Joubert Syndrome

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Joubert Syndrome Information Page.

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 -- as well as a malformed brain stem (molar tooth sign). The most common features of Joubert syndrome in infants include abnormally rapid breathing (hyperpnea), decreased muscle tone (hypotonia), abnormal eye movements, impaired intellectual development, and the inability to coordinate voluntary muscle movements (ataxia). Physical deformities may be present, such as extra fingers and toes (polydactyly), cleft lip or palate, and tongue abnormalities. Kidney and liver abnormalities can develop, and seizures may also occur. Many cases of Joubert syndrome appear to be sporadic (not inherited). In most other cases, Joubert syndrome is inherited in an autosomal recessive manner (meaning both parents must have a copy of the mutation) via mutation in at least 10 different genes, including *NPHP1*, *AHI1*, and *CEP290*.