

Neuronal Ceroid Lipofuscinosis, *CLN5*-Related

What Your Results Mean

Test results indicate that you are a carrier of neuronal ceroid lipofuscinosis, *CLN5*-related. Carriers typically show no symptoms. You and your partner would both have to be carriers of neuronal ceroid lipofuscinosis, *CLN5*-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.



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, *CLN5*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Neuronal Ceroid Lipofuscinosis, *CLN5*-Related Explained

What is Ceroid Lipofuscinosis, *CLN5*-Related?

Neuronal ceroid lipofuscinosis, *CLN5*-related is an inherited disease that causes degeneration of the brain, leading to progressive loss of mental and motor skills. Neuronal ceroid lipofuscinosis, *CLN5*-related typically causes blindness and leads to early death. Symptoms begin between the ages of four and seven, usually with clumsiness, progressing to vision loss, seizures, developmental delays, muscle stiffness, balance problems and involuntary movements.



Prognosis

Prognosis is generally unfavorable. Neuronal ceroid lipofuscinosis, *CLN5*-related causes blindness and progressive loss of mental and motor functions. Death usually occurs between 10 and 30 years of age.

Treatment

Treatment is supportive as there is no cure for neuronal ceroid lipofuscinosis, *CLN5*-related. Physical and occupational therapy help retain physical ability.



Resources

Hide and Seek Foundation for Lysosomal Disease Research

<https://hideandseek.org/>

National Society of Genetic Counselors

<https://www.nsgc.org/>