Inheritest℠
CARRIER SCREEN

What is D-Bifunctional Protein Deficiency?
D-bifunctional protein (DBP) deficiency is an inherited disease characterized by neonatal low muscle tone, seizures, visual and hearing loss, developmental delays, and death usually by two years of age.[1] DBP deficiency involves defects in the D-bifunctional protein that is involved in the breakdown of a specific type of fatty acids in the body called "very long-chain fatty acids" (VLCFA). The symptoms of DBP deficiency are due to the toxic build up of VLCFA, which causes damage in the cells, especially in the brain and liver.[2] DBP deficiency is also known as peroxisomal bifunctional enzyme deficiency and pseudo-Zellweger syndrome.[3]

What are the symptoms of D-Bifunctional Protein Deficiency and what treatment is available?
Signs and symptoms of DBP deficiency are usually evident within the first month of life and may include:[1,4]

- Hypotonia (low muscle tone)
- Seizures
- Hepatomegaly (enlarged liver) and liver disease
- Vision and hearing problems
- Unusual facial features
- Mental retardation

There is no cure for DBP deficiency. Treatment is supportive care for symptoms. D-bifunctional protein deficiency is usually fatal by the age of two years, with survival rarely to later childhood.[1]

How is D-Bifunctional Protein Deficiency inherited?
D-bifunctional protein deficiency is an autosomal recessive disease caused by mutations in the *HSD17B4* gene.[4] An individual who inherits one *HSD17B4* gene mutation is a "carrier" and is not expected to have related health problems. An individual who inherits two *HSD17B4* gene mutations, one from each parent, is expected to be affected with DBP deficiency.

If both members of a couple are carriers of an *HSD17B4* gene mutation, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for D-Bifunctional Protein Deficiency?
D-bifunctional protein deficiency can occur in individuals of all races and ethnicities, and it is estimated to affect 1 in 100,000 individuals.[2]

Having a relative who is a carrier or is affected can also increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?
If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

LabCorp
Laboratory Corporation of America

Integrated GENETICS
LabCorp Specialty Testing Group

Inheritest℠
CARRIER SCREEN

What does a negative test result mean?
A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

- Genetics and Rare Disease Information (GARD): http://rarediseases.info.nih.gov/GARD/Condition/4539/Dbifunctional_protein_deficiency.aspx/Showall

References:

1. Ferdinandusse, S *et al.* Mutational spectrum of D-bifunctional protein deficiency and structure-based genotype-phenotype analysis. *Am J Hum Genet*. 2006; 78:112-124.
2. Ferdinandusse S *et al.* Clinical and biochemical spectrum of D-bifunctional protein deficiency. *Ann Neurol* 2006; 59:92-104.
3. D-bifunctional protein deficiency. Genetics and Rare Disease Information Center. Available at: https://rarediseases.info.nih.gov/GARD/Condition/4539/Dbifunctional_protein_deficiency.aspx/Showall. Accessed: May 13, 2012.
4. D-bifunctional protein deficiency. *OMIM.* Available at: http://omim.org/entry/261515#reference2 Accessed: March 23, 2012.

LabCorp
Laboratory Corporation of America

Integrated GENETICS
LabCorp Specialty Testing Group