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.
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):

References: