Neuronal Ceroid-Lipofuscinosis/Northern Epilepsy

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of neuronal ceroid-lipofuscinosis, also referred to as Northern epilepsy. Carriers typically show no symptoms. Risk for current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for 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 and their own personal clinical management.

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 neuronal ceroid-lipofuscinosis, each of your children has a 1 in 4 (25%) chance to have the condition.

Neuronal Ceroid-Lipofuscinosis/Northern Epilepsy

What is Neuronal Ceroid-Lipofuscinosis/Northern Epilepsy?

Neuronal ceroid-lipofuscinosis (NCL), or Northern epilepsy, is an inherited condition that causes recurrent seizures and progressive loss of intellectual function. All types of NCL are caused by defects in the process that help break down granules made of fat and protein called lipopigments. As a result, lipopigments accumulate in a person's tissues and lead to tissue degeneration over time. This leads to cognitive and motor function decline, seizures, loss of vision, and reduced lifespan. NCL, caused by variants in the *CLN8* gene, cause two different types of NCL: the EPMR (progressive epilepsy with mental retardation) type and the late infantile type, which has an earlier onset and a more rapid progression. Both forms result in cognitive and motor function decline, seizures, loss of vision, and reduced lifespan.

Prognosis

Prognosis is generally unfavorable. The NCL late infantile variant has an onset around two to four years of age. Patients show a rapid progression with speech delay, developmental regression, visual failure, balance problems, and eventual loss of ambulation. Death occurs between ages 10-30 years.

NCL EPMR presents with normal early development and onset of seizures between the ages of five to 10 years. Seizures increase in frequency until puberty. Mental retardation begins two to five years after the onset of seizures. Vision loss is not characteristic of this subtype. Death usually occurs after the fifth decade of life.

Treatment

There is no specific treatment for NCL, but symptomatic treatment can be used along with counseling. Seizures can be controlled with medication; however, this will not slow the progression of the disease towards mental disability. Care focuses on behavioral problems and depression. Physical and occupational therapy help retain physical ability.

Resources

Hide & Seek Foundation for Lysosomal Disease Research
https://hideandseek.org/

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

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