We are fund-raising for some life-changing, vein-finder machines!

Please follow the link to donate to us:

https://www.paypal.me/teamukts

What is thalassaemia?

Thalassaemia major is a serious, genetic blood disorder. People with the condition cannot make enough haemoglobin in their red blood cells and are dependent on regular blood transfusions throughout their lives. If left untreated, a child born with thalassaemia would die in early infancy. Thalassaemia developed as an evolutionary response to malaria which is why it mainly affects those coming from regions such as Asia and South East Asia, the Mediterranean, South America, the Caribbean, Northern and Central Africa and the Middle East. Whilst initially prevalent in these regions, due to the migration of communities over the centuries, anyone can be at risk. In fact, it is estimated that 300,000 babies, severely affected by thalassaemia, are born worldwide every year. With better education of those at risk and more effective screening, this inherited condition could be eradicated.

What does UKTS do?

Provides counselling, support and information to patients and families

Produces specialist educational material for patients and healthcare professionals

Organises national events including medical conferences, training days and support groups

Raises funds for vital medical research and life-saving equipment

Thalassaemia Matters is produced (articles - unless otherwise credited, interviews and design) by Neelam Dongha.

www.ukts.org
Letter from the Editor

Welcome to the last issue of 2020, a special edition packed with everything you need to know about screening - the process of identifying whether a person has a form of thalassaemia. This information is so important before starting a family and all it takes is a simple blood test! In a year where it feels like things are not within our control, this is something we can do, and really make a difference. Please spread the word amongst work colleagues, family and friends.

In fact, taking control is a common theme in the real-life stories featured in this issue. So too is the theme of openness. Read how Farzana has always been open about her son’s condition because she doesn’t want him growing up feeling he has anything to hide. Hear Cile talk about her upfront approach about having thalassaemia trait when meeting a potential partner. Likewise, Zeshan asked Farah to get screened before they started their family and you can experience their utter joy on the birth of their new baby. Could their smiles be any wider? Totally heart-warming stuff - just what we need right now. Although we will happily leave the nappies and sleepless nights to them...

If you are reading this magazine, it is likely that you or a family member has thalassaemia. But do you know how widespread the condition is? Or even why you have it? It is all explained in the article about the screening pathway and I found it particularly fascinating doing the research for this piece. The article on the screening programme throws up some interesting facts and charts its significant progress since inception - thank you to Dr Dormandy for her wealth of knowledge and input.

To learn about our screening partners and the future aims of the Programme, read the interviews with Amanda Hogan, who has recently joined at the helm at PHE and Iyamide Thomas from Sickle Cell Society.

There is so, so much more! So as the nights draw in (not to mention the 10pm curfew) and the autumn chill starts to bite, find a cosy corner and get reading...

Stay safe and see you in 2021!

Neelam
Message from the Chair

How are we already in October? When we started out in January, we had such exciting plans for our members and events for the year. However, Covid had other plans in store for us. To combat this and whilst it has not been the same, we tried to have more virtual events which I am pleased to say were well attended and appreciated.

We enjoyed receiving your feedback and comments! Please do continue to get in touch with us! I must say the entire UKTS team is very excited to continue with these new initiatives until its safer to have face-to-face events again.

In August we sent UKTS ‘goodie packs’ with copies of our magazines, standards of care and UKTS face coverings to every patient attending one of the 61 thalassaemia units across the UK and we hope you have received yours by now. We are also providing masks to our members and now selling them so please get in touch with us if you would like one!

We are celebrating National Thalassaemia Day on 19 October and we would like to encourage everyone to support the activities being planned! This day is incredibly special to us and we feel there is no better day to make the focus solely on thalassaemia. We are championing for more awareness, more action, better services and more compassion! I am asking you to please get on board and let’s flood social media with our voices and stories to make our strength fully known! Please do get in touch with us to share your stories, experiences and ideas!

I do hope that you enjoy the articles included in this edition. We strongly believe that timely access to screening and informed personal choice is vital and may help reduce the number of babies being born with thalassaemia in the UK. Lastly but most importantly, I would also like to remind members to follow the guidelines and advice in order to continue staying safe and keeping healthy throughout the winter period.

Until then, best wishes.
Gabriel Theophanous
Meetings and events attended by UKTS (July - September 2020)

July
2  UKTS team meeting
3  Screening update session
6  Screening group - Year 2 report and Year 3 Planning meeting
6  UK Forum for Haemoglobin Disorders meeting
7  ReSPoND project and research meeting- parents and carers
9  Sickle Cell and Thalassaemia Counselling, Skills & Knowledge Manuscript Meeting
9  FAIR – Steering Group meeting
9  UKTS online yoga session with Shayla Ghelani
13  ReSPoND project and research meeting- health care professionals
14  DWP PIP appeal meeting
16  UKTS online yoga session with Shayla Ghelani
21  Board meeting
22  Meeting with STANMAP, Sickle Cell Society and Health Education England
23  FAIR research investigators training
23  Screening PAG Group meeting
23  UKTS online yoga session with Shayla Ghelani
25  Instagram Live collaboration with the Wishing Factory
29  Breaking Down Barriers Project Resources webinar
29  Meeting with Bambos Charalambous MP
30  UKTS online yoga session with Shayla Ghelani

August
3  Patient Support meeting
4  NHSBT Joint Charity Stakeholder meeting
6  UKTS online yoga session with Shayla Ghelani
7  Health Education England meeting
10  Meeting with NHSSCTSP
10  Meeting with CEO Haemochromatosis Society
12  UKTS summer camp for children
13  UKTS online yoga session with Shayla Ghelani
14  Air Glove meeting
17  Meeting with CNS Whittington Hospital
18  Meeting with DKMS
18  Meeting with leadership team North Middlesex Society
19  Meeting with Paediatric Consultant and CNS Newham Hospital
19  Meeting with Paediatric CNS – Queen’s Hospital
20  Patient Support meeting
20  Ambassador meeting
21  Meeting with Paediatric CNS Royal London Hospital
21  QuickBooks training exercise
24  FAIR research investigators training

September
1  UKTS Board meeting
2  National Haemoglobinopathy Panel Business meeting
3  Meeting with Vertex
3  APPG For Thalassaemia AGM
3  Meeting with EURODIS
4  EURODIS CAB discussion
8  Meeting with Royal College of Pathology
8  NHS BT Steering Group meeting
8  Air glove demonstration by Health Care 21
11  Sickle Cell and Thalassaemia Manuscript meeting
12  TIF Board meeting
14  UK Forum for Haemoglobin Disorders meeting
15  Patrons’ meeting
16  Haemoglobinopathy CRG
17  Meeting to discuss PAG - agenda
17  Breaking Down Barriers meeting
18  Catch-up meeting with Sickle Cell Society and PHE
18  Patient Support meeting
21  Counselling Competencies Manuscript Committee meeting
21  Meeting with Department for Work and Pensions
22  Webinar Exploration meeting with TIF
23  Screening Catchup meeting
23  NHR Steering Group meeting
23  NHS Blood and Transplant SABTO discussion
23  Meeting with NHS Blood and Transplant Corporate Communications
24  Meeting with Health Education England
25  Prescription Coalition Group meeting
28  Thalassaemia National Day planning meeting
30  UK Forum HD Peer Review Steering Group meeting

Photo: September meeting with our wonderful patrons
APPG for thalassaemia

At the end of July, Bambos Charalambous, MP for Southgate and Chair of the All Party Parliamentary Group (APPG) for thalassaemia met with us to discuss APPG matters and our new fundraising venture to provide 20 hospital units with ultrasound vein finders. Then on 3 September, we had a virtual AGM for the APPG for thalassaemia.

The MPs involved in the APPG are:

Chair - Bambos Charalambous (Labour MP for Enfield Southgate)
Vice Chairs -
- Bob Blackman (Conservative MP for Harrow East)
- Feryal Clark (Labour MP for Enfield North)
- Alberto Costa (Conservative MP for South Leicestershire)
- Judith Cummins (Labour MP for Bradford South)
- Barry Gardiner (Labour MP for Brent North)
- Fabian Hamilton (Labour MP for Leeds NE)
- Kate Osamor (Labour & Co-op MP for Edmonton)
- Taiwo Owatemi (Labour MP for Coventry NW)
- Catherine West (Labour MP for Hornsey and Wood Green)

It was agreed that our work for this upcoming year will be focussed on addressing:
- The inequalities and inconsistencies in thalassaemia care across the UK.
- The issues people with thalassaemia are facing with DWP for Personal Independent Payment.
- Research into gene therapy and other developments for thalassaemia.
- The unfairness of prescription charges for adults with thalassaemia.

UKTS needs you to get involved with us to fight these inequalities! Please write to your MP and ask them to join the APPG. Please contact office@ukts.org if you would like help drafting a letter.
Prescription Charges Coalition (PCC)

The PCC is a group of 48 organisations calling on the Government to scrap prescription charges for people with long-term conditions in England. The coalition has had a response to the Parliamentary questions asked of the Secretary of State for Health and Social Care. These will be collated and sent out to all members of the coalition shortly.

As you know, prescription charges have recently increased and this can make life difficult for those with thalassaemia and other conditions not covered by the medical exemption. We want to make people aware of the low income scheme that is available so please do have a look here to see if you qualify www.nhs.uk/using-the-nhs/help-with-health-costs/get-help-with-prescription-costs/

IMPORTANT - PLEASE READ: GDPR

We are striving to ensure that we stay in contact with all of our members and would encourage everyone to complete a membership form. This ensures that we adhere to GDPR guidelines and only hold the information you would like us to have. It also helps us to plan events and activities relevant to our members. Forms can be found on our website here: https://ukts.org/member/

NCEPOD Transition study: Online young person and parent carer scoping survey

Young people and parent carers are being invited to share their experiences of transitioning from child to adult healthcare services. This study has been commissioned by the Healthcare Quality Improvement Partnership as part of the Child Health Clinical Outcome Review Programme. The aim is to help improve the transition process for both thalassaemia and sickle cell adolescents. Further information can be found on their website here www.ncepod.org.uk/transition.html

At this stage the survey is for completion by young people/parent carers of young people:
• Under 25 years of age
• With any condition that requires ongoing healthcare management
• At any stage of transition from child to adult health services

Please see below a link to this survey: www.surveymonkey.co.uk/r/ncepodtransitionsurvey

Royal College of Pathologists

On 10 September, UKTS met virtually with Dr Shubha Allard (Consultant Haematologist - Barts Health NHS Trust and NHS Blood and Transplant) and Penny Fletcher (Public Engagement Manager, Royal College of Pathologists) to discuss ways in which both organisations could support each other. We are really excited about this collaboration and initially, we will be working together to provide educational materials on thalassaemia within the Royal College and externally in schools and colleges.
Infected Blood Inquiry resumes hearings

The autumn hearings started on 22 September and you can keep updated with all the developments here: https://www.infectedbloodinquiry.org.uk/news

We are grateful to everyone who took their time to speak to the Inquiry team to share their story. We will never fully understand how difficult recalling and reliving these traumatic events were, but we appreciate you for being a part of this process to help bring justice and help the Inquiry hold those who were responsible to account.

The Infected Blood Inquiry team is still very interested in people with thalassaemia who contracted blood-borne viruses due to blood transfusions. They feel that our members’ stories have not been well represented and they are very keen to learn more. We would really be grateful, if you have been infected or affected by any of the viruses due to blood transfusions, to get in touch with us. We will support you as best as we can throughout this process. We need to get justice for what has happened, and YOUR story is invaluable to this process. Please remember that you can share your story anonymously – it is so important to be heard. You really can make a difference.

A fun way for children to learn about thalassaemia

UKTS is very excited to be collaborating with Jules Ventriloquist to create an educational series for children about thalassaemia. This is the first of its kind and a great educational resource for children - a fun way of learning about thalassaemia! Catch the first episode on our youtube channel here: https://www.youtube.com/watch?v=j6yDgi-J5sA

A huge thank you to the talented Jules for volunteering her time to do this! See more of her work on her website: https://julesventriloquist.co.uk and on youtube: @JulesVentriloquist

Latest Covid-19 advice

In early summer, the National Haemoglobinopathy Panel updated its patient information on Covid-19 infections for people with thalassaemia and other anaemias. You can read the latest guidance from the NHP and UKTS for people with thalassaemia across the UK here: https://ukts.org/wp-content/uploads/2020/07/UKTS-Patient-Info-30620FINAL.pdf
In July, UKTS welcomed Zoe Panaretou to the team as our Information Officer.

Zoe recently graduated from the University of Reading with a degree in Nutrition and Food Consumer Science. She has accreditation with the Association of Nutrition and is a Registered Associate Nutritionist (ANutr). We are very lucky then that, in addition to her role as Information Officer, Zoe will be providing guidance about nutrition for health - specific to the needs of thalassaemia patients. You may have already tried some of her wonderful recipes (shared via social media and our weekly updates), and there are plenty more to come. Healthy and tasty..? That’s got to be Foodie Heaven!

In this issue, Zoe has written about what foods to eat and supplements to take before, during and after pregnancy (see p36).

Screening and genetic counselling: we want to hear from you!

UKTS attended two online focus groups facilitated by the ReSPoND knowledge Exchange Forum looking at positive newborn outcomes, and how the results are relayed to parents. These meetings provided relevant and important feedback on experiences towards improving services.

We are especially interested in hearing from members who are interested in taking part in screening-based focus groups. We will be collating anonymised information on the experiences of parents and parents-to-be, to identify the needs and shortfalls in the service towards improving and proposing possible solutions. We will be looking for participation in the following categories:

Experience before/ during pregnancy
- To collect feedback about whether you knew you were a carrier prior to pregnancy;
- How you found out, what support you were offered;
- How your experience could be improved.

Experience after pregnancy
- What were you told about the heel prick tests?
- When were the results shared with you?
- If positive, how were the results shared with you?

Genetic counselling
We also attended a meeting about genetic counselling offered to parents and patients.
- Is this something that you have had?
- Did you find it helpful?
- Would any family member with trait benefit from finding out what it means to be a carrier and how it could impact their family?

We would also like to ask anyone who is pregnant or has had a baby in the last few years who would like to participate in a screening focus group to please get in touch with us.

To participate in any of these topics, or for more information/ questions please contact us at office@ukts.org.

Join the NHR steering committee

The National Haemoglobinopathy Registry (NHR) is a confidential database comprising of people living with sickle cell disease and thalassaemia (see more about this on p24). The NHR is looking for patient representatives to join their steering committee. It is a great opportunity to become involved and have your say, so if you have thalassaemia and live in England follow the link to apply:

https://www.england.nhs.uk/wp-content/uploads/2020/08/nhs-steering-group-application-information-pack.pdf?fbclid=IwAR0FjCrl-IRWwSznJwUjTgOdFmEeoa0FJkdyfWRnuY2e9vyWJkdCQsYDTM
**UKTS goody bag road trip!**

We clocked up the miles in August by delivering over 1,700 care packages for patients around the country! We are still in the process of sending more out to members. Each package included a copy of the magazine, a membership form and a UKTS branded mask. We have also sent copies of the UKTS standards of care for adults and children with thalassaemia, colouring books, story books and other information so please ask your teams! Remember to fill out the membership form. Send us pictures in your masks - we want to see your selfies!

We delivered to 61 hospitals in total:
Addenbrooks, Alder Hey Liverpool, Bedford, Birmingham Children’s, Birmingham City, Bradford Adults, Bradford Children’s, Buckinghamshire, Cardiff, Coventry, Croydon, Derby, East Lancashire, Glasgow children’s, Glasgow Royal Infirmary, Great Western, Guys and St Thomas, Homerton adults, Homerton children, Hull and East Yorkshire, Imperial College, James Cook, Kings College, Leeds Children’s Hospital, Leeds St James University, Leicester Royal Infirmary, Lewisham, Lister, London North West, Luton and Dunstable, Manchester Children’s, Manchester Royal Infirmary, Milton Keynes, New Cross Hospital, Newcastle, Newcastle Children’s, Newham Hospital, Norfolk and Norwich, North Middlesex, North Staffordshire, Northampton, Nottingham Children’s, Nottingham Hospital, Oxford Adults, Oxford Children’s Hospital, Queens Elizabeth Hospital Woolwich, Queens Hospital, Royal Berkshire, Royal Free, Royal Liverpool, Royal London, Sheffield, Sheffield Children’s, Southampton University Hospital, St Georges Hospital, University College London, University Hospital of Bristol, University Wales, Wexham Park, Whipps Cross and Whittington Hospital.

**Let’s talk thal!**

Roanna Maharaj (UKTS) was live with Partth Thakur, founder of The Wishing Factory, exploring the ways they’ve coped with having beta thalassaemia through various stages of their lives. Watch and be inspired! [https://www.instagram.com/tv/CDERxxRFN8H/](https://www.instagram.com/tv/CDERxxRFN8H/)

You can learn more about The Wishing Factory at [www.thewishingfactory.org](http://www.thewishingfactory.org)
Recognition for art

It was lovely to see that our recent art competition to encourage blood donation was featured in a national Iraqi Newspaper, Al-Sabah. The paper wanted to recognise the efforts made by talented young Iraqi artist, Sama Al Ameer. Well done Sama for your amazing artwork and helping to raise awareness!

And breathe...

A huge thank you to Shayla Ghelani from Yoga Logic, who very kindly ran a complimentary 6-week Yoga for Wellbeing programme for UKTS over summer. The sessions were online via Zoom and focused on yoga and breathing exercises. Perfect for relieving the stresses and strains of everyday life!
You can find out more about Shayla here https://yogalogic.co.uk/profile/shayla-

Trying in vain to find...veins!

The UKTS team has been hosting brainstorming sessions and discussions to identify ways to improve patient care and thus quality of life. Many of you will know how elusive veins can be, so it is not surprising that a key issue expressed by many of our members are the problems encountered finding viable veins for transfusion. Numerous cannulation attempts, intense pain and nerve damage are common experiences. To combat this, we have identified two possible solutions:

Sonosite IVIZ ultrasound machine: an expensive but effective way to prevent multiple attempts as the machine provides the user with a clear picture of the veins.

Air Glove: a much cheaper option whereby the patient is provided with a disposable plastic long sleeve glove. This glove is connected to a machine using hot air to warm the arm/ help the veins to pop up.

Meanwhile we have already started a fundraiser and would like to encourage everyone to help us achieve our goal (see supporter activities section on p55). Please let us know what you think!

And breathe...

A huge thank you to Shayla Ghelani from Yoga Logic, who very kindly ran a complimentary 6-week Yoga for Wellbeing programme for UKTS over summer. The sessions were online via Zoom and focused on yoga and breathing exercises. Perfect for relieving the stresses and strains of everyday life!
You can find out more about Shayla here https://yogalogic.co.uk/profile/shayla-

Photo: Airglove demonstration on our Chair, Gabs!
Introducing Amanda Hogan

Amanda Hogan is the National Programme Manager of the NHS Sickle Cell and Thalassaemia Screening Programme. She assumed this role in July 2019 after her predecessor, Cathy Coppinger, retired. With the pandemic to contend with after less than a year in the role, Amanda has certainly been thrown in at the deep end but has more than risen to the challenge. We spoke to her about what she brings to the role, the ins and outs of the programme and what the future holds for screening.

Before becoming National Programme Manager of the NHS Sickle Cell and Thalassaemia Screening Programme, what did you do?

I have 22 years’ midwifery and screening experience working in the NHS and PHE. I have built a professional career working both clinically on the front line and in senior leadership and strategic roles.

As a ‘Head of Midwifery’ I held 24/7 accountability for the safe delivery of maternity services to a high professional standard. In this role, I acted as the Trust expert on maternity issues to make sure a culture of proactive professionalism was embedded and promoted. I was accountable for setting, measuring and improving standards of care and the patient experience to meet NHS agreed objectives, targets and quality standards.

I became a Supervisor of Midwives in 2009 and converted to a Professional Midwifery Advocate (PAM) in 2017. As a registered midwife, I always promote professional standards in line with the Nursing and Midwifery Council (NMC) code. In all my roles, I strive for a continuous improvement process to enhance the quality of care for pregnant women, babies and their families.

Working as Senior QA advisor, I was the team leader providing screening specialist advice. My role was to provide a regional quality assurance service for the antenatal and newborn NHS screening programmes. I was the West Midlands Lead on the delivery of a QA function across the geographical area. I was responsible for liaising with provider organisations and commissioners to monitor organisations against minimum standards.
of service performance, and quality across all elements of antenatal and newborn NHS Screening Programmes.

Career Detail:

Dec 2013 to present: Public Health England - Senior Quality Assurance Manager Regional lead for antenatal and newborn screening (midwife clinical)

Jan 2012 to Dec 2013: Heart of England NHS Foundation Trust - Associate Head of Midwifery

Sept 2010 to Jan 2012: Ealing Hospital NHS Trust - Head of Midwifery and Assistant Nurse Director

Sept 2009 to Sept 2010: Supervisor of Midwives University Hospital Coventry and Warwickshire NHS Trust - Acting Head of Midwifery

Feb 2009 to Sep 2009: Supervisor of Midwives University Hospital Coventry and Warwickshire NHS Trust - Modern Matron, Inpatients Services

Mar 2007 to Feb 2009: Supervisor of Midwives Birmingham Women’s Hospital NHS Foundation Trust - Birth Centre Manager

Jan 2004 to Mar 2007: Supervisor of Midwives Birmingham Women’s Hospital NHS Foundation Trust - Community Team Manager

Sept 2000 to Jan 2004: Birmingham Women’s Hospital NHS Foundation Trust - Midwife (started as Student Midwife)

Please describe any personal experience you have had with thalassaemia.

As a community midwife working in a high prevalence area in Birmingham, I cared for pregnant women who were thalassaemia carriers. As a community midwife, I didn’t have in-depth knowledge of the condition, because I would fast-track women to a specialist nurse counsellor for follow-up and management. What was important for me to know was that thalassaemia is common in families where members are originally from:

- around the Mediterranean, including Italy, Greece and Cyprus
- India, Pakistan and Bangladesh
- the Middle East
- China and southeast Asia

Six years ago, when my sister was screened in pregnancy, my family were surprised to find out that she was a haemoglobin D carrier and that there is a chance that she could pass on the gene to her children. It was so unexpected, but a potent reminder that anyone can be a carrier of thalassaemia.

When did the screening programme originally start?

In 2001 the National Health Service (NHS) Sickle Cell and Thalassaemia (SCT) screening programme was established in England, and implementation was completed in 2008. Before that, it was only implemented in specific regions since 1981. It is the world’s first programme linking antenatal and newborn screening for SCD.

What attracted you to your current position and when did you start?

I completed a piece of qualitative research for my Masters Public Health, entitled ‘A qualitative study to explore the challenges that midwives face in offering antenatal sickle cell disease and
thalassaemia (SCT) screening in early pregnancy (within first 10 weeks of pregnancy) to parents in Birmingham West Midlands UK’.

From then I was hooked, my research findings were similar to the themes in the ‘Parents Stories’ book, that some healthcare professionals did not assess the information given to them by pregnant women and families 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 who was more knowledgeable.

Briefly describe your typical day.

I am leading and overseeing several formal strategic projects, so there are daily meetings with the SCT team. There are also detailed planning and management sessions almost every day. Depending on the scale of the project, this may also include meeting with the ANNB finance and administration team to ensure adherence to programme budgets and forecasting requirements. I will collect and analyse data in preparation for writing programme reports and so on. Meeting internal and external stakeholders, for example QA, laboratory scientists, clinical leads, societies and users. There is always a meeting with either maternity providers, child health, laboratories or the societies. As a programme manager I work with QA acting as an expert and will review and give advice on SCT screening incidents. Most days, there will be screening helpdesk enquiries and emails to answer. I have to be proactive and quickly address often complex, sensitive issues.

What would you say have been the major achievements of NHSSCTSP to date?

In my first year, approximately 670,000 pregnant women were screened for thalassaemia and sickle cell, and other haemoglobin variants, and around 626,000 newborn babies were screened for sickle cell disease. Not every new manager has a pandemic to cope with in the first year, but I believe I’ve met the challenge and have advised the NHS on managing SCT antenatal and newborn screening.

The programme has worked with African Research and Innovative Initiative for Sickle cell Education ARISE project helping them to understand newborn screening in England. We have also worked with the Sickle Pan-African Research Consortium (SPARCO), which is a collaborative project between Tanzania, Ghana and Nigeria, for years 2017-2021, with Tanzania as the lead institution.

I have been involved in the review and update of:
- SCT screening laboratory requirements
- Antenatal laboratory handbook
- Newborn laboratory handbook
- Laboratory e-learning
- Programme e-learning
We have made huge progress in making all the SCT programme information meet the accessible standard.

What are you currently working on and what are the short-term goals of the project?

I am continuing to roll out the newborn outcomes system to all providers. The system supports the referral of screen-positive infants from screening laboratories into treatment services. This project is important because it improves the safety of infants by allowing health professionals to view the status of babies along the screening/care pathway.

Longer-term, what would you describe are the main aims of the programme and how do you hope to achieve them?

I have used my first year in this fantastic role to fully review the programme. My review has centred around building key relationships, identifying, flagging, prioritising improvement opportunities and sharing information on the SCT screening pathway. I have a 5-year plan in place that sets out specific development work to promote measurable quality improvements.

What are the main challenges and how can they be overcome? (The pandemic obviously poses a significant challenge, but please also talk about other ones).

One of the main challenges has indeed been the Covid 19 pandemic, the team has worked tirelessly to make sure that business as usual continues whilst advising clinical teams on SCT screening during very difficult circumstances. The exciting
part of the new role was identifying interesting development work and setting objectives for the next five years. The future challenge will now be getting thoughtful development and solution plans in place, getting the priority order right and working on these to deliver positive change for users of the SCT screening service. I am here for the long run and will enjoy meeting this challenge.

The specific focus this year is improving data quality issues, so collecting newborn data for performance standard 8 and 9. This challenge will be met as soon as we complete the implementation of the Newborn Outcome System.

How do you find collaborating with UKTS and SCS?

I am delighted to be working and collaborating with the societies, it’s a real privilege. I value their knowledge and experience and I have learnt so much by working with them. They are great to work with because they are dedicated to their cause. They have helped enormously with my 5-year plan and setting key objectives.

Screening currently focuses on pregnant women (and partners where necessary) but ideally people should know their carrier status much earlier. What steps can we take to do this?

At the moment this isn’t in scope for the programme.

Are you a member of any professional groups or bodies? If so, which ones?

I am a registered midwife with the NMC.

What advice would you give to UKTS and SCS in their efforts to support people with thalassaemia and SCD?

I couldn’t give the societies advice on this; they are the experts on supporting people and families with thalassaemia and sickle cell disease. I look forward to working with both societies; their expertise is highly valued by the programme. I am pleased that our collaborations have already stimulated considerable discussion about quality improvement in the SCT screening pathway for women and their families. For the future, working in partnership, we will lead improvements to drive quality for all those who we are privileged to support in screening.

Tell me a little about your personal interests – what do you like to do in your free time?

My eldest son Aden lives in China and has been teaching there for the last 8 years, I really miss him. My husband and I have visited China twice and planned to go again in 2020, but Coronavirus ruined our plans. My youngest son Dominic lives in London and has established a career in Marketing. I tend to meet up with Dominic in London and we find somewhere new to try for a meal. I also really love spending time with my 3 little nieces, they are full of energy and so much fun to be with.

Other interests include yoga (I’m not very good, but I keep going) and running (I have competed in many 5K, 10k and half marathon events) and I like to relax by working in my garden.

Any longer-term personal aspirations?

I have found a home in the SCT programme, my longer-term goal is to make a difference for women, babies and families.

Thank you Amanda and wishing you continued success in your new role!
Before joining SCS, please can you describe your previous work experience?

Most of my work experience has been geared towards health as I grew up with a dad who was a medical doctor. He had dreams of me following in his footsteps; however, dissecting a rat during ‘A’ Level biology sealed the fact that I was too squeamish for medicine, so I opted to read Chemistry at University instead! Having a scientific background, I first worked in the mid-80s as the Research Development Officer at the Sickle Cell Anaemia Research Foundation, a voluntary organisation in Hammersmith. Then in 1988 I worked at Newham Health Promotion, as a Health Education Officer to promote Breast Cancer Screening amongst their Black, Asian and Minority Ethnic (BAME) community which was mainly Asian. In 1991, I joined the NHS Cancer Screening Evaluation Unit at the Institute of Cancer Research as the Trial Coordinator of a National Breast Cancer Screening Trial which was investigating the effect of annual mammographic screening of women starting at ages 40-41. I then joined the Sickle Cell Society (SCS) in October 2004, initially as a Regional Care Advisor.

What attracted you to work at SCS and what does your job involve?

I have always been interested in doing outreach and research work on health issues particularly affecting BAME communities so when my contract at the Institute of Cancer Research finally ended, I was thrilled that the job at the SCS came up.

I have actually had three job titles since I joined the Sickle Cell Society almost 16 years ago! I joined the Society as Regional Care Advisor for South London and that was mainly casework with individuals /families affected by sickle cell. My next two roles as NHS Outreach Project Lead and my current role as NHS Engagement Lead which began in 2016 were both on projects commissioned by the NHS Sickle Cell and Thalassaemia Screening Programme (NHSSCTSP).

My current role is to work collaboratively with UK Thalassaemia Society and the Screening Programme to provide service user input and ensure the Programme provides an improved and user-focussed service.

Please describe any personal experience you have had with SCD / thalassaemia.

I don’t have any personal experience of sickle cell besides knowing that my haemoglobin genotype is AA and that of my late dad was AS. Growing up in Sierra Leone I had two friends with sickle cell, one of whom I knew had the condition. The other was someone I knew at primary school who was always absent and had extremely yellow eyes and sallow-looking skin and I now know that was because he had sickle cell.
When did you get involved with the Screening Programme and what was your role?

I became involved in 2009 as NHS Outreach Project Lead for a project the Screening Programme commissioned to raise awareness of sickle cell, screening and associated myths using a DVD called ‘The Family Legacy’. It involved a lot of outreach leading a small team that conducted facilitated DVD film sessions at innovative places like barber and hairdresser shops, mosques, restaurants and even in family homes. That was something new for the Programme and they particularly liked the fact I organised sessions in barber shops to reach the men, who incidentally were not always there to have their hair cut!

Please summarise the main work that SCS/UKTS have done for the Screening Programme to date.

In the first SCS /UKTS collaboration which commenced in 2016, the main remit was to investigate barriers affecting the timeliness of the offers of screening and prenatal diagnosis (PND). SCS /UKTS conducted interviews with women /couples who had recently gone through antenatal screening for sickle cell or thalassaemia and documented their experiences in a publication known as ‘Parents’ Stories’. These stories raised a number of screening issues for: the public (present early in pregnancy, contact maternity and specialist counselling centres directly or your GP); and for screening providers (provide direct access to Specialist Nurse Counsellors to known ‘at-risk’ couples). As a result of parents’ feedback, the Screening Programme was able to revise its system of ‘fast-tracking’ the screening pathway for couples at risk of having an affected baby as well as its screening handbook.

The main work for the current commission has been to do a lot of community outreach to raise screening issues from the Parents’ Stories, raise awareness of newborn care for affected babies and the National Haemoglobinopathy Registry. We have also conducted consultations with health professionals and other stakeholders to include their feedback in a new edition of Sickle Cell Disease in Childhood: Standards and Recommendations for Clinical Care (‘Paediatric Standards’) which is currently on our website. We also did another consultation with parents and are using their feedback now as we update a Parent’s Guide to Managing Sickle Cell Disease’. SCS /UKTS helped review and update sickle cell and thalassaemia counselling competencies. We produced four lovely posters to raise general awareness of the two conditions and screening but cannot distribute these yet because of the pandemic!
What would you say have been the major achievements of NHS SCT Screening Programme to date?

In 2001, the NHS SCT Screening Programme was established as the world’s first linked programme of antenatal and newborn screening for sickle cell and thalassaemia. That’s a major achievement to develop and implement a successful Screening Programme that is respected and used as a model for other screening programmes across the world! 2016 was the 10th anniversary of full roll-out of newborn screening and 7 million newborn babies had been screened for sickle cell and just over 3,600 diagnosed with sickle cell. I think another achievement is the fact that the Screening Programme invested in outreach (via SCS/UKTS) which has really raised awareness of sickle cell and screening among high risk populations who as a result are able to make informed reproductive choices.

When I started working with the Programme, 360 babies were born annually with sickle cell and that number is now down to 270. I believe a lot of that is due to couples at risk of having an affected baby being able to access screening, counselling, prenatal diagnosis and good information provided by the Programme which has enabled them to make their own informed choices.

What are you currently working on in relation to screening?

Currently I am working on finalising the publication and launch of new editions of the Parent’s Guide to Managing Sickle Cell Disease’ and ‘Sickle Cell Disease in Childhood: Standards and Recommendations for Clinical Care’.

Jointly with UKTS, we are helping the Screening Programme to revise an E-learning resource that was produced for health professionals working on the screening pathway. We are part of a group writing a publication on the sickle cell and thalassaemia counselling competencies which is to be submitted to ‘Nursing Times’.

Longer-term, what would you describe is the main aim of the programme?

Some time ago, a slogan associated with the Programme was ‘screen to care’ which in a way meant that screening was the precursor to affected babies ultimately going into appropriate care. As such, I would say the Programme’s long-term aim is to ensure that, as a result of the various screening pathways, they achieve the lowest possible childhood death rate and minimise childhood morbidity.

What do you think are the main challenges and how can they be overcome?

The pandemic has definitely created challenges for our work as social distancing and shielding directives have meant us having to create new ways of working virtually to engage our user group. This has its advantages and disadvantages in that as no travelling out of your home is involved we get more attendance, however not everyone is familiar with Zoom or Microsoft Teams and nothing beats face-to-face in my opinion! With regards to our project, we produced something called a ‘restore’ document outlining how we planned to mitigate against the changes brought by the pandemic. I also think the pandemic has created funding challenges not only for charities like ours whose community fundraising was terribly affected by lockdown but also for the NHS as a whole.
How do you find collaborating with UKTS and PHE?

My working relationship with UKTS and PHE has been good, despite my having to build new relationships during the project as a result of staff changes in both UKTS and PHE.

Screening currently focuses on pregnant women (and partners where necessary) but ideally people should know their carrier status much earlier. What steps can we take to do this?

Yes, it’s good for people to know their ‘carrier status’ or haemoglobin genotype much before the antenatal screening stage as this enables more informed choice and for some even when choosing partners perhaps! This is particularly relevant if you know of family members who carry the sickle cell gene and you are thinking of starting a family yourself. In which case one should speak to their GP about being tested or go to one of the NHS Sickle Cell & Thalassaemia Screening Centres listed on our Society website.

Some churches (certainly in Africa) do pre-marital counselling to raise this awareness of knowing your carrier status before marriage and some years ago I worked with a church in South London which had a ‘JB4’ ‘Just Before Marriage’ campaign, although I still think that’s too late as you are at the verge of walking down the aisle by then! On 4 and 5 November 2008, the Screening Programme hosted an international residential conference in Greenwich to discuss the feasibility of developing ‘pre-conception’ screening in the UK. I remember the date as that was when Obama got elected and many of us hardly slept on the 4th! The Programme did come up with a preconception screening policy and I recall it had clear recommendations on how and when it is most effective to screen pre-conceptually for genetic disorders like sickle cell.

Tell me about your personal interests.

If I were to use two words to define my personal interests I would say ‘heritage’ and the ‘arts’, mainly performing arts for I have always liked theatre, mostly African plays which you find mainly in the fringe theatre sector, although more recently these plays are put on at the National Theatre. With ‘heritage’ I mean history and culture and I go to museums such as the British Museum, Horniman and Museum of London if they hold artifacts or exhibitions I am interested in. In fact my interest in my Sierra Leonean ‘Krio’ heritage and its important link to British history meant I initiated and co-curated an exhibition currently at the Museum of London Docklands in their London, Sugar and Slavery gallery. The display is called ‘The Krios of Sierra Leone’ and I even managed to sneak in a bit on sickle cell too to raise awareness, definitely to lots of people who would never have heard of it! I hope your readers will visit it.

Additionally, I like writing articles as I used to be on the editorial team of a Sierra Leonean magazine many moons ago. My interest in the performing arts meant I wrote reviews of plays and musical and pop concerts and often got press tickets. I remember one concert at Westminster Abbey being given by the visiting choir of the Sierra Leone Blind School (it was initially meant to be in Methodist Central Hall but the venue got upgraded) and when I mentioned I was covering the concert in our magazine, I was marched right up to the front and told I was being sat where Tony Blair normally sits!

Any longer-term personal aspirations?

Still too young to think of retirement plans! I try to fulfil some of my aspirations as I go along. However, having an exhibition at a mainstream museum took a few years to come to fruition so in 2017 that was a ‘long-term aspiration’ which I fulfilled in 2019. Not sure of the next long-term goal yet!

Photo (courtesy of Museum of London): Iyamide preparing for the Krio exhibition
The NHS Sickle Cell and Thalassaemia Screening Programme

Before we begin, you may actually be wondering: “What is screening?” According to a definition provided by PHE, screening is defined as “identifying apparently healthy people who may be at an increased risk of a disease or condition, enabling earlier treatment or informed decisions.” PHE is the home of all the NHS-led national screening programmes. The NHSSCTSP is one such programme. As most of you know, SCD and thalassaemia are severe genetic blood conditions that can be passed on from parents to children through altered haemoglobin genes.

The NHSSCTSP offers antenatal screening to identify carriers of SCD and thalassaemia. They are then able to facilitate counselling and PND and ensure affected babies are seen by health care professionals in a timely manner. This programme is unique as it is the only screening programme which allows collaboration between national patient societies and Public Health England. UKTS and SCS work closely with the NHSSCTSP in areas such as patient engagement, public outreach, lobbying and campaigning, media support, policy and resource development.

The over-riding aim of NHSSCTTSP is to improve the quality of care of pregnant women, babies and families with sickle cell or thalassaemia in England. This is broken down into the following programme objectives:

- ensure a high quality, accessible screening programme throughout England,
- support people to make informed choices during pregnancy and ensure timely transition into appropriate follow-up and treatment,
- improve infant health through prompt identification of affected babies and timely transition into clinical care, and
- promote greater understanding and awareness of the conditions and the value of screening.
User focus

One of the key benefits of involving SCS and UKTS in the screening programme is having access to their strong relationships within their respective patient populations. This helps to ensure user needs are heard and met. According to PHE: “Patient Societies are a valuable resource for health care professionals as they can work very flexibly within their respective communities, from a culturally sensitive perspective, thus gaining trust and depth of information from their service users. This provides assurance to NHSSCTSP that work streams are informed by the user perspective.”

In other words, the collaboration ensures the screening programme provides a service that is underpinned by the needs of its users. In order to improve the service user experience, facilitate informed decision making and minimise childhood morbidity, some projects such as The Parents’ Handbook (sickle cell) and updating Counselling Competencies, aimed at haematology nurse specialists, midwives and genetic counsellors (guidelines and criteria used to access the counsellors’ competence in providing counselling/advice to their patients) have been undertaken.

Get screened NOW!

At the moment, screening for thalassaemia in the UK focuses on pregnant women. However, testing for thalassaemia can be done at any time in a person’s life. At UKTS, we feel you should know your thalassaemia status before you embark on parenthood; we would like to encourage everyone to ask their GP for a test. It really is that simple!

Programme background and milestones

Before the programme was established, some regional health centres attempted to offer local screening programmes and counselling services. This led to a disjointed, ad-hoc service with many cases of babies with thalassaemia not being diagnosed during pregnancy. The Programme has successfully brought these under one umbrella with a coherent and nationwide strategy for antenatal and newborn screening for SCD and thalassaemia. It has not been an easy journey for two pivotal figures in the screening world; Dr. Allison Streetly OBE former Programme Director and the RT Hon Dr John Sentamu, former Archbishop of York and first Chair of the Advisory Group from 2003 to 2013. Together, they were relentless in their quest to get the NHS to recognise the importance of regulated haemoglobinopathy screening.

The very first sickle cell and thalassaemia centre was set up in Brent, North London in 1979. In 1981 Central Middlesex Hospital became one of the first hospitals in the UK to implement universal newborn screening. This was after the hospital found that selective antenatal screening led to some babies not being identified as having thalassaemia. Progress continued into the 1990s with universal screening being recommended in high-prevalence areas. In 2000, the NHS Plan outlined proposed changes to the healthcare systems which stipulated that by 2004, there would be a national linked antenatal and neonatal screening programme for haemoglobinopathies.
The NHS Haemoglobinopathy Screening Programme was formally set up in 2001 and in 2003, it was rebranded as the NHS Sickle Cell and Thalassaemia Screening Programme to encourage inclusion and understanding. By 2004, universal antenatal screening was being rolled out across the country.

The focus of 2005 was communications. The Programme developed an integrated communications strategy which involved: producing publications to reach out to the general public; strengthening media links to lobby around the importance of screening; and raising awareness of thalassaemia and SCD. The year 2007 saw the introduction of the Public Outreach Strategy which prompted the Screening Programme to enlist three health promotion agencies to target Pakistani, Bangladeshi and African communities. Another key year was 2008, as antenatal screening was formally offered in all trusts throughout England. In order to assess its effectiveness, in 2009 the Screening Programme commissioned an independent evaluation of its operations. The findings described the programme as “complex, innovative and dynamic.”

In 2010, a new project was started to monitor and improve the linked antenatal and newborn screening programme for thalassaemia and SCD. During the next three years, annual reports were completed along with expanding literature for service users, rewriting of the laboratory handbooks and programme standards. In 2013, the screening programme was restructured as part of Public Health England, with Cathy Coppinger as Programme Manager. In 2016, PHE decided to group together the projects for the screening programme. With both UKTS and SCS in agreement, a new partnership between the societies was formed. Following this, the societies then applied for a bid to collaborate with the NHSSCTP and once approval was granted, the new project began.

Part of the initial work required UKTS and SCS to conduct interviews with a cross-section of families with newborn babies and recent pregnancies. The rationale behind the ‘Parent Stories’ publication was to share the experiences - whether good or bad - faced by parents who had undergone a recent diagnosis. The Parent Stories publication unearthed heart-breaking stories which exposed the failures in the system. A conference was then held in Birmingham in March 2018 to disseminate the findings and further investigate the causes of late offers of antenatal screening and prenatal diagnosis. The outreach work was re-advertised in 2018 and the tender was again awarded to UKTS and SCS.

As well as the extended work identified in Patient Stories, the new focus also included newborn screening. In 2019, Cathy Coppinger retired, and Amanda Hogan was appointed as the new Programme Manager.

Challenges

As mentioned previously, it hasn’t always been a smooth journey and there have been numerous obstacles along the way. Some of the most significant ones are:

Recognition

When it was first introduced, in low prevalence areas, screening was not seen as being of major importance. In order to combat this, the programme advocates had to find local clinicians to champion the cause in their areas. Whilst there have been some improvements noted, there are still regional variations resulting in inconsistencies. Eradicating these differences remains a priority of the NHSSCTSP.

Meeting timelines

Timeliness of screening has been a challenge historically. The aim is for the blood test for thalassaemia to be administered before the mother reaches 10 weeks of pregnancy. Obviously, this gives time to test the father and to then conduct diagnostic tests on the unborn baby if necessary. The standard set by PHE for the entire screening process to be completed within is 12 weeks + 6 days from the date of conception. Perhaps, just as importantly, it
allows the couple time to consider their options and make informed decisions at what may be a difficult time. If the unborn baby is identified as having thalassaemia major and the couple decide to terminate the pregnancy, this process of abortion needs to be done as soon as possible. However, as mentioned, there are still regional variations in early access to screening and addressing this issue is a current priority for NHSSCTSP.

**Father engagement**

It is not always easy to involve fathers in the screening process which can pose problems if they do not get screened. There are several factors which play a part: fathers are often receiving information via the mothers and it may not always be clear to them why they need to be screened; they may be registered at a different GP which can pose practical problems; information about antenatal screening tends to be aimed at women; and there may be cultural factors at play.

The table below (provided by PHE) shows the number of father samples requested, available and the percentage uptake of father testing for 2017-18.

<table>
<thead>
<tr>
<th>Region</th>
<th>Father samples requested</th>
<th>Father samples available</th>
<th>Uptake (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>London</td>
<td>5,361</td>
<td>2,947</td>
<td>55.0</td>
</tr>
<tr>
<td>Midlands and East</td>
<td>3,580</td>
<td>2,449</td>
<td>68.4</td>
</tr>
<tr>
<td>North</td>
<td>2,375</td>
<td>1,681</td>
<td>70.8</td>
</tr>
<tr>
<td>South</td>
<td>1,823</td>
<td>1,367</td>
<td>75.0</td>
</tr>
<tr>
<td>England total</td>
<td>13,139</td>
<td>8,444</td>
<td>64.3</td>
</tr>
</tbody>
</table>

According to PHE, the uptake of biological father testing was stable between 2016 to 2017 and 2017 to 2018, following a small increase in uptake in both high and low prevalence areas in 2016 to 2017.

**Inadequate IT infrastructure**

This has been a major challenge for the programme historically as the success of the national programme was also highly dependent on a robust IT system. Additionally, some hospitals use different IT systems and programmes which impact on the delivery, the ability to properly monitor and quality check the programme; resulting in inconsistencies in reporting. Work has been done in this area, but it is still very challenging.
Recent and current projects

With the coronavirus pandemic, there has been considerable disruption to the Outreach Project workplan. However, based on the scope of work identified in the weekly e-meetings held with the SCS, UKTS and PHE (led by Amanda Hogan), the group have managed to adapt the workplan to continue to prioritise prenatal and newborn screening in England.

National Haemoglobinopathy Register (NHR)

The NHR is a confidential database comprising of people living with sickle cell disease and those living with a moderate to severe form of thalassaemia in England. It is optional for patients to be included on the Register and for various reasons, many patients and their families are not willing to be included on the register. However, it is intended that going forward, patient consent will only be required if their data is going to be used in research. By having more patients on the register means that data is more meaningful and an accurate representation and enables services to be commissioned that meet the needs of users.

Considerable work (health professional talks, parent support groups, patient education days) has been undertaken by the charities to make patients aware of the benefits of enrolling on the NHR and how their personal data will help commissioners to target resources and plan better services around their care.

Social media campaigns

Since lockdown, the group has met virtually on a weekly basis to discuss ways to adapt projects to ensure core milestones are still met. The major impact of the coronavirus has been to temporarily halt any planned outreach projects involving face-to-face contact. To combat this, the charities have developed posters and run social media campaigns to raise awareness and successfully increased their online presence.

In fact, as digital technology and the use of social media continues to rise, the charities have increased their social media posts on Facebook, Twitter and Instagram to target activities outlined in the project such as:

• the importance of early screening in pregnancy,
• the importance of newborn testing, and
• the importance of having patients enrolled on the NHR.
Medical and support

Newborn Outcomes (NBO)
The system is an automated system that supports the referral of babies diagnosed with sickle cell or thalassaemia into treatment. SCS and UKTS have been using their various networks to raise public awareness of all the relevant NBO system issues such as the 3-month pathway into care, penicillin prophylaxis and immunisation, especially in high-risk communities. The NBO also links into the National Haemoglobinopathy Registry (NHR).

Revision of E-learning material
During lockdown, the decision was made to include SCS and UKTS in the revision of PHE’s e-learning material, which is used to train specialist haemoglobinopathy nurses, midwives, genetic counsellors and other allied health professionals. SCS and UKTS utilised their knowledge and experience gained over the years from speaking to families and healthcare professionals to provide valuable input. They also updated the e-learning units to include factors such as culture, ethnicity and religion.

Update of educational resources
Guidelines on the offer of counselling and prenatal diagnosis were revised, and the Programme introduced new measures to monitor the implementation of performance standards. UKTS and SCS have supported the Programme Standards by engaging with their extensive public and professional community networks to raise awareness of the pertinent issues and by bringing user expertise to policymaking committees.

Outlook
Clearly, much has been achieved by the programme over the years. Important work continues to be undertaken with critical future projects planned. The weekly e-meetings have ensured the important work of the screening programme has continued to progress into the next phase, in spite of the pandemic. Whilst the programme hopes to return to full, normal services soon, it is clear that the collaboration can operate successfully in the event of another lockdown or tighter restrictions.

Acknowledgements
Thank you to Dr Elizabeth Dormandy for fact-checking this article and also providing information about the history and development of the project.

Having completed a PhD in Health Services Research at King’s College London, Dr Elizabeth Dormandy started work in the laboratory at the Whittington Hospital where she first became interested in antenatal screening. As a consequence, Elizabeth became involved in research looking at how to support women and their families in making informed choices about screening decisions. This in turn led to her taking on Quality Assurance for the sickle cell and thalassaemia screening programme. She retired from the screening programme in 2015. Since then she has been the chair of the advisory group for the outreach work. Her contribution to the screening programme has been immense and we would like to thank her for her unfailing commitment and tireless work.

Thalassaemia and the Screening Pathway

In this article we look more specifically at why we should all be screened. In order to do this, we need to understand how a child is born with thalassaemia and also how prevalent thalassaemia is in England. We then turn our attention to the current screening pathway for thalassaemia and how the process actually works in practice, at the individual level.

How is a child born with beta thalassaemia major?

To really understand why screening is so important, it is worth recapping what the likelihood is, of having a child with beta thalassaemia major. Thalassaemia is a genetic condition, which means it is inherited from parents. It is passed from parents to children through genes. A gene is a small packet of information that controls a characteristic in your body, by making very specialised protein molecules. It is a section of DNA, the unique molecule that makes up your chromosomes. Genes make us who we are; they determine our characteristics, for example, the colour of our hair, our height and so on. Genes work in pairs: for everything we inherit, we get 1 gene from our mother and 1 gene from our father. In some cases, a gene can be missing, defective or altered from the usual form. This can cause disease. In the case of a person with beta thalassaemia major, they have inherited two altered ß-globin genes from their parents (one from the mother and one from the father). As a result of the altered – sometimes termed ‘mutated’ genes, the person cannot produce enough ß-globin, which forms part of haemoglobin. This is the substance in the red blood cell that carries oxygen.

Illustrated example:
Aisha and Hassan each have one gene that makes the usual amount of red blood cells and one altered or ‘thalassaemia’ gene that makes less red blood cells than usual. Aisha and Hassan are thalassaemia carriers. Being a carrier is sometimes also referred to as having thalassaemia trait or thalassaemia minor. People who are carriers are well in themselves. They are healthy and in fact they may not even know they are carriers. But they can pass on the altered gene to their children. A child can only get beta thalassaemia major if they get TWO altered genes - one from their father and one from their mother.

Each time Aisha and Hassan have a baby, there is a:
- 1 in 4 (25%) chance that their baby will inherit beta thalassaemia major.
- 1 in 2 (50%) chance that their baby will be a thalassaemia carrier.
- 1 in 4 (25%) chance that their baby will be completely free from thalassaemia.

These chances are the same each time Aisha and Hassan have a baby. Their next child could have beta thalassaemia major like their child Imran, be a carrier or be completely free from thalassaemia.
The illustration below shows how they passed on the condition to their son Imran:

How prevalent is thalassaemia?

It used to be the case that thalassaemia was confined to certain geographic regions. This is because it initially developed as an evolutionary response to malaria; thus it mainly affected people from malarial regions such as Asia and South East Asia, the Mediterranean, South America, the Caribbean, Northern and Central and the Middle East. If a person was a thalassaemia carrier, it appeared to give them some protection against malaria. The human body had developed an effective evolutionary defence but had not anticipated what might happen if two carriers had children. This is where the issue arose, in the form of beta thalassaemia major.

In fact, it is estimated that 300,000 babies, severely affected by thalassaemia, are born globally each year. Sadly, this underestimates the real number because in many poorer countries, cases are not always reported. Furthermore, there are a staggering 80-90 million thalassaemia carriers across the world, representing 1.5% of the global population.

Whilst it was initially prevalent in the regions mentioned, this is no longer the case and anyone can be at risk. Due to several socio-economic and demographic changes - for example, the migration of communities over the centuries and the rise of inter-racial families - thalassaemia is no longer confined to people from certain ethnicities. This is why everyone should get tested. As said earlier, because carriers are generally healthy, you may not know if you are a carrier unless you have the test.
According to the most recent figures produced by the National Haemoglobinopathy Registry (www.nhr.nhs.uk), in 2018-19, there were 1,516 people living with various forms of thalassaemia in England. Of these, 890 were reported as having beta thalassaemia major.

To see the full data report, visit http://nhr.mdsas.com/wp-content/uploads/2019/06/NHR_AnnualReport201819.pdf (Please bear in mind that these figures likely underestimate the total number as not all thalassaemia patients are recorded on the Register; it is optional for patients to be included and not all want to be).

**Thalassaemia patients by ethnicity 2018/19**

Looking at the bar chart which shows thalassaemia patients by ethnicity, it is most prevalent amongst the Pakistani community in England; this sub-group had 410 patients, accounting for 27% of all thalassaemia patients in 2018-19. Also significant were the Indian (171 patients, 11%) and Bangladeshi (102 patients, 7%) populations.

As the map shows, the distribution of thalassaemia among different ethnic groups is reflected in the geographic spread across England; in other words, there is greater representation in areas with higher concentrations of these ethnic groups – predominantly, London, West Midlands and Yorkshire.

None of this is surprising or telling us anything we did not already know. What is of interest, from the bar chart above showing a breakdown by ethnic groups, are the last four bars: White – British (63 patients); White- Irish (1 patient); White and Asian (9); and White and Black Caribbean (8). While these sub-groups only represent 5% of all thalassaemia patients, they show that you cannot assume that it is confined to certain groups or ethnic backgrounds. This minority reflects the demographic trends mentioned earlier; namely the increase in migration and inter-racial families. This is why we urge everyone to get tested.
In 2018, 15 babies were registered as having beta thalassaemia major in England. This compares to 24 in 2011 and hopefully reflects the success of the screening programme. Looking at the table below, overall, the trend has been downward with an average of 16.5 babies born with thalassaemia per year between 2011-2018.

<table>
<thead>
<tr>
<th>Year</th>
<th>No. born</th>
</tr>
</thead>
<tbody>
<tr>
<td>2011</td>
<td>24</td>
</tr>
<tr>
<td>2012</td>
<td>27</td>
</tr>
<tr>
<td>2013</td>
<td>19</td>
</tr>
<tr>
<td>2014</td>
<td>11</td>
</tr>
<tr>
<td>2015</td>
<td>7</td>
</tr>
<tr>
<td>2016</td>
<td>14</td>
</tr>
<tr>
<td>2017</td>
<td>15</td>
</tr>
<tr>
<td>2018</td>
<td>15</td>
</tr>
</tbody>
</table>

Source: NHR Annual Report 2018/19

The screening pathway

We have discussed how thalassaemia is passed on between generations and why we should all be tested. Let’s turn our attention now to the actual screening process and what is involved.

A full diagrammatic representation of the screening pathway is shown on page 31.

At her first appointment with the midwife, the pregnant woman will be asked to complete a Family Origin Questionnaire (FOQ).

The aim of the FOQ is to identify the population groups at highest risk of thalassaemia, sickle cell, and other haemoglobin variants. All women – whether they are in high or low prevalence areas – are offered screening for thalassaemia.

The actual screening test for thalassaemia, to find out if you are a carrier, is a simple blood test taking just a few minutes. The test should be offered to all pregnant women before they reach 10 weeks of pregnancy. It's really important the test is done early. The result of the blood test is back within a week.
If the test shows the mother is a carrier, it is important that the baby’s biological father has a test to see if he is also a carrier as soon as possible. Babies can only inherit the condition if both parents are carriers. If tests show the father is not a carrier, the baby will not have a severe form of thalassaemia and the mother will not be offered further tests in pregnancy.

However, it is important to note that there is still a 50% chance that the baby could be a carrier. This is extremely important because when the baby is older, he or she could pass on the unusual gene to his/her children. It is therefore worth trying to find out if the baby is a carrier early on in his or her life. Parents can discuss the implications of their baby being a carrier with their GP or a healthcare professional at their local sickle cell and thalassaemia centre.

**What happens if both parents are carriers?**

If the blood test reveals the father is also a carrier, the parents will be offered pre-natal diagnosis (PND) in the form of diagnostic tests to determine if their unborn child will have beta thalassaemia. As we have seen, when both parents are carriers, there is a 1 in 4 (25%) chance that the baby could inherit the condition. A diagnostic test will show if the baby has thalassaemia, is a carrier or if the baby is completely unaffected. Ideally, PND should be performed by 12 weeks and 6 days of pregnancy. The PND method chosen will depend on the gestational age of the foetus (how many weeks of pregnancy have passed) and will be one of the following:

**Diagnostic test: Chorionic villus sampling (CVS)**
This is usually performed from 11 to 14 weeks of pregnancy. A fine needle, usually put through the mother’s tummy, is used to take a tiny sample of tissue from the placenta. The cells from the tissue can be tested for thalassaemia.

**Diagnostic test: Amniocentesis**
This is done from 15 weeks of pregnancy. A fine needle is passed through the mother’s tummy into the uterus to collect a small sample of the fluid surrounding the baby. The fluid contains some of the baby’s cells, which can then be tested for thalassaemia.

Results should be communicated to the woman or couple within 5 working days of the PND test. As these diagnostic tests are invasive, they carry risks and about 1 in 100 diagnostic tests result in a miscarriage (the loss of the pregnancy in the first 23 weeks). There are also some other risks, such as infection or needing to have the procedure again because it was not possible to accurately test the first sample.

If diagnostic tests show the baby has beta thalassaemia major, the parents then have to decide whether to terminate the pregnancy (have an abortion) or to continue and give birth to a child that will have a serious, lifelong health condition.

The sickle cell and thalassaemia screening pathway

Sickle Cell and Thalassaemia screening (linked antenatal / newborn programme)

Identify eligible population

Provide information and offer screening

Decline

Accept

Low prevalence trust

High prevalence trust

Known carrier couples

Send completed FOQ marked ‘declined’ to the laboratory

Take sample and send to laboratory with completed family origin questionnaire (FOQ)

Laboratory tests samples as per national guidelines and reports results as per local arrangements

Nothing abnormal detected

Inconclusive result

Carrier result

Offer screening to baby's father

Baby's father available and gives consent

Laboratory tests sample and reports results as per local arrangements

Nothing abnormal detected on screening of baby's father

Confirmed carrier or affected result in both parents - at-risk couple

Offer pre-natal diagnostic (PND) testing

PND accepted/contact PND laboratory to arrange for analysis and send samples

Decline

Inform parents, community midwife and GP

Result reported to referring clinician

Carrier or normal result

Baby affected by a major haemoglobin disorder

Provide information and offer choice

Continue pregnancy - include results on blood spot card

Go to Newborn Blood Spot Screening

Offer counselling and follow up support

Discontinue pregnancy

Source: PHE
Counselling and support

These are not easy choices or decisions to make and it is undoubtedly a very stressful time for the parents. Some parents decide not to conduct PND for a number of reasons. For example, it may go against their cultural beliefs, or they may not want to take the risk, albeit small, of miscarriage. Support is available throughout the screening process. If the mother is found to be a carrier of thalassaemia, she will be contacted by a specialist nurse or midwife counsellor for genetic counselling to discuss the result and implications. If the blood tests reveal that both parents are carriers, then they will be offered further counselling to help them through a very difficult time. If the result shows the baby has thalassaemia, the parents will be offered an appointment with a health professional to get information about thalassaemia and talk through their choices. There is also support available from the charity Antenatal Results and Choices https://www.arc-uk.org

Timing is critical

Early access to screening and PND can be critical for couples at increased risk of having a child with thalassaemia. By having the test early, expectant parents can explore and understand all possible options and make informed decisions if their baby is at risk of being born with beta thalassaemia major.

The NHS Newborn Blood Spot Screening Programme

The NHS Newborn Blood Spot Screening Programme uses the heel prick test – where a few drops of blood are taken from the baby’s heel when it is 5 days old – to test for 9 different health conditions, of which sickle cell disease is one. By identifying any of these conditions early on, treatment can be given promptly. Although thalassaemia is not one of the conditions tested for in the Newborn Programme, if a baby has beta thalassaemia major, it will be detected and reported as part of the process. However, the Newborn Screening Programme cannot identify carriers of thalassaemia until they are over approximately one year of age because their carrier status is often

Summary: why is screening so important?

By screening you are essentially providing people with important, possibly life-changing, choices:

• It allows parents to find out in the early stages of pregnancy if there is a chance their child could be born with thalassaemia. As described earlier, this chance would arise if both parents are tested and found to be thalassaemia carriers.
• If both parents are found to be carriers, they can then make the choice of whether to proceed with PND to ascertain if their unborn child will have thalassaemia.
• If they find out that their unborn baby will have thalassaemia, they then have the option to terminate the pregnancy. In this way, it can help to eradicate this serious, lifelong condition.
• They can choose to take up the offer of counselling to advise them through this difficult process and ease the stress at what can be an extremely difficult time.

Additionally, screening enables healthcare professionals to make special arrangements for care during pregnancy if necessary, and for couples to prepare and plan ahead if they know they are going to (and continue to) have a baby with additional medical needs in the form of beta thalassaemia major.
The aim of sickle cell and thalassaemia screening in pregnancy is to identify people who are carriers of unusual haemoglobin genes and other haemoglobin disorders so that counselling and screening of the father of the baby can be offered. Following on, diagnostic tests can then be offered to couples at risk of having a child with a major haemoglobin disorder in a timely manner, to find out if the baby has inherited the condition. The parents can then make an informed decision whether to continue with the pregnancy or terminate.

Anyone can be a carrier for a haemoglobin disorder but it is more common among people whose ancestors are from Africa, the Caribbean, the Mediterranean, India, Pakistan, South and Southeast Asia and the Middle East. Sickle cell disease and thalassaemia are genetically inherited and affect haemoglobin, the substance in the blood that carries oxygen around the body. People who have these conditions need specialist care throughout their lives.

In Leicester, sickle cell and thalassaemia counselling is offered in our Specialist Nurse-Led Clinic which is run from Monday to Friday in the Hospital Outpatients’ Department. Referrals to the service are received from several different routes, such as: the newborn screening laboratory in Sheffield; antenatal screening / booking blood results from the laboratory; self-referrals; fertility clinics; local GPs; and any abnormal haemoglobinopathy result from the laboratory.

The Sickle Cell and Thalassaemia Service is based within the University Hospital of Leicester at the Leicester Royal Infirmary. Leicester is a high prevalence area, therefore all pregnant women booking with a midwife are offered screening for SCD and thalassaemia at their very first point of contact. This screening blood test is offered by the community midwives along with the other routine antenatal screening tests. The blood samples are then processed by the Special Haematology Laboratory. All results that are clinically significant, including unidentified haemoglobin variants are securely sent by the laboratory to the Sickle Cell and Thalassaemia Service.

The Sickle Cell and Thalassaemia Team invite the woman to attend a nurse-led clinic with our Specialist Nurse Counsellors - Rebecca Sekyere and Sarah Barnes to discuss her result, within five working days. The appointment letter advises that she attends the appointment with the father of the baby so that he can be
offered haemoglobinopathy screening. The NHS Sickle Cell and Thalassaemia Programme information booklet for fathers invited for screening is enclosed with the appointment letter. The Specialist Nurse Counsellors ensure that counselling about the implications of the result, and screening of the father is offered and performed as soon as possible.

If the father of the baby’s screening result suggests clinical risk to the baby, the Specialist Nurse Counsellor will invite the couple to an appointment usually on the same day, but within three working days. During this appointment, the Specialist Nurse Counsellor will discuss with the couple the risk of their baby having a major haemoglobin disorder and the choices that are available to them, including prenatal diagnostic testing. To recap, when both parents are carriers of unusual haemoglobin there is a: 1 in 4 chance the child will not have the disease or carry the gene; 2 in 4 chance the child being a carrier of the gene; and 1 in 4 chance the child having the disease.

If the biological father of the baby is not available for screening, a discussion takes place with the pregnant woman, of the risk of having a major haemoglobin disorder along with the various choices that are available.

The couple are given the PHE Screening Programme booklet – ‘information and choices for women and couples at risk of having a baby with thalassaemia major’ (or sickle cell version as appropriate). Having supported the couple to make their own informed choices for their pregnancy, the Specialist Nurse Counsellor will then coordinate prenatal diagnostic testing, if this is the preferred option. The Counsellor will do this by liaising with the obstetric and fetal medicine team within the trust, the cytogenetics laboratory (also within the trust), and Oxford National Haemoglobinopathies Reference Laboratory - the latter being where Leicester PND samples are sent for analysis. The results
of the prenatal testing are sent back to the Specialist Nurse Counsellors, who will then inform the couple and support them through their next decisions.

In Leicester, we are fortunate to have continuity of care as it is the same Specialist Nurse Counsellors that are sent the Newborn Screening Results from our regional Screening Service in Sheffield, and who would counsel the families regarding the results in the nurse-led clinic. Although beta thalassaemia carriers are not identified in The Newborn Screening, the newborn haemoglobin screening can often identify babies born with thalassaemia major. When a baby is identified with a major haemoglobin disorder such as beta thalassaemia through the newborn screening programme – the Sickle Cell and Thalassemia Nurse Counsellors are notified directly by the newborn screening laboratory. The specialist nurse counsellors will then contact the parents and arrange a home visit to discuss the result. The counsellors will have met the parents in early pregnancy at the Haemoglobinopathy Counselling Clinic and supported them through the discussions regarding their haemoglobin carrier status, reproductive implications and possible prenatal diagnostic choices.

This means the nurse counsellors are in the privileged position to have built a relationship with the parents from the beginning in early pregnancy, which can enhance communication when discussing the positive newborn result. This relationship, familiarity and trust can help reduce the anxiety of parents. It also puts us in a good position to support transition and hand over to paediatric services. The Specialist Nurse Counsellors liaise with the Specialist Paediatrician and the Paediatric Haemoglobinopathies Specialist Nurse to ensure the smooth transition into specialist haemoglobinopathy care within 90 days. However, in practice, here at University Hospital Leicester, this is usually done very quickly as we all work on site together. The Specialist Nurse Counsellor will attend the family’s first hospital appointment and introduce them to the paediatric team—a familiar face in an unfamiliar place.

Apart from the two main screening programmes, the Specialist Nurse Counsellors and team support officer (Daxakumari Parmar) offer counselling and family screening to individuals who are screened and found to be a carrier for any unusual haemoglobin gene. They also offer preconception screening and genetic counselling.

Haemoglobinopathy Counselling in Leicester and COVID Impact

During the coronavirus pandemic and COVID-19 lockdown, many services replaced face-to-face appointments with virtual clinics. However, our Nurse Specialists in Leicester continued to see the antenatal women who were found to be carriers of unusual haemoglobin, in order to offer screening blood tests to the fathers of the babies in a timely manner.

There were anxieties amongst some pregnant women, fearful of catching COVID-19. However, with reassurance and information about the safety measures put in place, most of them felt safe to come into hospital for their appointments despite the city being the first to have a localised lockdown in the country. The service has noted an increase in the number of pregnant women seen over the same period in 2019.
Vitamin D

Vitamin D plays an important role in regulating calcium and phosphate in the body. These nutrients are needed to keep bones, teeth and muscles healthy. This is especially important for women with thalassaemia, who are often osteoporotic and vitamin D-deficient. Vitamin D levels should be optimised before pregnancy and thereafter maintained within the normal range. It is advised to get a blood test pre-conception to ensure your vitamin D levels are within the correct range and therefore taken the necessary actions in response to the results.

Food sources of Vitamin D are limited and those that have vitamin D, have minimal amounts such as oily fish, some red meats and eggs. Some fortified products add extra vitamin D such as milk, dairy products, dairy alternatives, breakfast cereals and bread. Please be mindful that people with thalassaemia should limit their iron intake and these foods are high in iron, such as red meat, thus consumption of these foods should be moderated. Foods such as green leafy vegetables are a source of iron too, however you may not be so concerned with these as they have a lower bioavailability of non-haem iron. The main source of Vitamin D is from sunlight; therefore, between the months of October and March when it is not as sunny, a Vitamin D supplement is strongly recommended. Women with darker skins, those who don’t expose skin and those who use sunscreen regularly through the year, are recommended to have daily 10 micrograms all year round.

Calcium

Calcium has several important functions including helping to build strong bones and teeth for both the baby and the mother, regulating muscle contractions and ensuring blood clots normally. It also plays a major role in bone health and strength as a lack of calcium can lead to osteoporosis, which people with thalassaemia are already prone to. The recommended calcium intake can be met from the diet but if you are struggling to reach the recommended intake, supplements can be taken.
Dietary sources of calcium include milk, cheese and other dairy food, soya drinks with added calcium, calcium fortified milk alternatives, tofu and tahini, bread and anything made with fortified flour, fish where you eat the bones, such as sardines and green leafy vegetables such as kale, okra and spinach.

**Folic acid**

Folate demand in pregnancy is normally increased, and all women with thalassaemia are advised to receive folic acid supplementation at a dose of 5 mg/day, in order to prevent foetal neural tube defects, as well other complications. It is recommended to take a folic acid supplement as soon as you are trying for a baby and up to 12 weeks of the pregnancy. Food sources of folic acid include spinach, kale, brussels sprouts, cabbage, broccoli, beans and legumes (eg, peas, blackeye beans), yeast and beef extracts.

<table>
<thead>
<tr>
<th>Supplement</th>
<th>What is the dosage required?</th>
<th>When should it be taken?</th>
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<tbody>
<tr>
<td>Vitamin D</td>
<td>10μg</td>
<td>Once a day</td>
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<tr>
<td></td>
<td></td>
<td>Before pregnancy, during pregnancy and continued during breast feeding</td>
</tr>
<tr>
<td>Calcium</td>
<td>1250mg/day</td>
<td>Once a day</td>
</tr>
<tr>
<td></td>
<td>Supplements are normally 400-800mg - which one you buy depends on how much calcium is in your diet</td>
<td>Before pregnancy, during pregnancy and continued during breast feeding</td>
</tr>
<tr>
<td>Folic acid</td>
<td>5mg/day supplement</td>
<td>Once a day</td>
</tr>
<tr>
<td></td>
<td>This is recommended alongside a diet containing folate</td>
<td>It is recommended to take a folic acid supplement as soon as you are trying for a baby and upto 12 weeks of the pregnancy</td>
</tr>
</tbody>
</table>

*Please check with your haematology team as the recommended dosage may be different*

If you have thalassaemia and are thinking of having a baby, before you take any supplements, please speak to your GP and haematology team who can advise you on your health and nutritional needs, specific to you and your condition. Alternatively, you can ask to be referred to a registered dietician.

**Sources:**

[www.nhs.uk](http://www.nhs.uk)

Pregnancy in women with thalassemia: challenges and solutions (Petrakos et al., 2016)

[https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5019437/](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5019437/)

Effect of folate supplement on pregnant women with beta-thalassaemia minor (Leung et al, 1989)

Thalassaemia: the practical and emotional journey from screening to diagnosis

by Njinga Kankinza, Dr Chinea Eziefula and Grace Adjei-Chukwu

As part of this screening special issue, it felt important for us to address both the emotional as well as the practical steps that occur during the journey from screening for thalassaemia to receiving results that might indicate whether or not parents, children or newborns have a haemoglobin variant for thalassaemia. We have done this through exploring the emotional challenges that have been identified over the years by Grace Adjei-Chukwu, in her work as a community-based Senior Haemoglobinopathy Nurse Specialist and Genetic Lead Counsellor at the Whittington Health NHS Trust’s Sickle Cell and Thalassaemia Centre (https://www.whittington.nhs.uk/default.asp?c=10885).

This centre predominantly offers support to patients from Whittington Hospital and University College London Hospital; in particular, those living in the boroughs of Camden and Islington.

Amongst many other responsibilities, Grace coordinates and provides thalassaemia screening for mothers-to-be during pregnancy, for partners, for newborns and for members of the public who self-refer. In conversation with Grace, we gathered information about standard screening processes but also learned, through her vast experience, about some common responses people have about screening, and their results, and about the factors that influence these responses. We have also shared some information about what to hold in mind when navigating the screening process and where to seek help if you are interested in screening or any of the themes highlighted in this article.

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Njinga Kankinza is a Trainee Clinical Psychologist specialising in Haematology at Whittington Health.

Grace Adjei-Chukwu is a Senior Haemoglobinopathy Nurse Specialist and the Genetic Lead Counsellor at Whittington Health.

This article was written by Njinga Kankinza and edited by Chinea Eziefula. Valuable insights were gained through an interview Njinga conducted with Grace Adjei-Chukwu.
What do we mean by ‘screening’?
The process of testing for haemoglobin variants of thalassaemia is called ‘screening’; this is offered as standard to all pregnant women in England as part of the national antenatal screening programme. Another form of screening is also offered to newborns*. Screening is also offered to fathers of the unborn children when a mother-to-be is found to carry any thalassaemia haemoglobin variants. Additionally, screening is available to interested members of the general public who can self-refer to any screening centre or discuss the option of screening with their GP.

(For more information visit: https://www.nhs.uk/conditions/thalassaemia/carriers/.)

Although screening during pregnancy is routinely offered, it is an individual choice whether or not to take this up. Screening aims to provide parents-to-be with further information and support about the various options that are available to them during the pregnancy journey and beyond if they are identified as thalassaemia ‘carriers’ or ‘trait’ (this is someone who does not have thalassaemia major disease themselves but have genes for a thalassaemia haemoglobin variant that they inherited from their parents).

| Step 1: Identification for screening | • Anyone can request a ‘haemoglobinopathy’ screening to identify haemoglobin variants such as thalassaemia.  
• All pregnant women in the England are offered a referral for screening by their GP or midwife as part of the national antenatal screening programme.  
• Newborns are tested via the newborn screening programme. The newborn screening programme cannot identify carriers until they are over approximately one year of age but babies with thalassaemia major can be identified.  
• Biological fathers can be tested when results indicate that a pregnant woman is a carrier of thalassaemia or any other haemoglobin variant. |
|---|---|
| Step 2: Screening methods | • A blood sample is collected via a blood test (often with a genetic counsellor at a screening centre) and tested in a lab for haemoglobin variants of thalassaemia. The result are usually available within three working days, although it can take longer to feed this information back to parents-to-be.  
• For newborns, blood is taken via a heel prick to collect some drops of blood for testing. |
| Step 3: Screening results | • Results are usually available within three working days, although it can take longer to feed this information back to parents-to-be.  
• Results indicate whether or not the person tested has ‘thalassaemia trait’ or is a ‘carrier’ (meaning the person does not have thalassaemia major disease but does carry genes for a thalassaemia haemoglobin variant that were inherited from his/her parents).  
• If both parents-to-be are thalassaemia trait carriers, then their unborn child has a 1 in 4 risk of being born with thalassaemia. |
| Step 4: Support and advice | • If results indicate that both parents-to-be are carriers, then a genetic counsellor or specialist nurse/ midwife will arrange to discuss further options including information about further testing options. |
An interview with Grace Adjei-Chukwu

Who is most likely to attend screening?
Generally, pregnant women are the main clients seen for screening. We tend to screen far fewer fathers and this has been the case for quite some time - we have a poor uptake of screening offers for partners.

How has this changed during the COVID-19 pandemic?
Grace reported noticing an overall increase in screening since the COVID-19 crisis and thinks it might be due to the flexibility of telephone appointments rather than travelling to a face-to-face appointment. She also noted that some individuals have preferred to attend screening at the community centre rather than at the hospital due to concerns about COVID-19 risk.

What are some of the common reactions to screening?
There is a mix of both positive and negative responses to screening. While the reaction is not the same for everyone, here are some of the most common reactions reported by Grace:

**Denial**: Refusal to accept results that indicate a person/child does have a haemoglobin variant of thalassaemia. Grace explained that some people believe: “If I was sick, this would have already been picked up in previous blood tests – I have had many blood tests in my life”. She told us that in some instances, parents deny a thalassaemia diagnosis even when their child’s symptoms begin to show.

Grace has had to advocate for children to get the treatment and care they need; sometimes this has required involvement from children’s social care services. She described this process as extremely challenging and reported how this can potentially have a negative impact on the parent-clinician relationship. It can disrupt relationships with parents that she needs to continue to work with, in order to support their child’s long-term treatment.

**Anger**: Grace mentioned that some parents express this when questioning why their second child has thalassemia major when the first one does not.

**Blame**: A diagnosis sometimes causes tensions between couples with some partners blaming each other: “You brought it, my family doesn’t have this disease” or “You had the disease and did not tell us”.

What mostly influences these reactions?

**Knowledge and experience of thalassaemia**: According to Grace, there is a range with regard to prior information or knowledge about thalassaemia during the screening process. Some individuals have minimal to no prior understanding about thalassaemia for a wide variety of reasons. Others are interested and somewhat anxious, with some individuals consulting “Dr Google” (looking online to educate themselves) leading them to “imagine the worst possible scenarios”. These individuals are left feeling incredibly anxious because of the beliefs and images they have about their imagined future for what life with thalassaemia might mean. Some individuals have knowledge of thalassaemia because of having a child or other family member living with the condition.

**There are many individuals who lack the knowledge that 1 in 1000 Europeans carry a thalassaemia trait and that those who tend to carry the gene are not exclusively from the known world regions with high thalassaemia prevalence.**

A common misconception is the incorrect belief that a result that indicates someone is a thalassaemia carrier means they have beta thalassaemia major. Grace described often using a ‘chronological order approach’ to explain the biological aspects of thalassaemia, including: a start with the origins for the disease; explanations about how the cells of someone with thalassaemia function and the formation of haemoglobin; and finally an explanation of inheritance patterns. In doing so, she hopes to debunk any misunderstandings or misconceptions that people might hold, and support people to gain accurate and up-to-date knowledge about thalassaemia.
**Stigma and shame**: In some cases, there are those individuals who do not want to associate themselves with thalassaemia because of negative perceptions that they or those around them might have about the disease. Some individuals hold beliefs that thalassaemia is restricted to people who look a certain way for instance. Some also hold beliefs that thalassaemia only comes from those countries where thalassaemia is known to be highly prevalent (eg, Mediterranean countries such as Italy, Greece and Cyprus; India, Pakistan and Bangladesh; the Middle East; China and southeast Asia).

Such beliefs can cause people to believe that they themselves cannot carry a thalassaemia genetic variant and therefore avoid or refuse screening, or blame their partner when their child is diagnosed.

**Religion and culture**: For some individuals, the option of terminating a pregnancy is not possible because such an option would go against their religious beliefs and cultural practices.

**What are your views about the screening process and its importance?**

Grace told us she enjoys the informative, sensitive and non-judgemental space she offers to the people who come for screening. She described wishing that thalassaemia would not be associated with ethnicity because “genetically, we are all mixed”. She called on a “national campaign” to educate people on this. She repeatedly empathised with the added pressures of pregnant women undergoing screening on top of their “pregnancy journey which in itself is an exciting and anxious time”.

She emphasised her view that “screening should be done much earlier in people’s lives” and proposed screening once a child has left secondary school, college or university so that they have full information to aid with future family planning.
Navigating the screening process: Key things to remember

1. It is important to know that there is no right or wrong way to feel. The screening process can bring up a variety of intense and difficult feelings, as we have heard from Grace’s account. Although it can be helpful to hear about common reactions, it is important to also recognise that not everyone will experience the same feelings or reactions. For some, emotions may include fear, anger, sadness, guilt, shame, hopefulness, helplessness, relief, resentment, isolation or resignation. For others, none of these will apply and they will experience other responses. It is entirely normal to feel a range of emotions because they are common and natural responses to any potentially life-changing situation.

2. Coming to terms may take time and help is out there. Hearing that an individual and/or their child may be at risk of thalassaemia or receiving a result that is suggestive of a haemoglobin variant for thalassaemia can be shocking and may be challenging for a number of reasons. As such, it might also be difficult to absorb the information provided by healthcare professionals when they are talking about thalassaemia and what the screening result might mean. It may help to have a friend, relative or advocate present who can support and take notes or it may help to request a follow-up contact for answering any further questions later down the line. Specialist nurses and genetic counsellors are there to guide and support individuals, so they fully understand what thalassaemia is and what support is available to help to make informed decisions.

3. Be aware of any negative beliefs and notice how this affects actions taken. Following a screening result, some individuals experience guilt because of beliefs that they caused the illness and passed on “bad genes”. Worry and uncertainty about the long-term impact of living with thalassaemia, including concerns about coping, looking after a child with thalassaemia major and their future chances to live out their hopes and dreams, are also common experiences. Similar to the experience of people with children diagnosed with a long-term health condition such as diabetes, the diagnosis can for some parents represent numerous losses. They can be in the form of: loss of freedom and certain lifestyles resulting from adaptations that will have to be made to support their child; loss of confidence in their ability to protect their child from danger; and fears about the potential loss of the child’s life (Lowes and Lyne, 2000; Lowes et al. 2005; Mednick et al 2009).

Beliefs about screening results and perceptions about what can or cannot be done about living with thalassaemia can influence coping and help-seeking. For instance, believing that nothing can be done to help someone with thalassaemia might lead to not accessing help that is out there. While there is no cure for thalassaemia, good support for lifelong management and treatment is still the most readily available option, allowing individuals to grow up to live productive and satisfying lives. There is also support for parents and families of children with thalassaemia major and intermedia in various forms.

Photo credit: Stocksnap from Pixabay
Support

It is important to acknowledge any challenges and ask for help if needed. There are various services available:

• In need of someone to talk to about any feelings or difficult experiences? Call the Samaritans www.samaritans.org on their 24-hour free helpline. The number is 116 123.
• Consider psychology services within haemoglobinopathy services (https://www.england.nhs.uk/commissioning/spec-services/npc-crg/blood-and-infection-group-f05/specialised-haemoglobinopathy-services/) designed to support and run alongside the medical care for people diagnosed with thalassaemia. Psychology services offer an initial assessment to understand any difficulties in the first instance and may signpost to a different service if that is better suited to an individual’s needs.
• Consider accessing support from a local Improving Access to Psychological Therapies Service (IAPT) (https://www.nhs.uk/service-search/find-a-psychological-therapies-service/) where support is available for a range of emotional challenges.

The following organisations/ websites offer advice and information about thalassemia and/or screening:

• United Kingdom Thalassaemia society (UKTS) (https://ukts.org)
• Thalassaemia International Federation (https://thalassaemia.org.cy)
• NHS Sickle Cell and Thalassaemia population screening programme (https://www.gov.uk/topic/population-screening-programmes/sickle-cell-thalassaemia)

References

Public Health England (2017) NHS Sickle Cell and Thalassaemia Screening Programme Information and choices for women and couples at risk of having a baby with thalassaemia major. London

Further reading/ online guides

Public Health England Guides
Screening tests for you and your baby: https://www.gov.uk/government/publications/screening-tests-for-you-and-your-baby-description-in-brief (translated versions available via this website)
Adult carriers: sickle cell, thalassaemia, unusual haemoglobin: https://www.gov.uk/government/collections/adult-carriers-sickle-cell-thalassaemia-unusual-haemoglobin (translated versions available via this website)
NHS sickle cell and thalassaemia (SCT) screening programme: https://www.gov.uk/topic/population-screening-programmes/sickle-cell-thalassaemia
What happens when love blossoms between two people; one has beta thalassaemia major and the other is an alpha thalassaemia carrier? This is the story of Zeshan and Farah who fell in love and wanted to start a family. They tell us about the steps they took to ensure their future children would be born, free from any health conditions.

Background and careers

Farah: I grew up in Greenwich, South East London up until I was 14 then my family relocated to Ilford, East London where I have been ever since. I am a Mortgage Consultant for a High Street Bank. I have been in this line of work for almost 16 years!

Zeshan: I grew up in Brussels, Belgium until 2019 when I got married and moved to London. In terms of my education and career, I started by studying Hospitality Management at university. After that, I worked for a couple of years in a hotel as a manager and then for two years in advertising before opening my first restaurant. I am now a restauranteur; I have a small burger chain called ‘Bro’s Burger and Kitchen’ with three branches in Belgium and one branch in Istanbul.

Getting together

Farah: Both Zeshan and I were both quite settled in our respective lives before we met. Zeshan’s Uncle and Aunty live three doors away from me and told me that they had a nephew that they thought would be perfect for me, this was around April 2018! For some reason this ignited my curiosity and I wanted to know more. So, they made us exchange pictures and phone numbers and the rest as they say is history! We had the most beautiful wedding in February 2019.

Zeshan: As Farah said, we were introduced to each other through my uncle. I was traveling to London in March 2018 for the engagement of my uncle’s son and they were talking about this beautiful neighbour who lived on their street. They asked me if I would be interested in meeting Farah and whether it would be possible for us to exchange our numbers and pictures. We started communicating with each other just after the opening of my new branch in central Brussels. It was lovely as every day I was going through a new challenge with the business, I always had Farah on the phone, encouraging me and giving me advice, with her beautiful smile motivating me to be better. After some time, I travelled to London for our first face-to-face meeting and we decided to take a step forward. We got married in February 2019. I moved to London just after my marriage in February 2019 and I live in East London now.
Thalassaemia

Zeshan: I have beta thalassemia major + HbE, it’s the common Asian one. I was diagnosed at the age of six months. It’s then that I started my first blood transfusion. It was difficult because the paediatrician couldn’t understand where my anaemia came from; thalassemia is not a condition common in Belgium among the local population. My parents had not heard about my condition before. It was very difficult for them, especially for my mum. For my dad it was a bit different – for him having a condition is like having a handicap, he was ashamed of me most of the time – putting the blame on my mum. It was not easy for her as she was all alone taking care of me, until I was fully independent. Both my brother and sister are carriers.

Currently, I have my blood transfusion every 3-4 weeks, dependent on my haemoglobin level before my blood transfusion. I like to keep a good balance between my quality of life and my iron chelation. Every day I take chelation medication – deferasirox (Exjade) and I also have levothyroxine for my hypothyroidism. Since July 2019, all my treatments are managed than the UK.

Pregnancy

Zeshan: I chose UCLH at the early stage of the pregnancy by doing a self-referral. I knew that they have one of the best antenatal screening tests for haemoglobinopathies. I did not want to let the GP manage this as I was already disappointed with the way they were taking care of my thalassemia.

Farah: We have been very happy with the care that we received at UCLH – it has certainly been a unique experience, managing the midwife appointments and scans during the recent lockdown. We actually fell pregnant in August 2019 but unfortunately had a miscarriage at six weeks which was very upsetting and disappointing. The doctors said that it was very common and to not be anxious about falling pregnant again. They reassured us that it was nothing to do with Zeshan’s condition. I had blood tests to see if I carried the gene that could possibly result in our child having the same condition.

Zeshan: Before getting married, I asked Farah to do a blood test with her GP to see if she’s a carrier of any thalassaemia traits or any other blood conditions. It was really important for us, especially for me.
I didn’t want to risk our future babies having any health conditions if we could avoid them by taking some precautions. The GP did the test and the result came back that she was an alpha carrier but needed further investigation to confirm this. I spoke to my haematologist in Belgium and did genetic counselling and they both said that we are at low risk of putting our future babies in danger as I didn’t have any alpha trait in my DNA.

Farah: Once pregnant, we saw the genetic counselling doctor to make sure that we were not going to put the baby in danger. We did experience some complications in the early stages of the pregnancy and my first trimester was not easy – but by the time we reached the second trimester I felt much more like myself and started to enjoy the pregnancy. The third trimester was a bit difficult as I have low blood pressure. I was advised to take aspirin throughout the pregnancy to prevent pre-eclampsia. I also took iron supplements as my pregnancy tended to push down my HB level. Ironically due to the Covid-19 lockdown I was able to get plenty of rest and even work from home. This enabled me to relax and be pampered by Zeshan! We did go for some light walking in the early evening as exercise is very important. Zeshan made sure that I had a healthy and nutritious diet through my pregnancy – with the occasional treat! One positive of the lockdown was that I was lucky to have my husband next to me 24/7.

Zeshan: A challenge for me during the pregnancy was with my iron chelation as my haematologist wanted me to stop my chelation for a period of time before conceiving. I didn’t want to take any chances and trusted Dr Shah’s expertise in thalassaemia, so I followed her advice and temporarily stopped my chelation treatment.

Just before the birth...

Farah: We are super excited but at the same time very nervous. We cannot wait to meet our baby and begin the journey of parenthood.

Zeshan: I feel very excited and also very emotional as it was very challenging for both of us. I was scared that because of my condition, it would be hard for me to conceive but it was all perfect. I have been waiting for this moment for a very long time and now I just can’t wait to see the little baby.

...and then they were 3!

The birth and parenthood

Zeshan: Our little baby boy was, born at 16.40 on 12 August 2020 at UCLH in Camden. His name is Ilhan Malik. At birth, his weight was 2.390kg and he measured 47cm in length. A tiny little baby but so beautiful and brave.

Farah had to be induced two weeks early upon the doctors’ advice due to Ilhan being considered a small baby; this had nothing to do with my condition. The labour lasted nine hours which was amazing because we were told to prepare for a three-day labour! Farah had a completely natural birth with only gas and air for pain relief. Fortunately, I was able to be with her for the labour, (we weren’t sure I would be as there are a lot of restrictions with COVID).
Ilhan’s delivery was both smooth and super quick, much to our surprise – including the midwife’s! It was an incredibly emotional experience seeing our perfect little boy for the first time. I was able to stay for a few hours after the birth until they were transferred to the post-natal ward, no visitors were allowed beyond this point. Farah and baby stayed in for three nights as they wanted to carry out extra checks on Ilhan again due to his size. By the grace of God everything came back fine.

Parenthood has been wonderful, albeit challenging. These tiny humans take over your life but in a positive way. Even the sleepless nights! The best bit about parenting? That’s too hard to answer, there are just so many best bits! The cuddles, the way your baby gazes at you lovingly, your baby’s smell, looking down at him and knowing that he is all yours! Ilhan has completed us in every possible way and is our dream come true.

Huge congratulations to Farah and Zeshan on the birth of their beautiful son Ilhan!
Cile Kusbeci has thalassaemia trait and tells us why she takes an upfront approach when she meets potential partners. She also urges other young people to get tested.

I am 29 years old and I work in an architectural practice as a personal assistant. I have been in this role for six years, prior to which I studied Business and Management at Queen Mary University of London. In my free time I like to spend time with my family and friends; as a foodie this often involves going out to eat. In my downtime, I also like to read self-development books. My 10-year plan is to have children, have a shift in my career whereby I am working for myself, and to continue to explore the various sights and cultures of the world.

I was 25 years old when I found out that I am a thalassaemia carrier, although I suspected that I was a carrier prior to my diagnosis.

My parents were not aware that they were carriers, as neither of them had been tested. They had both heard of thalassaemia prior to my diagnosis, as back in their homeland of Cyprus, it is very common. In addition, it is present on both sides of my extended family.

Being a carrier does not affect my health in any way. However, as I would like children in the future, I am very conscious of it in a relationship context. It is important for me to be with someone who is not a carrier. I have always mentioned my carrier status early on in a relationship.

“I have always mentioned my carrier status early on in a relationship. It can come across as forward, but I believe it is something that needs to be discussed and addressed early on.”
It can come across as forward, but I believe it is something that needs to be discussed and addressed early on. In the past I have been met with resistance regarding a partner being tested. I have been told: “There’s nothing wrong with me,” and “I’m not sick, I don’t have any symptoms”. I have found that it can be a struggle to get someone to agree to being tested, even though all it requires is a blood test.

My advice to young people would be to please get yourselves tested if you envision having children. You can save yourself a lot of future anguish by simply having a blood test. Whether from a higher risk background like myself or not, I believe everyone should get tested as we are becoming ever more interracial. Living in a country that is low risk doesn’t mean you aren’t a carrier. I would recommend people to not leave it too late, or until you are pregnant to be tested. This is a condition that will affect more and more people over the coming years as we become more interracial, yet the adverse implications can be drastically limited by just being tested.

Very few people have heard of thalassaemia even if they come from a higher risk background. I have found that little is done to raise awareness of the condition like other illnesses, particularly in the western world. I think it may be useful to use social media to help raise awareness of the condition. The outreach to younger people is likely to be larger, therefore hopefully encouraging more people to get tested.

Wise words and a sensible approach Cile - thanks for sharing!
Rafay’s amazing teen spirit

As a parent, finding out your child has thalassaemia is a life-changing moment. It is often a time fraught with fear and worry about what the future holds for your precious child. We thought it would be useful to speak to a young person with thalassaemia and their parent to gain perspectives on what life is really like growing up with the condition. Having met Farzana and her lovely 14-year old son Rafay at the UKTS London conference earlier this year, I asked them if they would like to feature in the magazine. They were more than willing, so I spoke to them both. Here’s what they had to say...

Hi, I’m Rafay...

All about me...

My birthday is October 7th and I’m 14. My friends would describe me as kind and friendly. I like playing the piano and watching videos on the internet. I am happy when I am meeting my friends and doing well in school. I am sad when I am feeling sick. My friends are very fun to talk to. And obviously my parents take care of me :) I always try to remain positive and have a positive mind set.

School life

I find school fine and I’m doing well. I like french, music, history, science and religious studies because I find them fun to learn. I dislike English and maths.

I’m not the only child in my school with thalassaemia and my friends do understand what it is. They are all very supportive and I’ve told them about my condition. To them I’m still me.

For transfusions, I take half a day off school every three weeks. I’m fine with it as when I’m in the hospital I can do my homework if I have any. It does not affect my learning much as I’m only missing half a school day. Also the hospital school is great too.

I don’t have anything I find difficult at school because of thalassaemia. Sometimes I do get tired especially towards the end of my three week period and just before my transfusion. So PE can be challenging.
My message to other young thals:

Thalassaemia does not control you, you can control it and live life to the fullest.

Goals

My current goals are to pass my piano exams and explore some new countries.

I don’t have any ideas of what I would want to do for a job as of yet! I’m sure though it will come to me soon.
A chat with Farzana...

I met Rafay and he seems like a really fantastic young man, you must be very proud of him! But please tell me in your words, what makes him special?

Even though Rafay has a lifelong condition, he’s always dealt with it very maturely. He has taken responsibility and ownership as soon as he was old enough in certain aspects. For example, taking his medication on time.

Please provide a little background to your family.

My husband and I are of Pakistani origin and live in East London with our two children; an older daughter and then there’s Rafay. I have taken some time off at the moment to study but worked in finance and my husband is a businessman. No one in the family other than Rafay has thalassaemia.

Please tell us about Rafay’s medical treatment to date and what it involves?

Rafay has a transfusion every three weeks. He has a cross-match two days before each transfusion. Then there are various scans that need to take place for heart and liver scars related to iron overload. We have regular hearing and sight tests because of the iron chelation medication. Since Rafay became a teenager, we also need to attend growth and endocrine clinics.

Does Rafay have any other health issues? If so, please tell us about these and what treatment he receives for them.

We recently went through a very daunting period. Rafay has developed gallstones because of the repeated blood transfusions. This unfortunately was causing him pain and he developed pancreatitis. We have just spent about four weeks in hospital and he will need an operation soon.
Being a parent is hard work! What is it like raising a child with thalassaemia?

I try to remain positive, because life itself is challenging but I truly believe that we are not burdened with more than we can handle. Recently we had a bit of a challenge as mentioned above, regarding the gallstones, which are a side effect of the thalassaemia. I think taking care of a child with thalassaemia isn’t easy but you have to take each day as it comes. Also, it does become easier as they get older and become more independent, because they then start to take on some responsibility. I won’t allow myself or Rafay to think negatively, as that achieves nothing.

Each day that he is leading a near normal life is a massive milestone and I’d like to thank all the health professionals involved in our care. In addition, I would like to add that having access to good health care is a blessing in itself, as the better the healthcare involved in the treatment of thalassaemia the better the outcome and quality of life. My hope for the future is that there is eventually a cure for thalassaemia. This looks promising as advancements in medical knowledge are bringing us hope, especially the gene therapy trials that are taking place.

As this magazine is a screening special issue, we want to explore how the medical system tests for thalassaemia and how it can be improved. Please describe how you arrived at Rafay’s thalassaemia diagnosis.

Rafay was born at Whipps Cross Hospital in London.I have an older daughter and I had found out that I was a thalassaemia carrier when I was pregnant with her. I had a routine blood test where it was discovered I had the trait. When I was informed I was a carrier I was told that my husband would need to be tested, although I was seven months pregnant by this time, but that was also because I was abroad for the first two trimesters of my pregnancy. There was a mix-up with my husband’s blood test and we were told he wasn’t a carrier. I should add the blood test was done abroad, but the genetic nurse in the UK established that my husband wasn’t a carrier, even though she was doubtful. Given that I was told my husband wasn’t a carrier, I therefore thought there wouldn’t be an issue with my next pregnancy.

We found out that Rafay had thalassaemia from the heel prick test that was performed shortly after he was born. He was only a few weeks old when we were told by a haemotology nurse that
visited us at home. We were told before any symptoms had appeared which was an advantage as then treatment could be planned according. Also at that time, my husband was tested again and he was confirmed to be a carrier, as we had assumed after Rafay’s diagnosis. I then contacted the UKTS as I wanted to gain as much information as possible. They were a great source of help as they put us in contact with the Royal London Hospital and we were able to get a hospital appointment to see a consultant very quickly, something I'm still grateful for to this day. Once the appointment had taken place and my questions answered it was a huge relief, as it was then that I realised this was something that I would be able to cope with and that I would try and do the best for my child.

What advice would you give to parents who have just received a thalassaemia diagnosis for their child?

The initial diagnosis will be somewhat of a shock as it was for me. But with good management and newer treatments becoming available, your child should lead a near-normal life. It's also a good idea to remain informed and to gain as much knowledge about the condition. Never be afraid to ask the medical professionals any questions you want answering.

There is a lot of stigma around having a condition in many cultures, so it is wonderful that you talk openly about Rafay having thalassaemia. Please explain why you take this approach?

Thalassaemia is a condition but it doesn’t define who Rafay is, it’s a part of who he is no doubt but he’s so much more than his thalassaemia. In my mind, to hide it or to not tell people, or tell Rafay to hide his condition, will cause him to think it’s something he should be ashamed of. And having thalassaemia is not something you should be ashamed of having. It’s not anybody’s fault when they are born with a condition or become ill later on in their lives. In a perfect world we would all be healthy. Rafay is mature, unique and kind. I actively encourage him to talk about his condition to his friends and have never hidden the fact that he has the condition as to me that would be negative. I do, on the other hand, understand why some people choose not to tell others about the condition, because in certain cultures there is a stigma and it’s totally unfair and unjust. But to educate others is also important and to stand and say I’m ok even though I live with thalassaemia is ok too.

Is there anything else you might want to say that hasn’t been covered?

Children are precious and are a blessing, and perhaps parents who have those challenges in life with children that may have long-term conditions should count themselves lucky...because they’re strong enough to deal with those challenges. And those challenges are what shape us as people.

Thank you Rafay and Farzana!
Facebook birthday fundraisers
A very happy birthday and a big thank you to UKTS Ambassador, Sajid Hussain, Michael Giammalvo and Charalambos Loizou - all long-standing supporters of UKTS - who organised birthday fundraisers on Facebook. Together they raised over £1,000 for the charity! Thank you also to those of you who raised money anonymously. We are truly very grateful for all donations.

Please help us fundraise to improve the transfusion experience!
We are now embarking on raising funds to purchase vein-finding machines for thalassaemia units scattered throughout the UK. The funds will also be used to ensure that correct training is provided to staff using these machines. Transfusion can be a difficult experience, made so much harder when veins are hard to find. We have heard from adults and parents of young children with thalassaemia that sometimes it is extremely difficult to find viable veins and can take multiple attempts. As you can imagine, this must be a very distressing experience, with acute pain, bruising and trauma from having to be cannulated multiple times every few weeks.

We have had some fabulous fundraising ideas from our members so please continue to put your thinking caps on and help raise funds for this worthy cause. It really will make a huge difference. We have set up a go fund me page: https://gf.me/u/yg8gdi

Easy FREE ways for you to help
There are other really simple ways to help raise funds for UKTS, without having to spend a penny yourselves! These simple steps will ensure we can continue our valuable work for people with thalassaemia and their families.

Amazon Smile: AmazonSmile customers can now support United Kingdom Thalassaemia Society in the Amazon shopping app on iPhones and Android phones. Simply follow these instructions to turn on AmazonSmile and start generating donations.
1. Open the Amazon Shopping app on your device
2. Go into the main menu of the Amazon Shopping app and tap into ‘Settings’
3. Tap ‘AmazonSmile’ and follow the on-screen instructions to complete the process
4. Get shopping and help UKTS!

Easy fundraising: Please remember to use easy fundraising every time you shop online. You can find our easyfundraising page here: https://www.easyfundraising.org.uk/causes/ukts/

Over 4,000 shops and sites will donate to us, including all the big names like eBay, John Lewis & Partners, Argos, ASOS, Expedia, M&S, Just Eat, Uswitch and many more! This means you can raise FREE donations for us no matter what you’re buying.

If you haven’t signed up to support us yet, it’s easy and completely FREE. These donations really help us, so it would be great if you could take a moment to get started!
# United Kingdom Thalassaemia Society

**-Membership Form-**

All members receive regular email updates to keep informed about national developments and our events. Full members also have the right to vote at our AGM to decide how the society is run. (T&C apply).

Join the UKs largest community of people affected by thalassaemia

## Personal Details

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- Parent: [ ]
- Organisation: [ ]
- Other (Please state): ____________

## If you are a patient or parent, please complete below:

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Important GDPR requirement: We can only keep your details if you give us permission. Please tick yes if you would like to receive any further contact from us.

Data will be kept on our files and only accessed by UKTS staff. It will not be shared with anyone outside of the UKTS.

I agree for my personal details to be held by the UKTS and for the UKTS to use these details to contact me about their work and issues relating to thalassaemia.

Signature: ____________________________

Yes: [ ] No: [ ]

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