Clarity Genetics

Genetics Uncoded:

Facts about

PPT1-Related Neuronal Ceroid Lipofuscinosis

What Your Test Results Mean

Carriers typically show no symptoms of *PPT1*-related neuronal ceroid lipofuscinosis; however, carriers are at an increased risk of having a child with neuronal ceroid lipofuscinosis. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

PPT1-Related Neuronal Ceroid Lipofuscinosis Explained

Neuronal ceroid lipofuscinosis is an inherited disorder characterized by intellectual and motor disability, muscle twitches, recurrent seizures, vision impairment, and a progressive loss of nerve cells. Variants in the PPT1 gene result in a decreased production of the enzyme palmitoyl-protein thioesterase 1. This impairs the removal of fatty acids from proteins. The fats and proteins then accumulate and damage cells throughout the body, particularly nerve cells. The disease can develop in infancy or later in life. Affected infants rarely develop the ability to speak or walk, and death before age 10 is likely. Affected adults will have milder symptoms and have a normal life expectancy. There is no specific treatment for this disorder, so management of the individuals is symptomatic.

How the Genetics Work

PPT1-related neuronal ceroid lipofuscinosis is an autosomal recessive disorder is caused by pathogenic variants in the *PPT1* gene. In general, individuals have two copies of the *PPT1* gene. Carriers of neuronal ceroid lipofuscinosis have a single variant in one copy of the *PPT1* gene, while individuals with neuronal ceroid lipofuscinosis have variants in both copies of the *PPT1* gene, 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.

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