Parents’ Guide to Hemoglobin E Disease

You have just learned that your infant has hemoglobin E disease. Naturally you are concerned and have many questions. This information sheet will help answer some of your questions. However, it should not take the place of an informed discussion with your baby’s doctor (primary care provider).

**What is Hemoglobin?**

Hemoglobin is a protein in the red blood cells. It carries oxygen from the lungs to all parts of the body and gives blood its red color. There are many hemoglobin types (this is not the same as a blood type). Hemoglobin is inherited through genes, one from each parent. Most people have hemoglobin A, also called adult or normal hemoglobin. The presence of hemoglobin A in red blood cells makes them smooth and shaped with a large, rounded depression on each side of the cell, known as a biconcave disk shape (see picture). This shape allows normal red blood cells to be very flexible, moving easily through the blood vessels to deliver oxygen to the body.

**What is Hemoglobin E Disease (EE)?**

Hemoglobin E in the red blood cells is responsible for causing hemoglobin E disease. Children inherit this disease from their parents as a recessive genetic disorder. This means a hemoglobin E gene is passed from both mom and dad to the baby causing hemoglobin E disease (hemoglobin EE). Persons with hemoglobin EE have only hemoglobin E and no adult or normal hemoglobin. When both parents have one hemoglobin E gene, there is a 1 in 4 or 25% chance with each pregnancy that an infant will inherit two hemoglobin E genes. There are no serious health problems associated with hemoglobin EE, but the gene for hemoglobin E is passed on from your child to your future grandchildren. Hemoglobin E disease is not contagious.

Most people with hemoglobin E disease have mild anemia and occasionally may have a slightly enlarged spleen; but usually they do not have disease symptoms and do not require treatment. Persons with hemoglobin E disease have red blood cells that are smaller than normal and have an irregular shape because particles inside the red blood cells draw together toward the center of the cells (contract). Up to 75% of the hemoglobin EE red blood cells look like a bull’s eye target with a dark center (pictured below on left).

**What Problems can Hemoglobin E Disease (EE) Cause?**

Hemoglobin EE red blood cells are not very flexible in moving through blood vessels and have a smaller outside surface area to carry oxygen because they do not have the biconcave disk shape of normal red blood cell (pictured above). Hemoglobin EE red blood cells are very small, unstable and have a...
reduced ability to hold onto oxygen. The lifespan of these red blood cell is also shorter than normal. (Normal blood cells live about 120 days).

**What is the Frequency of Hemoglobin E Disease (EE)?**

Hemoglobin E disease affects both sexes equally and is the second most common abnormal hemoglobin in the world. It is very common among persons from Southeast Asia or have ancestors from Cambodia, Laos and Thailand. Hemoglobin EE is also found in people who live in Vietman, Malaysia, northeastern India, Bangladesh, Pakistan and Sri Lanka and their descendants.

**What can be done to Treat Hemoglobin E Disease (EE)?**

Treatment is usually not necessary. Folic acid supplements may be prescribed by your child’s doctor to help his or her body to produce normal red blood cells and improve the symptoms of anemia. People with hemoglobin E disease can expect to lead a normal life.

**What is Hemoglobin E/Beta-Thalassemia Disease?**

Hemoglobin E/beta-thalassemia disease is a more serious disease than hemoglobin E disease (EE). Children with hemoglobin E/beta-thalassemia inherit one gene for hemoglobin E from one parent and one beta-thalassemia gene from the other parent. The beta-thalassemia gene causes the body to make less than the normal amount of hemoglobin. Persons who are affected may have a more severe anemia which can be life threatening, depending on the severity of the beta-thalassemia. If not treated, this disease can lead to heart failure from the severe destruction of red blood cells. It also can lead to severe enlargement of the spleen and liver, changes in bones and poor growth. Treatment may include repeated blood transfusions. Your baby’s doctor will do a complete blood count (CBC) and smear of the red blood cells to look for beta-thalassemia when your baby is approximately 6 to 9 months of age.

**What are the Most Important Things to Remember about Hemoglobin E Disease (EE) and Hemoglobin E/Beta-Thalassemia Disease?**

- Work very closely with your child’s doctor and hematologist (a doctor who is a blood specialist). Make sure your child has regular checkups with them.

- Call your child’s doctor when you have questions and have your child seen if you have any medical concerns.

**How Do I Get More Information about Hemoglobin E Disease?**

- Talk with your baby’s doctor. You may also want to have a genetic consultation for you and your family to see how these diseases might affect future children or grandchildren.