Genetics Uncoded:

NxGen MDx

Facts about

D-Bifunctional Protein Deficiency

What Your Test Results Mean

Carriers show no symptoms of D-bifunctional protein deficiency and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

D-Bifunctional Protein Deficiency Explained

D-bifunctional protein deficiency is an inherited condition that inhibits the cell's ability to break down fatty acids. This leads to severe neurodegeneration beginning in early infancy. Newborns affected by D-bifunctional protein deficiency have poor muscle tone and seizures. These symptoms continue and these babies typically reach very few developmental milestones. They often have severe mental and physical retardation and develop visual and hearing impairment. Most affected individuals die before reaching age two. There is no cure for D-bifunctional protein deficiency but research is ongoing to determine whether stem cell transplant may be beneficial.

How the Genetics Work

D-bifunctional protein deficiency is an autosomal recessive disorder caused by pathogenic variants in the *HSD17B4* gene. In general, individuals have two copies of the *HSD17B4* gene. Carriers of D-bifunctional protein deficiency have a single variant in one copy of the *HSD17B4* gene while individuals with D-bifunctional protein deficiency have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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