NxGen MDx

Inclusion Body Myopathy 2

What Your Results Mean

Test results indicate that you are a carrier of inclusion body myopathy 2. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of inclusion body myopathy 2 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 inclusion body myopathy 2, each of your children has a 1 in 4 (25%) chance to have the condition

Inclusion Body Myopathy 2 Explained

What is Inclusion Body Myopathy 2?

Inclusion body myopathy 2 is an inherited condition that slows the body's production of sialic acid, a sugar molecule that allows cells in tissues to signal and adhere to one another. This disease causes progressive weakening of the skeletal muscles, usually beginning in an affected individual's late teens or early twenties. About 20 years after symptoms appear, the disease progresses to the point that the person can no longer walk and must use a wheelchair. Inclusion body myopathy 2 does not weaken the eye and heart muscles and does not cause neurological symptoms.

Prognosis

Prognosis is generally favorable. Affected individuals usually require a wheelchair 20-30 years after disease onset, but lifespan is not shortened.

Treatment

There is no cure for this disease, but physical and occupational therapy can help individuals retain strength and mobility for as long as possible.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/inclusion-body-myopathy-2
National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

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