

# Hemoglobin E Disease Fact Sheet

## What is hemoglobin E disease?

Hemoglobin E disease is a condition found mostly in the Asian population. It appears most commonly in people from Thailand, Cambodia, Bengal, Vietnam, Laos, Malaysia, and southern China. Hemoglobin is the part of the red blood cells that carries oxygen through the body. The most common (“normal”) form is called hemoglobin A. Your child’s red blood cells contain a different hemoglobin called hemoglobin E.

## How does someone get hemoglobin E disease?

Hemoglobin E disease is an inherited condition. If a child has hemoglobin E disease, it usually means both parents have hemoglobin E trait. The trait for hemoglobin E occurs in up to 30% of people from certain areas of Southeast Asia. Hemoglobin E trait causes no problems. *When both parents have E trait, each baby they have together has a 1 in 4 chance of inheriting hemoglobin E disease.*

Another type of hemoglobin E disease is called hemoglobin E-beta thalassemia. This situation can occur when one parent carries hemoglobin E trait, and the other carries beta-thalassemia trait (a different hemoglobin disorder). Beta-thalassemia trait may cause a slightly low blood count (anemia), which usually produces no symptoms. Once again, the risk of disease for each baby born to such a couple is 1 in 4.

## What problems can hemoglobin E disease cause?

Most people with classic hemoglobin E disease (“EE”) have mild anemia, and the red blood cells are smaller than usual. Usually this low blood count does not cause serious symptoms or problems. People with hemoglobin E-beta thalassemia may have similar findings. However, the most serious form of the disorder (so-called hemoglobin E beta-zero thalassemia), causes a severe anemia that may lead to heart failure, enlargement of the liver and spleen, poor growth, and bone changes. Because the newborn screening test does not distinguish between “EE” disease and hemoglobin E beta-zero thalassemia, it is important to establish a correct diagnosis. This may require testing of both parents.

## Are there any special treatments for people with hemoglobin E disease?

Many people who have classic hemoglobin E disease or a milder form of hemoglobin E-beta thalassemia disease do quite well. Iron and vitamins will not raise the blood counts. No other treatments or special precautions are generally necessary. Since this is a lifelong condition, however, it is necessary to be aware of its existence.

For people with the severest form, hemoglobin E beta-zero thalassemia, there is no established cure, although research continues. Repeated blood transfusions are usually necessary.