

## IMAGING HIGHLIGHTS

# Molar tooth sign - Characteristic of Joubert's syndrome

Saumya H Mittal MD DNB, Rakshith Kedambadi MD DM, Misri Zulkifli MD DM, Shivanand Pai MD DM, Nisha Shenoy MD DNB, Gundabolu Gautam MD DNB

Department of Neurology, KMC Hospital, Mangalore, Karnatka, India

A 9 year old female child presented to the hospital with history of seizures. She was stable at presentation. The child, born of first degree consanguineous marriage was found to have mental retardation, abnormal eye movements, low set ears, truncal ataxia and hypotonia. Investigations revealed renal failure with creatinine levels of 4.1 mg/d. Her MRI showed absence of superior vermis, dilated fourth ventricle, bilaterally elongated cerebellar peduncles resulting in a characteristic molar tooth appearance (Figure 1). A diagnosis of Joubert syndrome was thus considered.

A French neurologist, Marie Joubert, first described this rare autosomal recessive disorder in 1969 with a locus on chromosome 9q. The disease is clinically characterized by ataxia, hypotonia, developmental delay, oculomotor and respiratory abnormalities secondary to cerebellar vermian and midbrain dysgenesis. Other features include microcephaly, polydactyly, low set ears, retinal dysplasia, renal and hepatic abnormalities, and soft tissue tumors.<sup>1,2</sup>

The disease may be classified as “pure Joubert syndrome”, “Joubert syndrome with ocular defect” (JS-O), “Joubert syndrome with renal defect” (JS-R), “Joubert syndrome with oculorenal defects” (JS-OR), “Joubert syndrome with hepatic defect” (JS-H), and “Joubert syndrome with orofacio-digital defects” (JS-OFD).<sup>2</sup>

The midbrain dysgenesis that results in the molar tooth sign on axial MRI has three components: deep interpenduncular fossa, elongated superior cerebellar peduncles and absence of vermis. This molar tooth sign is characteristic of Joubert syndrome and related disorders, and differentiates from other hindbrain malformation.<sup>1,2</sup>

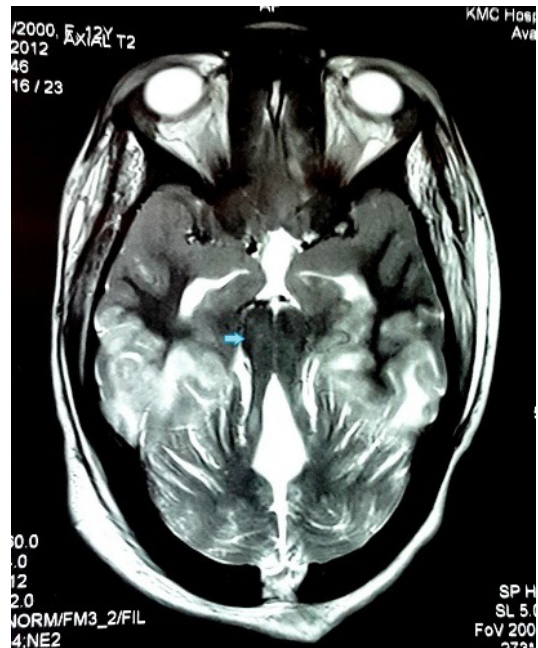


Figure 1. Patient's MRI showing absence of superior vermis, dilated fourth ventricle, bilaterally elongated cerebellar peduncles resulting in a characteristic molar tooth appearance (arrow).

## DISCLOSURE

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Conflict of interest

## REFERENCES

1. Byju N, Jose J, Saifudheen K, Musthafa M. Molar tooth sign with ataxia and see-saw nystagmus (Joubert syndrome). *Ann Indian Acad Neurol* 2011; 14(1): 62-3
2. Nagpal T, Pande S. Molar tooth sign: Neuroimaging characteristic of Joubert syndrome. *Neurol India* 2008; 56( 2): 223
3. Brancati F, Dallapiccola B, Valente EM. Joubert syndrome and related disorders. *Orphanet J Rare Dis* 2010; 5:20