NxGen MDx

Bardet-Biedl Syndrome, *BBS1*-Related

What Your Results Mean

Test results indicate that you are a carrier of Bardet-Biedl syndrome. Carriers are not expected to show symptoms of this syndrome. You and your partner would both have to be carriers of Bardet-Biedl syndrome for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Bardet-Biedl syndrome can be caused by changes in a variety of genes. Carrier testing for genes related to Bardet-Biedl syndrome is recommended for your partner or donor 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 are carriers for Bardet-Biedl syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

Bardet-Biedl Syndrome Explained

What is Bardet-Biedl Syndrome?

Bardet-Biedl syndrome is an inherited condition characterized by progressive vision loss, obesity, extra fingers or toes, intellectual disability, kidney disease, and abnormalities of the genitalia. Signs and symptoms of Bardet-Biedl can vary between affected individuals and even members of the same family. Complications from obesity can include type 2 diabetes, high blood pressure, and high cholesterol. Affected males produce reduced amounts of sex hormones (hypogonadism) and are usually infertile. Bardet-Biedl syndrome can additionally affect the heart, liver, and digestive system.

Prognosis

Prognosis varies for individuals with Bardet-Biedl syndrome as symptoms and severity can vary. Most affected individuals are legally blind by early adulthood. Kidney failure is the most common cause of death, but with early diagnosis and consistent management, some individuals can have a normal life expectancy.

Treatment

Treatment for Bardet-Biedl syndrome is symptomatic as there is no known cure. Dialysis and/or kidney transplantation can be considered for kidney disease. Diet and exercise can be beneficial to manage obesity. Extra fingers and toes can be removed by surgery if desired. Some affected individuals may also benefit from early intervention or attending a school for the visually impaired.

Resources

Bardet Biedl Syndrome Family Association
https://www.bardetbiedl.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/bardet-biedl-syndrome
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/bardet-biedl-syndrome/
National Society of Genetic Counselors
https://www.nsgc.org/

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