Joubert Syndrome

Joubert syndrome is a rare, genetic disorder that affects the area of the brain that controls balance and coordination. The disorder is characterized by absence or underdevelopment of a part of the brain called the cerebellar vermis and a malformed brain stem. The most common features include ataxia (lack of muscle control), an abnormal breathing pattern called hypernea, sleep apnea, abnormal eye and tongue movements, and hypotonia. Other malformations such as extra fingers and toes, cleft lip or palate, tongue abnormalities, and seizures (convulsions) may also occur. There may be mild or moderate retardation.

TREATMENT

Treatment for Joubert syndrome is symptomatic and supportive. Infant stimulation and physical, occupational, and speech therapy may benefit some patients. Infants with abnormal breathing patterns should be monitored.

WHAT WILL HAPPEN

The prognosis (likely outcome) for individuals with Joubert syndrome varies. Some patients have a mild form with minimal motor (movement) disability and good mental development, while others may have severe motor disability and moderate mental retardation.

RESEARCH BEING DONE

The NINDS supports research on the development of the nervous system, especially the cerebellum. The goals of this research are to improve scientific understanding and medical treatment of developmental disorders, and to disover methods of prevention. NINDS, in conjunction with the NIH Office of Rare Diseases, sponsored a satellite symposium on Joubert Syndrome in conjunction with the Child Neurology conference. Research priorities for the disorder were outlined at this meeting.

Research is continually being done to learn more about this problem.

Provided by: The National Institute of Neurological Disorders and Stroke

Document Released: 12/08/2003 Document Re-Released: 03/16/2010

ExitCare® Patient Information ©2011 ExitCare, LLC.