International Journal of Contemporary Pediatrics
Ahmad M et al. Int J Contemp Pediatr. 2026 Jun;13(6):1011-1013
https://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20261550

Case Report

Joubert syndrome with typical magnetic resonance imaging findings

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Received: 08 February 2026
Revised: 07 March 2026
Accepted: 15 May 2026

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ABSTRACT

Joubert syndrome (JS) is an autosomal recessive disorder characterized by hypotonia, ataxia, specific breathing irregularities such as episodic apnea and hyperapnea (which improves with age), global developmental delay, nystagmus, strabismus, ptosis, and oculomotor apraxia. A 4 months old male child presented in ER as a case of acute respiratory illness. There was no h/o any seizure in the past or any family history of seizure with history of 3rd degree consanguineous marriage. On examination there was no head control with unilateral Ptosis in right eye and horizontal nystagmus as shown in figure 1 along with global developmental delay. Patient was managed for Acute respiratory illness but on magnetic resonance imaging (MRI) brain which was done in view of global developmental delay with hypotonia and nystagmus showed classical molar tooth appearance and was diagnosed with JS. As this patient had abnormal head movements and MRI brain showing Joubert like presentation, so patient was labelled with diagnosis of JS and was discharged after counselling.

Keywords: Joubert, Molar tooth malformation, Nystagmus, Ataxia, Global developmental delay

INTRODUCTION

Joubert syndrome (JS) is a rare autosomal recessive neurodevelopmental disorder first described by Joubert in 1969, characterized by a distinctive constellation of clinical and radiological findings. The hallmark neuroimaging feature, known as the "molar tooth sign," was later recognized as a defining diagnostic criterion and reflects a combination of cerebellar vermis hypoplasia, thickened and elongated superior cerebellar peduncles, and deepened interpeduncular fossa on MRI.[1-2]

JS has an estimated incidence of approximately 1 in 80,000-100,000 live births and demonstrates significant phenotypic variability, often leading to underdiagnosis or delayed recognition.[3] The disorder primarily affects the development of the cerebellum and brainstem, resulting in characteristic neurological manifestations including neonatal hypotonia progressing to truncal ataxia, global developmental delay, and intellectual disability.[4]

A distinctive clinical feature of JS is abnormal respiratory regulation in infancy, manifesting as episodic apnea and hyperpnea, which typically improves with age.[2] Ocular abnormalities are also prominent and include nystagmus, strabismus, ptosis, and oculomotor apraxia, contributing significantly to diagnostic suspicion in early life.[4]

JS is now recognized as part of a broader group of disorders known as ciliopathies, with multisystem involvement reported in many patients. These may include retinal dystrophy, renal cystic disease, hepatic fibrosis, and skeletal abnormalities, reflecting the underlying genetic heterogeneity of the condition.[4] The wide clinical spectrum has led to the use of the term "JS and related disorders" (JSRD), emphasizing the variability in phenotypic expression and associated organ involvement.[4]

Early diagnosis is crucial for appropriate management, genetic counseling, and surveillance for systemic complications. However, due to its rarity and overlapping

clinical features with other neurodevelopmental disorders, JS remains diagnostic challenge.[5] Recognition of the molar tooth sign on MRI plays a pivotal role in confirming the diagnosis and guiding further evaluation.[6]

CASE REPORT

A 4-month-old male infant presented to the emergency room with complaints of difficulty in breathing and fever for the past three days. There was no history of frothing from the mouth, fisting of hands, deviation of the angle of the mouth, or up-rolling of the eyeballs, and there was no prior history of seizures or family history of seizures. A history of third-degree consanguineous marriage between the parents was present. On admission, the child had a temperature of 100.1°F, respiratory rate of 55/min (regular), pulse rate of 140 bpm, and random blood sugar of 70 mg/dl. On clinical examination, the infant had poor head control, unilateral ptosis of the right eye, and horizontal nystagmus, as shown in Figure 1, along with features suggestive of global developmental delay.

Management and outcome

Investigations

Investigations revealed a total leukocyte count of 18,040 and positive C-reactive protein, while blood culture was sterile. Urine routine examination and culture, liver function tests, kidney function tests, and serum electrolytes were within normal limits. Fasting blood sugar was 84 mg/dl. On cardiovascular examination, S1 and S2 were present with no murmur. MRI brain showed diffuse vermian hypoplasia with cerebellar hemispheres apposed in midline without fusion, producing characteristic molar tooth appearance of the midbrain and bat-wing appearance of fourth ventricle, suggestive of JS, as shown in Figures 2 and 3. Ultrasound whole abdomen, ophthalmological examination, and 2D echocardiography were normal.

Treatment

The patient was managed with intravenous antibiotics, intravenous antipyretics, intravenous fluids, and other supportive treatment, and was monitored closely during the hospital stay.

Figure 1: Infant with poor head control.

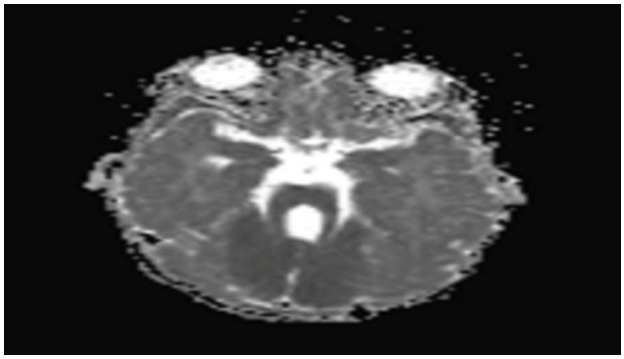

Figure 2: Brain MRI.

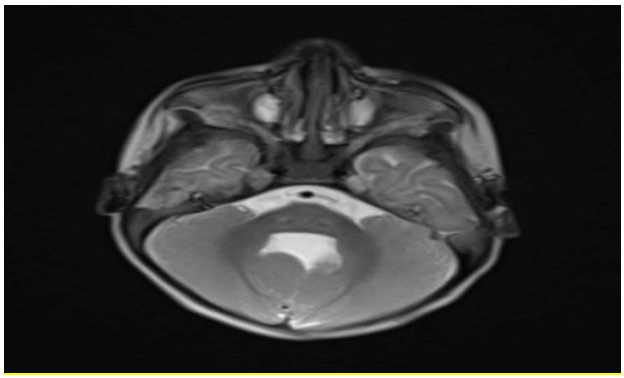

Figure 3: Brain MRI.

DISCUSSION

Joubert syndrome is a rare autosomal recessive neurodevelopmental disorder characterized by hypoplasia or aplasia of the cerebellar vermis and a distinctive neuroradiological feature known as the “molar tooth sign.” This hallmark imaging finding, best appreciated on axial neuroimaging, comprises a deepened posterior interpeduncular fossa, thickened and elongated superior cerebellar peduncles, and a hypoplastic or absent cerebellar vermis.[2,4]

Clinically, Joubert syndrome presents with neonatal hypotonia that gradually evolves into truncal ataxia, along with global developmental delay and varying degrees of intellectual disability. Additional neurological manifestations include abnormal eye movements such as nystagmus, oculomotor apraxia, strabismus, and ptosis, as well as characteristic neonatal breathing abnormalities, including episodic apnea and hyperpnea, which often improve with age.[7]

Joubert syndrome demonstrates significant phenotypic heterogeneity and is frequently associated with multisystem involvement, reflecting its classification among ciliopathies. Common extracranial manifestations include retinal dystrophy, nephronophthisis, congenital hepatic fibrosis, and polydactyly, with considerable inter- and intrafamilial variability in clinical expression.[8] Furthermore, a range of associated central nervous

system anomalies may be observed, including cerebellar dysplasia, polymicrogyria, agenesis of the corpus callosum, and cortical or brainstem heterotopia.[7] Recognition of these characteristic clinical and radiological features is essential for early diagnosis, appropriate management, and genetic counseling.

CONCLUSION

As this patient had abnormal head movements and MRI brain showing Joubert like presentation, so patient was labelled with diagnosis of JS and was discharged after counselling.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Ahmad M, Goyal A. Joubert syndrome with typical magnetic resonance imaging findings. Int J Contemp Pediatr 2026;13:1011-3.