



**NEWBORN SCREENING
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Hemoglobin D Disease
(D-Los Angeles, D-Punjab, D-Ibadan)
Information for Physicians and Other Health Care Professionals

Definition

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

Clinical Symptoms

A mild hemolytic anemia develops in the first few months of life as fetal hemoglobin decreases and hemoglobin D increases. Individuals with hemoglobin D disease may develop an enlarged spleen; however, most have no serious health problems.

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 D. **If results indicate presence of hemoglobin D, referral should be made to a pediatric hematologist.** Confirm diagnosis with infant blood tests and test the parents to rule out hemoglobin D thalassemia disease.

Treatment

Treatments are available to relieve symptoms caused by the disease.

Incidence

Hemoglobin D disease is seen most frequently among people of Asiatic Indian heritage and those of European descent; however, anyone can have the disease.

Inheritance Patterns

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

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

Physiology

There are several hemoglobin variants that cause hemoglobin D disease. In D-Los Angeles and D-Punjab, the disease is caused by a glutamic acid substitution (in place of glutamine) at the 121st position of an individual's beta globin chains. In D-Idaban, lysine is substituted for threonine at the seventh position. Microcytic red blood cells with reduced oxygen affinity are seen in individuals with hemoglobin D disease.

Key Points for Parents

Hemoglobin D disease is a mild hemolytic anemia. Avoid overly alarming the child's parents, especially if the diagnosis has not yet been confirmed.

If results indicate presence of hemoglobin D, 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 D disease has been confirmed:

- 1) Follow-up with the child's hematologist as recommended.
- 2) Parents should understand that hemoglobin D is a mild disease; symptoms may not appear in every affected individual and treatment is not always 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.