

# 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/>