Genetics Uncoded:

NxGen MDx

Facts about

CLN3-Related Neuronal Ceroid Lipofuscinosis

What Your Test Results Mean

Carriers typically show no symptoms of *CLN3*-related neuronal ceroid lipofuscinosis (NCL); however, carriers are at an increased risk of having a child with NCL. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

CLN3-Related NCL Explained

CLN3-related NCL is an inherited lysosomal storage disorder that impairs the cell's ability to break down proteins and primarily affects the nervous system. Symptoms typically begin around ages 4-10 with vision loss, blindness occurring within 2-4 years. This is followed by progressive motor deterioration, developmental regression, and increasing intellectual disability. Lifespan is shortened, but some individuals live to early adulthood. Clinical trials surrounding stem cell therapy and other treatments are currently underway.

How the Genetics Work

NCL can be explained by variants in one of thirteen genes. *CLN3*-related NCL is an autosomal recessive disorder caused by variants in the *CLN3* gene. In general, individuals have two copies of the *CLN3* gene. Carriers of NCL have a single variant in one copy of the *CLN3* gene while individuals with NCL have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder 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.

NxGenMDx, LLC
801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504

Revised 12/15/16