out that we would be terminating the pregnancy if the baby was affected and that it had to be done before 120 days that they woke up and took notice. Khadijah had an amniocentesis at 13 weeks and within a week we received the welcome news that the baby was a healthy carrier. I feel that if I had not gone to the scan appointment with Khadijah and been insistent about PND and the 120-day limit it may not have been done until it was too late for us to have exercised our informed choice.

Interview 3 - Neelam and Raj's story

My name is Neelam and I have been married to Raj for 10 years. Our families are from India but we were both born in the UK. We were both tested for thalassaemia before we got married as it runs in Raj's family; he has a cousin in India who has thalassaemia major. Therefore, when we decided to start a family, we knew we were both carriers. We arranged to see a specialist genetic counsellor privately so we could gather all the necessary information and thoroughly discuss the implications of the fact that we were at risk of having a child with thalassaemia. We decided to opt for privately funded pre-implantation genetic diagnosis (PIGD) to avoid having an affected child. However, we experienced serious fertility problems and decided to adopt – only to conceive naturally shortly thereafter. We used private healthcare throughout the first pregnancy; which included referral to hospital for counselling about PND. After many discussions about all the options available to us, we declined PND. Our first daughter was born in 2012 and is healthy. Feeling unwell later in the same year I went to see my private obstetrician, who performed a scan and discovered I was 8 weeks pregnant with twins. The obstetrician recommended that, should both twins survive beyond 12 weeks, I should transfer antenatal care to the NHS as they are better equipped to care for a woman in a high-risk pregnancy. My obstetrician offered to arrange PND for us around the time of the transfer to NHS care but we declined both the test and any further counselling; as we felt that we thoroughly understood all the implications of our at-risk status.

When I entered NHS care at 12 weeks pregnant I clearly wrote in the front of my antenatal care book that we were at risk of thalassaemia, but the midwife who saw me did not pick up on it or discuss thalassaemia with me at all - she was fixated on it being a twin pregnancy. At 18 weeks' pregnant I had a phone call from the screening midwife who had just picked up on the fact that we were at risk from my medical notes – neither of us was re-tested. The screening midwife also wrote a letter to us about our at-risk status. I explained that we had already been through thorough counselling and wished to decline any further counselling and to decline PND. I had scans every 3 weeks and some of the sonographers upset me by asking whether I was aware the babies could be affected by thalassaemia; which was not appropriate. I also saw an NHS obstetrician who was very insensitive and tried to influence me towards finding out whether one or both babies were affected. He told me that children with thalassaemia often have heart problems and other "defects". Again, this was inappropriate behaviour and I was made to feel I was being socially irresponsible by exercising my fully informed choice to decline testing and continue with the pregnancy. I gave birth to fraternal twin girls and both were diagnosed with beta thalassaemia major. Raj and I are convinced that all the information we learned during our first pregnancy enabled us to accept the diagnosis and be very positive about our daughters' long term prospects.

Interview 4 - Abdul & Fozia's story

My name is Abdul and I have been married to Fozia for 4 years. I have lived in the London area since I was a child. Fozia moved from Pakistan to the UK after we were married. She was already pregnant when she arrived and saw a midwife when she was 25 weeks pregnant. I was asked to come in for a test because Fozia was a thalassaemia carrier. When she was about 30 weeks pregnant we found out that I too am a carrier. We saw a nurse counsellor who explained everything to us very thoroughly; what thalassaemia is; how it is inherited and that all our children would be at risk. She also explained that they can do a test in early pregnancy to find out whether the baby is affected. It was after this conversation that I realised it is the same illness that my cousin in Pakistan has, none of us in the family knew that it was genetically inherited until this time. In accordance with our Muslim beliefs Fozia and I would only be able to terminate a pregnancy up to 120 days after conception so with our first baby we could only pray for a healthy child. However, we decided that in any future pregnancies we would have the test as we did not want to bring up a child with a serious medical condition. Thankfully, when our daughter was born she did not have thalassaemia.

When Fozia became pregnant for the second time we went immediately to the GP – she was about 5 weeks pregnant. I went with her because my English is much better than hers. I explained to the doctor that we are thalassaemia carriers and we want the special test. While we were there the doctor sent a fax to the hospital and said they will send you an appointment. This was fine but then weeks went by and I started to worry. I phoned the surgery many times and I even went back in and spoke to the receptionist. They told me that the hospital is very busy and we just had to wait. I was panicking but I did not know who I could speak to. When we finally got the antenatal appointment Fozia was 16 to 17 weeks pregnant. I told the midwife we need the test right now but by the time the test was arranged Fozia was already 20 weeks pregnant and it was too late for us to have any choice other than to proceed with the pregnancy.

When Fozia was 22 weeks pregnant the hospital telephoned her at home and told her the news that the baby has thalassaemia. She phoned me at work in a hysterical state, I rushed home immediately and she was so

distraught I was afraid and rang the doctor's surgery. I don't understand how they can give that kind of news to a pregnant woman who is all alone; the hospital should have telephoned me. We went to the GP and he told us about UKTS and gave us their contact details. I phoned them and they sent us a lot of information, books and films to watch about people who have thalassaemia. Fozia was very depressed for the rest of the pregnancy. When she was about 30 weeks we saw a doctor at the hospital who told us we could still choose to have a termination because they could inject something into the baby's heart to kill it. We were very upset at this suggestion; we thought it was a very insensitive thing to say as we had already explained about our beliefs and that we could not terminate after 120 days.

When our son was born we went to see a specialist who gave us her number and told us that if Fozia gets pregnant again we should call her as soon as we know and she will arrange the test. If only we had known about this specialist before. Now I have to watch them sticking needles into my son and my wife cries all the time.

Interview 5: Asad and Nadia's story

My name is Asad and I am from a country in the Middle East. I came to the UK in 2012 with my wife Nadia and my son who was 2 years old at that time. We initially lived in Scotland before moving to the North of England in 2014. Nadia and I had our blood tested before we got married, which is standard practice in our home country. I do not know what tests were done but we were told that there was no problem for us. Our son was born in 2010 and everything was fine. Neither of us knows of any family history of any medical condition.

Nadia became pregnant in 2013 while we were living in Scotland. We went to the GP and got an appointment with a midwife when she was 12 weeks pregnant; this was the first time they took her blood. I took Nadia to all her appointments as she does not speak English. We saw the midwife again at 16 weeks and she told us that Nadia is a thalassaemia carrier but it was nothing to worry about. When Nadia was 22 weeks pregnant we had a hospital appointment with a different midwife. She saw in the notes that Nadia is a thalassaemia carrier and told me I should be tested immediately. I had blood taken that same day. About 3 weeks later I went to the GP to ask about my test result. The GP told me that my test had come back positive and Nadia and I should see a genetic counsellor. Looking back, I feel guilty that I did not look into it further at the time; but the GP did not seem to think it was very urgent and we were in the process of packing up and moving the family to England so it was a very busy and stressful time.

Once we were settled in England I went to register with a local GP and I remembered to ask him about thalassaemia, telling him that we are carriers and had been told to see a genetic counsellor as Nadia was pregnant. We had a Chinese GP and he googled thalassaemia in front of me. He got very agitated and told me that this means the child has no chance of life and you have to terminate the pregnancy as soon as possible. I now know that the GP made a mistake and he was thinking of another kind of thalassaemia, but at the time it was very distressing and Nadia was distraught at the thought of having a late termination. We are Muslims and we can only terminate a pregnancy up to 120 days after conception. He referred us to a local sickle cell & thalassaemia centre to see a specialist counsellor. She was very nice and spent over 2 hours with us, explaining that there would be a 25% chance that the baby would have thalassaemia; and that children who have thalassaemia can be treated so it is not fatal. She also explained that a diagnostic test can be done during the pregnancy; but by now Nadia was already 29 weeks pregnant. After our daughter was born we found out that she has thalassaemia.

Our daughter has had a lot of medical complications and we are determined not to bring another child with thalassaemia into the world. I know now how to contact the specialist nurses in the sickle cell & thalassaemia centre, so when Nadia becomes pregnant again we will go straight to them to arrange the test.

Interview 6: Fathima and Ali's story

My name is Fathima and I have been married to my husband Ali for 12 years. We are Muslims. Our families are aware of thalassaemia and its inheritance pattern. Ali has a brother in his thirties who has thalassaemia major; and Ali knew he was a carrier long before we got married. I was tested when we got married, so we knew we were at risk before I became pregnant with my first son, who was born in our home country. While I was pregnant we discussed the risk of thalassaemia with our obstetrician who recommended PND to us. I had an amniocentesis at 18 weeks which revealed that the baby had thalassaemia. The obstetrician offered us a termination; but we were unsure about this because of our strong religious beliefs. Ali consulted our imam who advised that it would only be permissible to terminate if continuing with the pregnancy would put the mother's life at risk; so we went ahead and had the baby. I know that not all Muslims share the same views on termination of pregnancy but we have chosen to accept the advice of our imam. We faced the fact that all our children would be at risk of thalassaemia with acceptance; as God only gives the burden to he who can bear it.

In 2009 Ali successfully applied for work in the UK and we moved here later that year. In 2010 I became pregnant for the second time. I reported the pregnancy to the GP and saw a midwife at about 10 weeks. Neither the GP nor the midwife picked up on the fact that we already had a child with thalassaemia; but the consultant who was treating our son suggested that the new baby might be a stem cell donor for him. The consultant arranged for me to speak to a nurse specialist who explained that I could have PND and find out whether the baby was affected at the same time as finding out whether s/he could be a donor. I had an appointment for PND at 16 weeks. Ali and I thought very carefully about the PND; but when discussing the matter between ourselves we both felt certain that we would not choose to terminate our baby whatever the outcome of the test should be; so there was no need to go through with the procedure. We came to this decision the day before the appointment. On the day we went together to explain that we had decided against the test; as we thought that would be more courteous to the staff rather than to just cancel by a phone call. We appreciate that the medical and nursing staff conducted themselves in the best possible manner; they made sure we were fully informed, provided all the options for us to consider and were respectful and understanding of our decision.

After this Ali and I decided that in future we would only consent to minimal, medically necessary tests during pregnancy and would not take any tests designed to diagnose conditions affecting the baby. When my second baby was born, we had the heel-prick test, but we specifically said that we did not want the baby tested for thalassaemia so we were not given any result. The consultant treating my older son was concerned about the baby and asked to see him in clinic, and he too was diagnosed with thalassaemia at around 4 months old.

When I became pregnant for the third time I saw the same midwife, I have had the same midwife for all 5 of my pregnancies in the UK. Before meeting me, she had never dealt with a family affected by thalassaemia. We are the only family with the condition in our area. During this pregnancy we declined all screening tests for medical conditions affecting the baby and declined the heel-prick test after birth. The consultant treating

our older 2 boys tested the new baby's blood at 5 months and she proved to be a thalassaemia carrier. Our fourth child was born a year later and again we declined all screening tests. Once again the consultant tested the baby when a few months old; and he proved to have normal blood.

Two years later we had our fifth child. The consultant treating our 2 oldest children spoke to us during the pregnancy and offered us PND and possibly stem cell collection but we declined. After the baby was born the consultant called when he was 2 or 3 months old and told us that, unknown to us, the heel prick test had been done "automatically" while the baby was still in hospital; and it revealed that he had thalassaemia.

Our sixth child, a daughter, was born a year later and has thalassaemia. We declined all screening tests throughout the pregnancy. The midwife and I discussed the possibility that the baby would have thalassaemia; but she did not attempt to persuade me to have any screening or PND in this pregnancy. She understands that we do not want any tests and we will serenely accept God's will.

We are a couple who fully understand that we are at risk of having a child with thalassaemia with every pregnancy – we have six children and four of them have beta thalassaemia major. In accordance with our religious beliefs and personal ethics we do not believe in antenatal screening (for the baby) or in termination of pregnancy.

Parents' stories: Sickle Cell Society

Interview 1a Rachel's story

I am Rachel and live in London. My ancestors come from West Africa and my husband and I both carry the sickle cell trait. We knew before the pregnancy that we wanted to have PND and would have a termination if the baby had sickle cell disease. If we needed a termination we wanted it early, as this would make it a bit easier emotionally. We have one child who also carries the trait.

I attended the GP when I was 5 weeks pregnant, he seemed reluctant to refer me and didn't seem to understand the urgency or know much about sickle cell disease. Twice I completed the same form for a booking appointment, first at the GP surgery and again at the early pregnancy assessment unit. I waited another 7 weeks before getting my bloods done and was 12 weeks pregnant when I finally saw a sickle cell specialist nurse and got the help I was looking for. I was 14 weeks pregnant by the time I had the test. Our baby had sickle cell disease and because I was late had to have a medical termination of pregnancy.

Interview 1b Rachel's story

I am Rachel and want to tell you about my third pregnancy having had a termination for my previous pregnancy as the PND showed the baby was affected. This time I was aware of all the processes and at 4 weeks pregnant I completed the midwife referral form at the early pregnancy unit and also called the sickle cell nurse specialist to inform them. My antenatal appointment with the midwife was at 8 weeks but the nurses at the sickle cell and thalassaemia centre had already done my bloods the week before. This time I was fast

tracked for PND and had the test done by 12 weeks. As it was early this time around I was able to have the chorionic villus sampling (CVS) procedure which was not as painful as the other test. We were pleased the baby was only trait. My only complaint is that I felt I was given too many consent forms to sign just before the procedure and I think these should have been sent to me earlier.

Interview 2: Ekanem's story

I am Ekanem and my husband and I are of West African heritage. We both knew we were carriers as my eldest child has sickle cell disease. I returned from Africa about 6 months pregnant and could not do a PND test then. We were both tested again for this my 4th pregnancy. I had already had two previous PND's and knew I wanted to do a PND again. I told my GP about my pregnancy quite early and that we wanted a termination if the baby was affected. We were keen to do this before anyone realised I was pregnant. Blood tests were done around 3 weeks of my pregnancy but the midwife had little knowledge of sickle cell. She did not seem to understand the urgency when I told her I was at risk and already had a child with sickle cell. She seemed more concerned and knowledgeable about Downs's syndrome. She gave me the screening booklet and said she would speak to a more senior midwife about sickle cell. I waited but when no midwife contacted me I then went to see the sickle cell nurse specialist who looks after my child. The nurse specialist faxed the PND request and everything was fast tracked from there. The test was carried out around 12 weeks of my pregnancy and happy to say my baby was just a carrier.

Interview 3: Ola's story

I am Ola of West African heritage. I have two children with my previous partner; they were born in Ireland and I was not screened. My current partner and I did not know we both had sickle cell trait until I was screened for this baby when I was about 12 weeks pregnant and they called him for screening too. I first attended my GP when I was 4 weeks pregnant so really the screening test was late right? After the blood test results they told us we were at risk of having a child with sickle cell disease and when I was about 20 weeks they invited us for counselling about PND. We did not accept the test as we were both scared of having a miscarriage. I did not even read the leaflets they gave me. My baby was born with sickle cell disease. I had my other two children tested after that but they are alright. My baby looks fine and you wouldn't even know he has sickle cell disease so only my immediate family here in England know, not even my family back home know as there is so much stigma about the condition. They have seen his picture but know nothing about his condition. My children are very caring towards the baby. I would say everything about the antenatal service was alright but after my baby was born and they did the heel prick test a nurse came to the house and after a few visits I asked if my baby was ok as it was after 2 weeks and I had no results yet. The midwife said since no news then it was good news! My mind was at rest then after 6 weeks I got a call saying my baby had sickle cell disease and I was very upset. A senior healthcare professional visited me that same day and said she was sorry that I got given wrong information but my baby had sickle cell disease. She booked an appointment for me and my baby to see a haematologist to discuss the results. I didn't know much about sickle cell disease but saw people in Africa who didn't live long so was scared. The midwife should not have told me my baby was ok.

Interview 4: Lorraine's story

I am Lorraine, I am from the Caribbean and my partner is British Caribbean. We live in the north of England and I have a child with sickle cell disease. As a child I knew I had the trait as my mum also had it but I was not aware of the implications. I was about 4 weeks pregnant when I told my GP. I had my first antenatal appointment when I was about 8 weeks pregnant but no bloods were taken. I transferred hospitals in early pregnancy as I was not happy with the care and was screened at about 10 weeks. My partner could not attend screening but the hospital found his test results from pre-operative records. They acted on the offer of PND so quick I thought they were being too pushy but now talking to you they were probably trying to meet the deadline. I felt overwhelmed though as there were 4 or 5 health professionals in the room telling me about PND. We decided not to go for PND because of the risk of miscarriage. The after care service for my child with sickle cell disease at my hospital was not very good. Antenatal services at the GP were fine but care of my child with sickle cell was not, they knew very little about the condition. I was advised by a friend with sickle cell disease of a GP practice with an 'advanced nurse specialist' who had kids with the same condition.

Interview 5: Pamela's story

My name is Pamela and my husband and I are from East Africa. I am currently in my third pregnancy; I have had 3 PND's in total. We found out we were an 'at-risk' couple when we were screened for our first child. I can tell you about my second and current pregnancies. In my second pregnancy I first reported to my GP and I booked online. The appointment with the midwife was quick and bloods were done within ten weeks of pregnancy. I wanted PND and the midwife referred me to the sickle cell centre. I had PND by 11 weeks. Our baby was affected so we had a termination of pregnancy by 13 weeks.

In this pregnancy the midwife told me to contact the centre myself so things could move quicker and they did. I liked the counselling and that their telephone number is direct and not all around the houses. The sickle cell centre was very helpful and got me a quick appointment for PND. I hid the news of my pregnancy from my siblings and mum until I got my PND results because the last time I had told them about the pregnancy and it had ended in termination. It took me a while to believe the results for this pregnancy as I was worried they made a mistake but was happy when it was confirmed my baby was only a carrier. Things for this pregnancy were all timely since I referred myself directly to the nurse specialists. I am looking forward to my baby which is due in two months.

User needs

In order to learn and improve practice on the basis of the parent's stories in this section we draw out the implications for practitioners of meeting parents in different circumstances:

Example 1

As parents we fully understand that we are at risk of having a child with thalassaemia with every pregnancy. In accordance with our religious beliefs and personal ethics we do not believe in antenatal screening or in termination of pregnancy unless the mother's life is at risk. We need the healthcare professionals to:

- be respectful of our beliefs

- treat an informed decision to decline screening and PND with sensitivity and respect
- tell us about the positive outcomes for children being treated for thalassaemia in this country
- put us in touch with patient organisations so that we can:
 - i. find someone in our community to help explain genetic inheritance to our families
 - ii. meet young people/ adults living successfully with thalassaemia

Example 2

As a couple who understand the chance of having a baby with sickle cell disease who have decided we cannot bring up a child with the disease, we need healthcare professionals who:

- understand our situation and the screening and diagnostic pathway
- can advise on pre-implantation genetic diagnosis (PIGD)
- who are accessible; we need direct access to counselling and PND
- have knowledge of sickle cell disease and genetic inheritance

Example 3

As a couple at risk of having a child with thalassaemia who believe in “ensoulment” at 120 days’ gestation and have made the decision that we do not wish to bring another child with thalassaemia into the world, our needs are:

- self-referral to counselling and PND
- for healthcare professionals to be aware that timing of PND is crucial to informed choice
- for healthcare professionals to be aware of the Muslim belief of ‘ensoulment’, women who hold this belief may opt for PND and TOP of an affected child but only if this happened before the 120 days gestation

Example 4

As a couple unaware of the risk of having a child with thalassaemia, our needs are:

- healthcare professionals who understands the condition, can explain genetic inheritance, take the screening tests and refer promptly
- all screening tests and if required counselling and offer of PND and PND tests to be performed as soon as possible

Example 5

As a couple who understand the risk of having a baby with sickle cell disease who have declined PND we need healthcare professionals to:

- tell us about the positive outcomes for children being treated for sickle cell disease in this country
- put us in touch with patient organisations so that we can:
 - i. find someone in our community to help explain genetic inheritance to our families
 - ii. meet young people/ adults living successfully with sickle cell disease
- tell us how and when our baby’s heel prick result will be communicated
- advise on PIGD
- advise on direct access to counselling and PND in future pregnancies. Even though I did not accept it in my first pregnancy, I might reconsider and accept next time around

Discussion

The majority of women already knew they carried the gene for one of the conditions. Carrier status was also already known in the majority of men.

The belief that late testing is due to late presentation was not supported; all women presented early in pregnancy.

In 2015, the programme recommended that women and their partners are offered screening in every pregnancy. This was in response to patient safety incidents. Several fathers were not offered the test and decline was common in those offered who already knew their status. Careful wording in national guidelines is needed to ensure:

- fathers are offered the test in every pregnancy
- where status is known, the offer does not delay counselling and offer of PND
- the service knows how to proceed when fathers decline

A common theme in the interviews was an assumption on the part of the service users that the healthcare professionals would recognise the risks and act when carrier status was known. This was not the case even in families that already had a child or children affected by the condition.

There was an expectation that information told to one healthcare professional would be recorded and passed on to others. In contrast, some service users did not pass on information about carrier status or affected children possibly due to denial or fear of raising a difficult subject. Midwives must ask specific questions about risk of inherited conditions when taking medical and family history.

Several healthcare professionals lacked knowledge of the conditions and did not recognise the need for prompt referral to counselling and PND.

All women and couples who were counselled by specialist haemoglobinopathy nurses and midwives reported positive experiences. Direct access to specialist SCT centres was really valued. These healthcare professionals had the expertise and really understood the issues of an at-risk pregnancy and the need to expedite the screening process. Too often referral processes blocked rather than facilitated access to specialists and for some it came too late in the pregnancy to allow reproductive choice.

Unacceptable delays were encountered. Three of the families interviewed said that they would opt for PND and termination of an affected baby, but only if it could be done within 120 days in accordance with the Muslim belief in "ensoulment" at 120 days' gestation. Of these three families, only one had PND within the 120 days. The other 2 families had affected babies. In contrast, it is also important to treat people as individuals. The Qur'an does not explicitly mention termination of pregnancy, Islamic theologians have differing points of view and some Muslims will not contemplate terminating a pregnancy unless the mother's life is in danger. Healthcare professionals should be aware that beliefs regarding termination of pregnancy vary within the Muslim communities and should not harbour any preconceptions about the choices a couple will make. It should also be borne in mind that women or couples may not make the same choices in every pregnancy.

Women who are new to the UK can be very vulnerable and isolated. In-laws and new family members may not understand (or refuse to understand) the inheritance pattern of the conditions. The difficulty with the issue of 'woman blaming' and refusal to accept that thalassaemia or sickle cell are inherited equally from both mother and father is not necessarily lack of information; rather it is the more complex situation of refusal to accept an unwelcome fact or the stigma that the condition might carry. Although there is still a lot of work to be done around raising awareness of thalassaemia and sickle cell by giving people information, there is the far more difficult task of changing entrenched attitudes to make way for a more enlightened approach to the possibility of inherited conditions. This is an issue which needs to be addressed over the longer term. Healthcare professionals should be mindful that the complex issues which affect young women in this vulnerable position can affect the way they communicate with other people, especially on sensitive subjects.

Lessons learned

Known at risk women should be given a direct contact to ensure early access to counselling in future pregnancies.

Known at risk women should be given information on PIGD.

Counselling should be offered in every pregnancy and it should be clearly understood that every pregnancy is treated as a new situation. No assumptions should be made based on decisions made in an earlier pregnancy.

Counselling must be provided by specialists who can explain the concept of risk, the difference between the carrier and the clinical state and take into account the cultural context of the lives of the couples involved.

Counselling must include information about living with an affected child in England and the treatment that should be available to their child. This may be very different from experience in their country of origin.

Women must be given the contact details of the relevant patient society UKTS or SCS.

The opportunity to meet other parents and understand the realities of coping with everyday life should be available – the aim being to present couples with factual information which is neither “sugar coated” nor overly pessimistic.

Guidance for community organisations and public

Community organisations

Raise awareness in your communities about the importance of early screening

Raise awareness in your communities that screening and PND are a personal choice and that parents' decisions are respected

Raise awareness in your communities about how the conditions are inherited, the importance of knowing the results of both parents and how to get tested

Public

Keep presenting early in pregnancy or contact maternity services/specialist counselling services direct. For a list of specialist centres see:

<http://www.sickle-thal.nwlh.nhs.uk/information/nationalsicklecellthalassaemiacentres.aspx>

Tell us about your family and medical history

Tell us that you want to have counselling and prenatal diagnosis; don't assume that healthcare professionals know what you want

Contact the Sickle Cell Society and UK Thalassaemia Society for information and support

Guidance for screening providers

Raise awareness in your communities about

- the importance of early screening
- that screening and PND are a personal choice and that parents decisions are respected
- how the conditions are inherited
- the importance of knowing the results of both parents and how to get tested

Find out about the local screening pathway and how to access specialised counselling; provide information and training to non-clinical administrators and GP receptionists.

Standardise the delivery of the screening pathway across the service (unless targeted action is needed). Develop and implement a referral template with a minimum dataset (preferably online). Provide secure online web portal for self-referral and GP referral.

Involve your Maternity Services Liaison Committee in service design. Be aware of and implement the:

- service specification for NHS England responsible population
- programme standards
- programme handbook (in development); includes new guidelines for counselling and referral prenatal diagnosis and sample letters and templates
- laboratory handbooks

Continually audit your service and implement SCT checks and audits to improve quality and reduce risks.

Provide direct access to specialist nurses midwifery counselling services to known at risk couples; circulate a list of centres that can provide both counselling and PND. When counselling include positive outcomes for children affected by sickle cell and thalassaemia who live in England; outcomes may be very different in the country of origin.

Provide information on pre-implantation genetic diagnosis.

Provide direct access to maternity services to arrange a booking appointment. Take the screening sample at first appointment / booking appointment. Triage referrals for booking to prioritise women who present late. Text message appointment alerts to reduce non-attendance.

Ensure you ask about family history; ask specific questions about risk of inherited conditions. Share important medical and family history with other professionals who have a duty of care to the patient. Don't hold any preconceptions about the choices a couple will make.

Service providers should keep up to date with the sickle cell and thalassaemia screening e-learning module and sign up to the blog phescreening.blog.gov.uk | @PHE_Screening.

For more information on training see the genetic risk assessment and counselling module at <http://cpd.screening.nhs.uk/sct-externaltraining>.

Appendices

Recruitment letter and survey

Introduction

The Sickle Cell and Thalassaemia Societies in partnership with the NHS Sickle Cell and Thalassaemia Screening Programme are looking for people to participate in a project investigating the experiences of couples who have gone through the antenatal sickle cell or thalassaemia screening process. This screening in pregnancy involves a blood test to find out if you (and in some cases the baby's father) carry a gene for sickle cell or thalassaemia. The main reason for screening is to enable parents-to-be to make informed choices about their expected baby based on information about the baby's risk of inheriting sickle cell or thalassaemia. In order to improve screening services nationally we are particularly interested in finding out the following:

At what stage in pregnancy the offer of screening was made.

For a couple who both carry a gene for sickle cell or thalassaemia and are therefore at risk of having an affected baby, at what stage was the offer of a diagnostic test made to determine whether the unborn baby was affected. This test is known as prenatal diagnosis (PND).

The level of support offered at various stages of the screening process.

We would like to conduct face to face interviews with women who have been pregnant within the last 5 years and who fall into any of the following categories:

Those who have accepted or declined prenatal diagnosis (PND).

Those who know that they (and the baby's father) are both carriers of a gene for sickle cell or thalassaemia.

Those who know that only they carry a gene for sickle cell or thalassaemia.

Antenatal services keep audits of whether screening is carried out in accordance with the recommended timelines and we will be examining these records. However, in order to get a full picture of the process, it is essential that we hear from people who have personal experience of the screening process and how it works in practice, not on paper. Screening during pregnancy is always stressful for parents-to-be; and delays in the system cause even more distress and worry. Your contributions will help us to improve the service for future couples.

The interviews will take approximately one hour and can take place on a date, time and venue convenient for you. As a token of gratitude for your help we will give a £50 gift voucher per interview plus public transport costs where applicable.

All data collected will be confidential and only the interviewer will know the identities of participants.

- 1) Would you like to participate in this interview? Yes/No

About the interviewee

- 2) What year did you find out you were pregnant (2010 – 2016)?
- 3) What year was your baby born (if different)?
- 4) Was this your first baby? Yes/No

- 5) If "no" please give baby's position in family (e.g. 2nd child)
- 6) If you have children with more than one partner, could you kindly let us have further details of your family?
- 7) How old were you when this baby was born?
- 8) What was the first half of your postcode when you first found out you were pregnant? (e.g. EN2)
- 9) If you moved house during your pregnancy, what was the first half of your postcode when the baby was born?
- 10) What is your family origin? (Use FOQ)
- 11) What is the family origin of the baby's father? (Use FOQ)
- 12) Did you know you might be at risk of having a baby with sickle cell disease or thalassaemia before this pregnancy? Yes/No
- 13) If yes, how did you know?
 - Family history of SCT (please describe)
 - From screening in a previous pregnancy (year?)
 - Was tested before starting a family (mother/father/both)
 - Other (please specify)

About the pregnancy

- 14) How many weeks pregnant were you when:
 - You first realised you were pregnant?
 - You first informed a health professional?
 - You had your first antenatal appointment with a midwife?
 - You first had bloods taken?
 - You received your SCT screen results (if applicable, i.e. carrier status not known before pregnancy)?
- 15) Would you like to tell us any further information about your first contact with antenatal services?
- 16) Which health professional did you first inform about your pregnancy?
 - GP
 - Midwife
 - Other, please specify
- 17) What action was taken by the health professional – did s/he
 - Refer you to a midwife
 - Take bloods on the spot or refer you to haematology to have bloods taken straight away
 - Record your family origins
 - Record your family history of any condition (please give details below)
 - Any other actions taken please give details

18) Would you like to tell us any further information?

19) Where did you have your first appointment with the midwife?

- Hospital
- Home
- GP surgery
- Children's centre
- Other, please specify

20) If you already knew you might be at risk of having a baby with sickle cell disease or thalassaemia, how many weeks pregnant were you when you first discussed this with a healthcare professional?

21) What was the outcome of this discussion?

- Referred to midwife
- Referred for counselling
- Referred directly for prenatal diagnosis (PND)
- Other, please specify

22) If you requested PND from the outset, how was your request dealt with?

23) Did you feel that the healthcare professional understood that your test should be carried out by 12+6 weeks gestation?

About your blood results (for couples whose carrier status not previously known)

24) How were you given your blood results?

- Letter
- Phone
- Face to face
- Other, please specify

25) Did you get a carrier result? Yes/No

26) How many weeks pregnant were you when you got your result?

27) If "yes"

- Were you given any written information? Yes/No
- Was the baby's father invited for testing? Yes/No

28) If the baby's father was tested:

- How many weeks pregnant were you when he was invited for testing?
- How many weeks pregnant were you when he was tested?
- How many weeks pregnant were you when you received his result?
- How did you receive his result? (e.g. phone, letter)
- Which healthcare professional gave you his result?
- Did he get a carrier result? Yes/No (if "yes" proceed to Q31)

29) If the baby's father was not tested, do you know if this was because?

- Carrier status already known
- Baby's father declined test offer
- Baby's father not available for testing
- Other, please specify

30) If the baby's father was unavailable or declined testing, how many weeks pregnant were you when this happened?

About prenatal diagnosis (PND)

31) Were you offered PND? Yes/No (if "no" go to question 41)

32) Were you offered counselling about PND? Yes/No (if "no" go to question 35)

33) If "yes" who provided the counselling?

- GP
- Midwife
- Screening midwife
- Counsellor/specialist nurse
- Hospital doctor
- Other, please specify

34) Were you given written information about PND? Yes/No

35) How many weeks pregnant were you when:

- You had counselling about PND (if applicable)
- You accepted or declined PND?
- You had the PND test if you accepted?

36) What was your eventual decision?

- Accept PND offer
- Decline PND offer (go to Q41)

37) Which PND test did you have?

- Amniocentesis
- Chorionic villus sampling

38) What was your PND result?

- Affected baby
- Carrier baby
- Baby not affected

Any further comments:

39) How many weeks pregnant were you when you got your PND result?

40) If your baby was affected, were you offered any further counselling when you received your result?

41) What was your pregnancy outcome?

- Miscarriage (at how many weeks)
- Affected baby
- Carrier baby
- Baby not affected
- TOP
- If TOP, at what gestation?

Any further comments:

About the service you received

42) In what ways do you think the service performed well?

43) In what ways do you think the service could be improved?

44) How did you hear about the Sickle Cell Society or the UK Thalassaemia Society? Was it during the pregnancy or did you already know about them?

Gestation at significant points in the pathway

1. Mother first informs HCP of pregnancy
2. First discussion of at-risk status (if applicable)
3. First blood samples taken from mother
4. Result reported to mother
5. Baby's father offered test (if different from 3)
6. Baby's father tested
7. When either
 - a) Conclusive screening result from both parents available OR
 - b) HCP concludes that father's carrier status cannot be determined
8. Offer of PND made
9. Offer of PND accepted/declined
10. PND carried out

Timeline: gestation at significant points in the pathway

client pseudonym	mother first presented to hcp (weeks of pregnancy)	mother carrier status known pre-pregnancy yes/no	first blood samples taken from mother (weeks of pregnancy)	father carrier status known pre-pregnancy 'yes' or gestation if tested this pregnancy	prenatal diagnosis accepted /declined (weeks of pregnancy)	pregnancy outcome
Amina	4	yes	5	yes	not offered	not affected or carrier
Khadijah	6	yes	7	yes	11 requested	not affected or carrier
Neelam 1	5	yes	not known	yes	6 declined	not affected or carrier
Neelam 2	8	yes	not known	yes	8 declined	twins both affected
Fozia	6	yes	17	yes	20 accepted result @ 22 weeks	affected
Nadia	6	no	12	22	29 declined	affected
Fathima	not known	yes	10 screening test declined	yes	10 declined	affected
Pamela 1	7	yes	8	yes	11 accepted	pregnancy terminated at 13 weeks
Pamela 2	5	yes	10	yes	11 accepted	not affected or carrier
Rachel 1	4	yes	<12	yes	>12 accepted	pregnancy terminated at 14 weeks
Rachel 2	4	yes	7	yes	12 accepted	not affected or carrier
Ekanem	2	yes	2	yes	<13 accepted	not affected or carrier
Lorraine	5	yes	10	>10	11 declined	affected baby
Ola	4	no	>12	>12	20 declined	affected baby
Subi	3	yes	8	8	<13 accepted	not affected or carrier
Seye	4	yes	5	yes	<13 accepted	not affected or carrier

Resources

1. The service specification

The purpose of the service specification is to outline the service and quality indicators expected by NHS England (NHS E) for NHS E responsible population and which meets the policies, recommendations and standards of the NHS Screening Programmes available at <https://www.england.nhs.uk/commissioning/pub-hlth-res/>

2. Programme standards

This publication explains the programme standards for monitoring the SCT antenatal and newborn screening programme. The generic newborn blood spot screening standards also apply available at <https://www.gov.uk/government/publications/standards-for-sickle-cell-and-thalassaemia-screening>

3. Programme handbook (in development) provides supporting guidance for all health professionals

The content is based on evidence, healthcare professional enquiries to the programme, lessons from patient safety incidents, data collection and assessment of performance against standards and evaluation of external SCT courses and the programme's e-learning resources. It will include new guidelines on counselling and referral for prenatal diagnosis guidelines will be available at <https://www.gov.uk/government/publications/handbook-for-sickle-cell-and-thalassaemia-screening>

4. Sickle cell and thalassaemia screening e-learning module

Unit 1 AN and NB screening for sickle cell, thalassaemia and other haemoglobin variants

This unit gives an overview of the linked Sickle Cell and Thalassaemia Screening Programme, highlighting essential elements of the screening pathway.

Unit 2 understanding haemoglobinopathies

These inherited conditions cause significant health issues around the world. Understanding how and why they occur; the significance of family origins; and the pathophysiology allows the practitioner the knowledge to support families throughout, and following, the screening process.

Unit 3 about sickle cell disease

This unit explains sickle cell disease in detail, and how to care for individuals with this condition.

Unit 4 about Thalassaemia

This unit explains the significance of thalassaemia syndromes, and how to care for individuals with these conditions.

Unit 5 informed choice and understanding diverse needs in screening

Screening should be an informed choice. Making the decision to undergo the blood test is not always straightforward for individuals and may be influenced by a range of personal issues which practitioners should be aware of.

Unit 6 understanding the screening test and the FOQ

There are some variations in the way that screening is offered throughout England. This unit gives the background to how the screening processes should be conducted.

Unit 7 understanding antenatal screening results

Following screening, healthcare professionals must be capable of understanding results in order to explain the risks to parents. This unit provides essential knowledge for practitioners about the screening test and the significance to the pregnancy of parental results.

Unit 8 communicating and responding to screening results

Practitioners have a responsibility to inform parents about results that are sometimes unexpected, and how this may affect the pregnancy. This unit highlights how best to communicate these results, and the options available for present and future pregnancies.

Unit 9 screening the newborn infant

Babies born with a major haemoglobin disorder require lifelong care from parents and professionals. This unit explores newborn screening results, both carrier and affected, what these results mean, how to deal with them, and the referral pathway for those children who require further investigations or entry to the clinical care pathway.

Available at [http://www.e-lfh.org.uk/programmes/nhs-screening-programmes/nhs-sickle-cell-and-thalassaemia-\(sct\)-screening-programme/](http://www.e-lfh.org.uk/programmes/nhs-screening-programmes/nhs-sickle-cell-and-thalassaemia-(sct)-screening-programme/)

5. SCT checks and audits to improve quality and reduce risks

Each NHS screening programme has a defined pathway(s). The pathways show how the individual undergoing screening moves from one stage of the pathway to the next.

Checks are needed at each stage to ensure the individual moves seamlessly and safely through the pathway unless they chose not to. If these checks are not in place there is risk that an individual does not complete the pathway or the pathway is delayed unnecessarily.

Healthcare professionals are encouraged to work through the template and check if:

- local processes are in place to do these checks
- there are any gaps
- the checks are being done often enough

If the answer is no to any of questions above the last column (trust response) can be used to develop an action plan. Available at <https://www.gov.uk/government/publications/sct-checks-and-audits-to-improve-quality-and-reduce-risks>

6. GP Blog – what you need to know about sickle cell & thalassaemia screening

This blog looks at primary care issues relating to sickle cell and thalassaemia (SCT) screening and the GPs role to:

- deliver fast access to screening: either in surgery on confirmation of pregnancy or by arranging rapid referral to midwifery
- facilitate access to baby's father and inform him about screening
- refer known carrier couples directly for prenatal diagnosis (target by 12 weeks + 6 days gestation) and/or discuss option of preimplantation genetic diagnosis

Available at <https://phescreening.blog.gov.uk/2016/10/26/gps-what-you-need-to-know-about-sickle-cell-and-thalassaemia-screening/>

7. Sickle cell and thalassaemia screening: fathers matter

This blog focuses on offering screening to the baby's biological father; available at <https://phescreening.blog.gov.uk/2017/03/30/sickle-cell-and-thalassaemia-screening-fathers-matter/>

8. Take the test

Genetic tests can be used to determine whether a person is a carrier of a genetic condition – but is having a test always the best thing to do? In this lesson the students are presented with an intriguing dilemma about whether a boy should have a screening test after his fiancée has found out she is a carrier of sickle cell disease. Students use information presented by experts to weigh up the options and come to a reasoned decision.

Learning objectives

- use knowledge about inheritance to interpret genetic diagrams, including family trees
- make a decision by identifying issues that need to be considered in choosing to have a genetic test

Available at <http://www.engagingscience.eu/en/2014/07/03/take-the-test/>
and <http://www.engagingscience.eu/en/2014/09/12/making-decisions/>

