International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

A Case Report of Joubert Syndrome and Related Disorders

G. Harsha Vardhan, Preethi N.

Abstract: ***Joubert syndrome is a rare genetic heterogeneous disorder characterized by cerebellar and brain stem malformations and multisystem involvement in varying proportions. It is also known as cerebello-ocular -renal syndrome. The presentation of JS is variable. Diagnosis of "Classic JS" is based on the presence of a Molar tooth appearance on MRI, Hypotonia in infancy later progressing to ataxia, Development delay, abnormal breathing pattern, and abnormal eye movements. If JS is associated with additional features like retinal dystrophy, ocular coloboma, congenital heart disease, cystic kidney disease, liver fibrosis, polydactyly, and dysmorphic facies it is called as "JSRD"(JS and related disorders). Our case presents with JSRD with Molar tooth appearance on MRI, Molar tooth, cystic kidney disease, and dysmorphic facies. Affected babies require periodic follow-up visits with growth monitoring, neurological assessment, monitor for progressive JSRD complications. These babies often require early intervention through speech, occupational, and physical therapy. Once the diagnosis is made in a neonate, serial antenatal USG screening should be performed in subsequent pregnancies combined with fetal MRI at 20-22 weeks gestation to maximize the accuracy of prenatal diagnosis. In general Prognosis for JS is poor and depends on the severity of different organ involvement with 5-year survival rate is 50%. Death is usually due to feeding difficulties and respiratory infections.***

Keywords: Joubert syndrome, Classic JS, JSRD (JS and related disorders), Molar tooth, Molar tooth

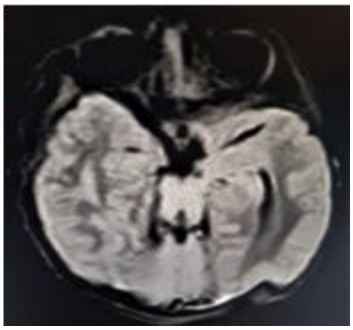
1. Introduction

Joubert syndrome is a rare genetic heterogeneous disorder characterized by cerebellar and brain stem malformations and multisystem involvement in varying proportions. It is also known as cerebello-ocular-renal syndrome. Reported incidence is 1/80, 000 to 1/1, 00, 000 live births.

2. Case Report

A 1 hr old MNB of 3.2 kgs born to primi mother through LSCSdidn't cried immediately after birth revived with bag and tube ventilation and admitted in NICU In view of Perinatal asphyxia. At admission, kept on a ventilator. The tone and activity were poor. Baby had dysmorphic facial features like a broad forehead, broad nose, depressed nasal bridge, and low set ears. Neurocutaneous markers were absent. on Day2 sepsis screen was positive started on antibiotics. Extubated on day 4. On Day 5-9 baby had a weak cry, generalized Hypotonia, hyporeflexia and incomplete Moro's reflex and intermittent hyperpnoea intensified whenever stimulated. Rest of the systemic examination was within normal limits. CBC is normal, Blood culture negative, LP done CSF analysis normal, Ophthalmic examination, Chest xray, and 2DECHO were normal. MRI brain was done which revealed cerebellar vermis hypoplasia with prominent thickened elongated superior cerebellar peduncles giving Molar tooth sign like appearance, 4th ventricle shows Bat wing configuration. USG abdomen suggests Bilateral medullary cystic kidney disease.

3. Clinical Imaging



4. Discussion

The presentation of JS is variable. Diagnosis of "Classic JS" is based on the presence of a Molar tooth appearance on MRI, Hypotonia in infancy later progressing to ataxia, Development delay, abnormal breathing pattern, and abnormal eye movements. If JS is associated with additional features like retinal dystrophy, ocular coloboma, congenital heart disease, cystic kidney disease, liver fibrosis, polydactyly, and dysmorphic facies it is called as "JSRD" (JS and related disorders). **Our case presents with JSRD** with Molar tooth appearance on MRI, Hypotonia, cystic kidney disease, and dysmorphic facies. Affected babies require periodic follow-up visits with growth monitoring, neurological assessment, monitor for progressive JSRD complications. These babies often require early intervention through speech, occupational, and physical therapy. Once the diagnosis is made in a neonate, serial antenatal USG screening should be performed in subsequent pregnancies combined with fetal MRI at 20-22 weeks gestation to maximize the accuracy of prenatal diagnosis. In general Prognosis for JS is poor and depends on the severity of

different organ involvement with 5-year survival rate is 50%. Death is usually due to feeding difficulties and respiratory infections.

5. Conclusion

JS is a rare genetic disorder associated with significant mortality and morbidity and diagnosis is often delayed due to its variable, non-specific presentation. MRI has an important role help not only in early diagnosis but will also help in ensuring appropriate counselling and proper rehabilitation of baby.

References

[1] Nelson 21st edition (vol.2, page 3073-3074).
[2] Parisi MA, Doherty D, Joubert syndrome orphaned J Rare Dis.2010
[3] Kohler S, Gargano M, Matentzoglu N, et al., The Human Phenotype Ontology in 2021, Nucleic Acids Research, Volume 49, Issue D1, 8 January 2021, Pages D1207–D1217.
[4] Downs SM, van Dyck PC, Rinaldo P, et al. Improving newborn screening laboratory test ordering and result reporting using health information exchange. J Am Med Inform Assoc.2010 Jan-Feb; 17 (1): 13-8.

Abbreviations

NICU-NEONATAL INTENSIVE CARE UNIT
LP-LUMBAR PUNCTURE
CSF – CEREBROSPINAL FLUID
ECHO – ECHOCARDIOGRAPHY
USG-ULTRASONOGRAPHY
JS-JOUBERT SYNDROME
MRI-Magnetic Resonance Imaging