

born around the world with the disorder each year.

The outlook for people with severe Thalassaemia is improving as treatment improves. A bone marrow/stem cell transplant is possible for some children with Thalassaemia but is not without risk. It is best done when the child is still young and the donor must be a close family member (e.g. brother, sister, mother or father) who is an exact tissue match.

### Pre-natal testing

Pre-natal testing is available for couples who may wish to know if their baby may be affected with thalassaemia. A very early test at 11 weeks called the chorionic villus sampling test (also known as a CVS) can be performed during pregnancy to test the baby. A test called the Amniocentesis is also available later in pregnancy.

If you wish to consider any of the tests above it is vital to contact your local Genetics Centre before nine weeks of pregnancy to make arrangements for the test.

### Pre-implantation genetic diagnosis

Pre-implantation genetic diagnosis also known as (PIGD) is available in some centres.

## Thalassaemia screening

The NHS Thalassaemia screening programme was set up in 2001 across England, Thalassaemia screening is offered during the antenatal and newborn periods for more details please view the website below.

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

### For more information

If you are interested in finding out more about Thalassaemia, you can contact the UKTS at:

UK Thalassaemia Society,  
19 The Broadway, Southgate Circus,  
London, N14 6PH.

Tel: 020 8882 0011  
Fax: 020 8882 8618  
E-mail: [office@ukts.org](mailto:office@ukts.org)  
Website: [www.ukts.org](http://www.ukts.org)

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**Birmingham Women's  
NHS Foundation Trust  
Edgbaston, Birmingham, B15 2TG  
Telephone: 0121 472 1377  
Fax: 0121 627 2602**

Reference Number: GG 15  
Author: Shagufta Khan, Genetic Counsellors  
Reviewed: June 2011 Next Review: June 2014

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# Thalassaemia

An information leaflet for  
patients and families

If you need more advice about any aspect  
of Thalassaemia please contact:

**Clinical Genetics Unit**  
Birmingham Women's  
NHS Foundation Trust  
Mindelsohn Way  
Edgbaston  
Birmingham  
B15 2TG

Telephone: 0121 627 2630  
Fax: 0121 627 2618

Email: [Clinicalgenetics.info@bwhct.nhs.uk](mailto:Clinicalgenetics.info@bwhct.nhs.uk)

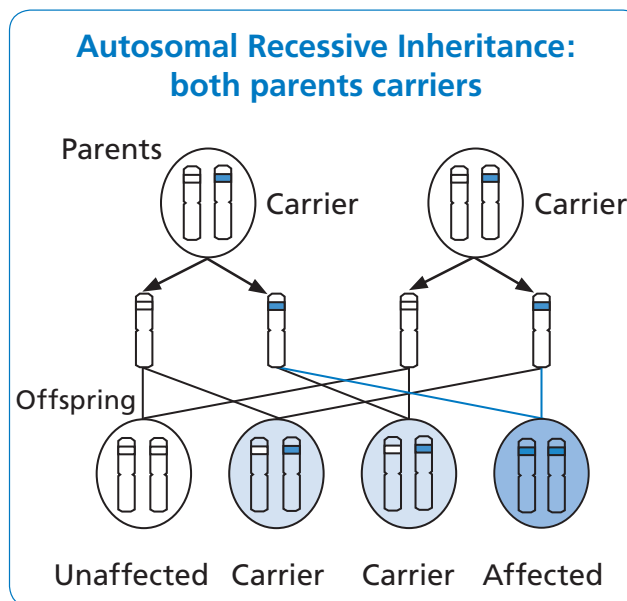
## What is Thalassaemia?

Thalassaemia (pronounced thal-er-seemeeya) is an inherited blood disorder in which there is a defect in the structure of haemoglobin. Haemoglobin is a protein which is contained in the red cells of the blood. It picks up oxygen from the air in the lungs and carries it to the tissues of the body where it is needed. A person who does not have enough haemoglobin is anaemic (pronounced er-nee-mick). There are different types of Thalassaemia. This leaflet is about Beta (pronounced beet-er) Thalassaemia.

## Is Thalassaemia passed on?

The pattern of inheritance for Thalassaemia is autosomal recessive. A person who inherits one altered gene for Thalassaemia will be a carrier. Carriers are unaffected but can pass the altered gene onto any children they may have. If one or both parents is a carrier, there is a 50% (1 in 2) chance that each child of theirs will also be a carrier. Carriers of Thalassaemia are sometimes said to have Beta-Thalassaemia trait (formerly referred to as thalassaemia minor).

A child who inherits two copies of the altered gene (one from each parent) will have Thalassaemia major. If both parents are carriers, there is a 25% or (1 in 4) chance of this happening.



## A person with Beta-Thalassaemia major may be affected in some, or all, of the following ways:

- Children with Beta-Thalassaemia major appear healthy at birth but become anaemic between the ages of 3 and 18 months. They become pale, irritable and weak, do not sleep well, lose their appetite and may vomit feeds.
- Once Beta-Thalassaemia major has been diagnosed, it will be recommended that the child has regular blood transfusions every four to six weeks for the rest of his/her life.
- Regular blood transfusions help by getting more red blood cells into the body, but these red cells contain iron and the body cannot easily get rid of excess iron. So treatment by blood

transfusion must be followed by a chelating drug such as Desferal to avoid damage to the heart, liver and other major organs. Desferal cannot be absorbed if taken orally and must be given by a slow infusion under the skin over 10-12 hours per day, at least 5 times a week.

- Oral preparations are becoming available, used singularly or in combination with desferal injections.
- Thalassaemia patients may be affected by complications such as diabetes, osteoporosis and endocrine problems.

## Other Information

Beta-Thalassaemia affects mainly people of Mediterranean, Middle Eastern or Asian origin, but is occasionally found within the Northern European Community. It is thought that Thalassaemia is more common in these parts of the world because people who carry the Thalassaemia gene are protected from the more severe forms of malaria. Thalassaemia is rare in North Europeans.

There are about 200,000 people in Britain who are carriers of Beta-Thalassaemia. They are healthy themselves but if their partner also carries the Thalassaemia gene, there is a 1 in 4 (25%) chance for each child that they will have Beta-Thalassaemia Major. At the moment, there are about 830 young people with thalassaemia in Britain, but at least 100,000 children are