

D-Bifunctional Protein Deficiency

What Your Results Mean

Test results indicate that you are a carrier of D-bifunctional protein deficiency. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of D-bifunctional protein deficiency for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.



Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for D-bifunctional protein deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

D-Bifunctional Protein Deficiency Explained

What is D-Bifunctional Protein Deficiency?

D-bifunctional protein deficiency is an inherited condition that leads to neurodegeneration beginning in early infancy. Newborns with D-bifunctional protein deficiency have poor muscle tone, seizures, structural brain differences, and characteristic facial features. Most infants are unable to acquire developmental skills. Some infants may achieve developmental milestones; however, they experience a gradual loss of these skills within a few months. Most affected individuals die before two years old.



Prognosis

Prognosis is considered poor. Most affected individuals die before the age of two.

Treatment

There is no cure for D-bifunctional protein deficiency. Treatment is mostly symptomatic.



Resources

Genetic and Rare Diseases Information Center

<https://rarediseases.info.nih.gov/diseases/4539/d-bifunctional-protein-deficiency>

Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/d-bifunctional-protein-deficiency>

National Society of Genetic Counselors

<https://www.nsgc.org/>