Genetics Uncoded:

Facts about Joubert Syndrome 2

What Your Test Results Mean

Carriers typically show no symptoms of Joubert syndrome 2; however, carriers are at an increased risk of having a child with Joubert syndrome 2. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Joubert Syndrome 2 Explained

Joubert syndrome 2 is an inherited multisystem disorder caused by a defect in the cilia. It is characterized by abnormal development of regions near the back of the brain (molar tooth sign), hypotonia, and developmental delays. Other signs of the disease may include neonatal breathing abnormalities, eye abnormalities such as retinal dystrophy, skeletal anomalies, and renal or liver disease. Joubert syndrome 2 is caused by a deficient amount of transmembrane protein 216. This protein is required for the assembly and function of cilia.

Treatment of individuals with Joubert syndrome typically includes supportive therapies. Surgical interventions may be required for anomalies including oral clefting, polydactyly, and hydrocephalus. Life expectancy may be shortened in the presence of renal or liver failure or breathing abnormalities.

How the Genetics Work

The clinical features of Joubert syndrome 2 (and a similar disorder called Meckel syndrome) can be explained by pathogenic variants in the *TMEM216* gene. In general, individuals have two copies of the *TMEM216* gene. Carriers of Joubert syndrome 2 have a single variant in one copy of the *TMEM216* gene while individuals with Joubert syndrome 2 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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