Bardet-Biedl Syndrome, *MMKS*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Bardet-Biedl syndrome, *MMKS*-related. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of Bardet-Biedl syndrome, *MMKS*-related 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 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 are carriers for Bardet-Biedl syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

Bardet-Biedl Syndrome, *MMKS*-Related Explained

What is Bardet-Biedl Syndrome, *MMKS*-Related?

Bardet-Biedl syndrome, *MMKS*-related, also known as Bared-Biedl 6 (BBS6), 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.

Variants in the *MMKS* gene can also cause an autosomal recessive condition called McKusick-Kaufman syndrome (MKS), an inherited condition that shares many of the same symptoms as Bardet-Biedl syndrome. It is characterized by a combination of three features: extra fingers and/or toes, heart defects, and genital abnormalities.

Most females with McKusick-Kaufman syndrome are born with hydrometrocolpos, which is a large accumulation of fluid in the pelvis. Genital abnormalities in males with McKusick-Kaufman syndrome can include placement of the urethral opening on the underside of the penis, a downward-curving penis, and undescended testes.

Bardet-Biedl syndrome has several features that are not seen in McKusick-Kaufman syndrome. These include vision loss, delayed development, obesity, and kidney (renal) failure. Because some of these features are not apparent at birth, the two conditions can be difficult to tell apart in infancy and early childhood.

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
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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. With early diagnosis and consistent management, some individuals can have a normal life expectancy.

The prognosis of McKusick-Kaufman syndrome differs from Bardet-Biedl syndrome as kidney failure is not noted in these individuals. There is limited data regarding life expectancy for individuals with MKS, but life expectancy is not known to be reduced.

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 wanted. Some affected individuals additionally benefit from early intervention or attending a school for the visually impaired.

Treatment for MKS focuses on surgically repairing a congenital heart defect if necessary as well as drainage of the fluid related to hydrometrocolpos. Extra fingers and toes can be removed by surgery if wanted as well.

Resources

Bardet Biedl Syndrome Family Association
https://www.bardetbiedl.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/bardet-biedl-syndrome
https://ghr.nlm.nih.gov/condition/mckusick-kaufman-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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