Facts about

**TPP1-Related Neuronal Ceroid Lipofuscinosis**

### What Your Test Results Mean

Carriers typically show no symptoms of *TPP1*-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.

### TTP1-Related Neuronal Ceroid Lipofuscinosis Explained

*TPP1*-related neuronal ceroid lipofuscinosis is an inherited condition that impairs the cell's ability to break down proteins and primarily affects the nervous system. In the late-infantile form, symptoms arise between the ages of two and four, while onset of symptoms in the juvenile form occurs between the ages of four and ten. Symptoms typically begin with seizures, followed by progressive motor deterioration, developmental regression, and increasing intellectual disability. Affected individuals suffer rapid vision loss, becoming completely blind about 2-4 years after seizures begin. Treatment for neuronal ceroid lipofuscinosis is symptomatic.

### How the Genetics Work

*TPP1*-related neuronal ceroid lipofuscinosis is an autosomal recessive disorder caused by pathogenic variants in the *TPP1* gene. In general, individuals have two copies of the *TPP1* gene. Carriers of neuronal ceroid lipofuscinosis have a single variant in one copy of the *TPP1* gene while individuals with neuronal ceroid lipofuscinosis have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

### Questions?

Contact us at 1-855-776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.