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Case Series

Joubert syndrome misclassified as cerebral palsy/global developmental delay: lessons from a case series

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ABSTRACT

Global developmental delay (GDD) is a frequent diagnosis in pediatric practice, often labelled idiopathic or sometimes attributed to perinatal asphyxia. However, a subset of these cases is caused by underlying genetic or metabolic disorders, some of which are treatable or have disease-modifying options. Joubert syndrome (JS), a rare ciliopathy, exemplifies such conditions that may initially masquerade as nonspecific GDD. In this case series, we evaluated four children who were initially labeled as having GDD. Two of them had a history of perinatal asphyxia. All children presented with developmental delay, ataxia, ocular abnormalities, and craniofacial dysmorphism. Neuroimaging revealed cerebellar vermis hypoplasia and the characteristic molar tooth sign (MTS) in two cases, while genetic testing identified pathogenic variants in AHI1, TMEM67 and TCTN3 genes in three patients. Systemic involvement included retinitis pigmentosa, chorioretinal colobomas, hepatic fibrosis and ventricular septal defect. Each child benefited from multidisciplinary management. This series highlights that even when perinatal asphyxia is documented, children with GDD should still undergo thorough evaluation for alternative etiologies. Early identification of such conditions enables timely intervention, targeted surveillance, family counseling, and better long-term outcomes.

Keywords: Joubert syndrome, Global developmental delay, Genetic syndrome, Cerebellar hypoplasia, Retinitis pigmentosa, COACH syndrome, Molar tooth sign

INTRODUCTION

Globally, nearly 52.9 million children reportedly had developmental delay. Although the exact prevalence of developmental delay remains unknown, according to the WHO, approximately 10% of the population in each country has some form of disability.[1] GDD is one of the most common reasons for referral to pediatric neurology clinics. While many children are initially labeled as GDD, this descriptive diagnosis often masks underlying neurological or genetic conditions that require specific recognition and targeted management.

One such entity is JS, a rare autosomal recessive neurodevelopmental disorder. First described in 1969, JS is part of the expanding group of ciliopathies, with multisystem involvement including renal, hepatic, and retinal abnormalities.

JS is caused by pathogenic variants in more than 40 genes, resulting in abnormal structure and function of primary cilia. Defective ciliary signaling disrupts embryonic development of the cerebellum, brainstem, retina, kidneys, and liver, leading to the characteristic malformations and multisystem involvement. The hallmark neuroimaging feature, the MTS, reflects hypoplasia of the cerebellar vermis, thickened and elongated superior cerebellar peduncles, and a deep interpeduncular fossa, representing abnormal axonal decussation and cerebellar dysplasia.

Clinically, JS typically presents in infancy with hypotonia, disordered respiratory rhythm, and abnormal ocular movements such as nystagmus and oculomotor apraxia. As

children grow older, hypotonia often evolves into ataxia, gait disturbances, and variable cognitive impairment.[2] Additional multisystem features include retinal dystrophy, coloboma, ptosis, and visual impairment; cardiovascular involvement in the form of VSD, renal involvement ranging from nephronophthisis to end-stage renal disease; and hepatic fibrosis, particularly in TMEM67-related subtypes. Craniofacial dysmorphism, oral-facial-digital anomalies, and skeletal abnormalities such as polydactyly may also be present.

Although these findings are not pathognomonic to Joubert syndrome, the essential features of Joubert syndrome have not been firmly established. Genetic studies have linked more than 40 different genes to Joubert syndrome, collectively responsible for up to 94% of cases.[3] It has distinctive radiological findings-most notably the MTS on MRI, which results from cerebellar vermis hypoplasia and brainstem malformations. In clinical practice, children with JS are often initially categorized under the broad umbrella of GDD, especially when initial neuroimaging is labelled normal. The wide phenotypic spectrum and early overlap with GDD often lead to delayed diagnosis, highlighting the importance of high-quality MRI and genetic testing in suspected cases. This misdiagnosis may postpone appropriate counselling, genetic testing, surveillance for systemic involvement, and timely interventions that can improve long-term outcomes.

Here, we present a case series of children who were first diagnosed as having GDD, but were later identified to have JS. This report highlights the diagnostic challenges, emphasizes the importance of neuroimaging and genetic testing in unexplained developmental delay, and underlines the need for heightened clinical suspicion to avoid overlooking this rare but important disorder. The primary etiology of a GDD should always be identified if possible. This is particularly important in the case of genetic or metabolic disorders that have specific disease-modifying treatment.

CASE SERIES

This is a prospective analysis of children presenting to the Pediatric Neurology clinic in a tertiary care hospital who were labelled with GDD and had occulo-motor concerns during a one-year period from September 2024 to September 2025. Clinical details including perinatal history, developmental milestones, family history, and consanguinity were recorded.

All children underwent a detailed anthropometric and neurological examination, with particular attention to gait/coordination (ataxia), speech, ocular movements, visual acuity, presence of saccadic eye movement and fundus examination. Each patient underwent a thorough ophthalmologic evaluation. This included visual acuity, cycloplegic refraction, dilated fundus and retinal examinations. Ocular alignment was measured by a cover test using prisms. It was followed by detailed systemic examination. Based on clinical suspicion, magnetic resonance imaging (MRI) of the brain was performed to assess for structural malformations, particularly the MTS characteristic of JS. In children where MRI did not reveal diagnostic findings or was reported as normal, genetic testing was done with proper pretest counseling and informed consent. Children diagnosed with JS subtype were further evaluated in detail for specific system involvement, including detailed ophthalmological assessment for retinal dystrophy and abdominal ultrasound, fibroScan to detect hepatic fibrosis, respectively.

Case 1

A 12-year-old boy presented with difficulty in walking and poor vision. He was born at term via normal delivery, with a perinatal period complicated by birth asphyxia. There was a history of significant developmental delay, and he was initially labeled as having GDD because of perinatal asphyxia. On general physical examination, the child had facial dysmorphism in the form of broad forehead, hypertelorism (canthal index=0.44), deep-set eyes, prominent supraorbital ridges, depressed nasal bridge with broad nose and long philtrum.

On neurological examination, higher mental functions were appropriate for age. Cranial nerve examination revealed reduced visual acuity (RE:3/60, LE:6/60 with correction), right eye esotropia and rotatory nystagmus. Motor examination showed normal bulk, slight decreased tone and full power (5/5) in all four limbs. Deep tendon reflexes were 1+, and plantar responses were flexor bilaterally. Cerebellar testing revealed ataxia with a broad-based gait, scanning speech, and dysdiadochokinesia, though there was no titubation, intention tremor/dysmetria. Child was able to draw simple figures, albeit slowly-demonstrating difficulty with fine movements. Orthopedic examination revealed bilateral pes planus. Ophthalmology evaluation revealed advanced retinitis pigmentosa with macular sparing; electroretinography advised but not performed. Echocardiography showed a dilated coronary sinus with left ventricular systolic collapse. Abdominal ultrasound, renal, and liver function tests were normal.

Investigations included MRI brain, which showed hypoplasia of the superior cerebellar vermis with cerebellar dysplasia, though the classical MTS was not clearly appreciable. A prior neonatal CT brain was reportedly normal. Genetic testing revealed a homozygous pathogenic variant in exon 9 of the AHI1 gene, confirming JS type 3 (Figure 1). Management included physiotherapy for gait and balance training, corrective lenses for visual impairment, orthopedic shoes for pes planus, and supportive therapies. Ophthalmology follow-up was advised to monitor progression of retinal dystrophy, and cardiology follow-up was planned for echocardiographic findings. A multidisciplinary follow-up plan involving neurology, gastroenterology, ophthalmology, cardiology, and rehabilitation services was initiated.

FINDINGS RELATED TO PHENOTYPE

Gene&Transcript	Variant	Location	Zygosity	Disorder(OMIM)	Inheritance	Variant Classification
AHI1 NM_001134831.2	c.1051C>T p.Arg351*	Exon 9	Heterozygous	JOUBERT SYNDROME 3; JBTS3:608629	Autosomal Recessive	**Pathogenic**

* Genomic Position based on Assembly GRCh37, **Number of applied in silico programs predicting the effect of the variant on the protein outcome (CADD: Combined Annotation Dependent Depletion (v1.6), SIFT, PolyPhen-2, MT: Mutation Taster), N/A: Not Applicable, ***Minor Allele Frequency as described in GnomAD (Controls), ****based on ACMG Guidelines. het=heterozygous, hom=homozygous, hemi=hemizygous.

Figure 1: Genetic report of case 1-pathogenic AHI1 variant identified in a patient of JS.

Case 2

A 12-year-old male child, born of a third-degree consanguineous marriage via normal delivery, presented with developmental delay. There was a history of perinatal asphyxia requiring a 5-day NICU admission. Parents had noted microphthalmia of the left eye and saccadic movements of the right eyeball. He had poor vision in both eyes, especially minimal vision in left eye. He had GDDs which were addressed with physiotherapy and specialized therapies targeting language and fine motor skills. He attended a special school for three years before transferring to a regular school. At the presentation he complains of difficulty in walking, writing, involuntary movements of hands which got increased during stress and poor vision. On examination he had facial dysmorphism in the form of prominent supraorbital ridges, microphthalmia of the left eye with esotropia, and low-set ears (Figure 2). On neurological examination higher function revealed low average IQ (82). In the motor system he had normal bulk in all four limbs, slight decrease in tone, normal power DTR of 1+ and flexor plantar response. In cerebellar examination he had broad based gait with swaying during movement, he had kinetic and intentional tremors, dysdiadokinesia and dysmetria.

Ophthalmological examination revealed visual acuity of 6/36 in the right eye and 1 meter finger counting in the left eye. No improvement with the pinhole was seen. Ocular apraxia was present with left eye esotropia. Fundus examination revealed inferior chorioretinal coloboma in the right eye and an optic disc with choroidal coloboma in the left eye. MRI done at birth and at the time of admission was reported to be normal. In blood biochemistry liver function test mild transaminitis was present with raised lactate and LDH level. Abdominal ultrasound does not reveal a significant finding. Genetic testing was advised because of strong suspicion of JS. It revealed homozygous pathogenic variant in TMEM67 gene, Exon 18 associated with JS 6 COACH syndrome 1 (JBTS6, COACH1). It also revealed homozygous uncertain significance variant in the NRCAM gene, Exon 21 associated with Neurodevelopmental disorder with Neuromuscular and Skeletal abnormalities. Based on the report liver fibroscan was advised which showed elevated liver stiffness with moderate fibrosis (Figure 2-3).

The patient was managed conservatively with physiotherapy, occupational therapy and speech therapy. The patient was connected to a multidisciplinary team consisting of an ophthalmologist, neurologist, gastroenterologist and physical rehabilitation team.

Figure 2: Ocular and facial features of case 2-photograph demonstrating microphthalmia of left eye, deep set eyes and prominent supraorbital ridge.

FINDINGS RELATED TO PHENOTYPE

Gene&Transcript	Variant	Location	Zygosity	In silico Parameters**	Disorder(OMIM)	Inheritance	Variant Classification
NRCAM NM_001037132.4	c.2222C>T p.Thr741Met	Exon 21	Homozygous	CADD: 24.2 SIFT: Deleterious MT: Damaging	NEURODEVELOPMENTAL DISORDER WITH NEUROMUSCULAR AND SKELETAL ABNORMALITIES; NEDNMS:619833	Autosomal Recessive	Uncertain Significance
TMEM67 NM_153704.6	c.1847C>T p.Ala616Val	Exon 18	Homozygous	CADD: 27.3 SIFT: Deleterious MT: Damaging	JOUBERT SYNDROME 6; JBTS6:610688 COACH SYNDROME 1; COACH1:216360	Autosomal Recessive	Likely Pathogenic

* Genomic Position based on Assembly GRCh37, **Number of applied in silico programs predicting the effect of the variant on the protein outcome (CADD: Combined Annotation Dependent Depletion (v1.6), SIFT, PolyPhen-2, MT: Mutation Taster), N/A: Not Applicable, ***Minor Allele Frequency as described in GnomAD (Controls), ****based on ACMG Guidelines. het=heterozygous, hom=homozygous, hemi=hemizygous.

Figure 3: Genetic report of case 2-pathogenic variant identified in TMEM67 in a patient of JS.

Case 3

A 14-year-old girl, sibling of case 2, born out of a third-degree consanguineous marriage via normal delivery, presented with developmental delay. The perinatal period was uneventful. Early developmental delays were addressed with physiotherapy and specialized therapies targeting language and fine motor skills. She attended a special school for three years before transferring to a regular school. Parents reported persistent poor eye contact and limited social interactions, along with subtle facial dysmorphism in the form of prominent and broad forehead, hypertelorism, flat and broad nasal bridge, thickened lips with tented upper lip appearance, long philtrum. She also had upper labial frenulum. On examination, she had a low average IQ. Cerebellar signs included a wide-based gait, scanning speech, and dysdiadokinesia. The rest of the motor system, including tone, power, and reflexes, was normal. Ophthalmological evaluation revealed right eye exotropia and reduced visual acuity (RE:6/18, LE:6/24) which showed slight improvement with pinhole. Fundus examination revealed bilateral coloboma of the disc. MRI brain revealed the classic MTS, consistent with JS (Figure 3). Genetic testing was advised but refused by the parents. Abdominal ultrasound, renal, and liver function tests were normal.

Management included upper lip frenulotomy, continued physiotherapy, speech therapy and supportive therapies, educational support in regular school settings, and ophthalmology follow-up for visual impairment and exotropia. A multidisciplinary follow-up plan involving neurology, ophthalmology, and rehabilitation services was recommended.

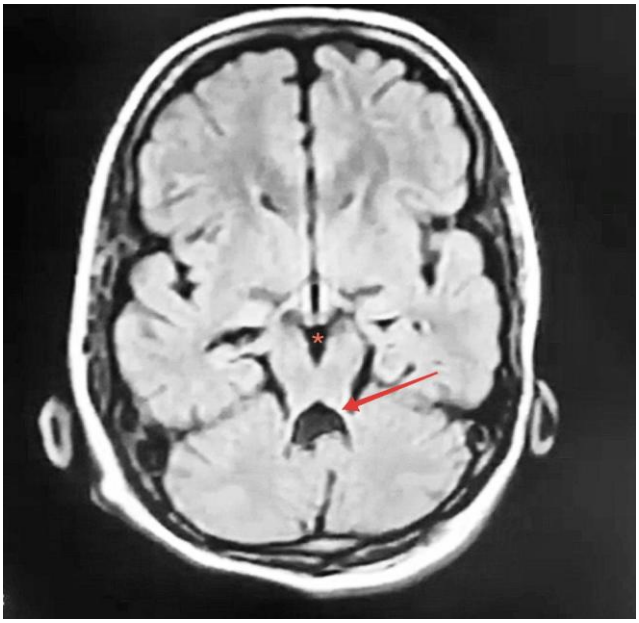

Figure 4: Axial T2/flair MRI brain demonstrating the characteristic MTS, with a deep interpeduncular fossa (asterixis) and thickened/elongated superior cerebellar peduncles (arrow).

Case 4

A 4-year-old female child was brought to the OPD with recurrent pneumonia, complaints of frequent falls while walking, and not looking at things around her. She was born out of consanguineous marriage at term via LSCS, with a history of delayed cry but no NICU admission. At the age of around 5 weeks the child developed pneumonia and since then she had multiple episodes of pneumonia. On evaluation the cause of recurrent pneumonia was found out to be ventricular septal defect. Parents also noted that a child is not able to hold the neck till the age of 7 months, not able to sit till the age of 13 months and not able to stand without support till 2 and half years and walk by 3 years of age. Regarding vision, mother complains of irregular jerky movement of eyes and not looking at the objects that are far from her. The child was labelled as GDD with ventricular septal defect. On examination the child had facial dysmorphism in the form of broad forehead, deep set eyes, prominent supraorbital ridge with broad nose and long philtrum.

On neurological examination, the child was interacting with the parents normally but not paying attention to the toys kept at distance from her. She was able to use 2–3-word sentences but the speech had scanning characteristics. Cranial nerves examination showed no abnormalities except 2nd cranial nerve. On functional visual assessment, she demonstrated rotatory nystagmus, fixation to her mother's face and a bright light at approximately 3 metres. Grossly horizontal and vertical tracking was present. Formal quantitative visual acuity could not be reliably assessed because of limited cooperation. Fundus examination revealed chorioretinal coloboma in both eyes. In motor system examination she had normal bulk in all 4 limbs, generalised hypotonia, normal power, Dtr 1+ and flexor plantar response. In cerebellar examination she has a broad based gait with swaying and repeated falls. When she was offered a toy past pointing was noted along with slight tremors.

On systemic examination a pansystolic murmur was appreciated at the lower left sternal border. Rest systemic examination was unremarkable.

On investigation blood parameters and biochemistry were normal. Abdominal USG was normal. ECHO revealed a small muscular VSD. MRI brain revealed classic molar tooth, the characteristic neuroimaging abnormality of JS. Based upon strong clinical suspicion genetic testing was advised to further characterize the Jouber syndrome and it showed heterozygous mutation in TCTN3, which is seen in JS 18. It also showed a heterozygous mutation in L2HGDH which is associated with L-2 Hydroxyglutaric aciduria. Further testing by TMS and GCMS was advised to investigate the L2HGDH finding.

The patient was managed by a multidisciplinary team and prenatal counselling was offered to parents.

Gene&Transcript	Variant	Zygosity	In silico Parameters**	Disorder(OMIM)	Inheritance	Variant Classifcation
TCTN3 NM_015631.6	c.776dup p.Lys260GlnfsTer 5	Heterozygous	CADD:34	JOUBERT SYNDROME 18; JBTS18:614815	Autosomal Recessive	**Likely Pathogenic**

Figure 5: Genotype of case 4 showing heterozygous mutation in TCTN3.

Table 1: Comparative clinical characteristics of the cases.

Variables	Case 1	Case 2	Case 3	Case 4
Age (in year)/sex	12/M	12/M	14/F	4/F
Age at diagnosis (in years)	12	12	14	4
History of perinatal asphyxia	Present	Present	Absent	Present
Consanguinity	Absent	Present	Present	Present
Weight(kg)	31	34	38	13 kg
Stature(cm)	147	148	152	96
BMI	14	15	15	14.1
Facial dysmorphism	Present	Present	Present	Present
Visual acuity RE	3/60	6/36	6/18	Grossly can look at object at 3 metres
Visual acuity LE	6/60	Finger counting at 1 meter	6/24	Grossly can look at object at 3 metres
Fundus	Advanced retinitis pigmentosa with macular sparing both eyes	RE- inferior chorioretinal coloboma, LE-choroidal coloboma	Coloboma of the disc in both eyes	Chorioretinal coloboma in both eyes
Miscellaneous occular findings	Ocular aparxia, rotatory nystagmus	Ocular aparxia, rotatory nystagmus, Microophthalmia	Ocular apraxia, Amblyopia, RE esotropia	Rotatory nystagmus
CNS	Generalized mild hypotonia, DTR 1+, Cerebellar signs +	Generalized mild hypotonia, DTR 1+, Cerebellar signs +	Generalized mild hypotonia, DTR 1+, Cerebellar signs +	Generalized mild hypotonia, DTR 1+, Cerebellar signs +
MRI brain	Hypoplasia of Cerebellar vermis with dysplasia of cerebellum (MTS+)	Normal	Hypoplasia of Cerebellar vermis with dysplasia of cerebellum (MTS+)	MTS
Renal function test	Normal	Normal	Normal	Normal
Hepatic involvement	Normal	Mild hepatitis, Moderate fibrosis	Normal	Normal
Genetic mutation	AHI1 (Exon 9) – c.1051C>T (p.Arg351*)	TMEM67 (Exon 18) – c.1849C>T (p.Ala617Val),	NA	TCTN3 (Exon 6)- c.776 dup (p.Lys260GlnfsTer5)
Mode of inheritance	AR	AR	NA	AR

DISCUSSION

JS is a rare autosomal recessive ciliopathy characterized by the classical MTS on neuroimaging, together with a wide spectrum of neurological, ophthalmological, renal, hepatic, cardiovascular and skeletal manifestations. The hallmark clinical features include developmental delay, hypotonia evolving into ataxia, ocular motor apraxia, and variable cognitive impairment.[3] All the neurological features were seen in our cases. Our series illustrates the diagnostic challenges that persist in routine practice. All of our patients were initially diagnosed with GDD, reflecting

the difficulty in differentiating JS from nonspecific neurodevelopmental disorders in early childhood. Early neuroimaging, particularly computed tomography, and even initial MRI may fail to detect subtle cerebellar vermian and brainstem abnormalities, leading to delayed or missed diagnosis of JS.[4] We encountered this directly: in one child from our series early neuroimaging (CT at birth) was reported as normal and in another, early neuroimaging including MRI did not show clear abnormalities, contributing to an initial label of GDD before repeat imaging and further workup clarified the diagnosis. Recent studies confirm that MRI remains the gold standard for detecting the MTS, while CT is frequently normal in neonates.[5]

In our cases, Ophthalmological manifestations were prominent, including retinitis pigmentosa, ocular colobomas, strabismus, and ocular motor apraxia. Recent studies report that up to 40% of JS patients develop progressive retinal dystrophy, often leading to severe visual loss.[6] Hepatic involvement, as noted in case 2 which showed moderate fibrosis, is well documented in TMEM67-related JS, where nearly one-third of patients develop clinically significant liver disease.[7] Craniofacial dysmorphism was another salient feature, with hypertelorism, broad nasal bridge, long philtrum, and abnormal upper lip frenulum attachment observed in our cases. Although not pathognomonic, such craniofacial features are increasingly recognized in JS cohorts and can serve as additional diagnostic clues when combined with neurological and ophthalmological features (Table 1).[4,8]

In the evaluation of patients presenting with developmental delay, hypotonia, and cerebellar abnormalities, several conditions can mimic JS and must be considered in the differential diagnosis. These include Dandy-Walker malformation, Meckel-Gruber syndrome, 3C syndrome and certain ciliopathies. Dandy-Walker malformation typically presents with cystic dilatation of the fourth ventricle and an enlarged posterior fossa, but unlike Joubert, the characteristic MTS on axial MRI is absent.[9] Meckel-Gruber syndrome often presents prenatally with occipital encephalocele, polydactyly, and renal cystic dysplasia, features not seen in Joubert.[10] 3C syndrome, characterized by heart defects, cerebellar hypoplasia, and cranial dysmorphism, can also present with overlapping features with JS.[11] However, the presence of heart malformations and distinct craniofacial abnormalities aid in distinguishing 3C syndrome from JS. Genetic testing plays a pivotal role in differentiating JS from other ciliopathies, as JS is associated with mutations in multiple genes (e.g., AHI1, CEP290, TMEM67, TCTN3), whereas other syndromes have distinct genetic markers. Thus, careful integration of neuroimaging, systemic examination, and genetic studies is essential for accurate diagnosis and appropriate counseling.

Genetic testing has become central not only to the diagnosis but also to the subtyping of JS. More than 40 genes are now implicated in JS, with pathogenic variants explaining most cases.[2] Our series identified mutations in AHI1, TMEM67 and TCTN3 consistent with previous literature that links AHI1 with retinal dystrophy, TMEM67 with hepatic fibrosis and ocular colobomas and TCTN3 with VSD.[2,7] The presence of other variants of uncertain significance (VUS) further highlights the challenges in clinical interpretation of genetic data.

At present, no curative therapy exists for JS. Management remains supportive and multidisciplinary, focusing on early intervention and prevention of complications. Treatment strategies include physiotherapy for hypotonia and ataxia, occupational and speech therapy for motor and communication deficits, visual rehabilitation and ophthalmologic interventions, orthopedic support for skeletal abnormalities, and regular surveillance for renal, hepatic and cardiovascular involvement.[2,3,6] Genetic counseling is a critical component of care, particularly in consanguineous families. Emerging research into ciliary biology and targeted molecular pathways may offer future disease-modifying therapies, but these remain investigational.

CONCLUSION

This case series underscores the clinical and genetic heterogeneity of JS and the risk of misclassification of idiopathic or perinatal asphyxia associated with GDDs, particularly in the presence of documented perinatal asphyxia. Our findings emphasize the importance of maintaining a high index of suspicion for genetic etiologies in children with unexplained developmental delay, especially when accompanied by ocular abnormalities, ataxia, or craniofacial dysmorphism.

Future research should focus on early diagnostic biomarkers, improved access to advanced neuroimaging and genetic testing, clearer genotype–phenotype correlations, and the development of disease-modifying therapies targeting ciliary dysfunction. Long-term prospective studies are needed to define neurodevelopmental outcomes and quality of life in affected individuals.

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