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Case Report

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Joubert syndrome: magnetic resonance imaging findings

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Abstract

Joubert syndrome is a rare posterior fossa malformation presenting with cerebellar and brainstem malfunction. Differential diagnosis should include other posterior fossa malformations. Typical magnetic resonance imaging (MRI) findings of Joubert syndrome including "molar tooth sign" and "batwing appearance" are discussed which strongly suggest the diagnosis.

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Introduction

Joubert syndrome is characterized by episodes of abnormal respiratory pattern, oculomotor findings, hypotonia, ataxia, developmental retardation with evidence of neuropathologic abnormalities of cerebellum and brainstem [1].

Herein we present a case of this commonly underdiagnosed syndrome in a 2.5 years old girl with magnetic resonance imaging (MRI) findings.

Case Report

A 2.5 year old girl with motor and mental retardation, nystagmus and ataxic movements was referred to our department for a cranial magnetic resonance imaging study. She was the first child of a second degree consanguineous marriage. The parents also informed that she had abnormal eye movements and delayed neurologic development. She was diagnosed in another medical center as having Dandy-Walker variant abnormality. An MRI study revealed vermian agenesis which resulted in median approach of the two cerebellar hemispheres. Isthmus was hypoplastic together with thickening and elongation of the superior cerebellar peduncles giving the "molar tooth" appearance (Figure 1). Also shape of the 4th ventricle had been changed into what described as "bat-wing" appearance (Figure 2). Size of the posterior fossa was normal with cerebrospinal fluid filling the ventricular space. All these MRI findings suggested "Joubert sydrome".

Discussion

Joubert syndrome first described by Marie Joubert and associates in 1969 in four siblings and one sporadic case that exhibited episodic hyperpnea, abnormal eye movements, ataxia and mental retardation with agenesis of cerebellar vermis [1]. In the syndrome, midline structures of the brain-stem have both anatomic and functional defects. Neuropathological studies reveal agenesis of cerebellar vermis, malformations of several brainstem nuclei and dysplasia of structures at the ponto-mesencephalic junction. Extensive brainstem malformation could explain the oculomotor apraxia and hyperpnea; anomalies of the gracile nuclei and solitary tract are thought to contribute to the abnormal respiratory pattern [2]. But the effortless hyperventillation which increases upto 200 breaths/min in the neonatal period usually wanes with age. The eye abnormalities observed in this disease include complete oculomotor apraxia in horizontal and vertical directions and ocular coloboma [3]. Hypotonia and mental retardation are nonvariable features of Joubert syndrome. Long-term follow-up of the children with this disease reveals majority having severe mental and motor developmental

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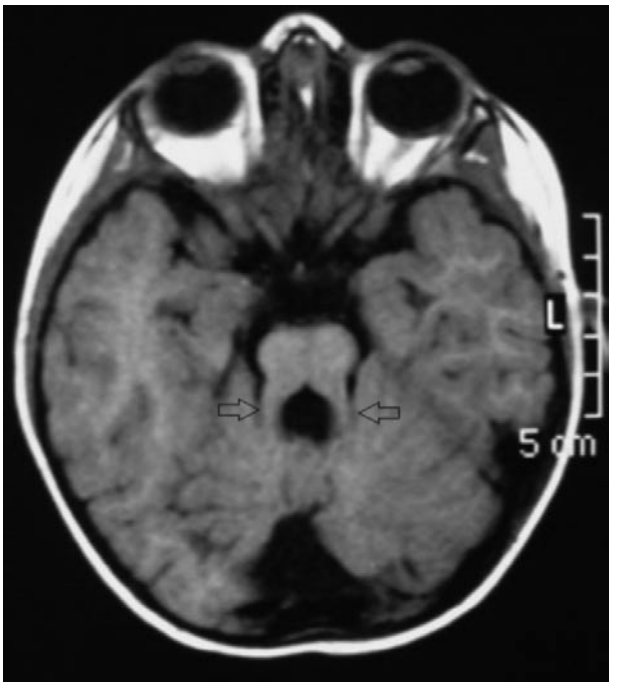

Figure 1. Molar tooth sign, note elongated and thinned superior cerebellar peduncles indicated by arrows.

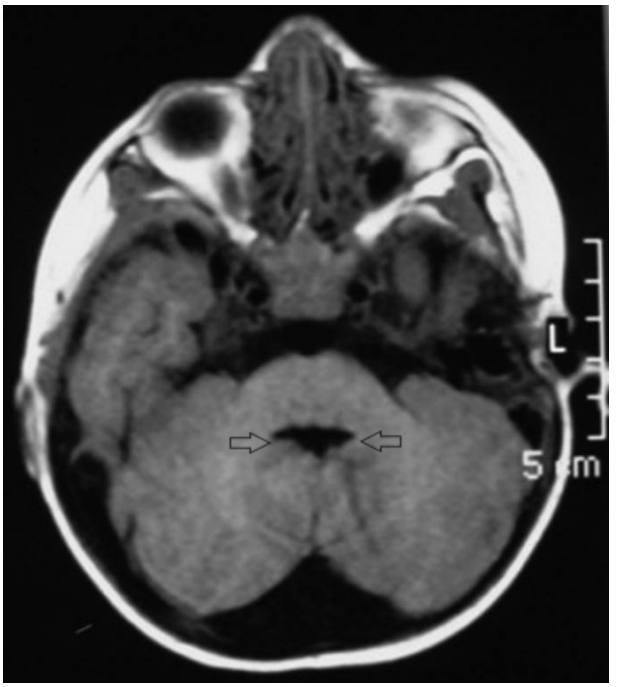

Figure 2. Batwing appearance of the 4th ventricle (arrows).

impairments [4]. Inheritance of this disease is said to be autosomal recessive. Recent studies have shown that it is a genetically heterogenous disorder with one locus pointing to chromosome 9q [5,6].

The striking structural defects of Joubert syndrome in imaging studies are dysgenesis of the isthmus (part of the brainstem between pons and inferior colliculus) which is seen as elongation and thinning of ponto-mesencephalic junction, and deep interpeduncular fossa; thickening of superior cerebellar peduncles; hypoplasia of vermis characterized by incomplete lobulation and enlarged fourth ventricles; incomplete fusion of the halves of the vermis creating a sagittal cleft seen on coronal MRI planes. Combination of the first three features produce the characteristic "molar tooth sign" on axial MRI or CT planes [7].

Other syndromes like Arima, Senior-Loken and COACH which have posterior fossa malformations frequently lead to confusions in the diagnosis of Joubert syndrome. But molar tooth sign is virtually diagnostic of Joubert syndrome. Tectocerebellar dysplasia and Dandy-Walker syndrome, other two situations which may be the source of confusion, also may accompany Joubert syndrome [8].

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