SYMPTOMS OF β THALASSAEMIA MAJOR

- Without regular blood transfusions, patients will be pale-looking and easily fatigued, 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.
**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.

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**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).

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**IT IS CLASSIFIED INTO TWO TYPES OF THALASSAEMIA TRAIT:**

<table>
<thead>
<tr>
<th>Alpha (α)</th>
<th>Beta (β)</th>
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<tbody>
<tr>
<td>5%</td>
<td>3.5%</td>
</tr>
</tbody>
</table>

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**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.

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**Severity Categories**

<table>
<thead>
<tr>
<th>Categories</th>
<th>Mild Form</th>
<th>Moderate Form</th>
<th>Severe Form</th>
</tr>
</thead>
<tbody>
<tr>
<td>α-Thalassaemia</td>
<td>α or β Thalassaemia Minor (carrier)</td>
<td>Haemoglobin H disease</td>
<td>Haemoglobin Bart’s Hydrops Fetalis</td>
</tr>
<tr>
<td>β-Thalassaemia</td>
<td></td>
<td>Haemoglobin Bart’s Hydrops Fetalis</td>
<td>Thalassaemia Intermedia</td>
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<tr>
<td></td>
<td>Minor (carrier)</td>
<td>Small amount of an unusual haemoglobin, Haemoglobin H can be found in the blood</td>
<td>Severe anaemia occurs early in the fetal life resulting in poor growth and hydrops (fetus puffed up with water)</td>
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<td>Usually moderate anaemic will be found in both children and adult</td>
<td>Usually results in fetal or newborn death</td>
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<td>The condition is only detected by blood tests</td>
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<td></td>
<td></td>
<td>The Red blood cells are much more smaller than usual</td>
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<td>One out of eleven people in Hong Kong is a Thal-carrier</td>
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<td></td>
<td></td>
<td>Degree of anaemic is between those thal-minor and thal-major</td>
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<tr>
<td></td>
<td></td>
<td>Most patients have moderate anaemia, jaundice and enlarged liver and spleen</td>
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<td></td>
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<td>Most do not need regular transfusion</td>
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<td>Serious form of disease, symptoms of anaemia start in early childhood (6 month to 1 year old)</td>
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<tr>
<td></td>
<td></td>
<td>Inherited a pair of defective genes, one from each parent</td>
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<tr>
<td></td>
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<td>Symptoms of pallor, poor appetite, enlarged liver and spleen, bone deformities and retarded growth</td>
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<tr>
<td></td>
<td></td>
<td>Need regular blood transfusion every 4 weeks to sustain life.</td>
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