

Photoclinic

Hosseininejad SM¹, Safaeian B (M.D)², Hosseini SA (M.D)*³

¹Student of Medicine, Student Research Committee, Golestan University of Medical Sciences, Gorgan, Iran.

²Assistant Professor, Pediatric Nephrologist, Neonatal and Children Health Research Center, Golestan University of Medical Sciences, Gorgan, Iran. ³Assistant Professor, Pediatric Neurologist, Neonatal and Children Health Research Center, Golestan University of Medical Sciences, Gorgan, Iran.



Figure 1



Figure 2

Case presentation: A 20 month-old hypotonic boy with a history of preterm labor at 32 weeks of gestational age, secondary generalized seizures in infancy, hypothyroidism (T4: 2, TSH: 14); motor and speech developmental delay, mild ataxia and renal failure (chronic kidney disease: CKD) was presented to the clinic of Taleghani pediatric hospital in Gorgan, Northern Iran during 2015. In further evaluation, the patient did not have fix and follow ocularly and the head of the left optic nerve was atrophic and congenitally malformed although it seemed the visual prognosis for the right eye was even good with just a slightly pale nerve. The renal ultrasonography showed an increase of general echo which was suggestive for congenital or parenchymal abnormalities. Dialysis was ordered but the patient's family declined. The Image of the infant brain in axial cut using MRI 1.5 Tesla marked molar tooth sign (MTS) (Figure 1). The schematic view is also given (Figure 2). Conservative treatment including Scholl, anti-convulsant solution, (30 cc daily), ferrous sulfate (20 drops), calcium tablets carbonate (2 per day) and phenobarbital (5 mg/kg/bw divided in two parts) was administered for the patient.

Diagnosis: Classic Joubert syndrome subtype: CORS (11q12.2 TMEM216)

Joubert syndrome is a rare autosomal recessive disorder affecting the cerebellum and characterized by abnormal respiratory pattern and eye movements, hypotonia, ataxia, developmental retardation with neuropathological defects of cerebellum and brainstem comprising inherited hypoplasia or aplasia of vermis. This clinical entity is possibly under-reported with a prevalence of less than 1 per 100,000. This syndrome is prototype of congenital vermian hypoplasia. Treatment for Joubert syndrome is symptomatic and supportive. Infant stimulation and physical, occupational, speech and hearing therapy may benefit some patients. Infants with abnormal breathing patterns must be monitored.

* **Corresponding Author:** Hosseini SA (M.D), E-mail: parnianah@yahoo.com

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