Joubert Syndrome 2/ *TMEM216*-Related Disorders

What Your Results Mean

Test results indicate that you are a carrier of Joubert syndrome 2/*TMEM216*-related disorders. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of Joubert syndrome 2/*TMEM216*-related disorders for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for Joubert syndrome 2/*TMEM216*-related disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

Joubert Syndrome 2/*TMEM216*-Related Disorders Explained

What is/are Joubert Syndrome 2/*TMEM216*-Related Disorders?

Joubert syndrome 2 and *TMEM216*-related disorders are inherited, multisystem disorders caused by a defect in the cilia. The diseases are characterized by abnormal development of regions near the back of the brain (molar tooth sign), hypotonia, and developmental delays. Other symptoms can include infant breathing abnormalities, eye abnormalities such as retinal dystrophy, skeletal anomalies such as polydactyly, and kidney or liver disease.

A condition called Meckel syndrome 2 has also been associated with variants in the *TMEM216* gene. Meckel syndrome is a disorder with severe signs and symptoms that affect many parts of the body. The most common features are enlarged kidneys with numerous fluid-filled cysts; an occipital encephalocele, which is a sac-like protrusion of the brain through an opening at the back of the skull; and the presence of extra fingers and toes (polydactyly). Most affected individuals also have a buildup of scar tissue (fibrosis) in the liver.

Prognosis

Prognosis for Joubert syndrome 2 is generally unfavorable. Affected individuals typically have significant psychomotor and cognitive delays, as well as oral motor difficulties resulting in swallowing and chewing difficulties. Some individuals experience end-stage renal insufficiency by mid-adolescence.

Prognosis for Meckel syndrome 2 is also unfavorable. Because of their serious health problems, most individuals with Meckel syndrome die before or shortly after birth. Most often, affected infants die of respiratory problems or kidney failure.

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Treatment

Treatment typically involves supportive care, as there is no cure for Joubert syndrome 2/*TMEM216*-related disorders. Surgical interventions may be considered for physical anomalies such as oral clefting, polydactyly, and hydrocephalus.

Resources

Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/10167/joubert-syndrome-
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/joubert-syndrome
https://ghr.nlm.nih.gov/condition/meckel-syndrome
National Society of Genetic Counselors
https://www.nsgc.org/

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