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The Molar Tooth Sign of Brain Mri: A Case Report of Joubert Syndrome

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Abstract | **Case Report**

Joubert syndrome is a rare genetic disorder of autosomal recessive inheritance characterized by congenital malformation of the brainstem and agenesis or hypoplasia of the cerebellar vermis resulting in respiratory disorders, nystagmus, hypotonia, ataxia and delayed motor development. Magnetic resonance imaging allows the diagnosis to be made by demonstrating the "molar tooth sign". We report the observation of an infant aged 1 year 4 months, from a 2nd degree consanguineous marriage, hospitalized for seizures. On examination, she presented with hypotonia. She had abnormal eye movements such as nystagmus. The cerebral magnetic resonance image revealed hypoplasia of the cerebellar vermis and enlargement of the superior cerebellar peduncles with a "molar tooth" aspect in favor of a Joubert syndrome. The evolution was marked by the installation of a delay in psychomotor development.

Keywords: brain MRI, Molar tooth sign, Joubert syndrome.

INTRODUCTION

Joubert syndrome (JS) is a recessive neurodevelopmental disorder defined by a characteristic malformation of the cerebellum and brainstem recognizable on axial Magnetic resonance imaging (MRI) of the brain by the "molar tooth sign [1].

This syndrome is rare with an estimated incidence rate of 1 in 80,000 to 1 in 100,000 live births [2].

It results from a combination of abnormalities: cerebellar vermis aplasia/hypoplasia, thick horizontally oriented upper cerebellar peduncles, and often a deep interpeduncular fossa.

In addition to these central nervous system (CNS) features, subsets of individuals with SJ have ocular (chorioretinal coloboma and progressive retinal dystrophy), renal (nephronophthisis), hepatic (ductal plate malformation spectrum and fibrosis), and/or skeletal (dystrophy and polydactyly) involvement [3].

PATIENT AND OBSERVATION

A male infant aged 1 year and 4 months was admitted to our facility for generalized tonic-clonic seizures, abnormal limb movements, generalized hypotonia, and abnormal head movements with nystagmus. There was also a history of intermittent feeding difficulties and frequent chest infections since the first months of life.

The history revealed that the child was born by vaginal delivery in a hospital to a full-term pregnancy, with a history suggesting birth asphyxia. His parents had a consanguineous marriage. From the first days of his life, his mother noticed feeding difficulties and frequent chest infections. She also noticed abnormal head movements and peculiar eye movements with deviation of the eyes. No similar illnesses were reported in any of the siblings in the family.

Physical examination revealed a hypotonic infant with abnormal head-to-side and limb movements. Head circumference and other anthropometric examinations were normal. No morphologic abnormalities were detected. Cardiovascular and respiratory system examinations were normal. Complete blood count, renal function, and liver function were normal.

Brain MRI Showed

- A hypoplastic cerebellar vermis with enlargement of the superior cerebellar peduncles with the characteristic "molar tooth" appearance.
- Atrophy of the corpus callosum.
- Cortical atrophy above the tentorial cortex, more marked in bilateral frontotemporal.

- Tetra ventricular dilatation with periventricular white matter abnormality in T2 and Flair hypersignal.

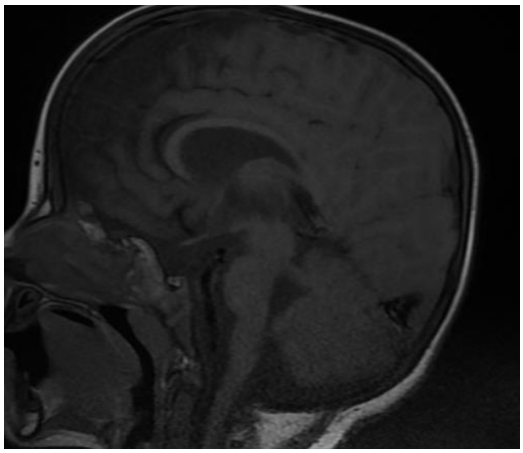

Figure 1: T1 sagittal section sequence; atrophy of the corpus callosum

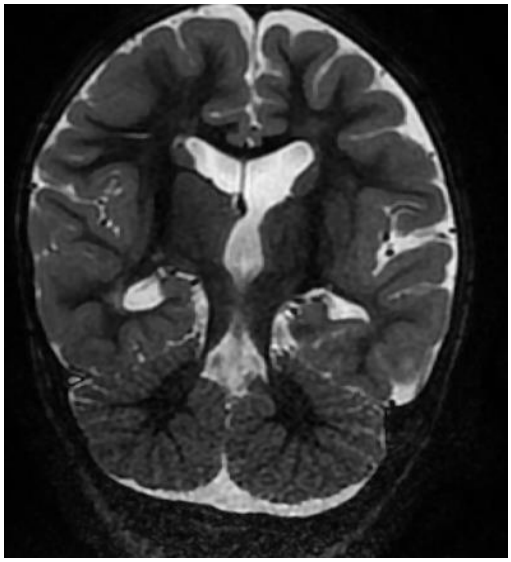

Figure 2: T2 coronal section sequence; cortical atrophy above the tentorial cortex, more marked in bilateral frontotemporal

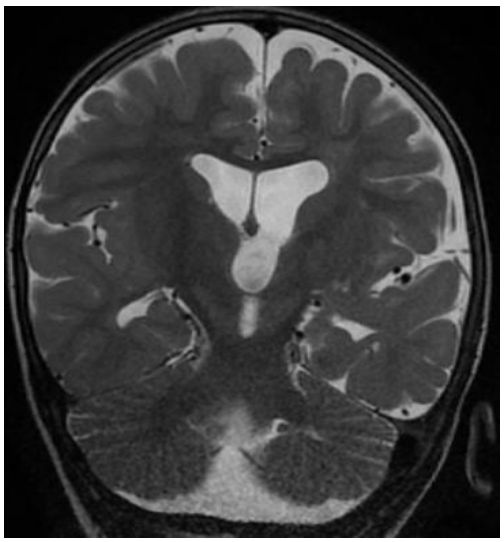

Figure 3: T2 coronal section sequence; hypoplastic cerebellar vermis

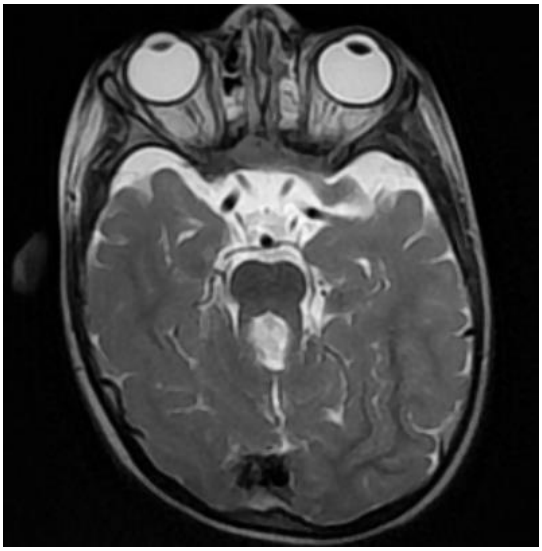

Figure 4

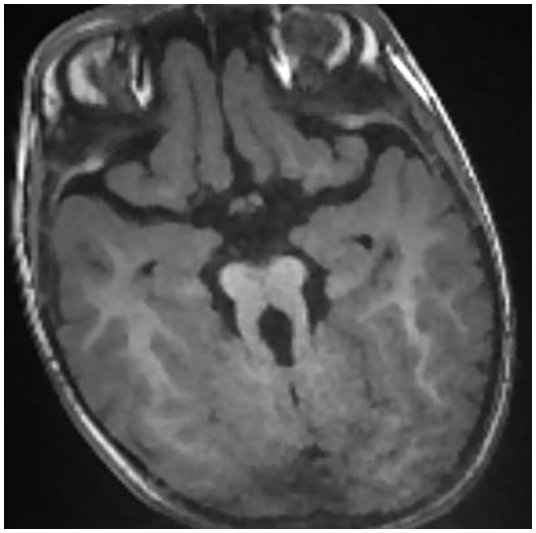

Figure 5

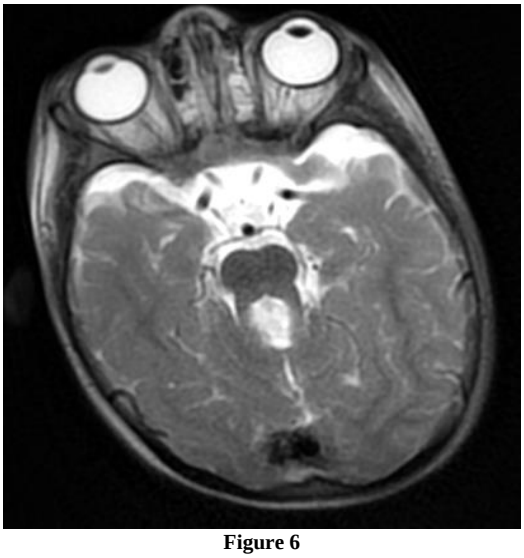

Figure 6

Figure 4, 5 and 6: T2 sequence and BRAVO in axial sections: hypoplastic cerebellar vermis with enlargement of the superior cerebellar peduncles with the characteristic "molar tooth" appearance

DISCUSSION

Joubert syndrome is a rare autosomal recessive disorder characterized by hyperpnea and eye movements, hypotonia, ataxia, developmental delay with neuropathological abnormalities of the cerebellum and brainstem [4].

In 1969, a French neurologist, Marie Joubert, reported a series of five cases of children with mental retardation, episodes of abnormally deep and rapid breathing, abnormal eye movements and ataxia that were associated with agenesis of the cerebellar vermis.

The name Joubert syndrome was given several years later when another group of patients with similar findings were identified. Since then, about 200 cases of this have been reported, revealing a variable phenotype [4].

Joubert syndrome is an autosomal recessive disorder with cerebellar vermilion hypoplasia with a median cleft.

The underlying abnormality of this syndrome may be an inability of posterior fossa fiber tracts to cross the midline.

Although the cause is unknown, vermian hypoplasia and pontomesencephalic junction abnormalities are the hallmarks that lead to the diagnosis of Joubert syndrome [5, 6].

The clinical presentation of the syndrome includes hypotonia, ataxia, developmental delay and cognitive impairment. Respiratory abnormalities usually present during the neonatal period with hyperpnea and intermittent apnea. Ocular abnormalities such as nystagmus, strabismus and oculomotor apraxia may also be observed. Autistic features have also been reported in Joubert syndrome [7].

There may be hydrocephalus, cystic hypertrophy of the posterior fossa, hypothalamic hamartoma and absence of pituitary gland [8].

The main imaging findings, present almost uniformly, are partial or complete absence of the cerebellar vermis, hypoplastic cerebellar peduncles, and deformation of the fourth ventricle. The cerebellar hemispheres are usually normal. The brain is unaffected, although moderate lateral ventricular hypertrophy due to atrophy has been described in 6% to 20% of cases, and dysgenesis of the corpus callosum was present in 6% to 10% of cases [9].

The absence of the vermis leads to a medial cleft between the cerebellar hemispheres. The combination of hypoplasia of the cerebellar peduncles leading to a molar tooth sign and severe hypoplasia of the vermis resembling the bat-wing appearance of the fourth ventricle on MRI are highly suggestive of JS [10].

Genetic counseling is important in a family with JS. Screening for a known genetic mutation can detect less than 50% of cases. In addition, the diagnosis of JS can be made before birth by looking for imaging findings on prenatal ultrasound. Prenatal magnetic resonance can also capture JS with caution [11, 12].

CONCLUSION

Joubert syndrome is a very rare autosomal recessive disorder. It is characterized by agenesis of the cerebellar vermis, abnormal eye movements with nystagmus, episodes of hyperpnea and apnea, generalized motor developmental delay, multi-cystic kidney disease. The importance of recognizing JS is related to the outcome and its potential complications. The appearance of the molar tooth is characteristic. Prenatal diagnosis by ultrasound and antenatal magnetic resonance imaging (MRI) is also possible.

CONFLICTS OF INTEREST

The authors declare no conflict(s) of interest.

AUTHOR CONTRIBUTIONS

All authors contributed to the conduct of this research work. The authors have read and approved the final version of the manuscript.

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