



JOUBERT SYNDROME & RELATED DISORDERS FOUNDATION

The faith to believe, the hope to dream, the love to see it through

COACH Syndrome

(Cerebellar vermis hypoplasia/aplasia, Oligophrenia, Ataxia, Coloboma, and Hepatic fibrosis)

COACH is a mnemonic, whose name refers to the different characteristics of the disorder. It is one member of a group of genetic conditions that result from an abnormality in the part of the brain called the cerebellar vermis. The disorders that share this cerebellar malformation are known as Joubert syndrome and related disorders (JSRD). These conditions have some characteristics in common, but there is a spectrum of symptoms and abilities in affected individuals. For additional information regarding this family of conditions, please refer to the Joubert Syndrome & Related Disorders Foundation website at www.jsrdf.org.

Patients diagnosed with COACH traditionally exhibit the following features:

- Underdevelopment (hypoplasia) or complete lack (aplasia/agenesis) of the cerebellar vermis, usually indicated by the “Molar Tooth” sign found on an axial view of a brain MRI scan.
- Developmental delays/mental retardation—variable severity (oligophrenia)
- Difficulty coordinating voluntary muscle movements; uncoordinated movements (ataxia).
- A malformation of the retina or other regions of the eye (coloboma)--nonprogressive and may not impact vision
- Abnormalities of the liver, including hepatic fibrosis—may have a delayed onset or be slowly progressive
- Decreased muscle tone (hypotonia).

While less common, the following features may also be present in some individuals:

- Facial features may be abnormal in appearance (eyes far set from each other, small ear lobes, broad forehead, arched eyebrows, broad mouth)
- Renal insufficiency, particularly increased urination and excessive thirst (nephronophthisis).
- Rapid, involuntary movements of the eyes (nystagmus).
- Abnormal breathing pattern with episodes of rapid breathing or panting (hyperpnea), which may be followed by pauses in breathing (apnea).
- Difficulty processing and reacting to information received through any of their five senses.
- Other conditions not listed here may also be observed

Explanation of features:

Individuals diagnosed with COACH syndrome have an absence or underdevelopment of part of the brain called the cerebellar vermis which controls balance and coordination. The severity of the resulting ataxia varies from person to person.

Decreased muscle tone is common in children with COACH. As a result of the poor muscle tone, developmental delay (usually in gross motor, fine motor and speech areas) is common. Some children have also been noted to have abnormal eye and tongue movements. Developmental delays are usually treated through physical therapy, occupational therapy, speech therapy, and infant stimulation. Most children diagnosed with COACH syndrome are able to achieve standard milestones, although often at a much later age.

Some individuals experience difficulties resulting from an inability to appropriately process information received through the five senses - hearing, seeing, tasting, touching, and smelling - as well as from their poor sense of

balance and muscle movement. Some families have found that sensory integration therapy can help to minimize these sensory issues.

Mild to moderate mental retardation is typical, but overall health and growth are not known to be severely affected by this condition unless significant liver or kidney failure occurs.

Management and treatment:

Presently, there is no cure for COACH. It is recommended that individuals with COACH see the appropriate specialists necessary to help monitor their various clinical features. Suggested specialists include a nephrologist (kidney specialist), ophthalmologist (eye doctor), gastroenterologist (liver specialist), geneticist and neurologist, as well as any others recommended by your doctor.

Screening for some of the complications associated with COACH syndrome, such as liver or kidney involvement that may become progressive over time, is recommended on an annual basis. Please refer to the Joubert Syndrome Foundation & Related Cerebellar Disorders website's "Evaluation Recommendations" link for a complete listing of recommended annual tests.

Inheritance and recurrence:

COACH syndrome is passed down from parents to offspring as an autosomal recessive trait, which means that both parents have one altered copy of the gene(s) responsible for this disorder in their DNA. (In order for a child to be born with COACH, both the egg and the sperm must contain the same altered gene in question). The odds of having a child born with COACH to parents who carry the altered gene(s) involved are 1 in 4, or 25%, in each pregnancy that they share.

Genetic cause:

Presently, no genes have been identified that cause COACH syndrome specifically. It is likely that alterations in multiple genes cause this condition. Research is currently underway to assist medical professionals in developing a greater understanding about this disorder. For more information about genetic research, please contact the Joubert Syndrome & Related Disorders Foundation.

Additional resources for families:

- Joubert Syndrome & Related Disorders Foundation: www.jsrdf.org
- The ARC, an advocacy organization for individuals with disabilities: www.thearc.org
- National Eye Institute: www.nei.nih.gov
- American Liver Foundation: www.liverfoundation.org/

Resources used in the creation of this document:

- Gleeson, J.G. et al. (2003). Molar Tooth Sign of the Midbrain-Hindbrain Junction: Occurrence in Multiple Distinct Syndromes. *American Journal of Medical Genetics*, 125A, 125-134.
- Joubert Syndrome & Related Disorders Foundation website: www.jsrdf.org
- Multiple Congenital Anomaly/Mental Retardation (MCA/MR) Syndromes Database: <http://www.nlm.nih.gov/cgi/jablonski/syndrome.cgi?index=96>
- ORPHA.NET database on rare diseases and orphan drugs: www.orphanet.net
- Parisi, M.A. and Glass, I. A. "Joubert Syndrome" GeneReviews, Online publication of expert-authored disease reviews: www.genereviews.org
- Satran, D, Pierpont, M. M., & Dobyns, W. B. (1999). Cerebello-Oculo-Renal Syndromes Including Arima, Senior-Loken, and COACH Syndromes: More Than Just Variants of Joubert Syndrome. *American Journal of Medical Genetics*, 86, 459-469.

The information presented is intended to summarize this condition as it is presently understood by medical professionals.

The statements included in this document are for information only and should not be considered as medical advice. Please always consult your physician for medical advice.