Neuronal Ceroid Lipofuscinosis, *CLN3*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of neuronal ceroid lipofuscinosis, *CLN3*-related. Carriers typically show no symptoms. You and your partner would both have to be carriers of neuronal ceroid lipofuscinosis, *CLN3*-related for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a 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 are carriers for neuronal ceroid lipofuscinosis, *CLN3*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Neuronal Ceroid Lipofuscinosis, *CLN3*-Related Explained

What is Ceroid Lipofuscinosis, *CLN3*-Related?

Neuronal ceroid lipofuscinosis, *CLN3*-related is often referred to as Batten disease. It is an inherited disease that causes degeneration of the brain, leading to progressive loss of mental and motor skills. Neuronal ceroid lipofuscinosis, *CLN3*-related typically causes blindness and leads to early death. Symptoms begin between the ages of four and 10, usually with vision loss, progressing to seizures, developmental delays, muscle stiffness, and behavioral and psychiatric symptoms, such as attention deficits and aggression.

Prognosis

Prognosis is generally unfavorable. Neuronal ceroid lipofuscinosis, *CLN3*-related causes blindness and progressive loss of mental and motor functions. Death usually occurs in the late teens to twenties. Some people with the disease have lived into their thirties.

Treatment

Treatment is supportive as there is no cure for neuronal ceroid lipofuscinosis, *CLN3*-related. Physical and occupational therapy can help retain physical ability.

Resources

Batten Disease Support and Research Association
https://bdsra.org/

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

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

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