

# About Thalassemia

## What is Thalassemia?

Thalassemia is a genetic condition affecting the red blood cells. Haemoglobin is a chemical found in the red blood red cells which carries oxygen from the lungs to all parts of the body. Thalassemia occurs when a person is unable to produce the required amount of haemoglobin.

## How does one get Thalassemia?

We all inherit many of our characteristics through the genes we take from our parents. For example, the shape of our nose, colour of our eyes and whether we are tall or short.

Similarly we inherit our haemoglobin type through the genes we get from our parents. We get one haemoglobin gene from our mother and the other from our father. The usual, most common haemoglobin is normal haemoglobin A. If a person inherits two haemoglobin A genes he/she will have haemoglobin AA. This is normal. If we inherit two Thalassemia genes we get Thalassemia major.

## What is Beta Thalassemia trait (minor)?

Some people inherit one haemoglobin A gene and one beta Thalassemia gene and have beta Thalassemia trait, also called beta Thalassemia minor. The red blood cells of these individuals are a bit paler and smaller than normal red blood cells. They are also a little anaemic, but this anaemia does not usually need treatment. However, they can pass on the beta Thalassemia gene to their children.

## What happens if a person has Thalassemia major?

People with beta Thalassemia major are unable to make the chemical needed for producing enough mature haemoglobin. Haemoglobin is essential for carrying oxygen around the body. Without sufficient haemoglobin the body cannot carry oxygen properly and after a while the body will not survive.

## What are the symptoms of Thalassemia?

Without regular blood transfusions, patients will be pale looking and fatigue easily. They have poor appetite and frequent infections. Their liver and spleen are often enlarged. Signs and symptoms of Thalassemia also include shortness of breath, slow growth, dark urine Yellow discoloration of skin (jaundice) and irritability. The signs and symptoms you experience depend on your type and severity of Thalassemia. Some babies show signs and symptoms of Thalassemia at birth, while others may develop signs or symptoms later, during the first two years of life.

## What are the essential treatments for beta Thalassemia major?

### Regular Blood Transfusion

- Usually once every 3 - 4 weeks to sustain life and to keep Hb > 10 gm
- Iron overload from regular blood transfusions may damage the heart and liver and result in stunted growth, delayed puberty, diabetes and other endocrine complications. Hence, drugs to reduce iron (iron chelation) are the most important aspect of Thalassemia care.

### Iron Chelation

- Desferal (iron removing drug) is administered over 10 hours each night, 5 to 6 nights per week injected slowly via an electrical pump
- Oral Chelator: Kelfer is an iron chelating agent that can be taken by mouth
- Exjade is another orally active iron chelator that will be available soon.

## What can you do to prevent Thalassemia major?

### Family Planning:

- Special blood screening test or premarital test is available to find out whether you and your partner are Thalassemia carriers
- Consult your doctor for more information about Thalassemia

### Prenatal Check up:

- If you and your partner are Thalassemia carriers, consult your obstetrician regarding prenatal diagnosis.

### Prenatal tests available are:

- **Chronic Villus Sampling:** This test is performed from 10th to 12th week of pregnancy. It looks at cells taken from the placenta.
- **Amniocentesis:** This is usually done between 16 th to 18 th week of pregnancy. Under ultrasound guidance, the doctor removes a small amount of the amniotic fluid for DNA testing.
- **Foetal Blood Sampling:** This can be carried out at 18 th to 20 th week of pregnancy. A sample of the baby's blood is obtained and tested to determine whether or not the baby has Thalassemia Major.

## Is there a cure for beta Thalassaemia?

Yes. **Bone Marrow Transplantation (BMT)** is the only definitive cure for Thalassaemia but it has its risks. These risks depend mostly on availability of a compatible family donor, generally a sibling and the age and health of your child at the time of transplant. It consists of replacing your child's faulty bone marrow stem cells, from which red cells originate, with those obtained from a healthy compatible donor. Umbilical cord blood can also be used as a source of stem cells for transplantation.

In low-risk cases (less than 10 years of age, having regular chelation therapy, non liver enlargement and no transfusion-associated diseases like hepatitis or HIV), BMT provides a 80-90% cure probability, with 5% mortality rate and a 10% chance of rejection (thus leaving the child thalassaemic). The cost of transplant varies and may range from 8 lakhs 15 lakhs.

## Support us

There are many different ways you can help in our ongoing fight against Thalassaemia.

- By creating awareness & forwarding this information to others
- Get your blood tested for Thalassaemia traits
- Donate your blood for Thalassaemia patients-
- Contact us at +91-452-2528520
- Donate money to NIVETHAN TRUST for treatment of poor Thalassaemia patients

**Donate generously and help us in our mission.  
Send your "Donations" to:**

### NIVETHAN

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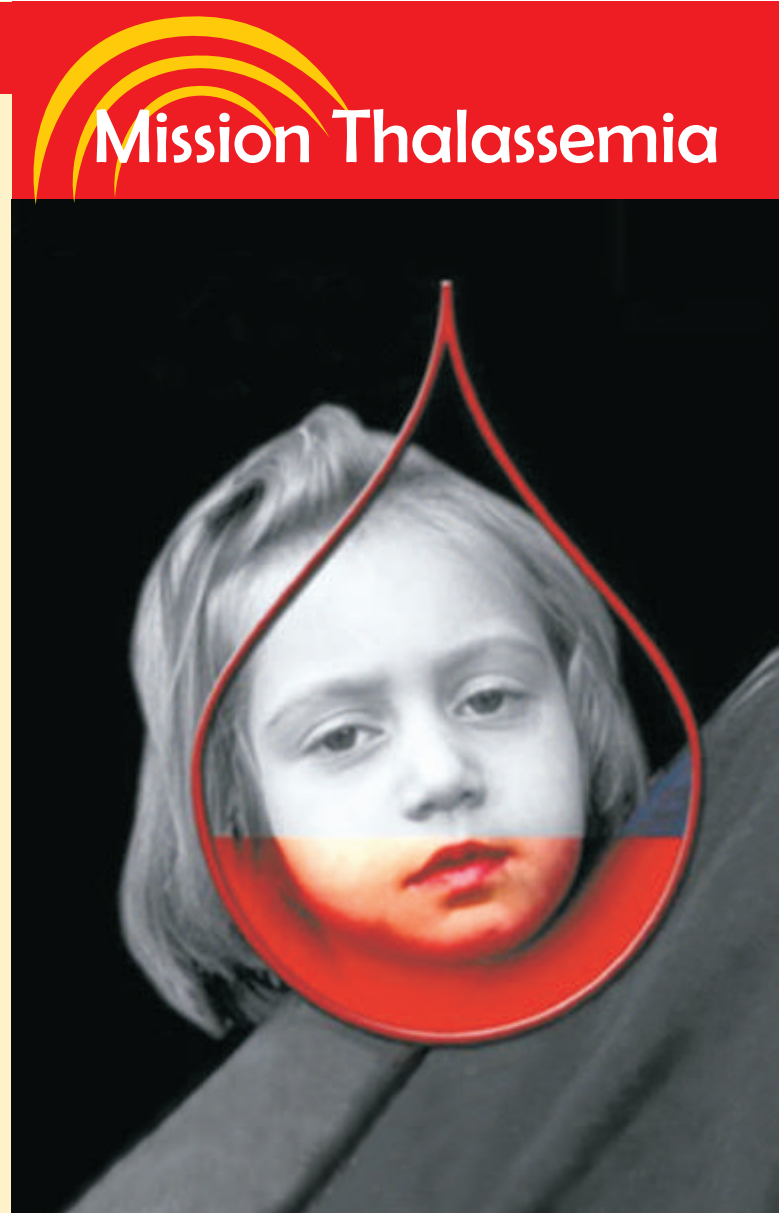
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Account name : Nivethan Trust  
Account number : 30437489626  
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All your donations to Nivethan Trust are exempted under 80 (G) of Income Tax Act. Vide No: 464/154/2008-2009/CIT-I

*To know more about Nivethan and its Mission Thalassaemia Initiatives, please visit [www.nivethan.org](http://www.nivethan.org) or mail us at [nivethan.mdu@gmail.com](mailto:nivethan.mdu@gmail.com)*



**Supporting children with Thalassaemia & Sharing knowledge and creating awareness**