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Joubert syndrome 2

What is Joubert syndrome 2?

Joubert syndrome 2 is an inherited disease characterized by brain malformations, developmental delay, low muscle tone, and breathing abnormalities.[1] Signs and symptoms of Joubert syndrome 2 are thought to be caused by the abnormal functioning of cilia, which are hair-like structures found on the surface of all cells of the body.[2] Joubert syndrome 2 is also known as cerebello-oculo-renal syndrome 2 (CORS2).[2,3]

What are the symptoms of Joubert syndrome 2 and what treatment is available?

Individuals with Joubert syndrome 2 have variable symptoms, including:[4]

- Brain malformations
- Mild to severe delayed development and intellectual disability
- Hypotonia (low muscle tone) in infancy, followed by ataxia (difficulty coordinating movements)
- Abnormal eye movements
- Abnormal breathing patterns
- Polydactyly (extra fingers and/or toes)
- Oral hamartomas (skin tags in the mouth)
- Abnormal kidney function

There is no cure for Joubert syndrome 2**. T**reatment includes supportive care for symptoms.[4]

How is Joubert syndrome 2 inherited?

Joubert syndrome 2 is an autosomal recessive disease caused by mutations in the *TMEM216* gene.[3] An individual who inherits one copy of a *TMEM216* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two *TMEM216* gene mutations, one from each parent, is expected to be affected with Joubert syndrome 2 or a related disease known as Meckel syndrome.[3,5]

If both members of a couple are carriers of a mutation in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Joubert syndrome 2?

Joubert syndrome 2 can occur in individuals of any ethnicity; however, it is most common in individuals of Ashkenazi (Eastern European) Jewish ancestry. In the Ashkenazi Jewish population, the carrier frequency has been found to be 1 in 92[1] and the incidence of Joubert syndrome 2 is calculated to be approximately 1 in 33,800.

Having a relative who is a carrier or is affected can also increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetic health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

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Where can I get more information?

Jewish Genetic Disease Consortium: http://www.jewishgeneticdiseases.org/diseases/joubert-syndrome-type-2/

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/joubert-syndrome

Center for Jewish Genetics: https://www.jewishgenetics.org/joubert-syndrome

References

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4. Parisi M and Glass I. Joubert syndrome. http://www.ncbi.nlm.nih.gov/books/NBK1325/. Accessed: Mar 2, 2012.
5. Transmembrane Protein 216; TMEM216. OMIM. http://omim.org/entry/613277. Accessed April 2, 2012.