Genetics Uncoded:
FACTS ABOUT D-BIFUNCTIONAL PROTEIN DEFICIENCY

AKA | 17-beta-hydroxysteroid dehydrogenase IV deficiency, Bifunctional peroxisomal enzyme deficiency, DBP deficiency, PBFE deficiency, Peroxisomal bifunctional enzyme deficiency, Pseudo-Zellweger syndrome, Zellweger-like syndrome

What Your Test Results Mean
Testing results indicate that you are a carrier of D-bifunctional protein deficiency. Carriers typically show no symptoms of D-bifunctional protein deficiency; however, carriers are at an increased risk of having a child with D-bifunctional protein deficiency. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor.

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 mutations in the HSD17B4 gene. In general, individuals have two copies of the HSD17B4 gene. Carriers of D-bifunctional protein deficiency have a single mutation in one copy of the HSD17B4 gene while individuals with D-bifunctional protein deficiency have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Recommended Next Steps
Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

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