**Primary Care Information on Thalassaemia and Rare Inherited Anaemia**

1. **Thalassaemia**

The term “thalassaemia” refers to a group of blood diseases characterized by decreased or absent synthesis of one or more of the normal globin chains. The most clinically relevant types are α and β thalassaemias.

Based on their clinical severity and transfusion requirement, thalassaemia syndromes can be classified into transfusion-dependent thalassaemias (TDTs) and non-transfusion-dependent thalassaemias (NTDTs).

Transfusion-dependent thalassaemias (TDTs) – mostly Beta Thalassaemia Major, HbE/Beta thalassaemia or severe HbH disease.

Non-transfusion-dependent thalassaemias (NTDTs) – mostly Beta Thalassaemia Intermedia and moderate HbH disease.

**Please note**: Thalassaemia trait / minor does not fall under this patient group. Thalassaemia trait is usually asymptomatic, and these patients are under their own General Practitioner.

**Main health conditions associated with thalassaemia:**

People with thalassaemia often experience:

* Anaemia – severe tiredness, weakness, shortness of breath, pounding, fluttering or irregular heartbeats (palpitations) and pale skin caused by the lack of haemoglobin.
* Iron overload – caused by regular blood transfusions used to treat anaemia and can cause problems with the heart, liver and hormone levels if untreated.
* Some people may also have delayed growth, weak and fragile bones (osteoporosis), and reduced fertility.

**Treatments:**

Regular or top up transfusions:

People with a more severe type of thalassaemia will need regular transfusions to treat the anaemia.

People with a less severe type of thalassaemia might only need occasional top up transfusions if severely anaemic e.g. during pregnancy or in adolescence.

Blood transfusions are safe, but when given regularly they will cause iron to build up in the body, so regular monitoring with blood tests and/or MRI T2\* scans are required. Iron overload requires treatment with medication called chelation therapy to remove the excess iron. This will be prescribed and monitored by the hospital.

Some children will have a port-o-cath insitu to facilitate regular blood transfusions. Parents/carers are taught to recognise the signs of port infection and advised to attend for medical review as a matter of urgency if they have any concerns.

Iron chelation:

There are three types of chelation therapies available, either used individually or in combination:

* Deferasirox (DFX)
* Desferrioxamine (DFO)
* Deferiprone (DFP)

Most children and young people will be treated with Deferasirox which is given orally. If there is significant iron overload other treatments may be added. Desferrioxamine is given either subcutaneously or intravenously by the parents/carer or via a home care provider. All prescriptions for iron chelation will be undertaken by the BRHC team, who will also undertake the necessary monitoring. If there are any problems related to this, please do contact our haemoglobinopathy team.

Stem cell or bone marrow transplants:

Stem cell or bone marrow transplants are the only curative treatment for thalassaemia but are only routinely offered if a fully matched full sibling (brother/sister) is available.

**Other health issues often associated with thalassaemia:**

Thalassaemia can also cause several other health problems that may need to be treated. Referrals for thalassaemia related health issues will be made directly from the thalassaemia outpatient clinic. This may include referrals to the endocrine, cardiac, ophthalmology and audiology teams.

**When to contact our Haemoglobinopathy team?**

We recommend that, if thalassaemia patients present with any infection or worrying symptoms (including severe anaemia) please contact the haemoglobinopathy team for further advice. Please see contact details below.

**Appropriate vaccination:**

Children should be vaccinated according to the routine national childhood vaccination schedule.

Influenza vaccination should be given annually from age 6 months. For children aged 6 months to 2 years, give intramuscular flu vaccine and for those children aged 2 years to 17 years, Fluenz tetra nasal spray (a live attenuated vaccine).

Splenectomy is now rarely performed in children with thalassaemia in the UK. It is quite widely used in resource–limited countries, to reduce blood product requirements in TDT or to try to avoid regular transfusions in patients with NTDT. Patients who have been splenectomised should receive specific vaccinations according to Green book Chapter 7 and should be given prophylactic Penicillin V.

<https://assets.publishing.service.gov.uk/media/5e18a52940f0b65dc1918763/Greenbook_chapter_7_Immunsing_immunosupressed.pdf>

**Sexual Health and Contraception**

There are no specific restrictions with regards to contraceptive use. Use of long-acting reversible contraceptive methods such as injectables, implants and intrauterine devices are more effective in preventing pregnancies than user-dependant methods such as oral contraceptive pills and barrier methods. Planned pregnancy is essential, both in spontaneous and assisted conceptions, since pregnancies in patients with TDT are high risk for both the mother and the baby.

**You and your team in primary care can help by:**

1. Remembering that symptoms which may be trivial in others [e.g. sore throat, fever of 38.0°C or higher] may warn of significant bacterial infection in those with thalassaemia: please give broad spectrum antibiotics early and refer to us promptly via Children’s Emergency Department (CED) if there is any possibility of sepsis. **NB If a child has a port insitu and presents a temperature of 38.0c this needs to be treated promptly and the child should be referred to CED for review.**
2. Being aware of the side effects of some of the medications he/she may require in the future: for example,
   1. ***Deferasirox*** is used to reduce iron levels in transfused patients, and it can cause abdominal, rash, and kidney and liver function abnormalities.
   2. If people on ***Desferrioxamine*** develop acute abdominal pain and diarrhoea, we will need to assess them in case of Yersinia bowel infection.
   3. ***Deferiprone***, sometimes used to reduce iron levels in patients on regular transfusions, can cause agranulocytosis. Anyone on this medication should be referred immediately to hospital if febrile.

**Please contact us immediately if you have any concerns about anyone on these medications.**

1. Encouraging/giving annual flu vaccine and other vaccinations as needed.
2. Prescribing regular medications including Folic acid and Vitamin D supplementation, and Penicillin V in splenectomised patients, as outlined in outpatient review letters.

In general, we advise patients under our care to contact us if they experience fever or feel unwell. Patients unknown to us can be referred to the haemoglobinopathy service directly via eRS, urgent enquires for unknown patients can be referred via the Paediatric Benign Haematology SpR on the number below.

1. **Rare inherited anaemia**

Types of rare inherited anaemias:

* Diamond Blackfan anaemia (Congenital pure red cell aplasia)
* Deficiency of Adenosine deaminase 2 (DADA2)
* Congenital dyserythropoietic anaemia (CDA) Type I, Type II, Type III, Type IV, Type VI, Type other
* Congenital Sideroblastic anaemia
* Transfusion dependent membrane disorders eg Hereditary Spherocytosis
* Some types of G6PD deficiency
* Pyruvate Kinase Deficiency
* Glucose Phosphate Isomerase Deficiency
* Hexokinase Deficiency
* Unstable Haemoglobins
* Congenital Methaemoglobinaemia

Patients with rare inherited anaemia are a diverse group of patients and in addition to anaemia there may be other associated features e.g. growth and developmental delay, immunodeficiency.

For the patients who receive regular blood transfusions please refer to the previous sections under thalassaemia. Splenectomy is relatively commonly performed in children with transfusion dependent membrane and enzyme disorders, as this may ameliorate or significantly reduce the transfusion burden.

**More information needed?**

Refer to the resources for healthcare professionals on thalassaemia on the Thalassaemia Society UK website <https://ukts.org/healthcare-professionals/>

**Contact details**

**Monday to Friday 09.00hrs – 17.00hrs**

Paediatric Benign Haematology CNS team - 0117 342 8721

Paediatric Haematology registrar - 0117 923 0000 (switchboard) Bleep 3495

Paediatric Benign Haematology SpR 0788016927

**Out of hours, weekends and bank holidays**

Paediatric registrar on call for oncology/haematology via switchboard – 0117 923 0000