Disability Awareness Begins With You:

Joubert Syndrome

What is Joubert Syndrome?

Joubert Syndrome is a rare brain malformation characterized by the absence or underdevelopment of the *cerebellar vermis* - an area of the brain that controls balance and coordination. The most common features of Joubert Syndrome in infants include abnormally rapid breathing (*hyperpnea*), jerky eye movements, mental retardation, and the inability to coordinate voluntary muscle movements (*ataxia*). Physical deformities may be present, such as extra fingers and toes, cleft lip or palate, and tongue abnormalities. Seizures may also occur. Most cases of Joubert Syndrome are sporadic (in other words, no other family member has the disorder), but in some families, Joubert Syndrome appears to be inherited via a recessive gene.

Is there any treatment?

Treatment for Joubert Syndrome is symptomatic and supportive. Infant stimulation and physical, occupational, and speech therapy may benefit some children. Infants with abnormal breathing patterns should be monitored

What is the prognosis?

The prognosis for infants with Joubert Syndrome depends on whether or not the cerebellar vermis is entirely absent or partially developed. Some children have a mild form of the disorder, with minimal motor disability and good mental development, while others may have severe motor disability and moderate mental retardation.

http://www.ninds.nih.gov/health_and_medical/disorders/joubert.htm

What are the characteristics of Joubert Syndrome?

The most typical features of Joubert Syndrome include:

- Absence or underdevelopment of part of the brain called the cerebellum vermis which controls balance and coordination.
- A malformed brain stem, which may cause an abnormal breathing pattern called episodic hypernea, in which babies pant, and may be followed by apnea (cessation of breathing).
- Abnormal eye and tongue movements.
- Decreased muscle tone. It can be marked in the neonatal period and in infancy.
- Seizures (less typical).

How is Joubert Syndrome inherited?

Joubert Syndrome is inherited in an autosomal recessive manner. Both parents are carriers of the gene. Genetic testing is not currently available to detect this condition. Parents who have a child with Joubert Syndrome have a 1 in 4 chance of having another affected child in another pregnancy. Prenatal testing with a level 3 ultrasound may be possible.

What is the prognosis for Joubert Syndrome?

While global developmental delay is frequent, health and growth are usually not severely affected. Commonly there is a degree of mild or moderate retardation. Severe mental retardation is uncommon. There are a few cases of affected individuals with normal intelligence or learning abilities.

What is the treatment for Joubert Syndrome?

Infant stimulation, physical therapy, occupational therapy, and speech therapy are valuable. Somewhat less traditional therapies such as auditory training, sensory integration, horseback therapy and water therapy have also been found to be very beneficial. Infants with abnormal breathing should have apnea monitoring.

http://www.joubertsyndrome.org/JoubertFAQ's.htm