UNITED KINGDOM
Thalassaemia SOCIETY
matters

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...working for the thalassaemia community

In memory of Pany Garibaldinos (27/08/57 – 04/12/17)
Sorrow follows celebration as 2017 closes

From l to r Prof Bernadette Modell; Dr Mary Petrou; Dr Beatrix Wonke; Dr Emma Drasar, Pany Garibaldinos; Dr Bernard Davis; Dr Farrukh Shah; Specialist Nurse Emma Prescott and Sister Niamh Malone-Cooke

On 27th August 2017, Pany Garibaldinos became the FIRST UK thalassaemia major patient to reach his 60th birthday! Naturally, Pany's friends and colleagues at UKTS celebrated this landmark occasion, little suspecting how soon it would be followed by mourning. As Pany was never one to feel sorry for himself and detested the thought of others feeling sorry for him, only his family and a very few of his closest friends knew that he had already been diagnosed with liver cancer and did not have much time left. Pany's long and wonderful life drew to a peaceful close on the morning of 4th December 2017. Knowing him as we did, we feel sure that Pany would not want us to look back on his life and achievements with sorrow…….(continued on page 3)

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UNITED KINGDOM
THALASSAEMIA SOCIETY
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Chairman's message

As we welcome in 2018, we are reminded of both the challenges and milestones our Society has faced since inception. The UKTS will be celebrating its 40th anniversary as a registered charity on March 1st, 2018 and I must say that I am proof of the remarkable strides the society has been able to accomplish over the period of time.

I feel blessed to have progressed into adulthood without incident, married the love of my life and now the proud father to twin boys who will both be turning two shortly. I was encouraged to be the best at everything I attempted and even challenged myself to complete the London Marathon twice

My decision to join the UKTS was a conscious one made to try and equalise the treatment not only for patients in the United Kingdom, but internationally as well as I have seen first-hand, some of the many challenges being faced by patients in many countries.

I am also grateful to all our supporters and volunteers over the years and hope that we can continue to work together towards even more dynamic changes in our healthcare and treatment options. My team at the UKTS remains committed to our cause and will be marking this special anniversary with various activities, starting with two patients' conferences and a walkathon.

Feedback is extremely important and we look forward to hearing from everyone. Feel free to visit our Facebook, Twitter and Instagram pages to add your own suggestions or updates of your own activity in celebration of our milestone anniversary.

Working together to 2020 and Beyond!

Gabriel Theophanous

UKTS Mission Statement

To be the definitive source of information, education and research for those affected by or working with thalassaemia

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Facebook: https://www.facebook.com/ukthalassaemiasociety

Twitter: https://twitter.com/UKThal

Instagram: https://www.instagram.com/teamukts/

Cover story continued...

The hallmarks of his character were courage, humour and great love for his family and friends; and it is these qualities which we celebrate when we think of Pany.

For those readers who did not know him, Pany was a former Trustee of UKTS. He served several terms on the Management Committee, where he was renowned for his decisiveness, humour and straight talking. His professional life has revolved around his love of driving, first as a driving instructor and then as a driving examiner. Pany had been married to Trisha since 1984 and they have a son, Alex. Pany had been a patient at the Whittington Hospital since his early teens.

UKTS could not let the great occasion of Pany's 60th birthday go by unmarked – however knowing his unassuming nature, we decided that whatever was done would have to be a surprise. Surreptitious invitations and emails flew around until fifty of Pany's family, close friends, UKTS colleagues and medical and nursing staff both past and present were gathered at Babinondas Greek restaurant in Winchmore Hill, North London. Pany, expecting to have a quiet dinner with his wife Trisha and another couple, walked in completely unsuspecting – we had worried that he might have guessed, but the astonishment on his face spoke for itself when the room erupted in cheers and applause. Pany was amazed to see that we had got so many people together, and was especially touched to see Professor Bernadette Modell and Dr Beatrix Wonke, who looked after him from his teens right up to 2004. Also present were Dr Farrukh Shah, Dr Bernard Davis and Dr Emma Drasar who were Pany's doctors at the Whittington Hospital in the present day – and nurses from the Whittington's Thalassaemia Unit, Emma Prescott, Niamh Malone Cooke and Sandy Brody.

The evening passed with wonderful food, speeches, toasts and a great deal of laughter from all, but most of all from our guest of honour Pany himself. His friends and colleagues at UKTS will always be grateful that we had that evening to look back upon, and to remember Pany in his element – as the life and soul of the party. As one of the most senior thalassaemia patients in the world, Pany had seen it all – from his childhood when thalassaemia was barely recognised, through the years of experimental treatment and the advent of chelation therapy in all its various forms – right up to the present day. You can read Pany's amazing life story on pages 8-17.

From l to r Andy Charalambous; Christos Sotirelis; Pany and George Constantinou

Pany with his birthday cake surrounded by family and friends

MEET THE TRUSTEES

Ashkaan Bandoui (Assistant Treasurer)

Ashkaan Bandoui is a 30 year old thalassaemia patient, originally from Iran but living in London since he was two months old. He majored in graphic design at university but is currently working as a estate agent. He also enjoys playing the Iranian drum called the DAF which originates from Kurdistan and has been performing with a band for over nine years.

George Constantinou (Assistant Secretary)

George is a beta thalassaemia major patient. He is a founding member of the UKTS, having served on the Management Committee from 1976-1985 and again from 1999 to the present day. George has been a tireless campaigner on behalf of thalassaemia all his adult life and has conceived and been involved with many UKTS projects including conferences and awareness projects. He is also serving on the board of Thalassaemia International Federation (TIF). George is a hotel manager by profession; is married and has a daughter.

Anand Singh Ghattaura (Secretary)

Anand has lived with thalassaemia major for the past 30+ years. Though it has been a challenge at times, it has spurred him to succeed in life, particularly through school, university and now in a professional career. Anand has had the experience of being treated at a regional hospital with limited expertise in thalassaemia. He has been fortunate to have a great support network around him in life, and this has encouraged Anand to help others with thalassaemia and to make a difference in the community. As a life member of UKTS, Anand has seen the great work the society has done. Career wise, Anand works in the City of London in brand, marketing and communications (BMC) for one of the Big-4 accountancy firms. In the past, Anand has worked for a number of global accountancy and law firms in BMC. He has also been an active member of the Sikh community in Berkshire.

Raj Klair (Treasurer)

As a 42 year old beta thalassaemia major patient, I understand what it's like to live with the condition and undergo treatment. I want to give something back to society and I believe this is the best way. I own and run a chain of estate agencies in Yorkshire; and as an entrepreneur I understand the importance of organisation, communication and networking. I believe I can bring these key skills from my professional life to make a valuable contribution to the governance and future development of the Society.

Romaine Maharaj (Vice Chair)

Romaine's daughter Roanna is a beta thalassaemia major patient. Prior to moving to the United Kingdom in 2004, Romaine served as the President of the Society of Severe and Inherited Blood Disorders, Trinidad & Tobago. Romaine was also the Head of a mortgage division for one of the major banks in the Caribbean, her financial background spanning a period of twenty-three years. Currently she is attached to the Trinidad and Tobago High Commission in London and is also a board member of Thalassaemia International Federation (TIF).

Rahmatullah Khan Mohammed (Trustee)

Thalassaemia has always been an important feature of my life as I am both a brother and a parent of thalassaemia major patients. Having been born outside the UK, I have seen at first hand the sacrifices parents are forced to make to pay for life saving treatment for their children; and I am grateful that my children are well cared for. The staff of UKTS have always done their best to assist my family when asked; and I would like to give something back by offering whatever assistance I can to the management committee. I understand that there are currently no representatives from the Muslim communities on the Board of UKTS and I am pleased to be able to bring this perspective along with my personal experiences of being a brother and a parent.

Gabriel Theophanous (Chair)

Gabriel is a 40-year-old beta thalassaemia major patient; and has served as President / Chair of the Society for the past 6 terms. He majored in accounting and works in that field for a leading firm in London. In 2006, Gabriel became the first person in the world with thalassaemia major to complete a marathon race, a remarkable achievement. In 2013 he repeated this feat when he again completed the London marathon, raising funds for the Society and raising the morale of thalassaemia patients everywhere. Gabriel and his wife became parents with the arrival of their twin boys in April 2016.

Staff

Elaine Miller National Coordinator

Elaine Miller joined the UK Thalassaemia Society in 2002. She originally trained as a nurse and after her 2 children were born returned to study, gaining a 2:1 degree in law. During her first 9 years with the Society she was based in the London office; and was the first contact for anyone getting in touch with the Society. During her time with UKTS Elaine has represented the Society on committees and consultations; and has worked extensively on the NHS Sickle Cell & Thalassaemia Screening Programme, the peer reviews of haemoglobinopathy services, the education/ awareness programmes and the production of all three editions of the Standards for the Clinical Care of Children and Adults with Thalassaemia in the UK.

Katerina Loizi-Read Administrator

Katerina Loizi-Read is a 58-year patient who has been a member of the UK Thalassaemia Society for the last 38 years. Katerina has been a previous board member for many years and has been the Office Administrator for the past 9 years. She is the proud mother of a 20 year old daughter.

PLANNING FOR PARENTHOOD – sooner rather than later!

In 2018 more and more people with thalassaemia major are living very full and active lives– following their dreams by studying, pursuing their chosen careers and of course that which is of vital importance to most of us, marriage and children. Young people who have thalassaemia can become parents and have healthy children of their own – some of them need a bit of medical help along the way, but nowadays nearly every issue of TM features a couple of baby announcements. We have come a very long way since the days when parenthood was an impossible dream for men and women with thalassaemia. These opportunities have been made possible by advances in reproductive medicine, better chelating medicines and better diagnostic equipment such as scans which can tell us whether pregnancy is a good idea or whether there is any hidden iron lurking away in the heart and liver. And it is our old enemy, iron, which still makes the crucial difference.

Iron overload in children

As we all know, both male and female bodies go through changes when they reach puberty; and these changes are caused by the release of hormones, chemicals which prepare the body for the process of becoming a mother or father. The fertility hormones are produced by tiny glands in the brain called the hypothalamus and the pituitary; and the problem we have in thalassaemia is that these glands are extremely sensitive to iron –meaning that even slightly raised ferritin levels can cause problems. Even in childhood, long before puberty, iron overload can cause the glands to be damaged, meaning that when it is time for them to start producing the fertility hormones, they do not work properly.

Wanting children and grandchildren

This is one of the reasons that parents of children who have thalassaemia need to understand the importance of taking chelation medicine exactly as it is prescribed – and as children grow old enough, parents owe it to them to explain how important it is. In the words of one young woman: "I transitioned over to the adult hospital at the age of 16. I was taking Exjade but I would skip it quite often if mum wasn't watching me and thought nothing of it as I felt OK. The first time I saw my doctor on my own I was amazed when she told me my ferritin was too high and I would have to work hard at getting rid of the extra iron because (among other things) it could affect my chances of having children! I had no idea." So please bear this in mind - if you have a child with thalassaemia, the prospect of having grandchildren probably seems very distant – but when the time comes for your son or daughter to get married, he or she will want children and you will want grandchildren, just as much as anyone else does. You owe it to your son or daughter to explain why chelation now is so important for their future life.

Give yourself the best chance

The message is strong and clear that, for a person with thalassaemia to have the best possible chance of having children, iron loading must be kept at a minimum, even in childhood. There is no easy way around this and no alternative. Letting things slide and thinking that you'll knuckle down and start chelating properly later on when you actually want to have children is asking for trouble. The more iron loaded you are, the lower the chances of success.

Don't wait too long!

The other thing to bear in mind for women with thalassaemia is that if you are thinking of having children, do not put it off until you are older. Of course, this is not 100% within our personal control; because different people are ready to have children at different stages in life – but it is important for people like thalassaemia patients who maybe sub) ertile to know that fertility levels decline sharply from as early as the age of 35. In fact, statistically, age is the most important factor affecting a woman's fertility. At 30 the chance of conceiving naturally each month is about 20%; but by the age of 40 it is only 5%. So once again the message is clear – if you want to have a baby, don't put it off if you can possibly avoid it!

Young people in clinic

If your parents come to clinic with you and you don't wish to discuss fertility, contraception or any other subject for that matter, in front of them, ask if you can have five minutes alone with the doctor at the end of your appointment. If you don't want to suggest this yourself, by all means tell your nurse specialist when you are in for transfusion that you wo uld like to speak to the doctor in confidence then either the doctor or nurse can make the suggestion in clinic. Most doctors will suggest this anyway once their patients get to their middle teens; but don't be afraid to raise the matter yourself – it is not about shutting your parents out, but part of the process of taking responsibility for your own treatment.

Highlights of my Life - Pany Garibaldinos

First of all, let me say that I have done many things in my life but this is the first time I have been interviewed and told my life story from the beginning – it has been quite an experience and has brought back memories of things I haven't thought about for years. So here is the story of my sixty very full years.

At the start of my life no-one would have predicted that my story would fill so many pages. I was born in 1957 in Lagos, Nigeria – my father, an engineer, had left his native Cyprus to set up the first Coca-Cola factory in Africa. I was the fourth son born to my parents Andreas and Maria; but only the eldest, Costandino, was still alive at the time of my birth – the two middle boys had both died before the age of two. Now of course it is obvious that, like me, they had thalassaemia major – but at the time thalassaemia was little understood, there was no diagnosis and no real comprehension of the genetics – just a vague realisation that something "ran" in certain unlucky families whose children seemed to waste and die at a very young age. Two years later my sister Sodiroula was born, also in Lagos, so my parents had two healthy children – and me.

As time went by my father started to recognise in me the symptoms which had heralded the start of my two brothers' fatal illness. With a very heavy heart he made the arrangements for my mother to return to Cyprus with my sister and me (my eldest brother had remained in Cyprus with our grandparents). Poor Dad, he must have wondered whether he would ever see me again when he waved us off – it breaks my heart now to think of what my parents went through in those early years. In any event, when we returned to Cyprus in the early 60's doctors were starting to understand a little more about thalassaemia; and they told my mother I needed a blood transfusion – unfortunately however, one of my brothers had been given a transfusion just before he died, which meant that Mum did not trust the procedure.

I am told that I was crying for my father and the doctor told Mum in no uncertain terms that unless I got some blood I would never see my father again! Mum stood her ground however and I was not transfused. I would probably have gone the same way as my brothers if Dad had not at that point returned from Nigeria and taken our family off to Athens, where I eventually received my first transfusion at the age of 3 in the Children's Hospital.

My family continued their travels, returning to Nigeria in 1962 for Dad's work. In Lagos I was taken to see a doctor who must have had some vague idea of what was wrong with me (there was still no diagnosis, just "unexplained anaemia") and suggested that I be taken to see a paediatric haematologist at the Hammersmith Hospital in London. The next time we travelled back to Cyprus for a holiday, we stopped off in London to visit relatives and sure enough, Dad took me off to the Hammersmith. The doctor we saw there examined me; but the thing which impressed Dad the most was that he asked if he could bring a colleague into the consultation as he might know more about this kind of condition! Dad, used to dealing with doctors who thought their godlike opinions should never be questioned, saw this as a sign of wisdom and humility and decided on the spot that the Hammersmith was the place for me!

So, by now at the age of 5 having done my fair share of travelling back and forth, I was settled in London – Mum and my sister returned to Cyprus while Dad and I stayed with my uncle's family in Highbury. This brings me to one of the most interesting (and, looking back, alarming!) times in my history. The doctors at the Hammersmith decided they would try what was then the highly experimental technique of bone marrow transplantation. To this day I have no idea why, rather than any of my numerous family members, they chose an Irishman who was completely unrelated to me as the donor. I can remember very little about that time other than that I spent a long time in hospital in isolation – and that Mum came back from Cyprus to look after me, so I was very happy to have both my parents with me again. The transplant was duly carried out and I am told that my donor, an outgoing individual, enquired whether I had subsequently acquired a taste for Guinness and gambling on the gee-gees! Sadly, for him (and me) it was not to be – after a few months my own bone marrow kicked in again and I rejected the transplant. So unfortunately, I was not able to swap the blood bags for pints of Guinness at the tender age of six.

Pany, with mum, dad and sister at age 5 (1962)

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Dad had by now decided that the family should settle permanently in London – he was always in demand as an engineer and had no difficulty in obtaining work. He bought a house in Highbury near our relatives and we were able to be together as a family again. At the age of 7 my spleen was removed – standard procedure then of course – I don't think there are many thals around my age who still have their spleens rattling around inside them! But when I reached the age of 9 – and looking back I find this almost unbelievable – the doctors decided to carry out a second bone marrow transplant. I can only imagine that in those days it was thought inevitable that I would die at a very young age anyway and this procedure would at least give me a chance of living longer – but I don't think any of us had any idea of how dangerous it really was at the time. The second transplant was done in Guy's Hospital. This time they decided to take blood from my entire family and my older brother was determined to be the closest match. Once again, I was placed in isolation and given what were then the standard medicines to suppress the bone marrow – I remember it being the only time in my life when I ballooned in weight, due to all the steroids I was given. I looked like a chubby little teddy bear! The transplant went ahead and everyone hoped for the best; but it was not to be – once again I rejected the graft and was straight back to transfusions. I did have another bad scare while at the Hammersmith; or rather my parents did – I developed pericarditis (inflammation of the fluid-filled sac surrounding the heart) and had to be taken to theatre to have the excess fluid drained out – I can't remember much about it but my parents told me later that they were warned to prepare themselves as I was unlikely to live through the night! However, after the procedure I pulled round and was soon on the way to recovery. I should add that my transfusion regime in those days was completely ad hoc – nobody scheduled regular appointments for cross-match and transfusion, far from it – it was a question of "you let us know when you think you need it". Sometimes when they were busy I would be almost literally on my knees by the time I got my next transfusion – of course children nowadays are carefully monitored to make sure they get blood before their haemoglobin has dropped too low; but that was far in the future.

With friends at the hospital getting ready for transfusion

Some of my younger readers might find themselves thinking, hang on a minute – he's talking about transfusions, even transplants – what about chelation? You may well ask, we hadn't a clue about chelation, iron overload, all that fun stuff – I carried on for a few years with my transfusions, happily oblivious to the iron that was building up inside me. I think that Desferal must have been in development during these years, because I remember on one occasion my doctor at the Hammersmith tried it out on me while I was having blood – he gave me an injection and told me not to be scared if my wee turned red! But that was an isolated incident

Pany as a teenager

and I went on with my life, completely unaware that I was a walking iron time bomb.

During my time at the Hammersmith I very rarely came across anyone else my age who had thalassaemia – as I grew up a few other, much younger children started treatment (I remember that the hospital chef, an Italian chap, had two children who had thal) – but there was no-one else my age. I must have been about 14 when I was introduced to another family who had a boy similar in age to me who had thalassaemia. Little did I know what changes were in store when I collided with the force of nature that is George Constantinou! George was then being treated at UCLH by Dr (later Professor) Bernadette Modell; and was already on Desferal. If anyone out there is under the impression that George's intensity and drive only developed in later years, I'm here to tell them how wrong they are. I can see him now in that characteristic pose, hands up, fingers stabbing the air in front of my face, as he advised – no wrong word, insisted – that I change hospitals and follow him to UCLH. If I am honest what persuaded me was not the prospect of a longer life, but his stories of high jinks in the transfusion unit – the patients at UCLH were allowed to walk around while having their blood, go to the day room to watch TV and they even had free use of their own coffee machine – talk about luxury! Contrast this with the Hammersmith, where we were not allowed to move from the bed during transfusions, not even to use the toilet! (In fact, during all my time at the Hammersmith the only time I ever left my bed with blood up was when a nurse sneaked me into the day room to watch the historic Apollo moon landing in 1969! Having been made to think that the sky would fall in if I left the bed I was terrified of being found out, but we got away with it.)

Well, I may have the distinction of being the very first thalassaemia patient whose life was changed by George but I certainly wasn't the last! So off I went to UCLH, where I finally found other people around my own age who had the same condition. I'm not sure whether it was because of the influx of people from Cyprus coming to the UK in the 1960s, but UCLH seemed to be gaining more and more thalassaemia patients. It felt as though every time I went in there were more new faces. By the time I had been there for about a year, there were so many that Dr Modell decided to transfer half the patients to the Whittington Hospital; and as it happened George and I were in the transferred group. It was around this time that I was taken in hand by Sister Onufrak, who will be remembered fondly by the older cohort of Whittington patients. She was determined that I was going to chelate and get rid of all that iron. In those days there were no syringe pumps to maximise the effect of Desferal by giving it slowly, the recognised method was an intramuscular "bolus" injection – the needles were a terrifying size and the pain you would not believe. The idea was that we would learn to inject ourselves; and I remember Sister O making us practice using an orange. Well I had no problem in stabbing the orange but when it came to my own tender flesh that was a different matter! I remember sitting there holding the syringe for up to half an hour, trying to pluck up the courage to stab it into my leg. The Desferal was a thick liquid which stung and burned as it went in and for ages afterwards

Pany being presented with a signed football from Arsenal Players (1985)

- boy, were those injections painful! I remember that George had a "syringe gun" whereby you pressed a button and it "shot" the injection into your leg at high velocity – a bit like the guns they use now to pierce ears and other body parts! It was certainly quicker than my method but I never had the nerve to use it. It may be hard to believe for patients these days, but when syringe pumps came in and replaced the dreaded bolus injections, we thought all our Christmases had come at once!

For the most part, my memories of those early years have merged together into a blur; but I do remember us tearing up the ward when the nurses' backs were turned – water fights with stolen syringes were a popular game, as was pinching fruit to throw at each other. At the end of the day we were normal kids letting off steam, not saintly little invalids reciting psalms in preparation for the next world! I didn't really think about my long term prognosis to be honest and never discussed it with my parents. Poor Mum and Dad must have thought about it but in those days, it was considered better to protect children and young people from unpleasant realities. I was as happy as most youngsters despite having thalassaemia, transfusions, needles and all the tests – I won't say it was nothing to me because you never get 100% used to it – but it was the only life I knew. It was during my teens that it was finally brought home to me that my life could be in danger. I had gone to hospital for a clinic appointment and didn't feel well, nothing specific, I just knew that something wasn't right. Luckily they listened to me and kept me in for observation. About 11pm that night I felt much worse, I was freezing and wanted a bath to warm me up – the nurse humoured me by running the bath but she also phoned the doctor, who took one look at me and transferred me immediately to ITU with a diagnosis of septicaemia. For the first time, I truly felt I was dying; and asked for my father to come and give me the last rites – I told the doctor "I'm going to die" but he retorted "Not if I can help it!" Thankfully his efforts were rewarded and after several days in ITU I responded to antibiotics and started to recover. But that was a close call.

I left school at the age of 15 and went to work in my uncle's dress factory in North London – I soon became a dab hand at using the industrial sewing machines, overlocking fabric, making buttonholes – until one day my uncle ordered me to re-do a job I had finished, even though he himself had given me the wrong instructions – I told him where he could stuff his job and walked out then and there. This hasty decision actually worked in my favour, because the upshot of it was that I learned to drive, I always loved driving but little did I know at that time that it would become my lifelong profession!

Before I go on I should mention that my beloved Dad, who you will remember was very much in demand as an engineer, had become very religious over the years. He was a devout member of the congregation at St Andrew's Greek Orthodox Church in Kentish Town, London; where

Pany enjoying himself on holiday

he attended divine service every day. His outstanding devotion was noticed by the priest of the church and he encouraged Dad to take holy orders. So by my early teens Dad had left his engineering job with British Leyland and had become a Greek Orthodox priest!

Knowing how fond I was of driving, now that I was unemployed Dad suggested that I accompany him on his priestly duties and act as his chauffeur, which I was happy to do – driving around the streets of London chatting to Dad was a lot more interesting than being cooped up with a sewing machine! After a few months however, a jeweller friend of Dad's asked him if he might know a youngster who would be interested in training as his apprentice – so once again I was indoors, learning how to make and repair jewellery in the basement of the shop. It was meticulous work but I got to be quite good at it, staying for almost 5 years until the jeweller decided that he was going to close up the shop, go home to Cyprus and retire. Once again I was at a loose end; and once again, driving came to my rescue. By now we had moved to Hendon; and walking along the high street I saw a notice in the window of a driving school saying they were looking for new instructors – I went in and asked for an interview; it wasn't complicated, they said "As long as you have a clean licence you can start on Monday." I passed the instructor's exam and worked there for the next four years; eventually setting up my own business as a self-employed driving instructor in 1979. Over the next 20 years I taught thousands of people to drive, including the children of Dr Modell and Dr Wonke and heaven only knows how many of my fellow thals and their family members! To this day I am always being stopped in the street by people I taught to drive saying hello. Eventually I changed careers yet again but remained behind the wheel to become a driving test examiner, first in Pinner then moving to Hendon where I am to this day.

So my professional life was sorted out but what about my personal life? Well, when I was in my early twenties another thal patient called Kenny invited me to go with him to a party at the Whittington Hospital - yes, not content with going there for transfusions the hospital was part of our social life – the attraction of course was the endless supply of good looking nurses! While at the party I got chatting to a lovely girl called Trisha, a nurse of course. We hit it off straight away, we started going out and I even taught her to drive (she passed at the 2nd attempt!). Perhaps in spite of this, she accepted my proposal and we were married on 9th September 1984.

My health remained fairly stable through my twenties and thirties, except that as the years went by I gradually became so hypersensitized to Desferal that the injection sites were incredibly painful – I couldn't bear the touch of even the lightest fabric on my skin. It was

decided to place a Portacath in situ, at that time an unusual procedure but for me a godsend – I loved that Portacath! No injections for Desferal, no cannulae for transfusions – everything went into the Portacath. I had several ports over the years; and did so much Desferal that my ferritin level came plunging down. I thought I was doing extremely well, until I started to feel like my transfusions were not doing me any good – instead of feeling energized and full of life after blood I remained breathless and exhausted. By now I was under the care of Dr Wonke and she arranged for me to have a scan. After the procedure, I went home and decided to take the dog out for a walk. We were in the park when I got a call from a very agitated Dr Wonke, saying "Pany you have to get back here now – the scan shows you have a massive clot at the end of the Portacath – it must have been building up for years!" I said; "So what are you going to do?" Dr Wonke replied; "I'm not sure but you have to come straight in!" "No chance", I said "You figure out what you're going to do and then I'll come in." As soon as humanly possible, I was packed off to see the eminent heart and vascular specialist, Dr Malcolm Walker – a famous name in our thalassaemia world. Dr Walker explained that I would need surgery - and very difficult, delicate surgery - to remove the clot. The risk was that the clot could break apart during surgery, which could result in a catastrophic stroke. "So you mean I might not make it through the op?" I asked (I always like to call a spade a spade.) "There is a very real risk of that" he said gravely. "Then we can't do it yet," I told him. Here was the problem – after many years of trying to have a child, Trisha was 8 months' pregnant with our son. "After the baby is born you can take me straight to the theatre" I said, "But if there's a risk I might die, I'm not going to die without having held my son in my arms just once." Our son Alex was born the next month in July of 1999 – after his birth I felt ten feet tall, I was walking on air. I was so happy I convinced myself that I'd sail through the surgery – and I did.

Pany and Tricia on their wedding day

Then followed a quiet, happy few years, as Trisha and I enjoyed bringing up our little son, revelling in the novelty and pride of finally being parents. It was not to last, however. In 2003 repeated scans showed that I was suffering from severe pulmonary hypertension – dangerous

pressure in the heart was building up due to more clots narrowing and blocking the blood vessels. It wasn't looking good – Dr Walker, with that grave look I was beginning to recognise, suggested that I should be put on the waiting list for a heart and lung transplant. Well I didn't fancy that much as you can imagine! "If I don't have the surgery, how long have I got?" I asked; and was taken aback to get the reply "I'd give you two years, tops." I was sent to see a surgeon at the Papworth Hospital for a second opinion and he agreed 100% with Dr Walker about the prognosis. When it came to the heart and lung transplant though, he decided I was not a suitable candidate. Having looked at my medical history and performed various tests, his opinion was that there was only one operation which would extend my life. "There's a new procedure which was developed in the USA" he said. "I worked in the San Diego hospital that developed the technique, but it has only ever been done in two centres, San Diego and here at the Papworth." He was talking about a then new open-heart surgery technique called pulmonary thromboendarterectomy, or PTE for short. Basically, the chest is opened and incisions are made in the arteries allowing all the old, clotted material to be removed with meticulous care. At times during the procedure, the heart is stopped and the patient is kept alive by a heart-lung bypass machine, simultaneously being cooled down to hypothermic level to protect the organs from being damaged while the heart is not beating. The whole process takes 8-10 hours on the operating table, followed by 3-10 days in ITU. So, this was my alternative – PTE or knowing I had two years left to live. I thought about trying to live my life feeling that every day was bringing me closer to my "deadline"! "As much fun as that sounds" I told the surgeon "I think I'm going to go with the PTE." This time I went into the operating theatre with a deep feeling of trepidation – would I wake up? Would I ever see my wife and child again? Did the doctors know what they were doing? Then, mercifully quickly, the black curtain of anaesthetic came down and I knew no more.

Alex celebrating father's day with his dad

I'm not sure how many days and nights had passed until I finally came to my senses. I was lying in my hospital bed in a single room and the day was just about to dawn. I looked out of the window and watched as the grey light slowly changed. The sky filled with colours, soft at first then brighter, until finally shafts of sunlight reached the roofs, the walls, the trees, making them glow with a radiance I had never noticed before. I got out of bed and stood by the window trying to take in the beauty of the morning. Everything looked so clear and fresh I felt I was seeing it for the first time. I thought of the beautiful words of Eleanor Farjeon's hymn "Morning has broken" and understood them. A deep joy swept through me as I realised that I had pulled through and I was going to be OK. I was going home to Trisha and Alex, my friends and family. That sunrise taught me to appreciate the beauty of the world and what a blessing life is – if I ever took them for granted before I certainly never will again. Perhaps you need

to go into the dark to fully appreciate the light.

And since then life has been relatively uneventful, which is absolutely fine by me. I have enjoyed many years of happy marriage and seen my son grow up into a fine young man. I am still traipsing off to the Whittington every few weeks for my blood and still plugging away with the Ferriprox - I sometimes wonder whether I might have set a world record for blood transfusions! I look back at all the close shaves I have had over the years, the transplants, surgeries and various other complications too numerous to mention; and I have to say that thalassaemia today is a very different condition from when I was a youngster. It's very hard for me to understand why we still have problems with adherence to chelation when the options are so much easier than in my youth. I know older people always think that the younger generation has it easy, but compared to us, they really, really do. When I hear young thal patients whingeing that they don't take their chelating medicine because it "tastes horrible" I wish I could take them back in time to try those agonising bolus injections! Trust me, just one of those jabs and they'd be queueing up begging for the medicine.

Pany and Tricia attending the wedding of George and Mary

All in all, I can't complain – when I look back over my life so far what I remember best are the highlights, not the hard times. Maybe memory is kind to us that way, who knows – all I know is that what stand out to me are the happy landmarks of life – passing my driving test, my wedding, the birth of my son and his little face all lit up with excitement on Christmas mornings - and that first morning after my surgery. I mustn't forget the latest landmark, my 60th birthday and the parties given by the Whittington staff and my friends at UKTS – I can hardly believe how much trouble everyone went to, and I sincerely thank my wonderful friends for all their gifts, cards, good wishes and for helping me to celebrate. And thank you for reading my story.

Pany Garibaldinos

Alex, George and Pany at the Emirates stadium Oct 2017

This interview was conducted in mid-October 2017 when none of us, including Pany, had any idea how little time we had left with him. He passed away very peacefully on the morning of 4th December 2017, surrounded by his friends and family. Pany's wife Trisha and son Alex would like to thank everyone who visited him during his illness. They would also like to send very grateful thanks to all the medical and nursing staff of the Whittington Hospital; and the staff of St Luke's Hospice in Kenton for their expert, compassionate care which kept Pany comfortable and pain-free in his final days.

Parents' Stories – personal experiences of antenatal screening

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

The aim of the sub group was to identify all possible causes why screening and testing is not carried out in a timely fashion. According to the Screening Programme standards, all women at risk should receive an offer of prenatal diagnosis (PND) by the time they are 12 weeks' pregnant. In order to find out why the timeliness standards were not being met, we needed to examine the personal experiences of people who had been through the antenatal screening process. The Sickle Cell Society and the UK Thalassaemia Society were commissioned to carry out this research, which took the form of a series of interviews with women or couples who volunteered to share their experiences. The interviews with women and couples at risk of having a child affected by thalassemia were carried out by UKTS National Coordinator, Elaine Miller.

A personal thank you to the volunteers who shared their stories

I would like to thank the volunteers who came forward to share their memories of what were sometimes distressing and difficult times in their lives. Without your participation, we would never have obtained such rich evidence; which clearly illustrates not only the complexity and sensitivity of the decisions faced by couples at risk, but the difficulty of the task faced by health care professionals trying to meet a very tight deadline. Your stories will help to educate other parents and health care professionals at all levels. Thank you for your courage, your honesty, and for welcoming me into your homes. Elaine Miller, National Coordinator, UK Thalassaemia Society.

IMPORTANT MESSAGE FOR COUPLES PLANNING TO HAVE CHILDREN

The research clearly shows that in many cases, health care professionals are not picking up the fact that a baby may be affected by thalassaemia until many weeks into the pregnancy. Disturbingly, this seems to be the case even when a couple already has an affected child. If you want to be certain of having an early prenatal diagnosis test (to tell whether the baby has thalassaemia or not) make sure you raise the issue that thalassaemia runs in your family and you want to have a prenatal diagnosis at every antenatal appointment. Insist on your right to have the test carried out as early as possible in pregnancy – if you are pregnant and are worried that the test will not be done by 12 weeks 6 days (in accordance with the standard), please contact UKTS for advice.

The full report Parents' Stories contains interviews with couples who are carriers of thalassaemia and sickle cell disease; as well as an explanation of how the research was carried out and the valuable lessons which were learned from the evidence. The report can be found on the UKTS website at: www.ukts.org/parents-stories.html

Here we reproduce the stories of three couples who are carriers of thalassaemia. Names have been changed to protect the anonymity of the volunteers.

Iqbal and Khadijah's story

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

found Khadijah was pregnant again and the following year our son was born. We were devastated to find out that he too had thalassaemia.

The next few years were very difficult raising 2 children with thalassaemia; it was very stressful dealing with the hospital appointments and all the worry. The hospital explained to us about Prenatal Diagnosis (PND) and Khadijah and I agreed that we could not cope with any more children with thalassaemia. We decided that if any more children came along, we would have PND and terminate the pregnancy of an affected child. As Muslims, we believe that termination of pregnancy can only be done if it is carried out within 120 days of conception; so early PND is crucial for us. In our third pregnancy we had PND at 12 weeks. The haematologists treating our older children suggested that we also find out whether the baby was a human leukocyte antigen (HLA) match for stem cell transplant. Fortunately, the baby was healthy and proved to be an HLA match for his brother.

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

Abdul & Fozia's story

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

When Fozia became pregnant for the second time we went immediately to the GP – she was about 5 weeks pregnant. I went with her because my English is much better than hers. I explained to the doctor that we are thalassaemia carriers and we want the special test. While we were there the doctor sent a fax to the hospital and said they will send you an appointment. This was fine but then weeks went by and I started to worry. I phoned the surgery many times and I even went back in and spoke to the receptionist. They told me that the

hospital is very busy and we just had to wait. I was panicking but I did not know who I could speak to. When we finally got the antenatal appointment Fozia was 16 to17 weeks pregnant. I told the midwife we need the test right now. But, by the time the test was arranged, Fozia was already 20 weeks pregnant and it was too late for us to have any choice other than to proceed with the pregnancy.

When Fozia was 22 weeks pregnant, the hospital telephoned her at home and told her the news that the baby has thalassaemia. She phoned me at work in a hysterical state, I rushed home immediately and she was so distraught I was afraid and rang the doctor's surgery. I don't understand how they can give that kind of news to a pregnant woman who is all alone; the hospital should have telephoned me. We went to the GP and he told us about UKTS and gave us their contact details. I phoned them and they sent us a lot of information, books and films to watch about people who have thalassaemia. Fozia was very depressed for the rest of the pregnancy. When she was about 30 weeks we saw a doctor at the hospital who told us we could still choose to have a termination because they could inject something into the baby's heart to kill it. We were very upset at this suggestion; we thought it was a very insensitive thing to say as we had already explained about our beliefs and that we could not terminate after 120 days.

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

Asad and Nadia's story

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

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

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

Genome Editing for β-Thalassaemia By Dr Clare Samuelson

Genome Editing for β-Thalassaemia

In March 2018 Dr Clare Samuelson (MBChB(Hons) MA(dist) MRCPCH FRCPath), paediatric haematology trainee in Sheffield, will be travelling to Seattle, USA, to take up a research fellowship at the Fred Hutchinson Cancer Centre, working on genome editing for beta-globinopathies including beta-thalassaemia. This research is part sponsored by a grant from the UK Thalassaemia Society; and we look forward to seeing further hopeful developments in this fascinating field.

Background

While in recent decades there have been notable advances in the treatment options for patients with β- thalassaemia major, these still leave significant room for improvement. Patients receiving regular blood transfusions may run into problems with iron overload and subsequent organ damage; development of red cell antibodies or transfusion reactions. The need for regular blood transfusion is often inconvenient and limiting, interfering in education, work and travel arrangements. The only curative treatment currently available is allogeneic (donor) haematopoietic stem cell transplantation (bone marrow transplant). This is limited by lack of availability of a suitable donor in over half of patients; and in those patients who undergo such a transplant, the associated risks and toxicities – both short and long-term - are not insignificant.

Thalassaemias are genetic disorders, with β-thalassaemia being caused by mutations in the HBB gene. In the past few years there has been an exponential growth in interest in the potential for genetic correction of these conditions, without reliance on the need for donor bone marrow cells. The term 'gene therapy' was originally applied to techniques whereby a large number of normal copies of a gene were introduced into patient cells in the hope that they would be inserted into the patient's own genetic material and start working to produce a normal protein product. More recently such techniques have been refined to allow more precise correction or alteration of a patient's own genetic material using techniques known as 'genome editing'. It makes sense that a disease caused by faulty genes could be corrected by 'editing' those same genes or ones that interact with them, and such theories have now been successfully put into practice in many pre-clinical and animal studies in β-thalassaemia, along with a few early human clinical trials in related conditions such as sickle cell disease (which is also a disorder caused by a mutation in the HBB gene).

An Introduction to Genome Editing Techniques

A cell's genome is its complete genetic material, and genome editing describes making targeted changes to that genetic material. Genome editing is based on the introduction of a break in a cell's double-stranded DNA. The cell then seeks to repair the break and this can happen in one of two ways. The first mechanism is called non-homologous end joining (NHEJ), whereby the two broken ends are rejoined directly, a process during which small insertions and deletions (indels) of genetic material may be introduced. It is the introduction of indels during the rejoining process which is relied upon to disrupt the gene. The second mechanism is termed homology-directed repair (HDR), whereby the cell normally uses its own pre-existant genome as a 'template' for repair. It is this second mechanism which is employed in most genome editing techniques, as a new 'template' can be provided to the cell so that when the gene is repaired it is done so according to a new and pre-defined blueprint. (Figure 1)

There are 3 different types of technology (sometimes referred to as 'platforms') currently being used in genome editing techniques. The purpose of all 3 is the same: to introduce a targeted break in the cell's double-stranded DNA. They rely on nucleases: enzymes which break the DNA in specific sites. It is possible to decide which part of DNA should be broken (and therefore which part of which gene should be edited) by introducing the right nuclease into the cell. The 3 different methods being used at the moment are:

1 - Clustered Regularly Interspaced Short Palindromic Repeats/Cas9 (CRISPR/Cas9)

2 - Transcription activator-like effector nucleases (TALENs)

3 - zinc-finger nucleases (ZFNs)

The introduction of selected nucleases, along with the new template DNA or RNA where required, is usually accomplished using viral vectors (viruses that have been modified in the laboratory to carry these complexes into cells).

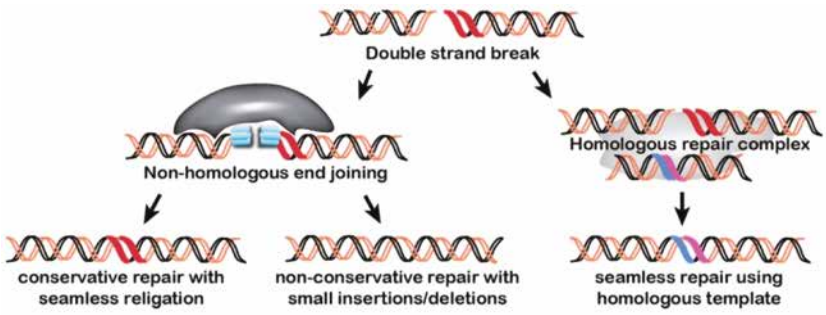

Figure 1. Schematic of double-strand break-induced gene editing: Top) A nuclease induces a double-strand break at a desired target site (red marked region of DNA) Left panels: Non-homologous end joining mechanisms are activated, which can rejoin the two ends of the broken DNA, with or without small insertions or deletions (indels). Right panels: Homology-directed repair mechanisms are activated. If a DNA template with regions of homology is provided to the cell, a new sequence (blue/pink section of short duplex) may be copied into the genome at the double-stranded break.

Reproduced with permission from Professor Hans-Peter Kiem, Fred Hutchinson Cancer Institute, Seattle, USA

Genome Editing Targets in β-Thalassaemia

The aim of genome editing in β-thalassaemia would be to convert the condition of a patient with β-thalassaemia major (where regular transfusion or allogeneic stem cell transplantation are required to sustain life) to that of someone who is only carrying the β-thalassaemia trait (where health is not affected). This would be accomplished by taking the patient's own haematopoietic stem cells (the early cells which have the potential to develop into all types of blood cells) and editing their genomes in the laboratory, then transplanting them back into the patient (autologous stem cell transplantation). This would provide a source of healthy blood cells without the need for a stem cell donor and avoids many of the complications of allogeneic (donor) stem cell transplantation.

There are multiple possible genetic targets which could be modified to ameliorate the clinical disease in β-thalassaemia. (Table 1)

Table 1. Genes currently being investigated as targets of genome editing in β-thalassaemia, along with rationale for inducing alterations.

Target Gene(s)	Gene Function	Desired Effect	Rationale
HBB	β-globin gene	Direct correction of the mutation causing β-thalassaemia	Correction of the gene defect to allow normal production of β-globin chains and therefore correction of β-thalassaemia
HBB	β-globin gene	Increase in HbF production by introduction of a new mutation known to cause HPFH (hereditary persistence of foetal haemoglobin: an inherited condition in which HbF continues to be produced beyond infancy in greater amounts than normal)	Increased production of HbF which compensates to an extent for the lack of HbA. Likely success of this approach is suggested by the protective effect in patients with β-thalassaemia mutations who have higher HbF levels.
BCL11A	HbF production control gene	Increase in HbF production by disruption of normal control processes	Increased production of HbF which compensates to an extent for the lack of HbA. Likely success of this approach is suggested by the protective effect in patients with β-thalassaemia mutations who have higher HbF levels.
HBA1, HBA2	α-globin genes	Introduction of deletions or disruptions resulting in decreased α-globin production, i.e. introduction of α-thalassaemia trait	Reduction in imbalance between α- and β-globin chain production, thereby reducing the number of excess, free α-globin chains which cause cellular damage. Success of this approach is predicted by the protective effect in patients with β-thalassaemia mutations who have co-inherited α-thalassaemia trait

Research Update and Future Directions

Successes reported so far have been mainly in vitro (in the laboratory only) or have involved transplantation of genetically edited haematopoietic stem cells into animals. While such reports are encouraging and point towards genome editing as a potential treatment for patients with β-thalassaemia in the future, there are many aspects of these methodologies which require further study before transplantation back into humans can be trialled.

Challenges include: selection of the optimal nucleases to target desired DNA sites, and improvements in the design of template genetic material to be provided to the cell to direct homology-directed repair; selection of edited stem cells in the laboratory and development of methods to encourage their multiplication so that an adequate stem cell dose of genetically edited cells can be provided back to a patient; and investigation into the optimal drug regimen which a patient should be given before such edited stem cells are re-implanted, to encourage their engraftment and growth in the bone marrow without causing excess toxicity to the patient. The long-term potential of these techniques is clear but investment into research and development is essential if they are to become safe and effective treatment options for patients in the future.

Ethical Considerations

Research involving genetic modification of cells is tightly regulated in the UK with stringent safeguards in place to ensure that work taking place is rigorously examined both scientifically and ethically. Work in this country and other Western centres has concentrated mainly on genetic modification of somatic cells (those which are not involved in reproduction), where any unforeseen side effects would be expected to be limited to an individual involved in the process and not passed on to future generations.

There are a smaller number of reports involving genome editing of embryos, which, theoretically at least, offers the potential to correct genetic conditions not just for that individual embryo but for his / her offspring and future generations. Only one report from China describes genome editing of embryos for thalassaemia. None of the genetically edited embryos from these studies have been re-implanted or even allowed to grow beyond a very limited number of cells so far. Research on embryos is a highly controversial ethical area, in part due to the fact that any unintended side effects from genome editing would be passed onto future generations if the edited embryos were to be re-implanted into a uterus and allowed to develop. While the long-term effects of such a new technology are unknown this must cause significant concern.

Another objection levelled by some who object to genetic modification of any sort is that this may be the start of a slippery slope leading to 'designer babies', where, for example, the hair and eye colour of a child or his/her IQ may be altered by genome editing techniques. While examples of such cosmetic selection are a far cry from treatment of life-threatening or life-limiting conditions, there will always be grey areas and such concerns point towards the importance of ensuring that ethical and legal debate do not lag behind scientific advances. Scientific and medical communities, in conjunction with patients and the general public, must always consider what should be done as well as what can be done.

Finally, a further concern with any new and expensive technology is the likely limitations in availability globally. Currently standard care for children and adults with β-thalassaemia major in western countries such as the UK is already good, whereas the majority of children affected by this condition across the world lack access to regular blood transfusion and die in early childhood. It is likely that once genome editing techniques have been optimised and can be offered as a treatment for β-thalassaemia, they will at least initially only be available in richer countries or to those patients who can afford to pay privately for such techniques, further widening inequalities in care.

Conclusions

In summary, progress towards genetic correction for patients with β-thalassaemia through genome editing of their own haematopoietic stem cells is advancing but there are still significant technical hurdles to overcome. Work continues to optimise such therapeutic strategies with the aim of providing safe and effective clinical cures for children and adults with this condition, without the need for donor bone marrow transplantation. Alongside scientific challenges in providing such treatment, the ethical considerations involved in genome editing, particularly where embryo research is involved, must also be thoroughly addressed.

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Recent events and meetings 129

Those who attended meeting on behalf of the UKTS are: Gabriel Theophanous *Chair*, George Constantinou *Assistant Secretary*, Elaine Miller *National Coordinator*, Katerina Read *Office Administrator*, Roanna Maharaj *Member*

01 April 2017	NHSE stakeholder testing workshop Gabriel Theophanous, George Constantinou
06 April 2017	North Middlesex Hospital SCT awareness event Katerina Loizi-Read
10 April 2017	SCT/PHE public engagement teleconference, streamlined screening pathway for at-risk couples, Elaine Miller
20-22 April 2017	TIF Board meeting, London Gabriel Theophanous, George Constantinou
13 May 2017	UKTS strategy meeting, UKTS Board members and staff
24 May 2017	SCT/PHE public engagement Advisory Group teleconference Elaine Miller
13 June 2017	presentation, health care professionals study day, Cardiff & Vale University Hospital Elaine Miller
19 June 2017	UKFHD committee meeting Elaine Miller
07 July 2017	STANMAP annual conference Elaine Miller
21 July 2017	NHSE communications and engagement group teleconference Elaine Miller
09 August 2017	SCT/PHE public engagement Advisory Group teleconference Elaine Miller
29 August 2017	NHSE communications and engagement group teleconference Elaine Miller
07 September 2017	APPG AGM, Portcullis House, London Elaine Miller
15 September 2017	Yorkshire & Humber haemoglobinopathy network meeting, Bradford Royal Infirmary Elaine Miller
18 September 2017	UKFHD committee meeting Elaine Miller
18 September 2017	UK Thalassaemia Society AGM
19 September 2017	SCT/PHE public engagement Advisory Group meeting Elaine Miller
20 September 2017	awareness event, Middlesex University Katerina Read
22 September 2017	awareness talk for nurses, Luton
27 September 2017	NHSE haemoglobinopathies focus group, London, Gabriel Theophanous, George Constantinou
4 October 2017	SCT/PHE public engagement Advisory Group teleconference Elaine Miller
9 October 2017	NHSE communications and engagement group teleconference Elaine Miller
11-13 October 2017	distribution of educational and awareness publications, 11TH Annual Sickle Cell Disease and Thalassemia Conference (ASCAT) 2017 (held by Guy's & St Thomas's Hospital), London Romaine Maharaj, George Constantinou, Elaine Miller, Katerina Loizi-Read, Roanna Maharaj
11 October 2017	Educational presentations (chelation therapy) to health care professionals, ASCAT conference, London George Constantinou, Roanna Maharaj
27 October 2017	distribution of educational and awareness publications, Chief Nursing Officer for England Black Minority Ethnic Nurses Conference, Brent Civic Centre, London Elaine Miller
6 November 2017	London Haemoglobinopathies Forum, Skipton House, London Elaine Miller
13 November 2017	SCT/PHE public engagement Advisory Group teleconference Elaine Miller
15 November 2017	SCTSP Advisory Group meeting, Euston London Elaine Miller
17-19 November 2017	TIF Thessaloniki, Greece – UKTS Board of Trustees and staff
23 November 2017	presentation, UKFHD scientific meeting, Birmingham – Elaine Miller
30 November 2017	SCT/PHE public engagement Advisory Group meeting Elaine Miller
11 December 2017	UKFHD committee meeting Elaine Miller
10 January 2018	NHSE communications and engagement group teleconference Elaine Miller
18 January 2018	NHSE communications and engagement group webinar Elaine Miller
23 January 2018	NHSE communications and engagement group webinar George Constantinou, Elaine

Acronyms

APPG – All Party Parliamentary Group for Sickle Cell & Thalassaemia
CRG – Clinical Reference Group for Haemoglobinopathies
GAUK – Genetic Alliance UK
NEBATA - North of England Bone Marrow & Thalassaemia Association
NICE – National Institute for Health & Care Excellence
NHSBT – NHS Blood & Transplant
NHSE – National Health Service England
NSC – National Screening Committee
PHE – Public Health England
RDUK – Rare Diseases UK
SCTSP – NHS Sickle Cell & Thalassaemia Screening Programme
SCS – Sickle Cell Society
SHCA – Specialised Health Care Alliance
STANMAP – Sickle Cell & Thalassaemia Association of Nurses, Midwives and Allied Professionals
TIF – Thalassaemia International Federation
UKFHD – UK Forum on Haemoglobin Disorders

VIRGIN LONDON MARATHON 2018

We are proud to introduce the six volunteers running for our cause in the Virgin London Marathon on 22nd April 2018. Please support them by visiting their online donation pages listed per runner.

Stephen Bobbin
www.justgiving.com/stephenbobbin

Owen Courtney
https://uk.virginmoneygiving.com/OwenCourtney

Lenos Kyriacou
www.justgiving.com/lenos/yriacou2

Ai Seng Paul
www.uk.virginmoneygiving.com/aiseng

Speros Spyrou
www.justgiving.com/speros spyror

Ben Winch
www.justgiving.com/benwinch1

Please Support our Runners

We will be offering Polo T shirts for sale which can be worn for the event to support our runners.
They will be available in adult and children sizes.
Please contact Katerina@ukts.org
or call 02088820011 to reserve your sizes.

BABY CONGRATULATIONS TO NOREEN

Another new baby to welcome to our UKTS "family" - this is definitely TM's favourite kind of feature! We are delighted to congratulate Noreen and Mohammed Mangera of Coventry on the safe arrival of their first child, Saarah Noor. Eagle-eyed regular readers of TM may remember issue 121 from 2013, in which we congratulated Noreen for – in the space of two months - obtaining her Master's degree in Health Psychology and her marriage to her husband Mohammed! In her own words she tells us about this latest and most precious development in her life.

Saarah Noor Mangera truly is our precious gift from God. She was sent to us when we were really not expecting it at all. (I was in the process of moving to another city for a job). She was born on Friday 30th June 2017, again another surprise as she decided to come 3 weeks early. She weighed 5lb 4oz. She has brought so much happiness and joy in to our lives. 2016 had not been a good year for my family, it was the year we lost my grandad - and exactly a year later we were blessed with Saarah. She is such a happy baby, growing fast and hitting all her milestones. She is everyone's little princess, she is the first grandchild on my side of the family and she is also the first great grandchild!

It is really so nice to see my mum so happy and so in love with her. With all the hardships and struggles she went through with me and my brother when we were children (my brother also has thalassaemia) she can now enjoy Saarah. They have a really good bond.

I would like to say a huge "thank you" and express how much I appreciate the wonderful care I received from my Consultant Dr. Nicolle and thalassaemia nurse specialist Melvis during my pregnancy; and also Dr. Dhingra my obstetrician. They monitored my pregnancy really well and put plans in place for every eventuality. They were down straight away for cuddles with Saarah !

Noreen, who has beta thalassaemia major, is under the care of Dr. Sarah Nicolle at Coventry & Warwickshire University Hospital. Many thanks to Noreen and Mohammed for sending the us their news and pictures of their beautiful daughter.

...and congratulations to Michael on becoming a father of two!

Many congratulations to our member (and thalassaemia patient) Michael Paraskeva and his wife Patricia on the birth of their second child, a son named Dempsey Kip Luka, who was born on 8th March 2017 weighing 8lbs 1oz. Little Dempsey is seen here with his proud parents and older sister Minnie, aged 3.

Thank you to Michael and Patricia for sharing their happy news and the delightful family picture we trust that both their children will bring them great joy over the years.

Michael is under the care of Dr Evely at Southmead Hospital, Bristol.

Anna's son Andrew with his baby sister Isabella

...and bringing you yet another beautiful "UKTS family"

Growing up with beta thalassaemia major, I didn't think that having children would have been possible. I had got used to the idea that I wasn't going to have any babies of my own. I thought that there were far too many health factors in the way. Slowly in time, I realised that it would be possible to have children. I saw that fellow patients had children of their own, this was very encouraging. Eventually, the time was right to start a family of my own. I'm so glad that I did as I now have two healthy, beautiful children.

By Anna Kakouris, who is under the care of Dr Farrukh Shah and Dr Emma Drasar at the Whittington Hospital.

Events

Wine Festival 2017

Members of the Metropolitan Police Service Greek & Cypriot Association (MPS GCA) with Katerina Read at the Wine Festival

ASCAT 2017

George Constantinou and Katerina Read at the ASCAT meeting with a presenter

The Board of Trustees would like to acknowledge and thank families and friends for choosing to donate funds the UK Thalassaemia Society in memory of their loved ones.

Stelio Arnaouti

16th March 1943 to 26th December 2017

Sofia Tryfonos

2nd February 1942 to 22nd December 2017

Pany Garibaldinos

27th August 1957 to 4th December 2017

Support the UKTS while shopping on Amazon.uk

The UK Thalassaemia Society would like to thank all members and well wishers for taking the time previously to Amazon UK, via our website. We recognise that this was problematic especially for those now using the Amazon app. We are happy to announce that with the new smile.amazon.co.uk incentive you are now able to list us as your preferred charity from March 1st, 2018 and any future purchases will be automatically included in the incentive.

So please ensure that you visit the website and choose us... then happy shopping!

The details are as follows:

amazonsmile

You shop. Amazon gives.

AmazonSmile is a simple and automatic way for you to support a charity of your choice every time you shop, at no cost to you. When you shop at smile.amazon.co.uk, you'll find the exact same low prices, vast selection and convenient shopping experience as amazon.co.uk, with the added bonus that Amazon will donate a portion of the purchase price to your se !ected charity.

How do I shop at AmazonSmile?

To shop at AmazonSmile simply go to smile.amazon.co.uk from the web browser on your computer or mobile device. You may also want to add a bookmark to smile.amazon.co.uk to make it even easier to return and start your shopping at AmazonSmile.

Which products on AmazonSmile are eligible for charitable donations?

Millions of products on AmazonSmile are eligible for donations to charities by Amazon. You will see eligible products marked "Eligible for smile.amazon.co.uk" on their product detail pages. Recurring Subscribe and Save purchases and subscription renewals are not currently eligible.

Can I use my existing amazon.co.uk account on smile.amazon.co.uk?

Yes, you use the same account on amazon.co.uk and smile.amazon.co.uk. Your shopping cart, Wish List, wedding or baby registry, and other account settings are also the same.

Thalassaemia: 2020 and beyond

Listen.
Learn.
Talk.
Inspire.

14th April 2018 Leeds

Presenting our 40th anniversary event

What's going on?
It's all about thalassaemia and the next generation.

- Come and hear about some promising new therapies.
- Meet some experts.
- Listen to some real life experiences.

When's it happening?
Saturday 14th April 2018

Where's it taking place?
Mercure Parkway Hotel, Leeds LS16 8AG

Who do I contact to confirm my place?
Email **Katerina** at The UK Thalassaemia Society, **katerina@ukts.org**, or call her on **0208 882 0011.**

Do I need to know anything else?
There's going to be a fully qualified crèche for small kids and you can get assistance with transport costs.

ukts.org
#ourthalconvo

Thalassaemia: 2020 and beyond

Listen.
Learn.
Talk.
Inspire.

Presenting our 40th anniversary event

10th March 2018 London

What's going on?
It's all about thalassaemia and the next generation.
- Come and hear about some promising new therapies.
- Meet some experts.
- Listen to some real life experiences.

When's it happening?
Saturday 10th March 2018

Where's it taking place?
Regency Banqueting Suite, 113 Bruce Grove, London N17 6UR

Who do I contact to confirm my place?
Email **Katerina** at The UK Thalassaemia Society, **katerina@ukts.org**, or call her on **0208 882 0011.**

Do I need to know anything else?
There's going to be a fully qualified crèche for small kids and you can get assistance with transport costs.

ukts.org
#ourthalconvo

NOTICE TO ALL MEMBERS C changes to UKTS membership

UKTS members are asked to note that changes to membership have been introduced further to constitu tional amendments which were approved by a members' vote at the Society's AGM on 18th September 2017. These changes are intended to increase revenue and to reduce the administrative burden on the UKTS office. We recognise that these changes will increase the cost of membership, however please note that this is the first increase in 15 years; during which time the running costs of the Society have increased enormously. Please read this notice carefully and if there is anything you do not understand, please con tact the UKTS office.

New members

From the date of the last AGM, 18th September 2017, all new members will be required to take out a standing order payment in favour of the UK Thalassaemia Society. The minimum standing order payment is £2 per calendar month or one yearly payment of £24. Members may choose to give more than £2 per month; and any additional sum will be counted as a donation. No renewal will be required, as your mem -ership will continue for as long as the standing order remains in force. If the standing order is cancelled, membership will cease on the cancellation date.

Annual members

Those who are currently annual members (i.e. who pay an annual subscription each January) will be re 7uired to take out a standing order payment in favour of the UK Thalassaemia Society. You will receive a standing order form by post in January 2018 when all annual memberships are due to renew.

Life members

From the date of the last AGM, 18.9.17, no new applications for life membership will be accepted. However, if you took out life membership before 18.9.17, your membership will not be affected and will continue during your lifetime.

LITERATURE

The following represents a list of the literature held in our offices presently. Please contact Katerina or Elaine should you require copies for distribution.

Name:	Language:	Type:
A guide for the haemoglobinopathy nurse	English	Book
About Thalassaemia	English	Book
All you need to know about being a carrier of beta thalassaemia	Bengali	Booklet
All you need to know about being a carrier of beta thalassaemia	Gujrati	Booklet
All you need to know about being a carrier of beta thalassaemia	Hindi	Booklet
All you need to know about being a carrier of beta thalassaemia	Urdu	Booklet
All you need to know about being a carrier of beta thalassaemia	English	Booklet
All you need to know about being a carrier of beta thalassaemia	Punjabi	Booklet
Alpha thalassaemia English	Booklet	
Alpha thalassaemia English &Chinese	Booklet	
Compliance to iron chelation therapy with deferoxamine	English	Book
Emergency management of Thalassaemia	English	Booklet
Guidelines for the clinical management of Thalassaemia	English	Book
Guidelines for the management of non-transfusion dependent thalassaemia	English	Book
If you teach a child who has Thalassaemia	English	Leaflet
Information for people who have haemoglobin H	English	Booklet
My baby has Thalassaemia	English	Book
My Thal English	CD	
Patients' rights English	Book	
Patients stories English	Booklet	
Prevention of Thalassaemia and other haemoglobin disorders	English	Book
Prevention of Thalassaemia and other haemoglobin disorders (second edition)	English	Book
Prevention of Thalassaemia and other haemoglobin disorders (volume 2)	English	Book
UK English	Book	
Thalassaemia major and me	English	Book
Thalassaemia major and me	Bengali	Booklet
Thalassaemia major and me	Punjabi	Booklet
Thalassaemia major and me	Urdu	Booklet
Thalassaemia major and me	Gujrati	Booklet
Thalassaemia major and me	Hindi	Booklet
Thalassaemia major and me	Chinese	Booklet
Thalassaemia your life, your choice, your test	English	CD
Thalassaemia your life, your choice, your test	English	Leaflet
Tied for life English	Leaflet	
UKTS colouring pages	English	Book

Help us to help you by supporting YOUR Society ...every £ is precious!

Please support The UK Thalassaemia Society by Making a Monthly Donation

STANDING ORDER MANDATE

To the Manager (Name of Your Bank)
Address:
City: / *Postcode:*

Please pay: *NatWest, 12 The Broadwy, Southgate, London N14 6PL*
For the credit of: *UK Thalassaemia Society, Registered Charity No: 275107*
Sort Code 51-50-00 Account Number 64949362

The sum of: *£2.00* ☐ *£5.00* ☐ *£10.00* ☐ *Other* ☐ *£* _____ *(amount)*
On the _____ *(day)* _____ *(month)* _____ *(year)*
And thereafter every month until further notice and debit my account accordingly.

Name(s) of account holder(s) to be debited:
Account Number:
Sort Code:

Signed _____ Date _____

Signed _____ Date _____

Your Address:
Tel Number:
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I would like tax to be reclaimed on my donation under the Gift Aid Scheme. I am a UK tax payer and pay an amount of income tax and/or capital gains tax at least equal to the tax that can be reclaimed on my donation. ***Please tick.***

YES ☐ NO ☐

Please call 020 8882 0011 if you have any queries. When completed please return to:
UK Thalassaemia Society, 19 The Broadway, Southgate Circus, London N14 6PH
We will then send this form on to your bank.

Thank you for your valued support.