



My baby has thalassaemia



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Living with thalassaemia



Foreword

This is a valuable little book that will be very helpful for parents who have just been told their child has thalassaemia. It is clearly written, with a very sympathetic and experienced tone, and provides a lot of support. I particularly like the quotes from other parents, the emphasis on taking all treatments as prescribed, and the assurance that the UK Thalassaemia Society is there to help.

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A UK Thalassaemia Society publication. Written by Elaine Miller

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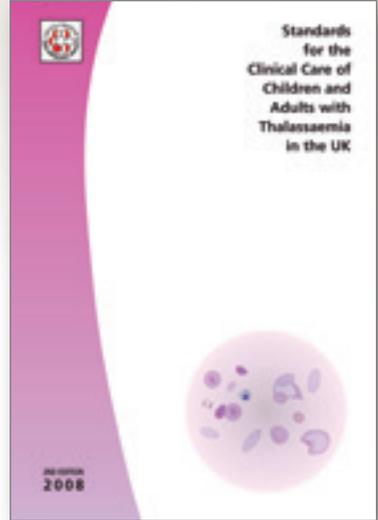
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All these publications are available FREE from the UK Thalassaemia Society



My baby has thalassaemia booklet



UKTS National Standards for the Clinical Care of Children & Adults with Thalassaemia in the UK



UKTS DVDs



Leaflet for teachers and carers

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Introduction

This is a booklet for the parents of children who have thalassaemia. It provides a basic guide to thalassaemia and signposts to sources of current information and appropriate services for patients, their families and carers. Health professionals may also find this information useful. In each section you will find references to where you can find further information.

The first part of the booklet applies for all types of thalassaemia. Most of the second part applies for types of thalassaemia with regular blood transfusions.

Beta thalassaemia major, beta thalassaemia intermedia and haemoglobin E/beta thalassaemia are all types of thalassaemia.

People with thalassaemia major need regular blood transfusions and other treatments from the first year or so of life.

People with thalassaemia intermedia need regular medical care, but do not always need regular blood transfusions, at least in the early years.

People with haemoglobin E/beta thalassaemia need regular medical care, and some need regular blood transfusions from early life.

There is more about different types of thalassaemia on pages 27-28.



What is thalassaemia?

How does thalassaemia affect the body?

Thalassaemia is a condition which affects the production of red blood cells. Children who are born with thalassaemia cannot make enough healthy red blood cells, so without medical treatment most would get weaker and weaker and would fail to develop and grow normally. Children who have thalassaemia will need medical treatment throughout their lives. Information about the treatment is provided on pages 14-18. It is very important for the parents of a child who has thalassaemia to follow their child's treatment programme carefully and understand as fully as possible the reasons for all the various tests, procedures and drugs which will be needed.

How does a child get thalassaemia?

Thalassaemia is an inherited condition, that is, it is something which is passed on from parents to children in a similar way as parents may pass on other physical characteristics like eye colour or the shape of a nose. Thalassaemia cannot be caught or acquired in any other way. Some people are healthy "carriers" of thalassaemia. Carriers are not ill in any way, but if a man and a woman who are both carriers have children together there is a risk that some of their children could be born with thalassaemia. If you are reading this book because your child has thalassaemia, both you and your child's other parent are carriers of thalassaemia. You can find more information about carriers and how thalassaemia is inherited on pages 23-26.

Can my child have a normal life?

It is very important to remember that, although thalassaemia is a serious, lifelong medical disorder, with modern medical treatment it is manageable. Children who are born with thalassaemia in the UK today should expect to reach their full educational potential, have careers, relationships and even healthy children of their own. They should expect to live a normal or near normal lifespan. All these things are possible provided that the treatment plan is followed closely.

My baby has thalassaemia



Diagnosis

Ideally, the fact that an unborn baby has thalassaemia would be detected in pregnancy and the parents would receive counselling about what to expect (see page 24). However this may not have happened for a number of reasons. The parents may have arrived recently in the UK, or may not have wished for any tests during pregnancy. In any event, all babies born in the UK are given a blood test (the “heel prick test”) when they are a few days old. This test, although it does not provide a definitive diagnosis, will usually “flag up” that there is a problem with the baby’s blood and further tests will be carried out. At this point, usually the parents will receive a visit from a specialist nurse counsellor who will explain what thalassaemia is. The nurse will also arrange for the baby to see a haematologist (a doctor who specialises in conditions of the blood) who will monitor the baby closely.

A parent says...

“I know that for us it felt almost like bereavement. It is like any loss or perceived loss, where only the passage of time and the realisation that your child can have a fairly normal life are the keys to getting over it.”

This is a very difficult time for parents. Having a child with thalassaemia comes as a severe blow to a family. Whether it is a first baby or the family already has healthy children, the parents face shock, disbelief and grief on finding out their child’s diagnosis. This is only

human and understandable; and it can take some time for parents to come to terms with thalassaemia and what it means for them and their child. However, although the parents of a child with thalassaemia will certainly face many challenges, there is no need to despair completely. With modern medical treatment, a child with thalassaemia should expect to live a normal or nearly normal life and have similar expectations as others in terms of education, career, social and family life.

At the UK Thalassaemia Society we have a film available on DVD called “My Thal”. It features interviews with people of all ages and backgrounds who have thalassaemia, speaking about how they manage their condition. They talk about their education, careers, relationships and having children. You may find it very reassuring to watch this film, which shows that a person with thalassaemia can have a normal life. The film is **FREE** so please don't hesitate to contact UKTS for your copy.

Guilt and/or shame?

As well as shock and grief, parents often experience guilt that they as carriers have passed the condition to their child. This again is natural but really there is nothing to feel guilty about. Every single one of us carries genetic abnormalities. Usually they do not cause any problems but occasionally they do. This is no-one's fault but is simply a sad fact of life.

Sometimes at UKTS we hear of parents who feel ashamed that they have a child with thalassaemia and wish to hide their child's condition from the world. They become very secretive and don't talk about their child's thalassaemia to their friends and sometimes even close relatives. This is very sad indeed as the feelings of the parents will certainly affect the child. Even very young children are extremely perceptive and they instinctively pick up on their parents' attitudes and examples. Imagine how awful it is for a child to grow up feeling that there is something shameful about them which must be hidden and never talked about. The damage done to the self-esteem and emotional well-being of the child can be irreparable and last throughout life. At the UK Thalassaemia Society's family conferences we see lots of good looking, lively, laughing young people who have thalassaemia – and their proud parents, who are happy to tell us of their son or daughter's plans for university, career, marriage etc. We in the UK thalassaemia community have every reason to be proud of our young people and their achievements.

A parent says...

“It is 35 years since my son was born and diagnosed with thalassaemia and yet it seems like yesterday. The initial reaction to the diagnosis was disbelief, grief, loss, anger, fear and still today a sense of guilt that we his parents don't need medical treatment but because of us we have passed on this condition to our son. However we have never hidden or denied that our son has thalassaemia, which has contributed to his acceptance of himself and his condition and the challenges he has faced over the years.”

You are not alone

Once the initial shock has worn off, many people find that it is helpful to be in contact with other parents who have children with thalassaemia. It can also be very reassuring for them to meet adults who have thalassaemia so they can see for themselves that the condition can be successfully managed. The UK Thalassaemia Society can help by putting parents in touch with local support groups if available; and if not by an informal “buddying” service in which we connect families who have no local source of support with others, so that they can maintain contact by telephone and email. If there are any language difficulties, the UKTS staff will try to connect the family with speakers of the same language. It is important to remember that you are not alone. At UKTS we hold family conferences and local meetings where you can meet parents of other children who have thalassaemia; and people of all ages and backgrounds who are living with the condition. *It is therefore very important that you get in touch with UKTS and let us have your contact details.* Many people assume that the Society will have been notified and given their details once their child with thalassaemia is born. This is not the case unfortunately. When we hold conferences we send an invitation to every household on our database where there is a thalassaemia patient; but we can only invite you if we have your address!

A parent says...

“It really is essential that parents and the child help each other to face these challenges and an important part of that has come from the UK Thalassaemia Society and meeting and building relationships with others who are either parents or people with thalassaemia”

(N.B. The UKTS database is entirely confidential and your details will NEVER be shared or disclosed to any other party.)

The importance of the parents' role

One vital thing to remember is that you as parents are just as important as doctors and nurses in helping your child to be as healthy as possible. Doctors and nurses can advise, prescribe medicines and give treatment, but it is the parents who are with the child all day, every day; and it is the parents who know their child best and who can influence their child better than any medical professional could do. They have a job to do in supporting and enabling their child so that he or she grows up to be a fully integrated, contributing member of society.

Long experience and many studies have shown that people who live with long term medical conditions need a positive attitude and positive expectations just as much as they need medical treatment if they are to achieve their full potential in terms of education, career and social life. This is where parents play such a crucial role. Whether a child grows up with this positive attitude or not is often dependent on his/her early years. It is vital that from a very early age, parents treat their child with thalassaemia just as they would any other child in terms of what they expect. For example, allowing your child to miss school (unless it is absolutely



An international group of thalassaemia patients enjoying the Thalassaemia International Federation conference, Singapore 2008.

necessary) is not advisable, nor is expecting a lower level of academic achievement than you would from other children. People with thalassaemia are as intelligent and capable as anyone else; and the experience of coping with their condition gives many of them an even greater determination to succeed. There are people who have thalassaemia in all professions and walks of life, including the medical professions, the law, teaching, accountancy and others. Many work in other fields such as the retail and service industries or work in their own businesses. You can give your child a good start in life by treating him or her as a normal child who faces some extra challenges, not an invalid.

A parent says...

“It is very, very important for these kids to be treated as normally as possible and make sure that life carries on as normal for them. I had many battles to overcome as, grandparents especially, thought that by treating my son as just a normal child I was being awfully hard on him. They felt I should make allowances for his condition, but you have to be strong, at times it was very hard, but I feel that by persevering, he is the clever, intelligent, lovable young man that he is today. A young man who can hold his own amongst his peers!”

Visits to the hospital

Visiting the consultant who is responsible for the care of your child can be quite a stressful experience for parents, especially in the early days. It is better if both parents can attend these first visits so they can support each other and learn as much as possible from the medical team. When your child is young there is a lot to learn and you will have lots of questions to ask. It is a good idea to write questions down in advance so that you don't forget – you may have a list of questions in your head, but once the doctor or nurse starts speaking to you about other things it is all too easy to become distracted and forget your questions until you are outside the doctor's office! Writing your questions down will make sure that you get an answer to everything. If you do not understand when it is explained to you the first time, do not be afraid to say so and ask for another explanation – you are entitled to have your child's condition and treatment explained to you fully so that you can understand it. There is no need to be embarrassed if you don't understand medical terms and language – remember that doctors and nurses feel exactly the same when they are talking to plumbers and mechanics!

A parent says...

“When my daughter was a baby I started the habit of keeping a notebook with me and writing down any questions about thalassaemia that came into my head. Then on visits to the doctor, we would go through the list of questions until everything had been answered. If I hadn't written things down as I thought of them, I would never have remembered everything. At first I was a bit worried about presenting the doctor with so many questions, but it was fine – in fact it soon became a standing joke with the medical staff and as soon as they saw me they would laugh and ask if I had my list!”

Starting school

If your baby has just been diagnosed, school probably seems very far away in the distant future! However, time goes by all too quickly and you will soon be planning for your child's first school days. Before your child starts school, it is a good idea to make an appointment to see your child's teacher and, if possible, the school nurse so that you can explain about thalassaemia, your child's treatment and the fact that he or she may need to miss some school time due to medical appointments. It is very important that the school staff have an understanding of thalassaemia as your child will be in their care for a large part of the day. At UKTS we produce a leaflet specially designed for teachers and carers of children with thalassaemia. You can access this leaflet as a pdf on our website at <http://www.ukts.org/pdf.html> and you can order as many free copies as you need from the UKTS office. The leaflet contains basic information about thalassaemia and its treatment and has space for emergency telephone numbers for parents and the child's medical team. However, the leaflet also stresses that a child with thalassaemia is a normal child and other than a few basic precautions (such as being tolerant of tiredness in the week before transfusion) should be treated as any other child. There is no need for a child with thalassaemia to be automatically excluded from school activities or trips.

Communication between parents and school is essential for any child but even more so for a child with a long term medical condition such as thalassaemia. You can help the school staff to support and care for your child by keeping them informed of any developments or changes in your child's treatment so that they can be on the look-out for any symptoms or changes in behaviour. If you have any concerns whatsoever about your child do not hesitate to discuss them with the school staff. In some cases, it is possible for a member of nursing staff from the hospital to visit the school and talk to the staff about thalassaemia; so if you are having difficulties, please discuss them with your nurse specialist.

School and education are just as important for the child with thalassaemia as for any other child. Thalassaemia, although a challenging condition, should not prevent your child from achieving his or her academic potential. Even though your child may have to miss some school due to transfusions or medical appointments, the vast majority of hospitals have teachers and well equipped schoolrooms available; so children can bring their schoolwork and can often receive one-to-one tuition during their transfusions. In many cases they can also access their schoolwork over the internet while in hospital. So do ensure that you take homework into hospital so that your child can keep up to date.

As previously stated, we have many people with thalassaemia who are university graduates and who work at the highest levels in their chosen professions. As a parent, you can encourage and support your child by having a positive attitude and talking to him or her about the many high achieving people with thalassaemia who are excellent role models. If you need evidence of positive examples, please contact the UKTS office and we will be happy to help!

Diet in thalassaemia

Children who have thalassaemia will benefit from a normal, healthy diet containing plenty of fresh fruit, vegetables and milk. Protein such as fish or meat (in limited quantities) is also important. Growing children need a wide variety of nutrients, so do introduce your child to lots of different foods. At UKTS we have a detailed dietary advice sheet written by an eminent dietician with wide experience of thalassaemia. You can request your free copy from the UKTS office; or click on the link to our website <http://www.ukts.org/diet.html?i6s3s2>

Benefits and allowances

The link below will take you to the NHS Choices website; which has advisory pages on benefits which may be available to persons living with long term conditions or their carers

<http://www.nhs.uk/Planners/Yourhealth/Pages/Benefits.aspx>

Treatment for children who need regular blood transfusions

Because no two people are the same, the details of the treatment may be different for different people. However all patients who need regular blood transfusions also need iron chelation treatment. These two treatments form the basis of successful management of thalassaemia.

Blood transfusion

Our blood looks red because it is full of red blood cells. These cells are very important because they carry the oxygen from the air we breathe around our bodies. Oxygen gives us energy and enables all the other cells in the body to work properly. If our red blood cells are not working we get weaker and weaker and eventually will not be able to survive. People who have thalassaemia cannot make red blood cells which work properly, so many need to be given blood transfusions at regular intervals. Blood lasts in the body for about 28 days, so transfusions are usually given every 3 or 4 weeks, although this may vary.

In the week before a transfusion, the person with thalassaemia can tire easily and may need more rest. After the transfusion the patient will usually feel much more energetic! Doctors and nurses carefully check all thalassaemia patients to make sure that they get transfusions often enough so that they have enough energy to live a normal life.

As well as being important for giving energy, transfusions are essential in order for the child to grow and develop normally. Just as children need enough food and nourishment in order to grow, they also need enough oxygen in their bodies. If a child does not have blood often enough and becomes anaemic, the growing body will not have enough oxygen to develop correctly. This is why young children are very carefully monitored to make sure that they are transfused often enough for normal growth and development.

Transfusions can be started from a very young age. They are usually started at some time in the first year of life (this may vary). Babies who have thalassaemia should be checked every month by their thalassaemia doctor so the doctor can decide if and when to start transfusions.

What happens during transfusions?

In order to give the blood, a nurse or doctor will place a cannula (a tiny tube which can be attached to the tube carrying the blood from the bag on the drip stand) into the vein. This is done by inserting a needle into the vein and obviously this can be uncomfortable for the child. Small children have tiny veins and they can be difficult to find. However, most parents find that the nurse or doctor quickly becomes used to their child and knows how to find his or her veins easily. Hospitals usually have a policy that no more than 2 attempts at cannulation will be made at one time. Some health professionals use a small hand-held device which shines



a light through the skin and this helps them to see the veins more clearly. Many hospitals also employ play specialists who are trained in how to calm and distract a child who is undergoing a medical procedure and they can be extremely helpful. Most parents hold their small child while the blood transfusion is being set up. You should not be afraid to do this – small children will understand that even if the needle is unpleasant, it is for their good because their mummy or daddy is holding them. Once the cannula is in place, the rest of the transfusion procedure is painless and children can play games, watch TV, do homework etc.

Taking your child to hospital for transfusion can be traumatic in the early days. Every parent knows that there is nothing worse than seeing your child suffer pain; and most children start transfusions at an age where they are too young to understand what is happening. You as a parent can help your child by remaining as calm as possible. If your child sees that you are upset he or she will become even more distressed; so try your best to remain calm and put on a brave face, even if you don't feel brave. We don't for one moment underestimate how difficult this is! There may be times when it is impossible and you have to step outside, take some deep breaths and compose yourself. This is completely normal. Try to tell yourself that as time goes by children who have thalassaemia become used to the fact that they have regular transfusions - and so do their parents. Many parents take favourite toys, games or DVDs to hospital so that once the cannula is in place they can quickly distract and calm their child. If you are at a hospital where other children with thalassaemia are treated you may find it helpful to talk to other parents and find out how they cope. If you do not have other parents to talk to however, you can always call us at the UK Thalassaemia Society.

Chelation treatment

Chelation treatment is the second essential step in treating thalassaemia.

When a person has a blood transfusion, along with the important red blood cells they also receive a lot of iron. We all need iron, but people who have regular blood transfusions have too much iron in their bodies because they cannot get rid of the extra iron which comes with the blood. For this reason they need special medicines to get the iron out of their systems. This is called “chelation treatment”. If the extra iron is left in the system, it will settle in important organs of the body such as the heart and the liver where it causes damage. It can also affect other things such as growth and development. Without chelation treatment the body gradually becomes overloaded with iron and the heart and other organs will start to fail. Almost all the complications which can cause problems and illness for people with thalassaemia are either caused or made worse by iron overload. It is just as important for children as adults because some of the damage done by iron overload can occur in childhood and is not reversible (for example, poor chelation in childhood can lead to later problems with fertility). **So chelation treatment is just as important as blood transfusions. It is the key to remaining as healthy as possible both in childhood and in future life. By ensuring that your child takes their chelation treatment properly, you are laying the foundation for his or her future.**

Chelation treatment is usually started once a child has received about 10 or 12 transfusions. Each child is carefully monitored so the doctor can be sure it is the right time to start chelation.

There are different types of chelation treatment. The treatment which has been around the longest is a medicine called Desferal (desferrioxamine). This is very safe and very effective at removing iron from the body. However because Desferal does not work if it is taken by mouth, it has to be given by a needle under the skin. It works best if it is given very slowly, so the needle is attached to a small pump which gradually pushes the Desferal under the skin over about 12 hours. This must be done between 5-7 times per week. Some people find it most convenient to wear their pump while they are in bed at night, while others (usually adults) prefer to attach it during the daytime while they are working or going about their lives. For children, nurse specialists will teach parents how to attach the pump and make sure it is working properly. They can use a special cream which numbs the area before putting the needle under the skin. As children get older, they are encouraged to learn how to attach the pump themselves.

Of course, having a needle under the skin for hours at a time is inconvenient and can be very uncomfortable. The good news is that we now have other chelation medicines which can be taken by mouth, either as tablets or a powder which can be dissolved in water or juice. At the moment the medicines available which can be taken by mouth are called Exjade (deferasirox) and Ferriprox (deferiprone). Some patients are treated with a combination of Desferal and medicines which can be taken by mouth. This usually means that they do not need to use the Desferal pump as often.

The doctor treating the person with thalassaemia will decide which chelation treatment is most suitable for that patient. However, you are entitled to receive a full explanation of which treatment has been chosen and why. All medicines work better for some people than others and have side effects which some, but not all, people may experience. You may therefore meet other people whose child's treatment is different from yours. If you have any questions about chelation treatment, do not hesitate to discuss them with your thalassaemia doctor or nurse.

As research progresses it is likely that more medicines which can be taken by mouth will become available and that doctors will use different combinations of medicines so that every patient has the treatment which suits him or her best.

Problems with chelation

Whichever chelation treatment is chosen for your child, it is extremely important that the medicines are taken **exactly as prescribed by the doctor**. That is, at the right time of day and in the correct amount. If your child does not take the medicines exactly as prescribed, they will not work as effectively and he or she risks becoming overloaded with iron. Iron overload happens slowly over time, so if the chelation medicines are not taken properly, there will be no immediate signs of harm. This means that it is easy for parents or patients to fool themselves that missing a day here and there does not make any difference – but inside the body, the iron level will be slowly increasing. So every bit of Desferal in the pump, every tablet and every drop of medicine is important (yes, even the dregs in the bottom of the glass!). **Remember that the chelation treatment your child takes now is laying the foundation for better health in future life.**

A parent says...

“Sometimes the negative reaction of your child to treatment (cannulas, needles etc.) can almost wear you out. The tears and screaming can almost make you give in and miss a night. However you must persevere for your child's health in the short term and in the realisation that it is your reaction and approach to treatment that will determine your child's adherence to treatment in later life.”

If you are having difficulty getting your child to drink all the medicine, take all the tablets or you are having problems with the Desferal pump, tell your nurse or doctor as soon as possible and they will be able to advise you. Also remember that other parents will have experienced the same difficulties that you are having and many of them will have handy tips and hints for you; so again, contact with other parents can be extremely helpful. And once again, you can always call the UK Thalassaemia Society. Most children and indeed adults experience times when they have problems with chelation treatment, so do not hesitate to ask for help if you need it.

Specialist reviews

All thalassaemia patients should be seen at least once a year by a haematologist who specialises in the thalassaemias. For those who are not normally treated at a specialist centre, this will probably involve travelling to another hospital, possibly some distance away. Although this may seem inconvenient, it is a vital part of treatment. Thalassaemia is rare in the UK, which means that many otherwise excellent doctors have not had the opportunity to build up the experience and expertise of those doctors who work in one of the specialist centres.

It is no disrespect to your usual doctor to ask for a specialist review, if it is not offered. In fact it is a vital part of the Clinical Standards for Children and Adults with Thalassaemia in the UK. (See page 22 for details of how you can get your own copy of the Clinical Standards.) Doctors are well aware that no-one can be a specialist in everything; and that people who have a rare condition will need their treatment to be reviewed by an expert in that condition.

During the annual review, the specialist doctor will check that all the necessary monitoring tests have been carried out and will order more tests if necessary. S/he will also check whether any drug treatment needs to be changed or adjusted.

A parent says...

“Parents shouldn't feel afraid to ask for a referral to a specialist. I am in no doubt that my son's good health is down to the excellent relationship we have had with our local doctors & nurses and their close liaison with his specialist consultant in London. We feel blessed because we know that sadly this is not always the case. It is a parent's responsibility to care for their child, to provide for them to the best of their ability and getting the best care for them is of paramount importance.”

Bone marrow transplants

Bone marrow transplant (BMT) from an identical tissue-type family donor offers the only cure for thalassaemia at the present time. Parents who have a child with thalassaemia should be offered the chance to discuss BMT when their child is around 12-18 months old. The discussion should be with a specialist who has experience of transplanting for thalassaemia.

Tissue matched donors are most commonly found among brothers or sisters of the affected child, but there is no guarantee that a match will be found. (In families which practise cousin marriage, sometimes a first cousin or other family member may be a tissue match.) Family members who may be donors will need to have a blood test which will tell if they are a tissue match. If a tissue match is found, the family will be given specialist counselling about the BMT procedure and further to this they can decide whether or not to proceed.

BMT has the best chance of success in a child between 18 months – 3 years of age who

has been very well chelated and has no existing complications. All patients are very carefully assessed before being considered for BMT. There are risks associated with the procedure and the parents will be fully informed as part of the counselling procedure before they make a decision.

If the BMT is successful, the child will no longer need transfusions or chelation treatment throughout life, although chelation may need to continue for a while until the iron levels in the body are back to normal.

BMT is not available to all patients as not everyone will have a matched donor. It is important to remember that modern treatment with transfusions and chelation therapy has generally excellent results.

Monitoring – avoiding complications

People who have thalassaemia are at risk of suffering from a number of complications. The most common of these include:

- Growth & development problems
- Fertility problems
- Diabetes
- Heart problems
- Liver problems
- Bone problems

Most of these problems are caused or made worse by iron overload. You can minimise the chances of severe complications by making sure that your child takes the chelation medicines exactly as your doctor prescribes. A list of monitoring tests is available from the UKTS office, or follow this link to the pdf library on our website <http://www.ukts.org/pdf.html>. The pdf library also has a range of articles by internationally renowned specialists on the various complications of thalassaemia. If you would like to have copies of any of these articles posted to you, please contact us at the UKTS office.

Going for monitoring tests

Most of the monitoring tests are blood tests which will be done during routine visits to the hospital, although some such as heart and liver scans will need a special appointment. The heart and liver scans are painless, but the child will be required to keep still during the scan. Don't be afraid to ask for more information if either you or your child feels at all apprehensive about the test.

A parent says...

“When my son had to go for a heart scan, he was a bit nervous about what would happen. I looked on the internet and found that there was a demonstration on YouTube. After watching it first myself, I watched it with my son and he felt very reassured once he knew what to expect.”

Growth & development

As we all know, around the approach to our teenage years our bodies begin to change; we grow taller and gradually become adults. These changes are caused by hormones which are produced by glands in our bodies. However these glands are particularly easily damaged by iron overload, so once again we are back to the subject of chelation. Although there are no guarantees, a well chelated child has a far better chance of producing a normal or near normal level of hormones. So yet again we must stress the importance of taking the chelation treatment exactly as the doctor prescribes it. All children who have thalassaemia should be regularly monitored for growth and development and referred to an endocrinologist (a specialist doctor who is an expert in hormones) at the appropriate time if there are any problems.

Fertility

In the past it was highly unusual for men or women who had thalassaemia to become parents. Thankfully, however, this has changed for the better and there is every reason for young people today who have thalassaemia to look forward to marriage and family just as other people do. In the UK we now even have some thalassaemics who are grandparents! Some people with thalassaemia (both male and female) need medical treatment to help them achieve parenthood, as do quite a significant number of people without thalassaemia - infertility is estimated to affect around one in six or one in seven UK couples at some point (ref. Human Fertilisation & Embryology Authority *Facts and Figures for Researchers and the Media 2011* www.hfea.gov.uk). A person's fertility is closely linked to growth and development (see above); so making sure your child takes all their chelation treatment will improve his or her chances of becoming a parent once they reach adulthood.

Diabetes

Diabetes is a problem caused by the body's inability to use glucose (sugar) in the blood. A lot of the food we eat is converted into glucose by the digestion. The body's cells need glucose to give them energy. Glucose remains in the blood until it is released into the body's cells by a hormone called insulin. In diabetes the body does not produce enough insulin or is not able to use the insulin it produces; so the level of glucose in the blood increases and the body's cells cannot get the glucose they need. Insulin is produced by an organ called the pancreas, which lies next to the liver. Like many other organs, the pancreas can be damaged by iron overload, so diabetes is a common complication of thalassaemia. Diabetes is managed by medicines which are given orally or by injection. People with thalassaemia can reduce their risk of developing diabetes by maintaining a healthy lifestyle (eating a healthy diet and taking regular exercise) and of course by adhering strictly to their prescribed chelation treatment.

Heart problems

Heart disease from iron overload is a very serious complication in thalassaemia, so yet again we are back to our old friend chelation treatment as the best way of preventing these

complications. A patient who is well chelated is far less likely to develop heart complications. Other things which can help to maintain a healthy heart are; encouraging regular exercise, a healthy diet and for adults, not smoking or drinking excessive amounts of alcohol.

Because heart complications are serious, every patient is carefully monitored from a young age, so that any problems can be identified and dealt with before they become severe. Therefore every child with thalassaemia should have a special heart scan (called a cardiac MRI T2*) every year from the age of 8 onwards. From the age of 10, every child should be regularly assessed by a heart specialist who is experienced in treating thalassaemia patients.



A doctor and patient discuss the results of a liver scan

Liver problems

Liver disease is common in thalassaemia, but again every patient is monitored and problems should be identified in the early stages. Liver function is tested every 3 months (this is a blood test). Special liver scans may also be carried out. As with all the other complications, good chelation can keep any iron loading in the liver to a minimum.

In the past liver problems were often caused by viruses which were caught from donated blood, but this is now very rare as all blood in the UK is screened to the highest standard and all patients receive vaccinations against blood-borne viruses.

Important note on travelling abroad: if you are travelling abroad it is not advisable to have a blood transfusion outside the UK where blood safety may not be as strictly monitored. Although there are clinics overseas which advertise their blood as being screened, the screening is often not done to the same rigorous scientific standards as in the UK. Any patient who has even one blood transfusion overseas runs the risk of exposure to dangerous viruses. For this reason it is safer to limit trips abroad to the time taken between transfusions (usually 3-4 weeks).

Bone problems

Osteoporosis is one of the commonest complications of thalassaemia. The causes are complex and may be linked to several factors, for example iron overload and delays in growth and development. Therefore bone mineral density scans are carried out regularly from age 10 onwards so that any problems can be detected at any early stage. Osteoporosis may be managed by hormone replacement treatment, calcium and vitamin D supplements, regular exercise and avoiding smoking.

FOR A FULL AND COMPREHENSIVE GUIDE TO THALASSAEMIA TREATMENT, PLEASE REFER TO THE *“STANDARDS FOR THE CLINICAL CARE OF CHILDREN AND ADULTS WITH THALASSAEMIA IN THE UK”*. YOU CAN READ THIS ON LINE AT <http://www.ukts.org/pdfs/awareness/standards/ukts-standards-2008.pdf>

OR TO REQUEST YOUR FREE COPY, PLEASE CONTACT THE UK THALASSAEMIA SOCIETY.



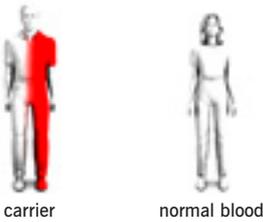
Children at a UK Thalassaemia Society conference

More information about thalassaemia

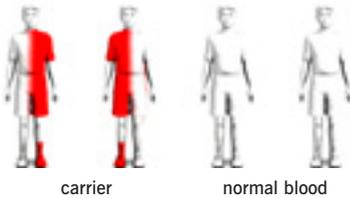
As previously stated on page 7, thalassaemia is an inherited condition, that is, it is something which is passed on from parents to children in a similar way as parents may pass on other physical characteristics like eye colour or the shape of a nose. Thalassaemia cannot be caught or acquired in any other way. Some people are healthy “carriers” of thalassaemia. Carriers are not ill in any way, but if a man and a woman who are **both** carriers have children together there is a risk that some of their children could be born with thalassaemia.

Carriers are perfectly healthy and most do not know that they carry thalassaemia. If a man and a woman who are **both** carriers have a child together, there is a 25% chance that their baby will be born with thalassaemia. The chances are the same with **every pregnancy**, so a couple who are both carriers are known as a “couple at risk”; because each time they have a baby together there is a 25% risk that the baby could be born with thalassaemia.

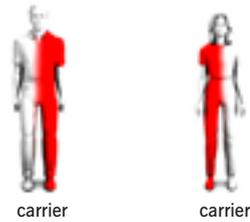
Diagrams of carrier possibilities



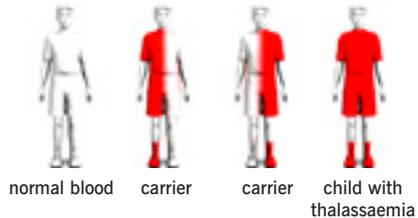
If one parent is a thalassaemia carrier...



No chance that any child will have thalassaemia
 50% chance that any child will be a thalassaemia carrier
 50% chance that any child will have normal blood



If both parents are thalassaemia carriers...



25% chance that any child will have thalassaemia
 50% chance that any child will be a thalassaemia carrier
 25% chance that any child will have normal blood

Testing and screening for thalassaemia

As mentioned above, carriers are perfectly healthy so they would have no warning that they carry thalassaemia. However, a simple blood test can tell you if you are a carrier. This test is called a “carrier test for haemoglobin disorders” or a “haemoglobinopathy screen”. **You should ask your GP to arrange for you to have this test before you have children, especially if:**

You, your parents or grandparents come from one of the parts of the world where thalassaemia is common (see below).

Map showing affected areas



Anyone can be a carrier of thalassaemia, but in some parts of the world there is more thalassaemia than others. The highest risk areas are:-

- All parts of Pakistan, Bangladesh and India
- All parts of South East Asia
- All parts of the Middle East
- All parts of Southern Europe and the Mediterranean
- Parts of North and East Africa

Antenatal screening for thalassaemia

In England all pregnant women are offered a test for thalassaemia and if they are found to be carriers, the baby’s father is also offered a test. This is called antenatal screening. If both parents are found to be carriers, the unborn child is at risk of having thalassaemia. The parents are then offered counselling and, if they wish, a test which will show whether the unborn child has thalassaemia. You can read more about antenatal screening on the website of the NHS Sickle Cell and Thalassaemia Screening Programme <http://sct.screening.nhs.uk/>



Although screening is done during pregnancy, it is very difficult for a young couple to find out that they are at risk when they already have a baby on the way. It is far better to find out beforehand so the couple have plenty of time to find out all the relevant information and talk about their options without the added pressure of knowing that their unborn child may be at risk.

Your extended family

If you already have a child with thalassaemia, you are a thalassaemia carrier. This means that at least one of your parents is a carrier, so your brothers, sisters and cousins may also be carriers. Please encourage your relatives to get tested before they have children, if they have not already done so. The UK Thalassaemia Society has a film on DVD about thalassaemia testing called *Thalassaemia – your life, your choice, your test*. It features interviews with thalassaemia patients of varying ages and backgrounds, parents of children with thalassaemia and experienced health professionals. The film explains how thalassaemia is inherited, what it means to be a carrier and shows that anyone can find out if they are a carrier or not by a simple blood test. If you would like to show this film to members of your family, you can order a copy **FREE** from the UK Thalassaemia Society.

If you or anyone you know has difficulty in getting a test please contact the UK Thalassaemia Society for advice.

Warning: do not assume that if your blood was tested in the past for some other reason, thalassaemia would have “shown up” on the test – you need to have a specific test to be sure.

Preimplantation genetic diagnosis and HLA tissue typing

Preimplantation Genetic Diagnosis (PGD) is an IVF technique, which allows couples where both partners carry thalassaemia, to select unaffected embryos before a pregnancy. Just like in IVF, a number of the mother's eggs are fertilised with the partner's sperm in the laboratory. The embryos are then tested for thalassaemia and unaffected, healthy embryos are selected and transferred back into the mother's womb.

HLA tissue typing uses the same technique, but as well as selecting embryos that do not have thalassaemia, the embryos are tested for bone marrow donation compatibility. This allows parents who have an affected child, and wish to have another healthy child, to ensure that their next child will be a suitable bone marrow donor for their existing affected child.

PGD and HLA tissue typing are not currently available as NHS services but are available privately. The funding arrangements may change in the coming years. Check with the UK Thalassaemia Society or Genetic Alliance UK for more information.



A UK Thalassaemia Society national conference

Other types of thalassaemia

Thalassaemia intermedia

Sometimes a child who has inherited thalassaemia from both parents will have a form of thalassaemia called thalassaemia intermedia. These patients do not have an absolute requirement for regular blood transfusions during childhood, although most will eventually require them. Thalassaemia intermedia is difficult to describe as it covers a wide range, from patients who are only just able to manage without transfusions during childhood to those who have almost no symptoms. About 10% of children who have inherited beta thalassaemia from both parents will have moderate to severe thalassaemia intermedia.

Although thalassaemia intermedia is seen as a milder form of thalassaemia, there are still a range of complications which may develop as time goes by. Therefore all children and adults with thalassaemia intermedia need to be monitored by a haematologist.

E/beta thalassaemia

E/beta thalassaemia is caused when a child inherits beta thalassaemia from one parent and a slightly different form of thalassaemia called haemoglobin E from the other parent. It is common among certain ethnic groups, e.g. people from Bangladesh where both thalassaemia and haemoglobin E are common in the general population. About half of those with E/beta thalassaemia need regular blood transfusions, and the rest have a moderate or severe thalassaemia intermedia.

Alpha thalassaemia

Alpha thalassaemia is extremely common in some parts of the world. The commonest form, **alpha plus thalassaemia**, is harmless even if it is inherited from both parents. About one in three people from Africa or the Indian sub-continent carry harmless alpha plus thalassaemia. It is also common in people from the Mediterranean area, the Middle East or South East Asia.

However there is another alpha type, **alpha zero thalassaemia**, which can cause serious problems. If a couple both carry alpha zero thalassaemia and their child inherits alpha zero thalassaemia from both parents, the unborn baby's blood cannot carry oxygen and the baby does not develop normally. This usually causes a late miscarriage or a stillborn baby. There are usually problems in pregnancy for the mother, who may develop high blood pressure and other problems. This condition is called **alpha thalassaemia major or Bart's hydrops fetalis**. It should be detected by antenatal screening and the parents are counselled and offered the option of whether to continue with the pregnancy.

In a few cases a baby with alpha thalassaemia major has been given blood transfusions while in the womb and this has enabled them to survive. These children require regular blood transfusions like patients with thalassaemia major. This type of treatment is very unusual. It must be done by an expert team, and is only possible if the baby is diagnosed very early in pregnancy.

Carriers of alpha zero thalassaemia are common among people of Chinese or South East Asian descent, and quite common among people of Cypriot origin. Fortunately cases of alpha thalassaemia major are extremely rare in the UK.

Sometimes a baby inherits alpha plus thalassaemia from one parent and alpha zero thalassaemia from the other parent. In this case the child will have a condition known as **haemoglobin H disease**. This is usually a moderate anaemia which causes very few medical problems and does not require blood transfusions. The name “haemoglobin H disease” is rather unfortunate as the great majority of people with this condition lead perfectly normal lives. They may occasionally need additional medical treatment, for example if they develop a bad infection. Children and adults with haemoglobin H disease should see a haematologist for a check-up once a year.

You can find more information on the alpha thalassaemias and other unusual haemoglobins on the website of APoGI (Accessible Publishing of Genetic Information) <http://www.chime.ucl.ac.uk/APoGI/>

List of website links you may find useful

- **APoGI** (*Accessible Publishing of Genetic Information*)
www.chime.ucl.ac.uk/APoGI
- **Contact a Family** (*for people who have children with disabilities or rare disorders*)
www.cafamily.org.uk
- **Genetic Alliance UK** (*national charity of patient organisations supporting all those affected by genetic disorders*)
www.geneticalliance.org.uk
- **NHS Choices** (*benefits and allowances*)
www.nhs.uk/Planners/Yourhealth/Pages/Benefits.aspx
- **NHS Sickle and Thalassaemia Screening Programme**
sct.screening.nhs.uk
- **Thalassaemia International Federation**
www.thalassaemia.org.cy
- **Thalassaemia Patients and Friends**
www.thalassemiapatientandfriends.com
- **Thalforum** (*online discussion forum for thalassaemia*)
www.thalforum.ca
- **UK Forum on Haemoglobin Disorders**
www.haemoglobin.org.uk
- **UK Thalassaemia Society**
www.ukts.org

About the UK Thalassaemia Society



The UK Thalassaemia Society was formed in 1976 by the parents of children who had thalassaemia. They were motivated to start the Society by Professor (then Doctor) Bernadette Modell, who was one of the earliest pioneers of thalassaemia treatment. Most of the parents were Greek or Turkish Cypriots who had settled in the North London area; and most of their children were treated at the Whittington Hospital; which is still one of the leading UK centres for thalassaemia treatment today. In the early days UKTS was a small support group, with parents taking turns to host meetings in their houses. Treatment in the 1970s was in its infancy, with blood transfusion being almost all that was available. Transfusions alone however were not enough and most patients died around the age of 20 from iron overload; so the parents in those days were faced with the terrible knowledge that their children would probably not survive to adulthood. With amazing courage, however, they were determined to pursue better treatment for their children; and with the help of dedicated doctors and researchers, thalassaemia treatment gradually improved until now in the UK, we regard thalassaemia as a serious but manageable disorder.

Some of the achievements of the UK Thalassaemia Society are:-

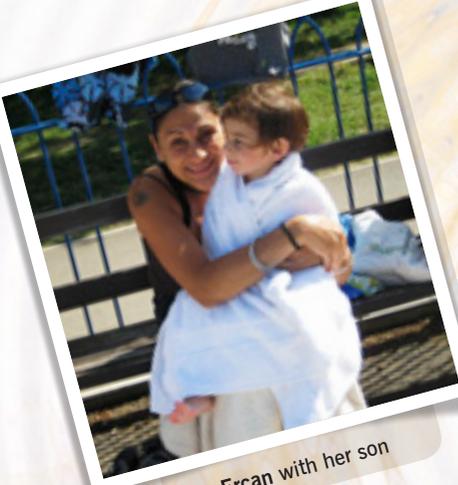
- Raising £750,000 for research leading to the development of the first oral chelating medicine deferiprone
- Fund raising for development of the infuser pump (for the administration of desferrioxamine)
- Funding for research into growth and development
- Funding for gene therapy research
- Producing the *Standards for the Clinical Care of Children and Adults with Thalassaemia in the UK* (1st edition 2005)

Today the UK Thalassaemia Society has members all over the world. The Society is represented on the Board of the Thalassaemia International Federation; and in the UK we work in partnership with: the UK Forum on Haemoglobin Disorders, the All Party Parliamentary Group for Sickle Cell and Thalassaemia and the NHS Sickle Cell and Thalassaemia Screening Programme. We produce information for families affected by thalassaemia, health care professionals and the general public; and organise meetings and conferences for both families and health professionals.

Our mission statement is **“to be the definitive source of information for those affected by, or working with thalassaemia”**.

Visit our website www.ukts.org for a wide range of articles and information on thalassaemia. We also have information and fact sheets in different languages. If you don't see what you need please contact our office (office@ukts.org or 020 8882 0011) and we will do our best to help.

Living with thalassaemia



Ercan with her son



Mumta with her husband and
twin daughters



Helen with her husband and
baby daughter



Tanver with his wife and
children



Gabriel on his wedding day

UNITED KINGDOM
THALASSAEMIA SOCIETY



An initiative of:

Genetic Alliance UK
Supporting. Campaigning. Uniting.

