IDPH
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NEWBORN SCREENING
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Hemoglobin E Disease

Information for Physicians and Other Health Care Professionals

Definition

Hemoglobin E is an inherited variant of hemoglobin A. Individuals with hemoglobin E disease do not produce hemoglobin A; they only produce hemoglobin E.

Clinical Symptoms

Hemoglobin E is considered to be benign; most individuals with the disease are asymptomatic. Some individuals may develop a mild thalassemia in the first few months of life as fetal hemoglobin decreases and hemoglobin E increases. Splenomegaly (enlarged spleen) is a rare symptom.

Newborn Screening and Definitive Diagnosis

In Illinois, high performance liquid chromatography (HPLC) is used to test for the presence of hemoglobin variants, such as hemoglobin E. **If results indicate presence of hemoglobin E, referral should be made to a pediatric hematologist**. Confirm diagnosis with infant blood tests and test the parents to rule out hemoglobin E thalassemia disease.

Treatment

Hemoglobin E disease is usually asymptomatic.

Incidence

Hemoglobin E disease is seen most frequently among people of Southeast Asian (Cambodian, Laotian, Vietnamese and Thai) heritage. It is also found in people of Chinese, Filipino, Asiatic Indian, and Turkish descent.

Inheritance Patterns

Hemoglobin E is an autosomal recessive disease. It occurs when an individual inherits two genes coding for hemoglobin E, one from each parent. Parents of a child with hemoglobin E disease are usually unaffected, healthy carriers of the disease. They are said to have hemoglobin E trait.

If both parents have hemoglobin E trait, there is a 25 percent chance that they will have a child with hemoglobin E disease; a 50 percent chance for a child with hemoglobin E trait; and a 25 percent chance for an unaffected child. **Genetic counseling is recommended for families planning future pregnancies.**

Physiology
Hemoglobin E disease is caused by a lysine substitution (in place of glutamic acid) at the 26^{th} position of an individual's beta globin chains. The substitution results in a slight decrease in production of the beta chain variant (thalassemia).

Key Points for Parents
Hemoglobin E disease is considered benign; most individuals with the disease are asymptomatic. Avoid overly alarming the child's parents, especially if the diagnosis has not yet been confirmed.

If results indicate presence of hemoglobin E, make certain the parents understand the importance of following the pediatrician's and/or pediatric hematologist's recommendations for additional testing and referrals.

Follow-up After Confirmation of Diagnosis
These guidelines should be followed after a diagnosis of hemoglobin C disease has been confirmed:

1) Follow-up with the child's hematologist as recommended.
2) Parents should understand that hemoglobin E is a benign disease; most affected individuals are asymptomatic and treatment is rarely necessary.
3) Recommend genetic counseling services to help the parents understand the complexity surrounding the carrier state and inheritance of this disease.
4) Additional information about newborn screening can be found at:
   - Baby's First Test: http://www.babysfirsttest.org/
     Health Resource and Service Administration (HRSA), Grant no. U36MC16509, Quality Assessment of the Newborn Screening System.

   - National Center for Biotechnology Information: http://www.ncbi.nlm.nih.gov/gtr/
     National Center for Biotechnology Information, U.S. National Library of Medicine 8600 Rockville Pike, Bethesda MD, 20894 USA.