FAMILIAL AGENESIS OF CEREBELLAR VERMIS: A SYNDROME OF EPISODIC HYPERPNEA, ABNORMAL EYE MOVEMENTS, ATAXIA, AND RETARDATION

M Joubert, JJ Einsenring, JP Robb, F Andermann

Neurology 1969;19:813-825

Some two years ago we investigated a child with profound psychomotor retardation who had had an occipital meningoencephalocele removed at birth. Contrast studies were carried out which showed a large midline defect in the posterior fossa and absence of the vermis. At the time we disregarded the nurses' comments about the child's abnormal breathing. A year later, Dr. P. P. Demers referred this patient's baby brother to **us** because he was concerned about his abnormal breathing and retarded development. It was then found that a third and older child in this family was retarded, ataxic, and breathing abnormally. Finally we were able to trace yet another sibling who had died in fancy and who, at autopsy, proved to have agenesis of the vermis. This diagnosis was then confirmed in the two affected living children by contrast studies.

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Comment from Jonathan W. Mink, MD, PhD, FAAN, Associate Editor: This is the original description of what has come to be known eponymously as Joubert syndrome, an autosomal recessive condition characterized by a distinctive cerebellar and brainstem malformation, hypotonia, developmental delay, breathing abnormalities, and atypical eye movements. Several genes have been identified in which mutations have been associated with Joubert syndrome.

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Neurology 2011;76;1837

DOI 10.1212/01.wnl.0000398450.03992.a8

This information is current as of May 23, 2011

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