Genetics Uncoded:
FACTS ABOUT JOUBERT SYNDROME 2

**What Your Test Results Mean**

Test results indicate that you are a carrier of Joubert syndrome 2. Carriers typically show no symptoms of Joubert syndrome 2; however, carriers are at an increased risk of having a child with Joubert syndrome 2. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

---

**Joubert Syndrome 2 Explained**

Joubert syndrome 2 is an inherited multisystem disorder caused by a defect in the cilia, the hair-like structures on the surface of cells. 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 mutations in the TMEM216 gene. In general, individuals have two copies of the TMEM216 gene. Carriers of Joubert syndrome 2 have a single mutation in one copy of the TMEM216 gene while individuals with Joubert syndrome 2 have mutations in both copies of their genes, one inherited from each parent.

---

**Recommended Next Steps**

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

---

**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.