

## Clinical Image

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# Molar tooth sign: Joubert's syndrome

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### Introduction

A 5-year-old female child from a well-monitored, full-term pregnancy with no specific history of ataxia, hypotonia and global developmental delay.

### Discussion

Joubert syndrome is an autosomal recessive malformation affecting the cerebellum and brainstem, with a pre- or neonatal set and with out clear gender predominance. It is a molar malformation of the midbrain and hindbrain.

The clinical picture is polymorphic; during the first months of life infants may present hypotonia, respiratory rhythm disorders, abnormal eye movements, or facial dysmorphism and in early childhood may appear ataxia, delayed motor development, intellectual deficit. Other manifestations are rare: retinal dystrophy, nephronophthisia, polydactyly [1].

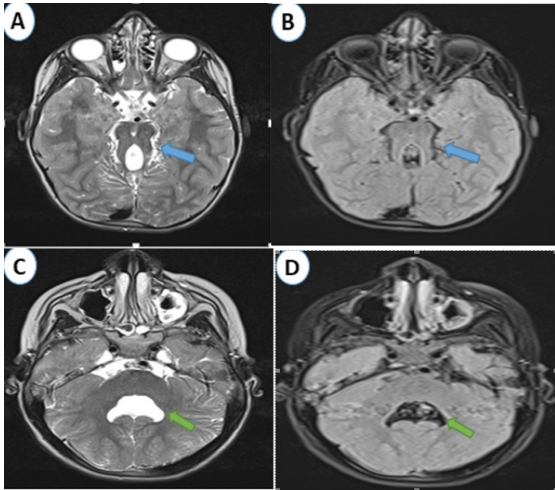
On MRI, the characteristic radiological sign is the molar sign [2] due to hypoplasia of the cerebellar vermis and malformations of the midbrain with a deep inter-peduncular fossa and thick and elongated upper cerebellar peduncles, as well as the bat-wing appearance of V4 (Figure 1).

Differential diagnoses arise when there is an abnormality of the vermis and V4. Joubert syndrome can be confused with Dandy walker malformation and Rhombencephalosynapsis, but the molar tooth appearance of the midbrain is pathognomonic of Joubert syndrome.

No known treatment has been developed for this disorder, although some organizations are currently studying the disorder, its prevention and treatment.

### References

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**Figure 1:** Brain MRI in slices: axial T2 (A,C) and T2 Flair (B,D): Deepening of the inter-peduncular fossa with elongation of the superior cerebellar peduncles associated with hypoplasia of the cerebellar vermis =>appearance of the molar tooth of the midbrain (blue arrow). Enlargement of the 4th ventricle producing a bat wing aspect (green arrow).