



JOUBERT SYNDROME &
RELATED DISORDERS FOUNDATION

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

Cogan-type Congenital Oculomotor Apraxia (Cogan Syndrome, Type II)

Cogan-type congenital oculomotor apraxia (Cogan-type OMA) refers to a specific eye movement abnormality. In some individuals, this type of OMA can occur as part 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 Cogan-type OMA traditionally exhibit the following features:

- Oculomotor apraxia (OMA), which is a specific eye movement abnormality in which it is difficult for children to track objects smoothly. Their eyes may appear to jump, with jerky eye movements.
- Symptoms are present from birth (congenital)

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

- 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.
- Difficulty coordinating voluntary muscle movements; uncoordinated movements (ataxia).
- Decreased muscle tone (hypotonia).
- Abnormal breathing pattern with episodes of rapid breathing or panting (hyperpnea), which may be followed by pauses in breathing (apnea).
- Rapid, involuntary movements of the eyes (nystagmus).
- Renal insufficiency, particularly increased urination and excessive thirst (nephronophthisis).
- Developmental delays/mental retardation—variable severity.
- 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:

Most children with this disorder are initially diagnosed with oculomotor apraxia. This means that when trying to look at an object, the child must turn his/her head to one side. Their eyes will “lag behind” and then move in the opposite direction. To compensate for this, infants will jerk their head past the object they are looking at in an effort to bring their eyes to a position where they can view the object. As affected children reach school age, this condition can play a role in poor reading skills and clumsiness. This response usually improves throughout the first two decades of life as the child learns to compensate for this condition, although it may not completely resolve.

Some individuals with Cogan-type OMA may also have aplasia (or hypoplasia) of the part of the brain called the cerebellar vermis, which can be detected on a brain MRI. The severity of the resulting ataxia varies from person to person. Some individuals with Cogan-type OMA do not have any cerebellar abnormalities or ataxia.

Children diagnosed with Cogan-type OMA may develop hypotonia. As a result of the poor muscle tone, developmental delays (usually in gross motor, fine motor and speech areas) may occur. Developmental delays, when present, can be treated through physical therapy, occupational therapy, speech therapy, and infant stimulation. Most children are able to achieve standard milestones, although some may do so at a later age.

Some individuals experience difficulties resulting from an inability to appropriately process information received through any of 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.

Renal insufficiency known as juvenile nephronophthisis has developed in some individuals with Cogan-type OMA during childhood. Initial symptoms may include excessive thirst and urination, and kidney failure may result.

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

Management and treatment:

Presently, there is no cure for Cogan-type OMA. Vision therapies may be helpful for some children with OMA. It is recommended that individuals with this disorder see the appropriate specialists necessary to help monitor their various clinical features. Suggested specialists include a nephrologist (kidney specialist), ophthalmologist (eye doctor), geneticist, and neurologist, as well as any others recommended by your doctor.

Screening for some of the complications associated with this disorder, such as renal involvement that may become progressive over time, is recommended on an annual basis. Kidney failure usually develops in childhood or early adulthood, and management may require medications, dialysis, and/or renal transplantation. 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:

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

Genetic cause:

One gene for this disorder has been identified, *NPHP1*, but it is unclear whether this gene is altered only in those individuals who have associated features of nephronophthisis and/or a cerebellar malformation. It is likely that alterations in other genes also can 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 Foundation & Related Cerebellar Disorders.

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
- Ocular Motor Apraxia Home Page (based in the UK): www.wwwweb.org/oma/index.html
- Scottish Sensory Centre: <http://www.ssc.education.ed.ac.uk/resources/vi&multi/eyeconds/OcMoAp.html>
(explains ocular motor apraxia (OMA) in easy-to-understand terms)

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
- National Organization for Rare Disorders, Inc.: #1038 "Apraxia, Ocular Motor, Cogan Type"
- 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.