

# Molar Tooth Sign in Joubert Syndrome: A Case Report

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**Abstract:** *The molar tooth sign refers to the appearance of the midbrain in axial section in which the elongated superior cerebellar peduncles give the midbrain an appearance reminiscent of a molar or wisdom tooth. Molar tooth sign is caused by lack of normal decussation of superior cerebellar peduncular fiber tracts which in turn leads to enlargement of the peduncles, which also follow a more horizontal course as they extend perpendicularly to the brainstem between the midbrain and the cerebellum. The absence of crossing fibers causes decreased anteroposterior diameter of midbrain and also causes interpeduncular cistern to be deeper than in the normal brain. It is seen in patients with joubert syndrome.*

**Keywords:** Jobert, Magnetic resonance imaging

## 1. Introduction

Joubert syndrome is first described by Joubert in 1969. It is a rare autosomal recessive disorder presenting with congenital hypotonia evolving into ataxia, developmental delay, and oculomotor apraxia or abnormalities of respiratory pattern or both. Molar tooth sign is a characteristic MR appearance of brainstem which results from cerebellar vermis hypoplasia and thick, elongated superior cerebellar peduncles and abnormally deep interpeduncular fossa. The presence of molar tooth sign and appropriate clinical features is almost diagnostic of Joubert syndrome. Joubert syndrome is a ciliopathy, characterised by defective primary ciliary function.

## 2. Case Report

A 3 year old baby boy born to a consanguineous married couple with negative family history brought by the parents with history of abnormal eye movement, inability to follow moving objects, 2 episodes of seizures and developmental delay.

By reviewing the medical history the child was a product of full term normal vaginal delivery cried immediately after birth but child had a poor cry and sluggish limb movement were present and the child did not attain developmental milestones for the present age (standing without support, walking, holding objects). The child was mentally retarded regarding family history the patient had 1 sister who is free from any illness and no similar presentations in the family.

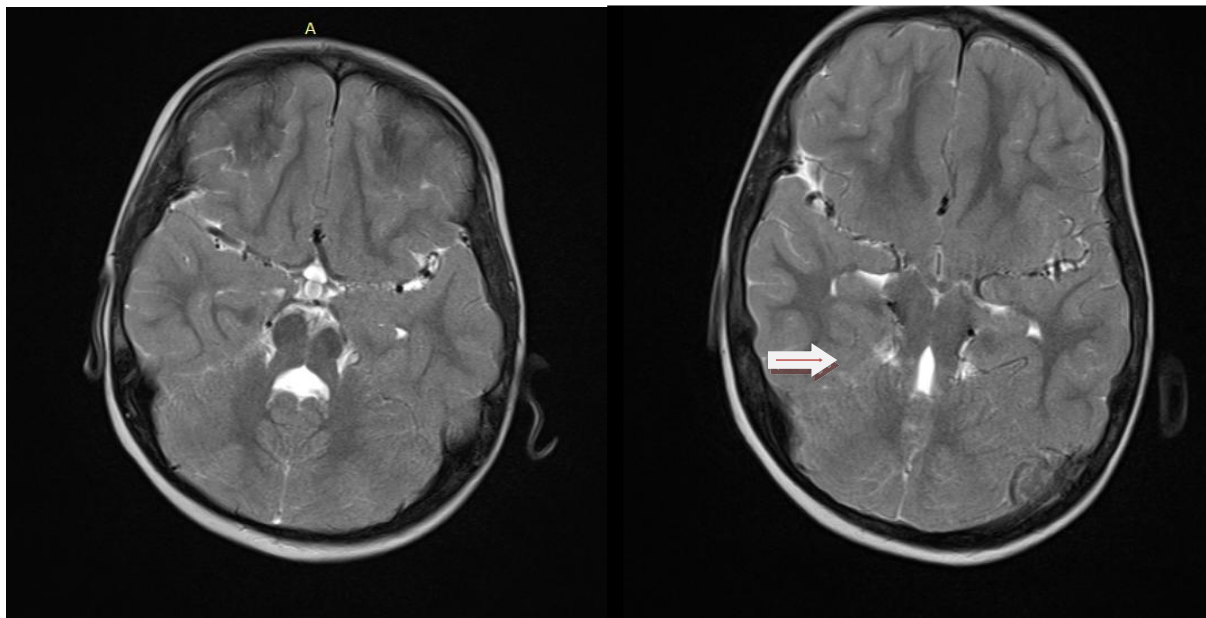
On examination, he was awake, alert, child had abnormal eye movements throughout the examination. Child did not have any neurocutaneous markers. He showed prominent forehead, deep-set eyes and low frontal hairline. There was no organomegaly. Cardiac and respiratory systems are

normal on auscultation. Neurological examination revealed normal cranial nerves and fundus. Motor examination revealed hypotonia with normal tendon reflexes.

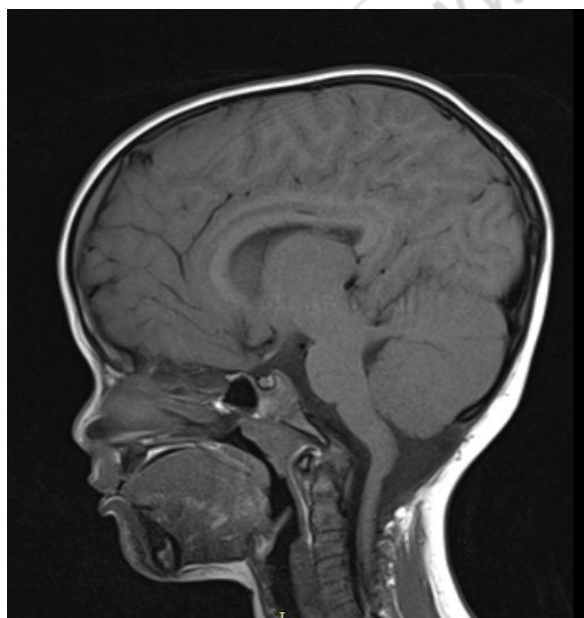
MRI - The axial T1-weighted and T2-weighted Magnetic resonance (MR) images showed bat shaped 4<sup>th</sup> ventricle and cleft communicating with 4<sup>th</sup> ventricle (FIG1), deep interpeduncular fossa and thickened superior cerebellar peduncles giving appearance of a molar tooth configuration (FIG 2). The more caudal T2- and T1-weighted MR images showed vermian hypoplasia, no evidence of any posterior fossa cyst (FIG3).



**Figure 1:** Axial T2WI Images showing cleft communicating with 4<sup>th</sup> ventricle and bat shaped 4<sup>th</sup> ventricle.



**Figure 3:** Axial T2WI showing deep interpeduncular fossa and thickened superior cerebellar peduncles giving appearance of a molar tooth configuration.



**Figure 4:** Sagittal T1WI showing vermian hypoplasia.

### 3. Discussion

Joubert's syndrome is a rare autosomal recessive disorder presenting with the congenital hypotonia evolving into ataxia, developmental delay, abnormalities of respiratory pattern and ocular apraxia. Both sexes are affected almost equally with onset early in the infancy. Most patients die in infancy or early childhood.

MRI is the main imaging tool. It has a characteristic MRI appearance of cerebellar vermian hypoplasia and brain stem abnormality giving appearance of molar tooth sign, which consists of thickened superior cerebellar peduncles, deep interpeduncular fossa. This appearance is due to lack of normal decussation of superior cerebellar peduncular fiber tracts which in turn leads to enlargement of the peduncles, which also follow a more horizontal course as they extend

perpendicularly to the brainstem between the midbrain and the cerebellum.

The absence of crossing fibers is responsible for the decreased anteroposterior diameter of midbrain and also causes interpeduncular cistern to be deeper than in the normal brain. Absence of vermis results in a midline cleft between the two cerebellar hemispheres resulting in a batwing appearance of the 4th ventricle on axial sections.

It is associated with nephronophthisis, retinal dystrophy, ocular colobomas, liver fibrosis and polydactyly.

Joubert syndrome is classified into two groups on the basis of the presence or absence of retinal dystrophy. In the group with retinal dystrophy, there is a high prevalence of multicystic renal disease, and they have the worst prognosis than in the group without retinal dystrophy.

Although Joubert syndrome is rare, it should be considered in the differential diagnosis of hypotonic cerebral palsy. The wrong or delayed diagnosis of Joubert syndrome may lead to delayed diagnosis of breathing difficulties and renal anomalies.

Supratentorial anomalies associated with Joubert's syndrome are uncommon, but may be associated with gray matter heterotopias and cerebral cortical dysplasia.

Cerebellar vermian anomalies have been associated with Dandy-Walker syndrome and rhombencephalosynapsis. In Dandy-Walker syndrome, the vermis is hypoplastic in its inferior aspect, the fourth ventricle is enlarged and communicates with a cyst in the posterior fossa, but the superior cerebellar peduncle and interpeduncular fossa are normal. Cerebellar hemispheres are fused in rhombencephalosynapsis.

Joubert syndrome has a variable outcome. Steinlin et al. suggested that outcomes in JS can be divided into three courses: first, children die young age; second, patients who

survive with delayed developmental milestones and have a variety of visual and motor handicaps; and third, patients whose developmental quotients fall within the mildly delayed range .

The prognosis is poor with 5 year survival rate of only 50 % .Genetic counselling is required for the parents because of the 25 % recurrence rate , screening and prenatal counselling is required. Prenatal diagnosis in high risk pregnancies is possible with serial ultrasound and fetal MRI at 20-22 weeks. Because of the respiratory involvement associated with these patients anaesthetic agents, like opiates and nitrous oxide should be avoided in these patients.

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