Nemaline Myopathy 2

What Your Results Mean

Test results indicate that you are a carrier of nemaline myopathy 2. Carriers typically show no symptoms. You and your partner would both have to be carriers of nemaline 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/donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner/donor are carriers for nemaline myopathy 2, each of your children has a 1 in 4 (25%) chance to have the condition.

Nemaline Myopathy 2 Explained

What is Nemaline Myopathy 2?

Nemaline myopathy 2 is an inherited muscle disorder that causes muscle weakness throughout the body, though most severely in the muscles of the face, neck, and limbs. Individuals exhibit weakness and feeding difficulties at birth or in the first year of life. Cardiac muscle and respiratory muscle weakness, which is life-threatening, is rare in nemaline myopathy 2. In general, weakness is usually static or very slowly progressive, and most individuals are able to lead independent, active lives.

Prognosis

Prognosis is generally favorable. Most children are eventually able to walk. However, 6% of affected patients die from respiratory failure in childhood. A more severe course has been described with certain variants more prevalent among the Ashkenazi Jewish population.

Treatment

There is no cure for nemaline myopathy 2, and treatment is based on symptoms. Management includes monitoring of nutritional status, special feeding techniques, aggressive treatment of lower respiratory tract infections, ventilator use, physical and speech therapy, and standard care for gastroesophageal reflux.

Resources

Building Strength: A Foundation for Nemaline Myopathy
http://www.nemaline.org/

Nemaline Myopathy Support Group
http://buildingstrength.org/

National Society of Genetic Counselors
https://www.nsgc.org/

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