

## Case Report

### Joubert Syndrome: A Case Report

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#### Abstract

Joubert Syndrome (JS) is a rare autosomal recessive genetic heterogeneously inherited disorder. In this case report, we have discussed a 9-month-old girl who presented with developmental delay, decrease vision and history of recurrent respiratory infection with respiratory distress. On examination, she had facial dysmorphism, decreased vision and hypotonia. Brain magnetic resonance imaging showed which showed thick, elongated and abnormally oriented superior cerebellar peduncle showing molar tooth appearance with elongated bat-wing shaped 4th ventricle & hypoplasia of vermis suggestive of JS. Patient has been treated at Garden Reach Institute for the Rehabilitation and Research (GRIRR), Kolkata, India by the multidisciplinary team of physiotherapist, speech therapist, special educator, orthotist, medical officer and social worker, special educator. In conclusion, knowledge of characteristic clinical and radiological findings in JS will help in early diagnosis, and successful rehabilitation.

**Keywords:** Genetic disorder; Joubert syndrome; Molar tooth sign; Respiratory distress

#### Introduction

Joubert Syndrome (JS) is a rare autosomal recessive genetic heterogeneously inherited disorder characterized by neurological features that includes hypotonia, ataxia, developmental delay, intellectual disability, abnormal eye movements, and neonatal breathing dysregulation [1]. It can be diagnosed on the basis of clinical presentation and presence of the Molar Tooth Sign (MTS) on Brain axial Magnetic Resonance Imaging (MRI) [1,2].

Reported prevalence of JS, commonly range of 1:80,000 to 100,000 livebirths 4 is probably underestimated due to lower number of case reports of MTS in the literature [2,3]. This signifies

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the case reporting of JS as epidemiology data such as population-based prevalence rates are almost completely lacking. As it represent like delayed developmental milestone so in most of the cases may not be diagnosed in early phase of childhood, however, presence of respiratory distress along with delayed developmental milestone may rise the suspicion and diagnosis of the JS [4]. Maria et al reported 33 months as an average age for its diagnosis [5]. Most children with this syndrome survive infancy to reach adulthood [6]. Early detection of this condition is essential so that appropriate intervention can be started as early as possible.

#### Case Report

A 9 months old girl child, presented to the Department of Pediatrics, IPGMER hospital, Kolkata, India with history of fever for 2 days and respiratory distress, where she was diagnosed with Joubert's syndrome with associated respiratory tract infection and recurrent seizure disorders. It was a known follow-up case of Hypoxic Ischemic Encephalopathy (HIE) Stage-II with Laryngomalacia and was on medication for the convulsions. The diagnosis of Joubert's syndrome was made on the basis of a Brain MRI, which showed thick, elongated and abnormally oriented superior cerebellar peduncle showing molar tooth appearance with elongated bat-wing shaped 4th ventricle & hypoplasia of vermis. Laboratory investigations, including CBC, blood sugar, electrolytes, C-reactive protein, liver enzymes, urea and creatinine, TSH, free T4 and T3 were all normal. There was term uneventful pregnancy delivered with no complications, no cesarean section, birth weight of 2.5 kg, and delayed birth cry. There was no history of consanguineous marriage. There was a history of recurrent respiratory infection at the age of 3 years also.

The child was regular medication and on-&-off physiotherapy treatment till the age of 3 years. At the age of 3 years, she was brought to Garden Reach Institute for the Rehabilitation and Research (GRIRR), Kolkata, an NGO working for the underprivileged and disable children. GRIRR has a multidisciplinary team of physiotherapist, speech therapist, special educator, orthotist, medical officer and social worker. An initial physical assessment and evaluation was made by the team that found facial dysmorphism, lack of neck & head control, unable to sit (supported or unsupported), unable to stand (supported or unsupported), hypotonic upper & lower extremities, unable to make sound even for cry, no vocal response to stimulus, drowsy eyes with no eye movement. No polydactyly has been noted. Initial goals and interventions were decided with the multidisciplinary team and rehabilitation was initiated in full-fledge manner.

The child was getting 90 minutes treatment session, 6 days a week that includes physiotherapy and in-between speech therapy. The physiotherapy intervention included positioning, sensory integration therapy, milestone developmental stages with functional activities, weight bearing & joint-compression techniques, stretching, motor control activities using gym ball, instrument-assisted ADL activities, hand function activities, and use of assistive devices and orthosis such as special chair, bilateral Ankle-Foot Orthoses (AFO), standing frame, balance board etc.

She has continued her treatment sessions at GRIRR from the age of 3 years to till date (Age 6 years) with the multidisciplinary team. She has shown excellent improvement in her condition and she has achieved good sitting balance, able to sit without support, stand with wall support, able to walk using bilateral AFO and reverse walker. She also responds to her name and other conversations, and also makes sound like papa, cha etc.

## Discussion

Hypotonia during infancy, developmental delay &/or mental retardation, associated with abnormal breathing pattern (hyperpnea and/or apnea or may be altered pattern of breathing) and abnormal eye movements (nystagmus) are the most important clinical signs and symptoms to suspect the case of JS [7]. However, radiological presence of 'molar tooth sign' on Brain MRI is an essential component to confirm the diagnosis. The present case had all the clinical symptoms with the exception of nystagmus and mental retardation which may have been overlooked.

The main findings from cranial MRI studies are:

1. Cerebellar vermis hypoplasia
2. Deepened interpeduncular fossa
3. Thick, horizontal enlarged superior cerebellar peduncles
4. The present case had all the findings radiologically

However, cerebellar vermis hypoplasia had been reported as a component of other disorders like trisomy 21, occipital encephalocele and dandy walker malformation [1,8,9]. Besides, hypotonia is also associated with other features to be considered as an essential clinical feature [10].

JS can be classified into 6 sub types: pure JS, JS with ocular defect (JS-O), JS with renal defect (JS-R), JS with oculorenal defects (JS-OR), JS with hepatic defect (JS-H) and JS with orofacioidigital defects. In our case, the child has some vision issue, but there is not a report of retinopathy and associated eye disorders. Moreover, the child has no pathogenomic features of suspected renal disorders. There is definitely presence of facial dysmorphism, though polydactyly is not present. There is lack of evidence to confirm or exclude hepatic defect as the child was not referred to specialist for it. This might be due to potential lack of clinical awareness or due to illiteracy of the parents' child.

Genetic counselling has been recommended as one of the important measure to prevent JS. Prenatal diagnosis is feasible through chorionic villus sampling at around eleven week gestation and diagnosis of JS has been documented [1,10]. Fetal ultrasound may be useful to diagnose high-risk pregnancies as it showed increased nuchal translucency. The prognosis of JS cases may be good, if diagnosed in early childhood and managed early through multidisciplinary intervention program for patients including physical therapy, special education, occupational, and speech therapy. The early intervention has shown significant benefits in advancement of developmental milestones for several patients with JS [11]. Unfortunately, in our case the child has received intervention late from the age of 3 years, though she had showed remarkable improvement in the condition and the parents are happy.

We have discussed it to summarize main clinical and radiological mile stones for its diagnosis, and approach of the rehabilitation for the better outcomes. We also emphasize to promote the awareness regarding disabilities of the child through this scientific article.

## Conclusion

Knowledge of characteristic clinical and radiological findings in JS will help in early diagnosis, and successful rehabilitation.

## Disclosure Statement

There are no conflicts of interest to declare.

## Funding Sources

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## References

1. Brancati F, Dallapiccola B, Valente EM (2010) Joubert Syndrome and related disorders. *Orphanet J Rare Dis* 5: 20.
2. Nuovo S, Bacigalupo I, Ginevrino M, Battini R, Bertini E, et al. (2020) Age and sex prevalence estimate of Joubert syndrome in Italy. *Neurology* 94: 797-801.
3. Boltshauser E, Isler W (1977) Joubert syndrome: episodic hyperpnea, abnormal eye movements, retardation and ataxia, associated with dysplasia of the cerebellar vermis. *Neuropadiatrie*. 8: 57-66.
4. Akcakus M, Gunes T, Kumandas S, Kurtoglu S, Coskun A (2003) Joubert syndrome: Report of a neonatal case. *Paediatr Child Health* 8: 499-502.
5. Maria BL, Quisling RG, Rosainz LC, Yachnis AT, Gitten J, et al. (1999) Molar tooth sign in Joubert syndrome: clinical, radiologic, and pathologic significance. *J Child Neurol* 14: 368-376.
6. Bandichhode ST, Anitha MS, Pandav A (2013) Joubert Syndrome- A Case Report. *JKIMSU* 2: 138-140.
7. Parisi MA, Glass IA. In: *Gene Reviews at Gene Tests- Gene Clinics: Medical Genetics Information Resource*. Seattle: Copyright, University of Washington; 1997- 2006.
8. Satran D, Pierpont ME, Dobyns WB (1999) Cerebello-oculorenal syndromes including Arima, Senior-Loken and COACH syndromes: more than just variants of Joubert syndrome. *Am J Med Genet* 86: 459-469.
9. Aslan H, Ulker V, Gulcan EM, Numanoglu C, Gul A, et al. (2002) Prenatal diagnosis of Joubert syndrome: a case report. *Prenat Diagn* 22: 13-16.
10. Saraiva JM, Baraitser M (1992) Joubert syndrome: a review. *Am J Med Genet* 43: 726-731.
11. Parisi MA (2009) Clinical and molecular features of Joubert syndrome and related disorders. *Am J Med Genet C Semin Med Genet* 15: 326-340.



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