

Imaging findings of Joubert syndrome: A Case Report

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Citation: Bennani H, Belkouchi L, Hassar S, et al. Imaging findings of Joubert syndrome: A Case Report. *Medi Clin Case Rep J* 2026;4(4):2020-2023. DOI: doi.org/10.51219/MCCRJ/Houda-Bennani/532

Received: 29 September, 2026; **Accepted:** 02 October, 2026; **Published:** 05 October, 2026

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ABSTRACT

We report the case of an 8-year-old boy, born to consanguineous parents, who was referred after a first episode of bilateral ocular deviation followed by involuntary shaking movements of the limbs that resolved spontaneously. His history included neonatal hypotonia and mild global developmental delay and neurological examination revealed ataxia with impaired motor coordination. The electroencephalogram recorded after the episode was normal and no antiseizure medication was started. Brain magnetic resonance imaging at 1.5 T demonstrated hypoplasia of the cerebellar vermis with a midline cleft, together with thickened, elongated superior cerebellar peduncles running horizontally alongside a deep interpeduncular fossa, forming the molar tooth sign. The fourth ventricle showed a “bat-wing” configuration, while no posterior fossa cyst or supratentorial anomaly was identified. The diagnosis of Joubert syndrome was established on these findings. This case illustrates a mildly expressed form of the disease that remained unrecognized until late childhood and was ultimately revealed by a first seizure. It shows that the molar tooth sign allows the radiologist to reach the diagnosis in the appropriate clinical context and that recognizing it should prompt systematic renal, hepatic and retinal screening, molecular testing where available and genetic counselling, which matters particularly in consanguineous families.

Keywords: Joubert syndrome; Molar tooth sign; Cerebellar vermis hypoplasia; Ciliopathy; Consanguinity; Magnetic resonance imaging

Introduction

Joubert syndrome is a primary ciliopathy characterized by a pathognomonic cerebellar and brainstem malformation, the molar tooth sign (MTS)¹. Its universal features are infantile hypotonia progressing to ataxia and developmental delay or intellectual disability; oculomotor apraxia and abnormal respiratory patterns are frequent^{1,2}. More variable features include renal cystic disease (typically nephronophthisis), retinal

dystrophy, congenital hepatic fibrosis and skeletal anomalies such as polydactyly².

JS shows marked genetic heterogeneity: more than 40 causative genes have been identified, together explaining up to 94% of cases^{2,3}. The first population-based study estimated its prevalence at 0.47 per 100,000 overall and 1.7 per 100,000 in individuals aged 0-19 years. The mean age at diagnosis in that cohort was 6.7 years, reflecting frequent diagnostic delay⁴.

We report the case of an 8-year-old boy with mild global developmental delay, born to consanguineous parents, in whom a first seizure led to the MRI diagnosis of JS.

Case Presentation

An 8-year-old boy was referred after a first episode of bilateral ocular deviation followed by involuntary shaking movements of the extremities. The episode resolved spontaneously.

He was born to consanguineous parents. His history included neonatal hypotonia and mild global developmental delay. There was no history of abnormal breathing pattern or previous seizures. His immunizations were up to date.

Neurological examination revealed ataxia with impaired motor coordination. An electroencephalogram (EEG) recorded after the episode was normal. Because this was a first, self-limited seizure with a normal EEG, no antiseizure medication was started.

Brain MRI was performed at 1.5 T, including axial T1-weighted, axial and coronal T2-weighted, axial T2-FLAIR, diffusion-weighted and 3D contrast-enhanced T1-weighted sequences. Axial T1- and T2-weighted images showed thickened, elongated superior cerebellar peduncles with a deepened interpeduncular fossa at the pontomesencephalic junction, producing the molar tooth sign (**Figure 1**). The sagittal contrast-enhanced T1-weighted image demonstrated a near-perpendicular reorientation of the superior cerebellar peduncles relative to the brainstem (**Figure 2A**), while the coronal T2-weighted image showed thickening of these peduncles together with a midline cleft (**Figure 2B**). Hypoplasia of the cerebellar vermis was evident on both coronal T2-weighted and sagittal contrast-enhanced T1-weighted images (**Figure 3**) and the fourth ventricle displayed a “bat-wing” configuration on axial FLAIR images (**Figure 4**). This configuration results from hypoplasia of the cerebellar vermis, which leaves the cerebellar hemispheres apposed in the midline and widens the superior part of the ventricle. There was no posterior fossa cyst, no signal abnormality, no restricted diffusion and no abnormal enhancement. Supratentorial structures were unremarkable, with a complete, normal-appearing corpus callosum, a normal cortex and no grey-matter heterotopia.

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The diagnosis of Joubert syndrome was based on the clinical and neuroimaging findings. The parents were counselled about the nature of the disease, its autosomal recessive inheritance and the need for regular multidisciplinary follow-up.

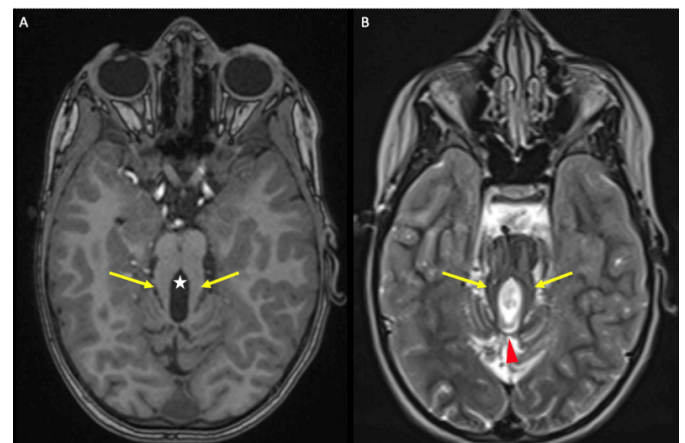


Figure 1: Molar tooth sign.

Axial T1-weighted (A) and axial T2-weighted (B) images at the pontomesencephalic junction demonstrating thickened, elongated superior cerebellar peduncles (yellow arrows) deep interpeduncular fossa (white asterisk) and midline cleft (red arrowhead).

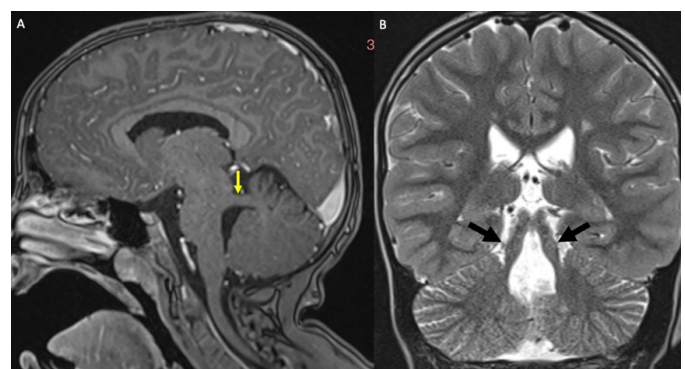


Figure 2: Superior cerebellar peduncle abnormalities in Joubert syndrome.

(A) Sagittal contrast-enhanced T1-weighted image showing near-perpendicular reorientation of the superior cerebellar peduncles relative to the brainstem (yellow arrow). (B) Coronal T2-weighted image showing thickened superior cerebellar peduncles (black arrows).

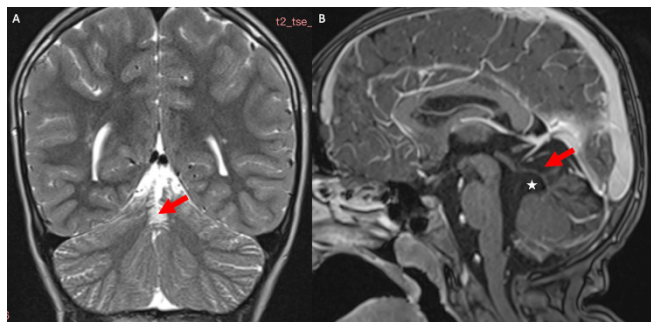


Figure 3: Vermis and fourth ventricle abnormalities in Joubert Syndrome.

Vermian hypoplasia on coronal T2-weighted (A) and sagittal contrast-enhanced T1-weighted (B) images (red arrow) associated with fourth ventricle deformation (asterisk).

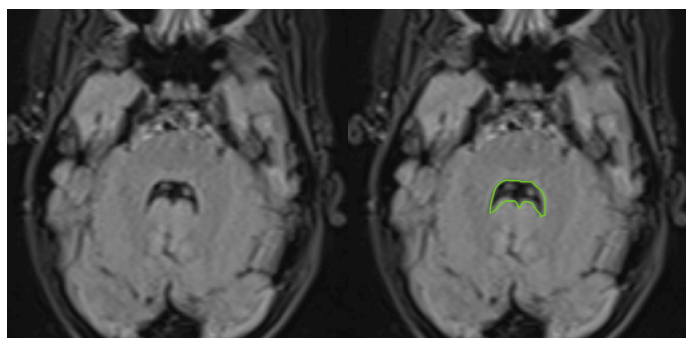


Figure 4: Bat-wing configuration of the fourth ventricle.

Axial FLAIR image at the level of the pons showing the enlarged, deformed fourth ventricle with the contour of the fourth ventricle outlined in green, highlighting its resemblance to a bat with outspread wings, known as the bat-wing sign.

Discussion

Imaging features

The MTS is the neuroradiological hallmark of JS. It reflects the lack of decussation of the superior cerebellar peduncle fibres, which become thickened, elongated and horizontally oriented, while a reduced anteroposterior diameter of the midbrain deepens the interpeduncular fossa⁵. Vermian hypoplasia or dysplasia produces the other typical features: a midline cleft between apposed cerebellar hemispheres, a «bat-wing» upper fourth ventricle and a triangular mid-fourth ventricle. All of these were present in each of the 12 children in the series by Liao and Zheng⁵. In a cohort of 36 children, 31 showed a typical MTS⁶.

MRI is superior to CT, on which posterior fossa artefacts can hide the midline cleft; multiplanar acquisitions also depict the peduncles and the vermis directly⁵. In our patient, the axial MTS, the abnormal course of the peduncles on sagittal and coronal images, the midline cleft, vermian hypoplasia and the bat-wing fourth ventricle made the diagnosis unequivocal and a 1.5 T examination was sufficient.

The main differential diagnoses are Dandy-Walker malformation (cystic dilatation of the fourth ventricle, enlarged posterior fossa, normal brainstem, no MTS) and rhombencephalosynapsis (fusion of the cerebellar hemispheres without a midline cleft)⁵. The absence of a posterior fossa cyst and the presence of a midline cleft in our patient argued against both.

Supratentorial anomalies may be associated with JS, including corpus callosum dysgenesis, grey-matter heterotopia, cortical dysplasia, polymicrogyria, encephalocele and ventriculomegaly⁵⁻⁷. Their absence in our patient is relevant, as it makes an underlying malformative epileptogenic lesion less likely.

Clinical Presentation

Our patient's course, with neonatal hypotonia followed by ataxia and mild global developmental delay, matches the typical natural history of JS^{1,2}. He also largely fulfilled the historical clinical criteria recalled by Liao and Zheng, which combine vermian hypoplasia, infantile hypotonia and cognitive impairment with breathing or oculomotor abnormalities⁵. This supports the diagnosis despite the absence of molecular confirmation.

Developmental delay is almost constant in JS, affecting 94% of children in one cohort, but its severity varies widely⁶. Purely neurological forms, without other organ involvement, have also been reported⁸. The mild degree of delay in our patient, together with the absence of respiratory abnormalities, probably explains why the diagnosis was made only at 8 years of age, consistent with the mean age at diagnosis reported in the Italian population-based cohort⁴.

Oculomotor involvement is very common in JS: in one series, oculomotor apraxia affected 75% of children and retinal dysfunction was sometimes present despite a normal fundus⁹. In our patient, the ocular deviation occurred during the paroxysmal episode and more likely corresponds to an ictal gaze deviation. Without an ophthalmological examination, however, underlying oculomotor apraxia or retinal involvement cannot be excluded.

Seizures are a less common but documented feature of JS. In the 36-child cohort, 3 children had seizures and an abnormal EEG was uncommon among those without epilepsy⁶. Presentations with seizures as early as the neonatal period have also been described¹⁰. The spontaneous resolution of the episode and the normal EEG in our patient justified not starting antiseizure medication, but clinical monitoring for recurrence remains necessary.

Prognosis and follow-up

The prognosis of JS depends largely on extra-neurological involvement, particularly nephronophthisis, congenital hepatic fibrosis and retinal dystrophy^{1,2}. Genotype-phenotype correlations have been described, which gives molecular diagnosis a prognostic value³. Current recommendations advise, at diagnosis and then periodically, assessment of renal function, urine concentration and renal ultrasound; liver function and abdominal ultrasound; ophthalmological examination; and neurodevelopmental evaluation¹.

Late-diagnosed forms can reveal themselves through severe renal failure, as in the 7-year-old girl reported by Montero Torres et al., who required peritoneal dialysis⁷. Children with purely neurological JS also have a better functional outcome after rehabilitation than those with multiorgan involvement⁶. Surveillance is therefore essential.

Genetic counselling

The consanguinity of our patient's parents is consistent with autosomal recessive inheritance, which implies a 25% recurrence risk for each pregnancy⁷. Molecular diagnosis would clarify

the prognosis through genotype-phenotype correlations, guide organ-specific surveillance and make prenatal or preimplantation diagnosis possible for future pregnancies¹⁻³.

Limitations

Extra-neurological screening (renal and hepatic work-up, ophthalmological examination) was not available, so the phenotype cannot be classified as purely neurological. No genetic testing was performed, so the diagnosis rests on clinical and radiological criteria. Diffusion tensor imaging, which could have shown the absence of decussation of the superior cerebellar peduncles, was not performed.

Conclusion

The diagnosis of Joubert syndrome rests on a constellation of posterior fossa findings rather than on a single sign. The molar tooth sign remains the hallmark on axial images, but a multiplanar assessment adds the near-perpendicular reorientation and thickening of the superior cerebellar peduncles on sagittal and coronal sections, hypoplasia of the cerebellar vermis with a midline cleft between the apposed cerebellar hemispheres and a bat-wing configuration of the fourth ventricle. Taken together and in the absence of a posterior fossa cyst, these features separate the malformation from Dandy-Walker malformation and rhombencephalosynapsis. Recognizing this combination carries the diagnosis even when neurodevelopmental features are mild and the presentation is delayed until late childhood. It should then prompt systematic renal, hepatic and retinal screening, molecular testing where available and genetic counselling, which matters particularly in consanguineous families.

Patient consent

Written informed consent for publication of this case and the accompanying images was obtained from the patient's parents.

Conflicts of interest

None.

Funding

None

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