

Routine investigations in the management of thalassaemia

All thalassaemia patients (children and adults) should be assessed at least once a year at a specialist thalassaemia centre by a paediatrician or haematologist experienced in the treatment of thalassaemia. This is intended to give a detailed assessment of every aspect of the patient's treatment and condition; to assess the patient's progress and identify any areas where treatment could be improved. Additional investigations and measurements should be kept between the annual review checks, and recorded and discussed. The results can be kept in a patient-held document and should be kept on a flow sheet in the hospital records which can accompany the patient to the annual review visit. This list does not include the usual clinic discussions which should take place about general health and wellbeing, any problems or side effects from medication or difficulty in adhering to prescribed medication.

Growth and development (children)

- The height of both parents should be recorded in the child's medical records.
- The child's height (both standing and sitting) and weight should be measured at least once every 6 months and plotted on growth charts.
- There should be an examination of how puberty is progressing, every year from age 10 onwards.
- For girls, the doctor should regularly check if periods have started and if so, that they are continuing.
- If the patient is growing more slowly than usual OR there has been no development of puberty (by age 13 in girls or age 14 in boys) OR puberty has started but ceased to progress, the patient should be referred to a paediatric endocrinologist who is experienced in the treatment of children with thalassaemia.
- Any hormone replacement treatment recommended by the endocrinologist should usually be started by the age of 16.

Iron levels

- Ferritin levels should be tested every 3 months.
- Heart and liver MRI scans (T2* for the heart, FerriScan for the liver where available) should be done yearly OR more frequently if levels are high and the patient is being treated with extra chelation drugs. Sometimes if levels are consistently good a longer interval between heart assessments may be satisfactory.

Liver function

- A liver function test should be carried every 3 months (this is a blood test).
- An ultrasound scan should be done if gallstones are suspected.
- Viral hepatitis testing (these are blood tests) should be done if there are abnormalities in the liver function blood tests which suggest liver inflammation.
- Regular (yearly or as indicated by the microbiology laboratory on the results of a previous test) blood checks should be made to ensure the child is immune to hepatitis B virus and if not then the child should receive immunisation for this.

Heart function

- Examination of the heart and circulation should be done at least every 6 months.
- Heart function should be assessed by a specialist (cardiologist) experienced in the treatment of thalassaemia patients, every year from age 10 onwards.
- If there are any suspicions of heart problems between these regular assessments the patient should be immediately referred to a specialist cardiologist.

Endocrine function

- Blood glucose test every 3-6 months and oral glucose tolerance test if results are high.
- An oral glucose tolerance test should be done every year from puberty onwards; or from age 10 onwards if there is a family history of diabetes.
- Thyroid function tests (these are blood tests) should be done every year from age 12 onwards or earlier if there is any suspicion of a problem.
- Calcium and phosphate measurements (blood tests) should be done every 3-6 months from age 12 onwards.
- Vitamin D deficiency is common and should be checked if other biochemical tests suggest it may be present (calcium and alkaline phosphatase levels).

Bone monitoring

- Advice about a calcium-rich diet, the importance of exercise and avoiding smoking should be given.
- A DXA (bone mineral density) scan should be done every 18-24 months (or annually if there is concern) from age 10 onwards.

Other tests

- Hearing and eye tests every year from age 10 onwards if patient is on Desferal (desferrioxamine).
- Frequent full blood count if patient is on Ferriprox (deferiprone) – the manufacturer's recommendation is for weekly checks.
- Assessment of zinc levels every 3 months (blood test) if taking Ferriprox.
- There should be an annual review of pre-transfusion haemoglobin levels as well as annual blood usage.