Genetics Uncoded:

NxGen MDx

Facts about

NEB-Related Nemaline Myopathy

What Your Test Results Mean

Carriers typically show no symptoms of *NEB*-related Nemaline myopathy (*NEB*-related NM); however, carriers are at an increased risk of having a child with NEB-related NM. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

NEB-related NM Explained

NEB-related NM is an inherited muscle disorder in which inefficient muscle contraction leads to muscle weakness and the other features of the disease. Individuals with *NEB*-related NM are not able to properly produce the protein nebulin. Without nebulin, abnormal proteins called nemaline bodies collect in muscle fibers causing inability of skeletal muscles to contract. There are several forms of Nemaline myopathy ranging from severe to adult onset. Most individuals with *NEB*-related NM have a more mild form of NM called typical congenital NM. Symptoms including hypotonia, weakness, and feeding difficulties usually present in the first year of life; however, weakness is typically very slowly progressive and most individuals are able to lead independent, active lives.

Treatment of individuals with *NEB*-related NM typically includes supportive therapies. Physical therapy to help with muscle weakness as well as speech and/or feeding therapies may be beneficial.

How the Genetics Work

NEB-related NM is the most common type of Nemaline myopathy and is caused by variants in the *NEB* gene. In general, individuals have two copies of the *NEB* gene. Carriers of *NEB*-related NM have a single variant in one copy of the *NEB* gene while individuals with *NEB*-related NM have variations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the condition is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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