

Thalassemia

A plain-English guide for you and your family

WHAT IS HAPPENING

Normally, your red blood cells carry oxygen to every part of your body using a protein called hemoglobin. Hemoglobin is made from four chains—two alpha chains and two beta chains—that must be perfectly balanced. In thalassemia, your genes do not give the right instructions to make one of these chains. When one chain is missing or low, the other chain piles up inside young red blood cells and damages them, so they break apart before they finish their normal 120-day life. This early death of red blood cells causes anemia, which means your body does not have enough healthy red blood cells to deliver oxygen where it is needed. Depending on how many genes are affected, thalassemia can be so mild you never know you have it, or severe enough to need regular blood transfusions for life.

What you might feel

- Pale skin and pale mucous membranes (pallor)
- Extreme tiredness and weakness (fatigue)
- Shortness of breath with activity
- Yellowing of skin and eyes (jaundice)
- Enlarged spleen (splenomegaly)
- Bone pain and facial changes (frontal bossing, prominent cheekbones)
- Slow growth and delayed puberty in children
- Severe chest pain or sudden difficulty breathing

Call the doctor right away if...

- Severe chest pain or sudden shortness of breath may indicate heart failure or acute chest syndrome
- High fever in a patient after splenectomy requires immediate antibiotics for possible overwhelming sepsis
- Sudden severe abdominal pain with rapid drop in hemoglobin suggests splenic sequestration crisis or rupture

- New irregular heartbeat or fainting may signal iron overload cardiomyopathy with dangerous arrhythmias

What the care team will do

- Monitor hemoglobin and hematocrit levels closely
- Assess for signs of iron overload
- Administer chelation therapy as prescribed
- Educate families about genetic counseling and inheritance patterns
- Monitor growth and development in pediatric patients
- Prevent and manage infections promptly
- Coordinate interdisciplinary care with hematology, endocrinology, and cardiology
- Support psychosocial needs and quality of life

Medicines you may be given — and why

deferasirox

Deferasirox is a molecule that wraps around iron atoms in the bloodstream and tissues. The drug-iron complex then travels to the kidneys, which filter it out into urine. This removes excess iron that builds up from repeated blood transfusions.

deferoxamine

Deferoxamine is given by slow injection under the skin or into a vein over many hours. It binds tightly to iron in the blood and tissues, especially in the heart and liver. The kidneys and liver then remove the iron-drug combination from the body.

deferiprone

Deferiprone is a small pill that absorbs quickly from the gut into the blood. It crosses into heart muscle very well and pulls iron out of cells where it has deposited. The liver processes the drug-iron complex and removes it in urine and stool.

luspatercept

Luspatercept is a protein given by injection under the skin every three weeks. It blocks certain signals (TGF-beta superfamily ligands) that stop red blood cell precursors from maturing. By removing this brake, more immature red cells in the bone marrow develop into healthy functioning red blood cells.

packed red blood cell transfusion

Donated red blood cells from healthy individuals are given directly into the patient's vein over 2-4 hours. The transfused cells immediately increase oxygen-carrying capacity by raising hemoglobin. Regular transfusions keep hemoglobin high enough to suppress the patient's own abnormal red cell production in the bone marrow.

folic acid

Folic acid is a vitamin needed to make DNA in all dividing cells, including red blood cell precursors. In thalassemia, the bone marrow works overtime to produce red cells, using up folate quickly. Daily folic acid supplements keep folate levels normal so DNA synthesis and red cell production do not slow down further.

Where this information comes from

- American Society of Hematology (ASH) guidelines on transfusion and iron chelation therapy in thalassemia major (2021)
- Thalassemia International Federation (TIF) Guidelines for the Management of Transfusion Dependent Thalassemia (2022)
- National Heart, Lung, and Blood Institute (NHLBI) Evidence-Based Management of Sickle Cell Disease and Thalassemia (2020)
- American Academy of Pediatrics (AAP) recommendations on genetic counseling and care coordination for hemoglobinopathies (2019)

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