Thalassaemia
Thalassaemia major is a genetic disorder that affects the production of haemoglobin, resulting in severe anaemia.

This disorder is usually diagnosed within the first six months of life.

Treatment options include regular blood transfusions and bone marrow transplants.

Couples should be tested for their thalassaemia carrier status.

Thalassaemia is the most common inherited blood disorder in the world. This condition is caused by changes to the genes for haemoglobin. Haemoglobin is a protein in red blood cells that carries oxygen around the body. Changes affecting haemoglobin result in severe anaemia.

Thalassaemia can affect people of any nationality and ethnicity. It is particularly common in people of Mediterranean ancestry and across a broad region extending through India, the Middle East, Asia and Latin America.

Thalassaemia is usually diagnosed within the first six months of life and can be fatal in early childhood without ongoing treatment.

There are two different types of thalassaemia: alpha (α) and beta (β). Alpha-thalassaemia involves genetic changes in copies of two genes, whereas beta-thalassaemia involves changes in copies of one gene. Alpha-thalassaemia is more common in countries in Africa, Asia and the Middle East. Beta-thalassaemia is more common in Mediterranean countries.

In Australia, beta-thalassaemia is the most common form because of the high number of people who have migrated from Mediterranean countries. However, more migration of people from Asia means that the number of people with alpha-thalassaemia has increased in Australia.

Thalassaemia minor

Thalassaemia minor refers to people who have genetic changes in one copy (for beta-thalassaemia) or in one or two copies (for alpha-thalassaemia) of the genes, but still have other copies of thalassaemia genes that don’t have genetic changes. This means that people with thalassaemia minor have mild or no symptoms, but they carry the genetic changes and can pass them on to their children. Thalassaemia minor can refer to people with either alpha- or beta-thalassaemia.

It is conservatively estimated by the World Health Organisation that five to seven per cent of the world’s population has thalassaemia minor. People who have beta-thalassaemia minor, for example, have one copy of the changed beta-thalassaemia gene and are healthy carriers. The only way to detect if someone is a carrier is by a specific blood test for thalassaemia.

Thalassaemia major

Thalassaemia major refers to the condition where people have inherited more than one copy of the genes with genetic changes. Thalassaemia major can refer to people with either alpha- or beta-thalassaemia. People with either alpha- or beta-thalassaemia major will have symptoms associated with the condition. Beta-thalassaemia major is more common than alpha-thalassaemia major.

Carrying the alpha or beta thalassaemia gene

There is a concern when two carriers of alpha or beta thalassaemia wish to start a family. If two carriers conceive a child, the child has a:

- 25 per cent risk of developing thalassaemia major because they inherited the thalassaemia gene from both parents
- 25 per cent chance of not inheriting the thalassaemia gene at all
- 50 per cent chance of inheriting the gene from one parent and becoming a carrier.

Symptoms of thalassaemia major

The red blood cells of a person with thalassaemia major can only survive for a few weeks, compared to normal red blood cells that survive for around four months. Lack of haemoglobin results in reduced oxygen to every cell in the body.

Symptoms appearing in early childhood include:
severe anaemia – red blood cells are produced without sufficient haemoglobin to carry oxygen
paleness
sleep difficulties
poor appetite
failure to grow and thrive
enlargement of organs – such as the spleen and liver.

Treatment for thalassaemia major

There is no cure for thalassaemia major and treatment must continue for life. Ongoing treatment includes regular transfusions to boost haemoglobin levels in the blood. However, these transfusions can lead to a build-up of iron and this can cause serious side effects, including diabetes, heart failure and liver disease.

Medications can be used to remove excess iron from the blood. These medications are called iron chelators. They work by binding with the iron and allowing it to be removed by the kidneys. Chelators can be oral or injectable, for example, Exjade is an oral chelator and Desferal is an injectable chelator.

Diagnosis of thalassaemia

It is suggested that all couples be tested for thalassaemia carrier status, especially before starting a family. In Victoria, testing can be arranged by your family doctor or is available free of charge at:

- Royal Women’s Hospital
- Monash Medical Centre
- Mercy Hospital for Women.

Options for a couple who are both carriers of thalassaemia

Carrier couples may elect not to have children, but choose to adopt, foster or consider using donor sperm or eggs. Others may elect to take the risk of having children with thalassaemia major. Couples may also choose prenatal diagnosis with the option of terminating the pregnancy if the foetus is diagnosed with thalassaemia major.

Pre-implantation genetic diagnosis (PGD) may also be used. This is where artificial reproductive technology is used to create an embryo that can be tested for thalassaemia before being transferred to the uterus of the mother.

Bone marrow transplants for thalassaemia major

A bone marrow transplant is the only chance of a cure, but the risks are considerable. The odds of finding a compatible donor are around 30 per cent and most donors are siblings. This operation is more successful in young children who don’t suffer from iron overload, but graft rejection or even death can still result.

The risks involved mean that bone marrow transplantation is not a viable option for some families. It should be remembered that a person with thalassaemia major can live a normal life with regular transfusions and medication. Genetic research is continuing in the hope of finding a cure.

Where to get help

- Your doctor
- Thalassaemia & Sickle Cell Society of Australia, Tel. (03) 9888 2211
- Thalassaemia Services Victoria, Monash Medical Centre, Medical Therapy Unit Tel. (03) 9594 2756
- Royal Women’s Hospital – Genetic Counselling Service Tel. (03) 8345 2180
- Mercy Hospital for Women – Clinical Genetics Service: Tel. (03) 8458 4250

References

- Alpha (a) Thalassaemia, 2012, Thalassaemia Australia.
- Beta (b) Thalassaemia, 2012, Thalassaemia Australia.
- Haemoglobin E (HBE), 2012, Thalassaemia Australia.
- Sickle cell disease, 2012, Thalassaemia Australia.
- Family planning, 2012, Thalassaemia Australia.
- Preimplantation genetic diagnosis (PGD) for single gene disorders, 2011, Monash IVF.
- Desferal (desferrioxamine mesylate), Consumer medicine information, 2011, TGA eBusiness Services.
- Exjade® (deferasirox), Consumer medicine information, 2016, TGA eBusiness Services.

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Blood and blood vessels explained

- Blood count
  The full blood count (FBC) test looks for abnormalities in the blood, such as unusually high or low numbers of blood cells.

- Blood groups
  A person's blood group is determined by a pair of genes, one each inherited from their mother and father.

- Bone marrow
  Bone marrow is the spongy tissue in the hollow centres of a person's long bones and is the blood cell 'factory'.

- Circulatory system
  The heart, blood and blood vessels work together to service the cells of the body.

- Heart explained
  The heart is about the size of a clenched fist and lies in the middle of your chest, behind and slightly to the left of your breastbone.

- Lipoedema
  Lipoedema is a painful, chronic, symmetrical swelling in the legs, thighs, buttocks and sometimes arms due to the accumulation of fat in the subcutaneous tissues. The onset often occurs during puberty.

- Lymphatic system
  The lymphatic system manages fluid levels in the body, filters out bacteria and houses types of white blood cells.

Cholesterol

- Cholesterol
  Your body needs cholesterol, but it can make its own. You don't need cholesterol in your diet.

- Cholesterol - healthy eating tips
  Replacing foods that contain saturated fats with foods that contain polyunsaturated and monounsaturated fats will help to lower your cholesterol.

- Genetic factors and cholesterol
  Familial hypercholesterolaemia is an inherited condition characterised by higher than normal levels of blood cholesterol.

- Triglycerides
  If a person habitually eats more kilojoules than they burn, they will have raised triglyceride levels in the blood.

Iron anaemia and blood disorders

- Haemochromatosis
  Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.

- Hughes syndrome
  Hughes syndrome is thickening of the blood caused by abnormal immune system cells.

- Iron
  Iron is important for transporting oxygen in the blood.

- Iron deficiency - adults
  Don't take iron supplements unless advised by your doctor.

- Iron deficiency - children
  Keep iron supplements away from children - as little as one to three grams can kill a child under six years.

- Porphyria

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Porphyria can affect the skin, nervous system, gastrointestinal system or all of these, depending on the specific type.

- **Thalassaemia**
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated.

**Bleeding clotting and infections**

- **Bleeding**
  Bleeding may be minor or it may be a life-threatening medical emergency.

- **Deep vein thrombosis**
  Long international flights are suspected of contributing to deep vein thrombosis in susceptible people.

- **Haemophilia**
  All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed.

- **Needlestick injury**
  A needlestick injury means the skin is accidentally punctured by a used needle. Diseases that could be transmitted by a needle or needlestick injury include human immunodeficiency virus (HIV).

- **Nosebleeds**
  Bleeding from the nose is common in children and is usually not severe or serious.

- **Septicaemia**
  Bacteria in the bowels, urinary tract, mouth and skin can cause disease if they get into the bloodstream.

- **Subarachnoid haemorrhage**
  A subarachnoid haemorrhage is any bleed located underneath one of the protective layers of the brain known as the arachnoid layer.

- **Subdural haematoma**
  Subdural haematomas are blood clots formed underneath one of the protective layers of the brain.

- **Travel tips for seniors**
  All travellers should plan carefully, but older people have a few extra concerns when travelling.

- **Von Willebrand disease**
  A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth.

**Blood pressure**

- **Blood pressure**
  Healthy eating and lifestyle changes can help to manage high blood pressure.

- **Blood pressure (high) - hypertension**
  Hypertension, or high blood pressure, can increase your risk of heart attack, kidney failure and stroke.

- **Blood pressure - keep your blood pressure down (video)**
  Heart Foundation of Australia warns of the risk of high blood pressure and tells you what you can do to keep your blood pressure down.

- **Blood pressure (low) - hypotension**
  Low blood pressure is only a problem if it has a negative impact on the body.

- **Dizziness - orthostatic hypotension**
  Postural hypotension is the lightheaded feeling you may get if you leap out of bed very quickly.

- **Fainting**
  Common causes of fainting include heat, pain, distress, the sight of blood, anxiety and hyperventilating.

- **Pulmonary hypertension**
  Pulmonary hypertension is high blood pressure on the lungs.

- **Shock**
  Shock is when there is not enough blood circulating in the body. It is a life-threatening medical emergency.

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Stroke explained
A stroke interrupts blood flow to an area of the brain and is a medical emergency...

Blood vessel and bone marrow conditions

- **Amyloidosis**
  A person with amyloidosis produces aggregates of insoluble protein that cannot be eliminated from the body...

- **Aneurysm**
  An aneurysm may have no symptoms until it is either very large or it ruptures...

- **Granulomatosis with polyangiitis**
  Granulomatosis with polyangiitis (GPA), formerly known as Wegener granulomatosis is a rare condition that targets the arteries, veins and capillaries of the kidneys and the respiratory system...

- **Henoch-Schonlein purpura**
  Henoch-Schonlein purpura causes a purple spotted skin rash which lasts around one to four weeks, and is often marked by relapses...

- **Leukaemia**
  Most children and many adults with acute leukaemia can expect to be cured, while chronic leukaemia can be successfully managed...

- **Peripheral vascular disease**
  Peripheral vascular disease is the reduced circulation of blood to a body part (other than the brain or heart)...

- **Polycythæmia vera**
  Polycythæmia vera is characterised by the production of too many red blood cells, caused by abnormal function of the bone marrow...

- **Raynaud's phenomenon**
  Raynaud's phenomenon can be a sign of a more serious underlying condition, so see your doctor if you experience it...

- **Thalassaemia**
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated...

- **Varicose veins and spider veins**
  Smaller varicose veins are usually treated by sclerotherapy - the injection of irritant chemicals into the affected vein...

Blood-donation-and-transfusion

- **Blood donation**
  Donated blood is used to help people who are sick or injured, or for medical research...

- **Blood transfusion**
  Donated blood is screened for blood-borne diseases such as hepatitis, syphilis and HIV...

- **Organ and tissue donation**
  Discover the facts about organ and tissue donation, decide about becoming a donor and discuss your decision with the people close to you...

Related Information

- **Blood and blood vessels**
  Bleeding, blood pressure, conditions, risks and blood products...

- **Haemochromatosis**
  Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses...

- **Haemophilia**
  All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed...

- **Down syndrome**
  With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community...
• **Tay-Sachs disease**

  Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians...

**Related information on other websites**

- Thalassaemia Australia - translated fact sheets.
- Victorian Clinical Genetics Services.

**Content Partner**

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