

# Positive Result:

## Blood Spot Screen Result Notification



### Hemoglobin E, no A (hemoglobin E disease or hemoglobin E-beta thalassemia)

#### What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby had only hemoglobin E and was missing normal hemoglobin (hemoglobin A).

#### What does this mean?

When only hemoglobin E is found, it is likely that your baby has either a condition called hemoglobin E disease or one called hemoglobin E-beta thalassemia.

#### What happens next?

Your baby's doctor or a specialist (pediatric hematologist) familiar with hemoglobin E and hemoglobin E-beta thalassemia will help arrange for more testing.

#### What is hemoglobin E disease?

Hemoglobin E disease is a disorder which affects the hemoglobin/blood. Hemoglobin is a protein in the red blood cells. The job of hemoglobin is to carry oxygen throughout the body.

Red blood cells containing normal hemoglobin (hemoglobin A) are round and flexible. People with hemoglobin E have red blood cells that contain no normal hemoglobin.

Most people with classic hemoglobin E disease ("EE") have mild anemia (drop in red blood cell count), and the red blood cells are smaller than usual. Usually this low blood count does not cause serious symptoms or problems. While usually without symptoms, this can be painful and cause health problems.

#### What is hemoglobin E-beta thalassemia?

People with hemoglobin E-beta thalassemia (E $\beta$ ) may have similar findings to hemoglobin E disease. However, those with hemoglobin E-beta thalassemia have a moderate to severe form of anemia.

It is important to have your provider or specialist do further testing to get a correct diagnosis. Newborn screening tests are not able to distinguish between hemoglobin E disease (EE) and hemoglobin E-beta thalassemia (E $\beta$ ).

#### What health problems can it cause?

Hemoglobin E disease or hemoglobin E-beta thalassemia is different for each child. This disease is a lifelong condition that may result in a mild to severe version of anemia. If untreated, it is possible for it to cause:

- Anemia
- Enlarged spleen

Children with hemoglobin E disease or hemoglobin E-beta thalassemia can benefit from prompt and careful treatment.

#### What treatment options are available?

Treatment for hemoglobin E disease or hemoglobin E-beta thalassemia throughout childhood will depend upon each child's unique case. Children with either of these conditions should see their regular doctor and a doctor who specializes in blood disorders.

#### Resources

**Genetics Home Reference:**  
<http://ghr.nlm.nih.gov>

**Save Babies Through Screening Foundation:**  
[www.savebabies.org](http://www.savebabies.org)

**Baby's First Test:**  
[www.babysfirsttest.org](http://www.babysfirsttest.org)