Joubert Syndrome: Radiographic Images

Reza Bidaki¹, Azam Ghanei², Mohammad Ebrahim Ghanei³, Ehsan Zarepur⁴

¹- Research Center of Addiction and Behavioral Sciences, Shahid Sadoughi University of Medical Sciences, Yazd, Iran
²- Department of Internal Medicine, School of Medicine, Shahid Sadoughi University of Medical Sciences, Yazd, Iran
³- Department of Radiology, School of Medicine, Shahid Sadoughi University of Medical Sciences, Yazd, Iran
⁴- Student Research Committee, School of Medicine, Shahid Sadoughi University of Medical Sciences, Yazd, Iran

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Corresponding author:
Mohammad Ebrahim Ghanei
Email: dr.ghanei62@yahoo.com

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Introduction

Joubert syndrome (JS) is a rare genetic disorder, first defined by Marie Joubert (1) with agenesis of the vermis of cerebellum presenting with episodic hyperpnoea, abnormal ophthalmic movements, ataxia, and intellectual impairment. A pathognomonic midbrain-hindbrain malformation, the molar tooth sign, or distinctive cerebellar and brainstem malformation on magnetic resonance imaging (MRI) were detected (1-3).

Our patient was a 19-year-old single man with history of mental retardation and developmental delay. He was concluded from normal and term delivery without asphyxia. The physical examination showed behavioral problem, aggression, drooling, protruded and large tongue, silly affect, broad forehead, unibrow, hypotonicity, severe bilateral myopia, nystagmus, and ataxia. The routine lab tests were normal.

Imaging Findings

Axial T2-weighted and FLAIR sequences of MRI showed hypoplastic cerebellar vermis with hypoplasia of the superior cerebellar peduncle resembling the molar tooth sign in the mid-brain (Figures 1 and 2). Bat wing 4th ventricle sign refers to the morphology of the fourth ventricle in the Joubert anomaly and related syndromes.
The absence of a vermis with apposed cerebellar hemispheres give the fourth ventricle an appearance reminiscent of a bat with its wings outstretched (Figures 1 and 2).

MRI of the axial T2-weighted image showed vermian agenesis and extension of the cerebrospinal fluid cleft through it (Figure 3).

MRI sagital T2-weighted image revealed partial agenesis of cerebellum and hypoplasia of the superior cerebellar peduncle (Figure 4).

MRI axial (at the level of post-fossa) T2-weighted image revealed hyperintensity of basal cisterns communicating with the fourth ventricle suggestive of vermian agenesis (Figures 2). Vermian dysgenesis was more depisite on coronal sequensis (Figure 5).

Renal ultrasound showed no abnormality.
Joubert syndrome

**Conflict of Interests**
Authors have no conflict of interests.

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**References**