Neuronal Ceroid Lipofuscinosis, *PPT1*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of neuronal ceroid lipofuscinosis, *PPT1*-related. 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, *PPT1*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Neuronal Ceroid Lipofuscinosis, *PPT1*-Related Explained

What is Neuronal Ceroid Lipofuscinosis, *PPT1*-Related?

Neuronal ceroid lipofuscinosis, *PPT1*-related (NCL) is an inherited disorder caused by defects in the process that helps 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. There are several forms of NCL, largely differentiated by the gene responsible and the age at which symptoms begin. Variants in the *PPT1* gene typically result in the infantile or juvenile form of NCL.

Prognosis

Prognosis is generally unfavorable. The severity of neuronal ceroid lipofuscinosis, *PPT1*-related is variable and depends on the specific mutation of *PPT1*. Most patients show symptoms within the first two years of life. Development slows down which affects motor skills, and there may be low muscle tone, decelerating head growth, and seizures. Sleep disturbances and irritability are common. Vision loss generally occurs between 16 and 23 months of age. Mobility and language skills are lost as vision loss progresses. Children will eventually enter a vegetative state and become totally dependent on others to care for them. Death usually occurs in childhood for children with infantile NCL. Among those with juvenile form of NCL, death usually occurs between one's late teens to thirties.

Treatment

There is no specific treatment for NCL, but symptomatic treatment can be used along with counseling and prenatal care. Care focuses on behavioral problems and depression. Physical and occupational therapy help retain physical ability.

Resources
Batten Disease Support and Research Association
https://bdsra.org/
Beyond Batten Disease Foundation
https://beyondbatten.org/
National Society of Genetic Counselors
https://www.nsgc.org/

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