



Genetics Uncoded:



Facts about

CLN5-Related Neuronal Ceroid Lipofuscinosis



What Your Test Results Mean

Carriers typically show no symptoms of *CLN5*-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.

● *CLN5*-related NCL Explained

CLN5-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 with vision loss 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. *CLN5*-related NCL is an autosomal recessive disorder caused by variants in the *CLN5* gene. In general, individuals have two copies of the *CLN5* gene. Carriers of NCL have a single variant in one copy of the *CLN5* 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.