Thalassaemia Society of NSW

The NSW Thalassaemia Centre

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Become a Member of the Society

☐ YES I would like to become a **Financial Member** $20
☐ YES I would like to become a **Lifetime Member** $200
☐ YES I would like **Lifetime Family Membership** $400
☐ YES I would like to **donate $**.....

Donations of $2 and over are tax deductible.

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Information & Privacy Policy:

The Thalassaemia Society of NSW Inc will use your information for the purpose of data processing, receipting of donations and to keep you informed of our programs and services.

If you wish to remain anonymous please tick this box ☐

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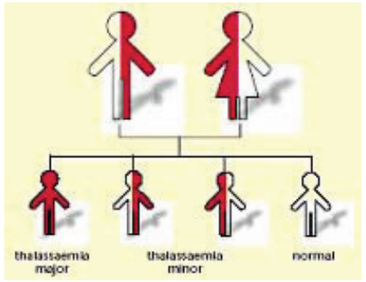
Both parents with β Thalassaemia Minor



If two people with β thalassaemia minor have a child, each pregnancy will have:

- A 1 in 4 chance that the child will inherit 2 copies of the β thalassaemia gene from both parents. This child would have β thalassaemia major
- A 1 in 4 chance that the child will not carry the β thalassaemia gene.
- A 1 in 2 chance that the child will be a carrier of the β thalassaemia gene (thalassaemia minor).

Therefore, in each pregnancy there is a 1 in 4 (25%) chance of having a healthy child, a 1 in 4 (25%) risk of having a child with β thalassaemia major and a 1 in 2 (50%) chance of the child being a carrier of the β thalassaemia gene.

α – thalassaemia

α thalassaemia is genetically more complicated than beta thalassaemia. Individuals normally have four α globin genes. Those with only three genes are normal but may have slightly smaller red cells than usual. Individuals with only two normal genes are affected in a similar way to those with β thalassaemia minor. Individuals with only one normal α gene are variably affected, and may have a severe transfusion-dependent disorder, while infants without any normal α globin genes will die before or shortly after birth.

Combinations of α and β thalassaemia

Because of the ethnic diversity of the Australian population, combinations of alpha and beta thalassaemia are being seen with increasing frequency, and the clinical consequences of these combinations may be difficult to predict. It is important to seek the advice of a specialist experienced in this area when couples are considering a pregnancy. In particular, it is important to seek advice prior to embarking on a pregnancy, as the tests that are required are complex and take a considerable time to perform and interpret.

What of the future?

The Society will continue in its mission to provide support for patients and carers of patients with thalassaemia, sickle cell anaemia, blackfan diamond disorder and other haemoglobinopathies, while providing funding for treatment, research and education.

What can you do to help?

Become a member of the Thalassaemia Society of NSW.

- Attend meetings and support our functions
- Make a donation of your time, money or ideas

As a member you will become part of a large network of individuals and families who have all been touched in some way by thalassaemia. You will receive regular newsletters on our activities and progress, as well as information on upcoming events. You also become eligible to provide input to the way the Society is managed through voting at our Annual General Meeting.

Becoming a financial member is easy and costs just $20 per year, $200 Single Lifetime Membership or $400 Family Lifetime Membership.

Support the Society

If you would like to become a member of our Society or make a donation and help us achieve our goals, please complete and send the application form by Fax: (02) 9519 3517 or mail to the address at the top of the application form.

inform

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research

Thalassaemia Society of NSW

Thalassaemia Society of NSW Inc.
ABN 23 221 006 171 CFN 14635

What is thalassaemia?

Thalassaemia is a blood disorder affecting red blood cells. The primary function of red blood cells is to carry oxygen around the body, binding the oxygen to a compound called haemoglobin, which it then delivers to each cell in the body. Red blood cells make up almost 45% of the body's total blood volume therefore playing an important part in transporting oxygen to main organs around the body.

Thalassaemia is an inherited (genetic) disorder passed from parents to children through the genes. It is found mostly in people whose ancestors come from countries around the Mediterranean Sea, the Middle East and Asia and across a broad region extending through India, Pakistan, Sri Lanka, Bangladesh, and South East Asia.

Thalassaemia is one of the most common genetic disorders worldwide. Anyone now living in Australia, regardless of their racial background can potentially inherit thalassaemia.

β – thalassaemia

This may occur as:

β thalassaemia major - the disorder
β thalassaemia intermedia - the disorder, and
β thalassaemia minor - the carrier state, also called thalassaemia trait.

β – thalassaemia major

In people who have β thalassaemia major, the body destroys red blood cells almost as soon as they are produced and the bone marrow cannot produce a sufficient number to replace them.

This is usually a severe chronic condition resulting in anaemia. The anaemia can cause bone marrow expansion and bone deformities, enlargement of the liver and spleen as these organs try to help bone marrow to make red cells, and poor physical development. Without treatment these patients suffer ill health and may die.

The **signs** of anaemia become apparent in early childhood where affected children become pale, do not sleep well, lose their appetite and fail to grow and thrive properly.

Treatment for β – thalassaemia major

Once the signs of anaemia develop and the child is diagnosed, treatment is commenced immediately. Treatment consists of regular red cell **blood transfusions** approximately every 3-4 weeks that are continued for the life of the patient.

Blood transfusions keeps the patient alive, however cause new problems. Deposits of iron from the blood transfusions build up in the patient's heart, liver, pancreas and other organs. These iron deposits must be removed or they will cause serious damage, which could result in heart failure, diabetes or liver disease.

One way of removing the excess iron is by using a drug called desferrioxamine. This drug is injected under the skin by a slow **infusion pump** over a 10 hour period 5-7 nights per week. Most patients who comply with this treatment will lead a near to normal life.

This treatment is often a great strain for the patients and their families.

Progress in clinical trials have lead to the availability of two oral chelators (taken by mouth) in Australia and are prescribed under strict medical guidelines. Patients must undergo suitable tests to be eligible to commence treatment with the oral chelators.

β – thalassaemia intermedia

Thalassaemia Intermedia describes a spectrum of clinical disease in patients who have a less severe condition than transfusion-dependent β thalassaemia major, but more severe than that of the assymptomatic heterozygous carriers.

There are a variety of genetic defects which lead to thalassaemia intermedia. The sufferers have inherited an affected β gene from both the mother and the father, one of which is a less severe thalassaemia mutation. In some cases they have inherited a genetic tendency to produce foetal haemoglobin which compensates for their lack of adult haemoglobin, and in other cases the inheritance of α thalassaemia together with β thalassaemia can "paradoxically" make the condition less severe. Such individuals often manage to maintain haemoglobin at satisfactory levels, and may not require regular blood transfusions.

Considerable research into the condition has demonstrated that thalassaemia intermedia covers a wide range of clinical symptoms. At the severe end of the spectrum, a patient may become transfusion dependent, similar to patients with thalassaemia major.

In thalassaemia intermedia, the most important question is when to begin blood transfusion therapy. The following medical conditions are indicative for initiating blood transfusion: delayed growth, pathological bone fractures, impact on cardiac function, facial deformities, decreased normal physical activity and hypersplenism.

Iron overload is a potential complication of thalassaemia, even in patients not requiring red blood cell transfusions. It results from an excessive absorption of dietary iron, a consequence of ineffective erythropoiesis and rapid turnover of plasma iron. Recent research has also indicated a role of newer mediators of iron turnover, such as a molecular called hepcidin.

β – thalassaemia minor (trait)

People with β thalassaemia minor may have a very mild anaemia, which does not stop them from enjoying good health. Most people with thalassaemia minor are unaware that they are affected unless specific testing has been performed.

This minor form of β thalassaemia, in which only one copy of β-globin allele bears a mutation, can never change into thalassaemia major or another blood disorder.

The only means of detection is by a specific blood test for thalassaemia.

People with thalassaemia minor should inform their doctor in order to avoid confusion with iron deficiency anaemia and to prevent any unnecessary treatment with iron tablets.

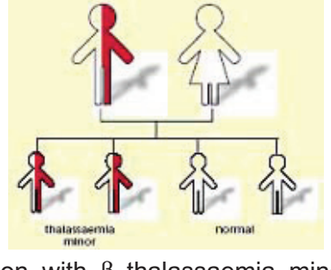
Testing for β – thalassaemia

All individuals are advised to be tested, in particular those from high risk backgrounds, and especially before starting their families. If a couple are found to both be carriers it is essential that they seek genetic counselling for information about their available options. The partner of an individual with thalassaemia major or minor should have a blood count, iron studies and a haemoglobin EPG — the latter is required to exclude the possibility of coexisting sickle cell disease, as a child with thalassaemia minor and a sickle gene may have a serious disorder.

β – thalassaemia minor and pregnancy

Thalassaemia minor does not affect the health of a woman during pregnancy although she should be carefully monitored to ensure that anaemia does not result. If anaemia does occur, it can be treated. If a person with β thalassaemia minor is considering starting a family, their spouse or partner must be tested for thalassaemia and other related blood conditions such as sickle cell trait. While β thalassaemia minor in itself does not affect the health of an individual, if both parents have β thalassaemia minor, there is a risk to their children as described in the next section.

One parent with β thalassaemia minor



If one person with β thalassaemia minor and one person without β thalassaemia minor have a child, each pregnancy will have:

- A 2 in 4 chance that the child will be a carrier of the β thalassaemia gene.
- A 2 in 4 chance that the child will not carry the β thalassaemia gene.