CONDITION

Thalassemia

Thalassemia is an inherited condition that reduces the body's production of healthy hemoglobin.

WHAT IT IS
Inherited condition reducing healthy hemoglobin production

BODY SYSTEM
Blood and oxygen transport

MAIN SYMPTOMS
Fatigue, pale skin, breathlessness, slow growth

WHAT CAUSES IT
Inherited changes in alpha or beta globin genes

HOW IT IS DIAGNOSED
Blood count, hemoglobin analysis, and genetic testing

MAIN TREATMENT
Monitoring, transfusions, iron chelation, and selected curative therapies

CAN IT BE CURED?
Sometimes - transplant or gene therapy for eligible people

IS IT AN EMERGENCY?
Sometimes - severe breathlessness, fainting, or transfusion reaction

LONG-TERM OUTLOOK
Normal lifespan for carriers; severe forms need lifelong care

Overview

Thalassemia is an inherited blood condition that affects hemoglobin. Hemoglobin is the protein inside red blood cells that carries oxygen around the body.

Hemoglobin contains protein chains called alpha globin and beta globin. Genetic changes that reduce production of these chains cause two main types:

Each type ranges from a carrier state, often called thalassemia trait, to a severe condition requiring regular treatment. Terms such as thalassemia major, intermedia, transfusion-dependent thalassemia, and non-transfusion-dependent thalassemia describe different levels of severity.

People with thalassemia trait usually have small red blood cells and mild anemia, meaning a

lower-than-usual level of healthy red blood cells. They often have few or no symptoms. More severe forms can cause significant anemia, slow growth, bone changes, and iron overload.

Thalassemia is not contagious. It is not caused by diet or something a person did during pregnancy.

- Alpha thalassemia , which affects alpha globin production
- Beta thalassemia , which affects beta globin production

Symptoms

Symptoms depend on the type of thalassemia and how much healthy hemoglobin the body can make.

Thalassemia trait often causes no noticeable symptoms. A routine blood count may show mild anemia and unusually small red blood cells. Some people have mild tiredness, but other causes of fatigue should also be considered.

Moderate or severe thalassemia may cause:

Severe beta thalassemia usually becomes noticeable during infancy or early childhood as the amount of fetal hemoglobin naturally falls.

Alpha thalassemia can range from a silent carrier state to hemoglobin H disease, which may cause ongoing anemia. The most severe form can cause profound anemia before birth and serious pregnancy complications.

Symptoms alone cannot confirm thalassemia. Iron deficiency and several other conditions can produce similar symptoms and blood-count results.

- Tiredness, weakness, or reduced exercise tolerance
- Pale skin or pale tissue inside the eyelids
- Shortness of breath
- Yellowing of the skin or eyes, called jaundice
- Slow growth or delayed puberty in children
- An enlarged spleen, which may cause fullness under the left ribs
- Changes in facial or other bones when severe anemia is not adequately controlled

Causes and risk factors

Thalassemia is caused by inherited changes in genes involved in hemoglobin production. Alpha thalassemia usually involves missing or altered HBA1 or HBA2 genes. Beta thalassemia results from changes in the HBB gene.

A person may inherit a changed gene from one or both biological parents. The combination of changes determines whether they are a carrier or have a more significant form.

Thalassemia is more common among people whose families come from the Mediterranean region, Middle East, South Asia, Southeast Asia, or Africa. However, it can occur in people of any ancestry.

The chance of having a child with significant thalassemia is higher when both biological parents carry related gene changes. The exact chance depends on whether alpha or beta globin genes are involved and which changes each parent carries.

A carrier can be healthy and not know they have thalassemia trait. Carrier testing and genetic counseling can explain reproductive chances when thalassemia is present in either family.

Diagnosis

Thalassemia is usually investigated with blood tests. A complete blood count measures hemoglobin and the size and number of red blood cells. Thalassemia often causes smaller-than-usual red blood cells.

Iron studies, including a ferritin test, help distinguish thalassemia from iron deficiency. The two conditions can also occur together, so red blood cell size alone is not enough to tell them apart.

Hemoglobin analysis measures different forms of hemoglobin. Methods include hemoglobin electrophoresis and high-performance liquid chromatography. These tests are especially useful for diagnosing beta thalassemia and identifying other hemoglobin conditions.

Alpha thalassemia trait may not produce a clear abnormality on routine hemoglobin analysis. Genetic testing can confirm alpha or beta thalassemia and identify the specific gene changes. It may be particularly helpful for pregnancy planning or when blood tests do not give a clear answer.

Additional tests may assess:

In some places, newborn screening can detect certain severe hemoglobin conditions. Screening programs do not identify every form of thalassemia.

- Red blood cell breakdown and jaundice
- Spleen and liver size

- Iron levels in the liver or heart
- Heart, liver, bone, and hormone health in severe thalassemia

Treatment and management

Treatment is based on the type, symptoms, hemoglobin level, growth, complications, and need for transfusions.

People with thalassemia trait usually do not need treatment. Iron supplements do not treat thalassemia itself and should not be taken for presumed iron deficiency unless testing shows that iron is low.

People with more significant anemia may need blood transfusions. Some need transfusions only during periods of increased stress, such as a serious infection, surgery, or pregnancy. Others need them regularly to maintain healthy red blood cell levels, support growth, and reduce complications.

Repeated transfusions gradually add iron to the body. Thalassemia can also increase iron absorption from food, even in people who do not receive regular transfusions. Iron chelation medicines bind excess iron so it can leave the body. Monitoring may include blood tests and MRI scans that estimate iron in the liver and heart.

Ongoing care may also include monitoring of growth, puberty, bones, the heart, liver, thyroid, and other hormone-producing organs. Vaccination and infection planning are particularly important for people whose spleen has been removed.

A stem cell transplant using blood-forming cells from a suitable donor can cure some people. It also carries significant risks, so eligibility requires specialist assessment.

Gene therapies are available for some people with transfusion-dependent beta thalassemia in certain countries. They use a person's own stem cells after laboratory treatment or gene editing. These treatments involve intensive preparation and are not suitable or accessible for everyone.

Complications

Complications are mainly related to chronic anemia, excess iron, increased red blood cell breakdown, or treatment.

Iron overload can damage the liver, heart, and hormone-producing organs. It may contribute to liver scarring, abnormal heart rhythms, heart muscle weakness, diabetes, thyroid problems,

delayed puberty, or fertility difficulties. Regular monitoring and iron chelation can reduce these risks.

Other possible complications include:

Not everyone develops these problems. Risk is influenced by the thalassemia type, anemia severity, transfusion history, iron levels, and access to consistent specialist care.

- Enlarged spleen or liver
- Gallstones caused by red blood cell breakdown
- Thin bones and fractures
- Abnormal expansion of bone marrow and changes in bone shape
- Blood clots or high pressure in lung blood vessels
- Reactions to transfused blood
- Development of antibodies that make blood matching harder
- Infection risks related to spleen removal or long-term intravenous access

Prevention

Inherited thalassemia cannot be prevented through diet, exercise, or lifestyle changes.

Carrier screening can identify people with thalassemia gene changes before or during pregnancy. If both biological parents are carriers, genetic counseling can explain the chance of different outcomes and discuss available reproductive testing options.

Early diagnosis and regular monitoring do not prevent the inherited condition, but they can help prevent or limit complications such as iron-related organ damage, growth problems, and severe untreated anemia.

Living with it and what to expect

Thalassemia trait generally remains stable throughout life. It does not usually develop into severe thalassemia. Mild anemia and small red blood cells may continue, but most carriers live normally without thalassemia-specific treatment.

Moderate forms can change with age or during physical stress. Infections, pregnancy, surgery, or other health problems may temporarily worsen anemia. Some people who did not need transfusions when young may need occasional or regular transfusions later.

Severe thalassemia is a lifelong condition unless successfully treated with a curative therapy. Care often includes scheduled transfusions, iron chelation, tests for iron overload, and monitoring of organ health. Children may need additional attention to growth, school attendance, puberty, and emotional well-being.

Treatment has greatly improved long-term health for many people with severe thalassemia. Outcomes are best when transfusions, iron control, and monitoring remain consistent. The long-term course is less favorable when severe anemia or iron overload cannot be adequately managed.

Pregnancy may be possible, including for some people with severe thalassemia, but it can place extra strain on the heart and increase transfusion or clotting concerns. Planning with a thalassemia and pregnancy team allows health risks, medicines, and iron levels to be reviewed in advance.

When to get medical help

Arrange routine medical follow-up if you have persistent tiredness, pale skin, unexplained anemia, or a blood count showing small red blood cells. Testing is also reasonable if thalassemia runs in your family or if a biological partner is a carrier.

If you already have thalassemia, contact your care team promptly about:

Seek emergency help for severe trouble breathing, chest pain, fainting, confusion, extreme weakness, or a rapid decline in alertness.

During or after a blood transfusion, immediately report fever, chills, breathing difficulty, facial swelling, chest or back pain, or dark urine. These can be signs of a transfusion reaction and require urgent assessment.

- Increasing fatigue, breathlessness, or reduced ability to exercise
- New jaundice or dark urine
- Increasing abdominal swelling or pain under the left ribs
- Fever, especially after spleen removal or with a central intravenous line
- Missed transfusions or difficulty taking iron chelation treatment
- Pregnancy or plans to become pregnant

Common questions

Is thalassemia the same as iron-deficiency anemia?

No. Both can cause anemia and small red blood cells, but their causes are different. Thalassemia is inherited, while iron deficiency means the body does not have enough iron. Blood tests are needed because taking iron does not correct thalassemia unless iron deficiency is also present.

Can thalassemia trait turn into thalassemia major?

No. The gene pattern inherited at conception does not change into a more severe pattern later. Illness, pregnancy, or another cause of anemia may make symptoms more noticeable, but trait itself does not become thalassemia major.

Can two people with thalassemia trait have a healthy child?

Yes, but there may also be a chance of a child having significant thalassemia. The chance depends on the exact alpha or beta globin changes carried by each parent. Genetic testing and counseling can clarify the possible outcomes.

Can I donate blood if I have thalassemia trait?

Some people with thalassemia trait can donate if they meet the blood service's eligibility rules, including its minimum hemoglobin level. Rules differ between services, and mild anemia may prevent donation.

Does thalassemia affect life expectancy?

Thalassemia trait usually does not shorten life. Severe thalassemia can affect life expectancy, especially when anemia or iron overload is not well controlled. Consistent transfusion care, iron management, and organ monitoring have improved outcomes substantially.

Is thalassemia curable?

Sometimes. A stem cell transplant can cure selected people, and gene therapies may offer a cure for some people with transfusion-dependent beta thalassemia. These are intensive treatments with important risks and eligibility requirements. Standard supportive treatment controls thalassemia but does not remove the inherited gene changes.

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