Joubert Syndrome in 10-Year-Old with Renal Involvement: A Case Study

Sagar Dabbara1*, Arya P2 and Laxmi Poojita AN3
1Sri Venkateswara Medical College, Andhra Pradesh, India
2Ramaiah Medical College, Bengaluru, India
3Kurnool Medical College, Andhra Pradesh, India

Abstract
Joubert syndrome (JS) is a rare recessive autosomal disorder in infants and children. JS is a ciliopathy with defects in the primary cilium. JS has mid-brain-hind brain malformation which includes: (i) cerebellar vermis hypoplasia, (ii) abnormal deep interpeduncular fossa at Isthmus and Pons, and (iii) Horizontally thickened and elongated Superior Cerebellar Peduncles. Diagnostic symptoms include hypotonia, ataxia, abnormal breathing patterns, atypical eye movements, and intellectual disability. Molar tooth sign on axial sections of MRI (magnetic resonance imaging) is a primary diagnostic criterion. Here we report a case of a 10-year-old female intellectually disabled child who was noted to have developmental delay and vision problems soon after 5 to 6 months of birth. The patient was diagnosed with JS based on an MRI brain finding of Molar tooth appearance.

Keywords: Hyperkalemia; Joubert syndrome; Classification; MRI brain

Introduction
The JS is an autosomal recessive disorder first described by Dr. Marie Joubert in 1969 is associated with agenesis of the cerebellar vermis characterised by hypotonia, ataxia, developmental retardation, oculomotor findings, and abnormal respiratory findings [1]. Joubert Syndrome and related disorders (JSRD) is the classification used to describe the disorders w that present with molar tooth signs on MRI brain and extra-central nervous system involvement. JSRD is classified into six phenotypic presentations (Table 1) [2]. JSRDs can be determined through the extra-central nervous system involvement, such as midline facial defects, polydactyly, hepatic fibrosis, nephronophthisis or cystic dysplastic kidneys, and retinopathy- each with its own set of symptoms and signs [3]. The spectrum of JS ranges from a mild form with minimal motor disability and normal mental development to severe motor disability and moderate mental retardation. Treatment of JS is symptomatic and supportive. JS is also associated with other syndromes like Dekaban-Arima syndrome, COACH syndrome, Varadi-Papp syndrome, and Senior-Loken syndrome (Table 1) [3, 4].

Case Report
A 10-year-old intellectually disabled female child presented to the hospital with chief complaints of decreased food intake and 1 - 2 episodes per day of vomiting for the past 15 days. She was drinking only coconut water (on a liquid diet) for many days. They consulted a nearby clinic and took supportive measures for gastric problems. Later, she developed rashes on both her upper and lower limbs (one week back). In view of persistent vomiting and rashes, they again consulted a nearby clinic and started on oral cefixime. On the day of admission, she has 3 episodes of vomiting followed by cardiac arrest in the clinic. She was resuscitated and was further evaluated and started treatment.

History: Second child of second-degree consanguineous marriage. Pregnancy was an uneventful and normal vaginal delivery. The child needed resuscitation for 3 minutes after which she cried. No history of NICU stay. The child was noted to have developmental delay and vision problems soon after 5 - 6 months of birth but was not evaluated for the same. MRI done after 1 year of birth suggested JS (molar tooth appearance) (Figure 1). The patient lost to follow-up after that. She was noted to have polyuria and polydipsia for 3 - 4 months before hospitalization.

She was admitted with the above-mentioned complaints. At the time of admission, the child was drowsy with GCS 12/15 with acidic breathing; immediate VBG was sent from the ER which was suggestive of Hyperkalemia.

Figure 1: MRI brain scan (a) Axial T2 weighted image showing opposition of cerebellar hemispheres in the middle with absent cerebellar vermis and (b) Axial T2 weighted image showing classical molar tooth appearance in the midbrain.
of severe acidosis with hyperkalemia (8.5 mg/dl). Peripheries were cold with feeble peripheral pulsations and BP was (85/45 mm Hg).

**Examination**

The patient was moderately built and nourished. She weighed 38 kg (75th percentile). She was pale and tachycardic with a heart rate of 188 bpm, cold extremities, and feeble peripheral pulsations with a BP of 90/60 mm Hg. She had no icterus/ cyanosis/ clubbing/ edema, Temp - 98.6 ºF, respiration rate - 24/min, SpO2 - 96% at room air, GRBS - 168 bpm, cold extremities, and of 90/60 mm Hg. She had no icterus/ cyanosis/ clubbing/ edema, Temp - 98.6 ºF, respiration rate - 24/min, SpO2 - 96% at room air, GRBS - 168 bpm, cold extremities, and feeble peripheral pulsations and BP was (85/45 mm Hg).

**Discussion**

JS is a rare disorder with a prevalence of 1 in 1,00,000. JS is a rare autosomal recessive disorder characterized by neuropathologic abnormalities of the cerebellum and brainstem, with hypoplasia or aplasia of the vermis [1]. JS is characterised by hyperpnoea episodes, hypotonia, ataxia, renal involvement, and hepatomegaly. The primary features of JS on MRI are (i) aplastic or dysplastic cerebellar vermis, (ii) absence of decussation of fibers in the superior cerebellar peduncles and the cerebellar tracts, (iii) abnormal inferior cerebellar peduncles [8].

The primary features of JS on MRI are (i) aplastic or dysplastic cerebellar vermis, (ii) absence of decussation of fibers in the superior cerebellar peduncles and the cerebellar tracts, (iii) abnormal inferior olivary nucleus, and (iv) dysplastic and heterotrophic cerebellar nuclei. Management of JS is mainly supportive treatment. Rehabilitative strategies are needed for cognitive defects and specific management is required for individual system involvement.

**References**


