JOUBERT SYNDROME FOUNDATION
& RELATED CEREBELLAR DISORDERS

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

WHAT IS JOUBERT SYNDROME?

Joubert Syndrome is a rare genetic condition characterized by a malformed brain stem and absence or underdevelopment of the cerebellar vermis. Joubert Syndrome affects such things as coordination, balance, breathing, eye movement, and muscle tone.

Lauren Gundling

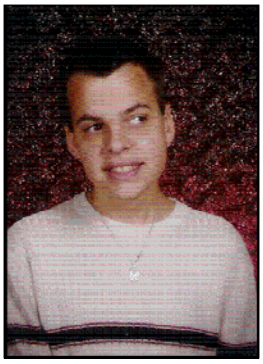
WHAT ARE THE POSSIBLE CHARACTERISTICS OF JOUBERT SYNDROME?

The most common features of Joubert Syndrome include:

- Absence or underdevelopment of part of the brain called the cerebellar vermis, showing the presence of a "molar tooth" on an MRI of the brain.
- A malformed brain stem which may cause abnormal breathing patterns, including episodic hypernea, in which babies pant or breath rapidly, and/or apnea, which is the cessation of breathing.
- Abnormal eye movements, which may include nystagmus and/or ocular motor apraxia.
- Abnormal tongue movements; rhythmic protrusion of the tongue.
- Decreased muscle tone.
- Developmental delay which may include gross motor, fine motor, and speech.
- Poor balance (ataxia).
- Facial features may include high rounded eyebrows, broad nasal bridge, "upturned" nostrils, and triangular mouth.
- Sensory sensitivities, including auditory and tactile.
- Retinal (eye), renal (kidney) and liver problems.
- Seizure activity.
- Behavior Issues

HOW IS JOUBERT SYNDROME INHERITED?

Joubert Syndrome is an autosomal recessive disorder. This means that both parents are carriers of the gene(s). Parents who have a child with JS have a 1 in 4 chance of having another affected child in another pregnancy. Researchers are working to identify the gene(s) involved in Joubert Syndrome. At this time, genetic testing is not currently available to detect Joubert Syndrome; however, prenatal testing with a level 3 ultrasound may be possible.



Timmy Duquette

FOR MORE INFORMATION CONTACT:

E-mail: info@jsfrcd.org
Website: www.jsfrcd.org

MEMBERSHIP APPLICATION

Office use only: FP _____ NLET _____

Last Name _____ Mother _____ Father_____
Other relation to child _____ Last Name _____ First Name _____
Affected Child _____ Date of Birth _____ Date of Death _____
Affected Child _____ Date of Birth _____ Date of Death _____
Street Address _____
City, State, Zip _____ Country/Providence _____
Phone Number_____ E-mail _____ Fax number_____

Yes, I want to be on the mailing list and receive The Rainbow, and receive other JSF & RCD mailings. Enclosed is my annual contribution:

____$35 Family Membership ____$40 International Membership (US currency only)

____I cannot afford the minimum contribution at this time, but would like to continue to be a part of the Foundation. Enclosed is $_____. Count me in as a JSF & RCD member.

____My gift will be matched by my company

If you would like to charge with your VISA or MasterCard, please fill out the information below:
Circle: VISA MasterCard Account Number_____ Expiration Date _____.

Tax Exempt #52-1871536

Tear off and mail to Send your payments to: **Janet Gundling, 8 Ash Road, Jackson, NJ 08527 USA**

Stephanie Duquette

WHAT IS THE TREATMENT FOR JOUBERT SYNDROME?

Children with Joubert Syndrome benefit from infant stimulation, physical therapy, occupational therapy, and speech therapy. Other therapies that have been successful for families include auditory training, sensory integration therapy, horseback riding therapy, and water therapy. Infants with abnormal breathing patterns should have an apnea monitor.

HOW IS JOUBERT SYNDROME RELATED TO OTHER CEREBELLAR DISORDERS?

Problems in the cerebellum of the brain may lead to specific symptoms. There are several known syndromes, like Senor-Loken and Dandy-Walker, which affect the cerebellum and share symptoms with Joubert Syndrome. Recent genetic research has shown that there are several different genes involved in the development of the cerebellum. The exact relationship between these genes and syndromes is not yet known.

WHAT IS THE JOUBERT SYNDROME FOUNDATION & RELATED CEREBELLAR DISORDERS?

The Joubert Syndrome Foundation & Related Cerebellar Disorders is an international network of parents who share knowledge, experience and emotional support. We offer the following services for our members:

- A family information packet with the latest information.
- News updates on breakthroughs and issues related to Joubert Syndrome.
- An active Scientific Advisory Board that reviews research proposals.
- Professional Advisors who help answer questions from our members and professionals.
- A quarterly newsletter called The Rainbow.
- A biennial JSF&RCD conference, bringing together families and experts from around the world.
- A message board, where families can communicate with other families from around the world.
- We play an important role in educating physicians and their support teams and in increasing public awareness