Genetics Uncoded:

NxGen MDx

Facts about Canavan Disease

What Your Test Results Mean

Carriers typically show no symptoms of Canavan disease; however, carriers are at an increased risk of having a child with Canavan disease. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Canavan Disease Explained

Canavan disease is an inherited metabolic disorder in which harmful amounts of N-acetylaspartic acid (NAA) accumulate within the central nervous system. Individuals with Canavan disease do not produce enough of one of the enzymes called aspartoacylase, which is needed to metabolize NAA. Over time, this excessive buildup of NAA in the brain causes demyelination and other signs of the disease.

There are two forms of Canavan disease. In the most common form, infantile Canavan, normal development is followed by regression of developmental milestones within the first year of life. With time, sleep disturbance, seizures, and feeding difficulties become apparent. Life expectancy is variable ranging from several years of age to teenage years. Individuals with the more mild form, juvenile Canavan, will have typical MRI findings and associated minor learning difficulties.

Treatment of individuals with Canavan disease is supportive and directed to providing adequate nutrition and hydration, managing infectious diseases, and protecting the airway.

How the Genetics Work

The clinical features of Canavan disease can be explained by variants in the *ASPA* gene. In general, individuals have two copies of the *ASPA* gene. Carriers of Canavan disease have a single variant in one copy of the *ASPA* gene while individuals with Canavan disease have variants in both copies of their genes, one inherited from each parent.

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.

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