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Beyond the Molar Tooth Sign: Joubert Syndrome Associated with Complete Agenesis of the Corpus Callosum in an Epileptic Child

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ABSTRACT

Joubert syndrome is a rare neurodevelopmental disorder characterized by cerebellar vermian hypoplasia and the pathognomonic molar tooth sign on magnetic resonance imaging (MRI). Although several supratentorial abnormalities have been described, complete agenesis of the corpus callosum remains an uncommon association. We report the case of an 8-year-old child with epilepsy and macrocrania whose brain MRI demonstrated classic features of Joubert syndrome, including vermian hypoplasia, horizontalized superior cerebellar peduncles, and the molar tooth sign, associated with complete agenesis of the corpus callosum and colpocephaly. This case highlights the broad neuroradiological spectrum of Joubert syndrome and the importance of comprehensive MRI evaluation for identifying associated cerebral malformations.

KEYWORDS :

Joubert syndrome, Corpus callosum agenesis, Molar tooth sign, Pediatric neuroradiology, Epilepsy.

MAIN ARTICLE

INTRODUCTION

Joubert syndrome is a rare autosomal recessive ciliopathy characterized by developmental delay, hypotonia, abnormal eye movements, respiratory dysregulation, and a distinctive hindbrain malformation visible on MRI [1,2]. The hallmark radiological feature is the molar tooth sign, resulting from vermian hypoplasia and abnormal configuration of the superior cerebellar peduncles [1].

Although infratentorial abnormalities predominate, associated supratentorial malformations have increasingly been recognized, including cortical malformations, ventriculomegaly, and abnormalities of the corpus callosum [2,3]. Complete agenesis of the corpus callosum remains an unusual association and may contribute to a more complex neurological phenotype.

CLINICAL PRESENTATION

An 8-year-old child with a history of epilepsy treated with antiepileptic medication was referred for brain MRI because of macrocrania and neurodevelopmental abnormalities.

Neurological examination revealed delayed psychomotor development without focal neurological deficit.

MRI was performed to investigate a possible structural cerebral abnormality.

MRI Findings

MRI demonstrated:

- Complete agenesis of the corpus callosum with absence of its normal anatomical components.
- Parallel and widely separated lateral ventricles producing the classic "racing car" appearance on axial images.
- Lateral eversion of the frontal horns resulting in the characteristic "bull-horn" configuration on coronal images.
- Dilatation of the occipital horns of the lateral ventricles consistent with colpocephaly.

- Vermian hypoplasia.
- Horizontalization and thickening of the superior cerebellar peduncles.
- Deepened interpeduncular fossa resulting in the classic molar tooth sign.

These findings were highly suggestive of Joubert syndrome associated with complete agenesis of the corpus callosum.

DISCUSSION

Joubert syndrome belongs to a group of inherited disorders known as ciliopathies and is characterized by abnormal development of the cerebellar vermis and brainstem [1,4].

The hallmark imaging feature, the molar tooth sign, results from a combination of:

- Vermian hypoplasia or aplasia
- Thickened and elongated superior cerebellar peduncles
- Deep interpeduncular fossa

These findings are considered essential for diagnosis [1,2].

Additional supratentorial abnormalities are increasingly recognized and may include:

- Corpus callosum dysgenesis or agenesis
- Cortical malformations
- Ventriculomegaly
- Hippocampal abnormalities

Complete agenesis of the corpus callosum is uncommon and may explain the associated colpocephaly and characteristic ventricular morphology observed in our patient [3,5].

The association of Joubert syndrome with complete corpus callosum agenesis expands the phenotypic spectrum of the disease and illustrates the importance of carefully evaluating both supra- and infratentorial structures on MRI.

CONCLUSION

This case illustrates a rare association between Joubert syndrome and complete agenesis of the corpus callosum in an epileptic child with macrocrania. MRI demonstrated both the classic molar tooth sign and characteristic features of callosal agenesis, including colpocephaly and abnormal ventricular configuration. Recognition of these combined imaging findings is essential for accurate diagnosis, prognostic assessment, and genetic counseling.

FIGURES:

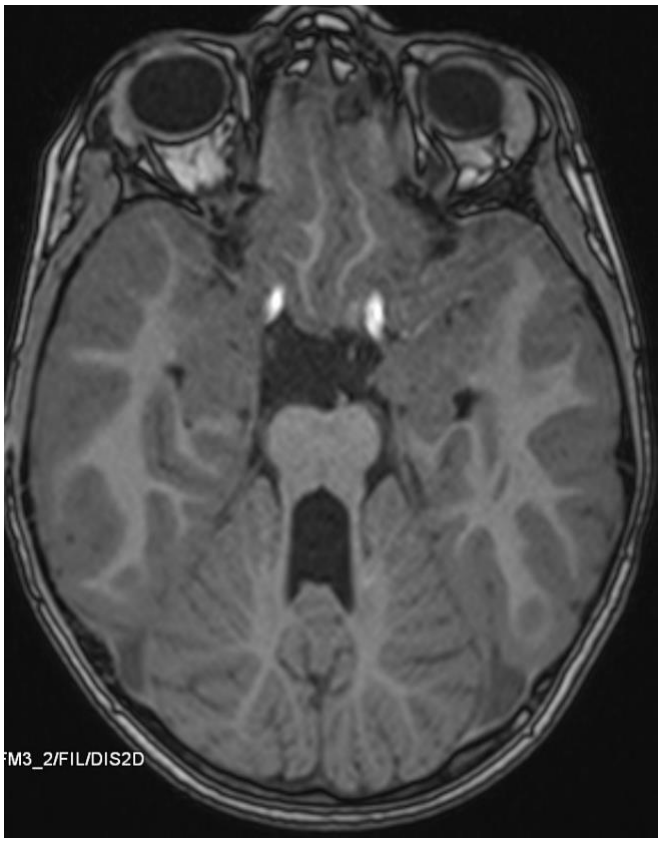

Figure 1: Axial T1-weighted MRI demonstrating the molar tooth sign with thickened and horizontally oriented superior cerebellar peduncles.

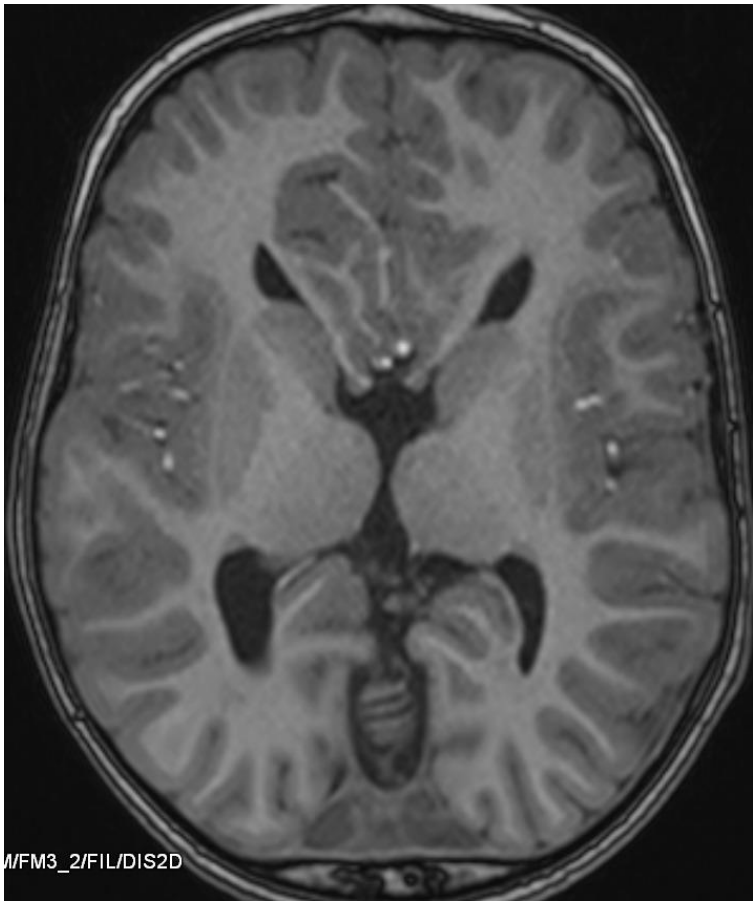

Figure 2: Axial Flair-weighted image illustrating the parallel orientation of the lateral ventricles, producing the classic "racing car" appearance.

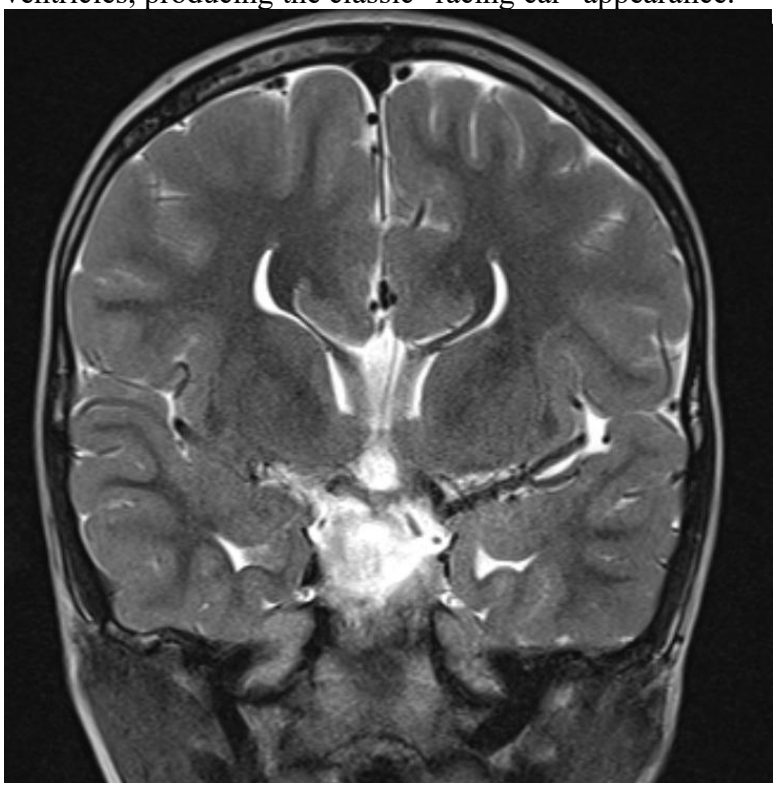

Figure 3: Coronal image demonstrating lateral eversion of the frontal horns ("bull-horn" appearance) associated with complete corpus callosum agenesis.

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