

# Molar tooth sign: Neuroimaging characteristic of Joubert syndrome

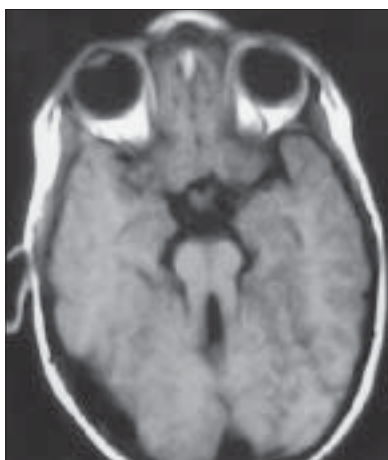
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A two-year-old boy, born of non-consanguineous marriage was brought with delayed motor and language milestones. The parents had also noted abnormal eye movements and periodic breathing difficulty since early infancy. The child was third of three sibs and the other two sibs were normal. On examination, the child was hypotonic and mentally retarded and had severe truncal ataxia and oculomotor apraxia. Axial magnetic resonance imaging (MRI) Brain showed split, segmented vermis and elongated fourth ventricle giving the midbrain an appearance of 'molar tooth' [Figure 1]. The child was diagnosed to suffer from Joubert syndrome and the prognosis explained.

Joubert syndrome is a rare autosomal recessive

disorder with a locus on Chromosome 9q characterized by ataxia, developmental delay and oculomotor and respiratory abnormalities in relation to cerebellar vermal and midbrain dysgenesis.<sup>[1]</sup> Joubert syndrome is often missed clinically and radiologically if not enough attention is paid to its subtle and variable clinical presentation and the imaging findings in the posterior fossa. A variety of abnormalities have been described in children with Joubert syndrome including delayed language, hypersensitivity to noise, autism, meningoencephaloceles, microcephaly, low-set ears, polydactyly, retinal dysplasia, kidney abnormalities (renal cysts), soft tissue tumor of the tongue, liver disease and duodenal atresia. Even within siblings the phenotype may vary, making it difficult to establish the exact clinical diagnostic boundaries of Joubert syndrome. The midbrain dysgenesis is responsible for the molar tooth sign on axial MRI which has also been documented in fetal brain MRI.<sup>[2]</sup>



**Figure 1:** Axial T1-weighted image MRI brain shows split, segmented vermis and elongated fourth ventricle. Midbrain appears as a molar tooth

## References

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