



JOUBERT SYNDROME & RELATED DISORDERS FOUNDATION

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

Dekaban-Arima Syndrome

(Cerebellar Vermis Hypoplasia with Leber Amaurosis and multicystic kidneys)

Dekaban-Arima syndrome refers to a disorder in which there is a visual deficiency and renal insufficiency. This disorder occurs 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.

Individuals diagnosed with Dekaban-Arima syndrome 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
- Retinal dystrophy, particularly increased pigmentation of the retina or flattened electroretinogram (ERG) traces. Some children may demonstrate repeated eye rubbing, poking, and pressing (symptoms of severe visual impairment, or Leber congenital amaurosis).
- Renal insufficiency, typically polycystic or cystic dysplastic kidneys
- Decreased muscle tone (hypotonia)
- Short stature/poor growth

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

- Difficulty coordinating voluntary muscle movements; uncoordinated movements (ataxia)
- Rapid, involuntary movements of the eyes (nystagmus)
- Malformations of the retina, optic disk, or other regions of the eye (coloboma)
- Facial features may be abnormal in appearance (eyes far set from each other, small ear lobes, broad forehead, arched eyebrows, broad mouth)
- Abnormalities of the liver, including hepatic fibrosis
- 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 Dekaban-Arima have an absence or underdevelopment of part of the brain called the cerebellum vermis which controls balance and coordination. The severity of the resulting ataxia (uncoordinated movements) varies from person to person.

Decreased muscle tone is common in children with Dekaban-Arima. 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. Some children diagnosed with Dekaban-Arima syndrome may be able to achieve some standard milestones, although these are likely to occur at a much later age.

Some individuals may also 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.

Individuals diagnosed with Dekaban-Arima syndrome tend to be more severely affected by their systematic abnormalities (brain, eye, kidneys, etc.) than others diagnosed with similar disorders. Renal impairment can be severe and life-threatening in infancy. As a result, higher death rates may occur for infants and children diagnosed with this disorder.

Management and treatment:

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

Monitoring of the complications associated with Dekaban-Arima, such as visual loss or kidney or liver involvement that may become progressive over time, is recommended on an annual basis or more frequently as needed. Kidney failure can result from cystic disease, and management may require medications, dialysis, and/or renal transplantation. Retinal dystrophy can be severe at birth and cause congenital blindness or can be relatively stable and progress slowly. 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:

Dekaban-Arima 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 responsible for this disorder in their DNA. (In order for a child to be born with this disorder, both the egg and the sperm must contain the same altered gene in question) The odds of having a child born with Dekaban-Arima syndrome to parents who carry the altered gene involved are 1 in 4, or 25%, in each pregnancy that they share.

Genetic cause:

To date, no genes known to be responsible specifically for Dekaban-Arima syndrome have been identified. However, it is possible that mutations in the *CEP290* gene, one of the three genes identified for JSRD thus far, may cause Dekaban-Arima syndrome in some children. However, this gene is not the cause in many individuals with JSRD, and the genetics of these disorders remain complex.

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 and Related Cerebellar Disorders.

Additional resources for families:

- Joubert Syndrome and Related Disorders Foundation: www.jsrdf.org
- National Eye Institute: www.nei.nih.gov
- National Kidney Foundation: www.kidney.org
- American Liver Foundation: www.liverfoundation.org

Resources used in the creation of this document:

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- 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.