

SYMPTOMS OF β THALASSAEMIA MAJOR

- Without regular blood transfusions, patients will be pale-looking and easily fatigued, they have poor appetite and frequent infections. Their liver and spleen are often enlarged.



ESSENTIAL TREATMENT FOR β THALASSAEMIA MAJOR

Regular Blood Transfusions

- Usually once every 4 weeks to sustain life resulting in iron overload which may:
 - a) damage the heart and liver.
 - b) cause stunted growth, delayed puberty, diabetes and other endocrine complications.

Iron Chelation

- Desferal (iron removing drug) is administered over 10 hours each night, 5 to 6 nights per week.
- Desferal is injected slowly via an electrical pump.

Oral Chelator – L1

- A new iron chelating agent that can be taken by mouth is now available. Due to consideration for safety and efficacy, it can only be used as a second-line treatment for older patients.

WHAT CAN YOU DO TO PREVENT THALASSAEMIA MAJOR?

Family Planning

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

Prenatal Checkup

- If you and your partner are Thalassaemia carriers, consult your obstetrician regarding prenatal diagnosis.
- Available prenatal tests are done at optimum time. The following will identify the fetus with Thalassaemia Major:

1. Chorionic Villi Sampling

This test is performed from 10th to 12th week of pregnancy. It looks at cells taken from the placenta.

2. Amniocentesis

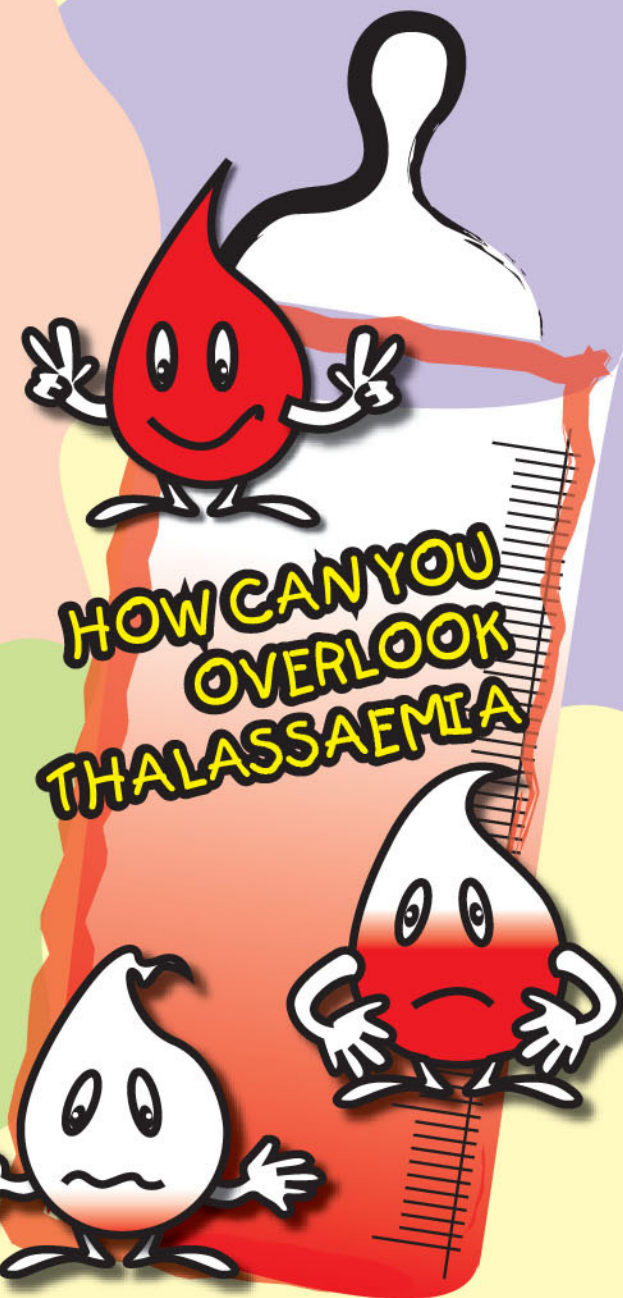
This is usually done between 16th-18th weeks of pregnancy. Under ultrasound guidance, the doctor removes a small sample of the amniotic fluid for DNA testing.

3. Foetal Blood Sampling

This can be carried out at 18th-20th weeks of pregnancy. A sample of the baby's blood is obtained and tested to determine whether or not the baby has Thalassaemia Major.

OTHER WAYS TO HELP TO PREVENT THALASSAEMIA

- Donate generously to support the public educational work provided by the Children's Thalassaemia Foundation.
- If you are a carrier, ask your family members and relatives to go for a blood test.
- Spread this message to your friends and relatives.



If you would like to know more about Thalassaemia, please contact:

Thalassaemia Education & Counseling Unit :
2986-3311

Children's Thalassaemia Foundation : 2523-5400

URL : www.thalassaemia.org.hk

The Thalassaemia Association of Hong Kong
2889-8399



WHAT IS THALASSAEMIA?

- It is one of the commonest inherited genetic blood disorders in the world. It is not acquired by social contact.
- “Thalassaemia” in Greek means anaemia around ‘the sea’ (‘thaloss’, Mediterranean)
- Prevalent in Mediterranean countries, the Middle East and Asia (including Hong Kong).

IRON DEFICIENCY ANAEMIA vs THALASSAEMIA

- ANAEMIA= Reduced number of red blood cells or low level of haemoglobin in the body.
- “Iron deficiency anaemia” is the commonest: which is due to insufficient iron in the diet. It can be cured by taking iron supplement.
- Thalassaemia is different, it cannot be treated by iron supplements.

TWO MAIN CATEGORIES OF THALASSAEMIA

Thalassaemia is a result of a reduced rate of synthesis of either the alpha (α) or beta (β) globin chains of haemoglobin which is the oxygen-carrying pigment in red blood cells.

Each individual has 2 sets of globin genes, one from father and the other one from mother. If one or more defective gene(s) is inherited from a parent, then there would be a reduction of either the α or the β globin chains, resulting in lowering of the haemoglobin level and Thalassaemia.

Clinically, there are 3 different forms of Thalassaemia which are classified according to the degree of anaemic. They are the Mild Form (Thal-minor), Moderate Form and Severe Form (Thal-major).

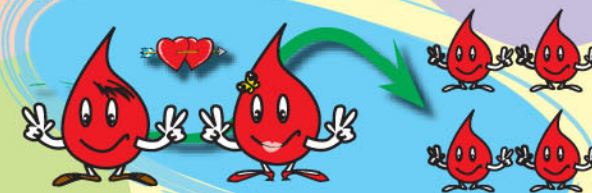
IT IS CLASSIFIED INTO TWO TYPES OF THALASSAEMIA TRAIT:



HOW THALASSAEMIA IS INHERITED?

The following are three examples:

If both parents are NOT carriers, all of their children will be 'normal'.



If one parent is a carrier and the other is 'normal', each child will have 50% chance of becoming a Thalassaemia carrier (Thal-minor).



If both parents are carriers of the same type of Thalassaemia, each of their children will have 25% of chance to be 'normal', 50% of chance of becoming a Thalassaemia carrier, and 25% of chance of becoming a patient with Thalassaemia Major.



Severity Categories	Mild Form	Moderate Form	Severe Form
α -Thalassaemia	<p>α or β Thalassaemia Minor (carrier)</p> <ul style="list-style-type: none"> • Most of the carriers has inherited one defective gene • Patient has no symptoms • May be mildly anaemic • No treatment is required • The condition is only detected by blood tests • The Red blood cells are much more smaller than usual • One out of eleven people in Hong Kong is a Thal-carrier 	<p>Haemoglobin H disease</p> <ul style="list-style-type: none"> • Small amount of an unusual haemoglobin, Haemoglobin H can be found in the blood • Usually moderate anaemic will be found in both children and adult 	<p>Haemoglobin Bart's Hydrop Fetalis</p> <ul style="list-style-type: none"> • Severe anaemia occurs early in the fetal life resulting in poor growth and hydrops (fetus puffed up with water) • Usually results in fetal or newborn death
β -Thalassaemia		<p>Thalassaemia Intermeida</p> <ul style="list-style-type: none"> • Degree of anaemic is between those thal-minor and thal-major • Most patients have moderate anaemia, jaundice and enlarged liver and spleen • Most do not need regular transfusion 	<p>Thalassaemia Major</p> <ul style="list-style-type: none"> • Serious form of disease, symptoms of anaemia start in early childhood (6 month to 1 year old) • Inherited a pair of defective genes, one from each parent • Symptoms of pallor, poor appetite, enlarged liver and spleen, bone deformities and retarded growth • Need regular blood transfusion every 4 weeks to sustain life.