Clinical Image

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Joubert Syndrome: Radiographic Images

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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.

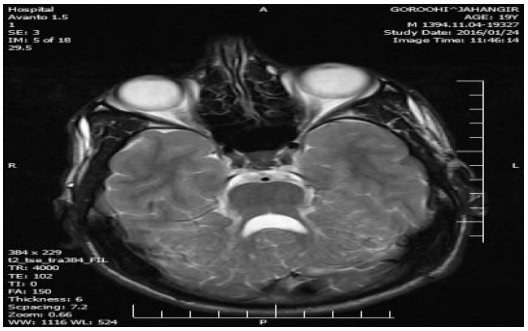

Figure 1. Axial T2-weighted magnetic resonance imaging (MRI) showing molar tooth sign in the mid-brain

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).

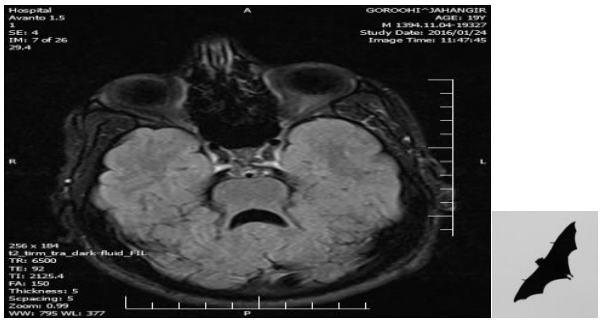

Figure 2. Magnetic resonance imaging of the axial T2-weighted image showing same molar tooth appearance of mid-brain and bat wing appearance of 4th ventricle

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

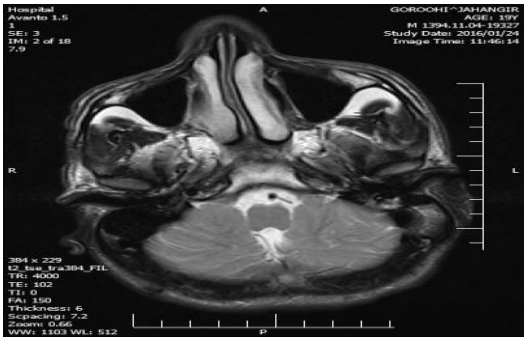

Figure 3. Magnetic resonance imaging of the axial T2-weighted image showing vermian agenesis and extension of the cerebrospinal fluid cleft through it

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

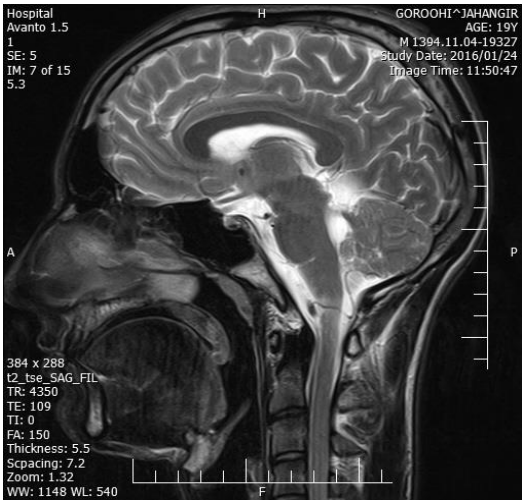

Figure 4. Sagital T2 demonstrate absent of vermis, elevation of 4th ventricle with normal appearing corpus callosum

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).

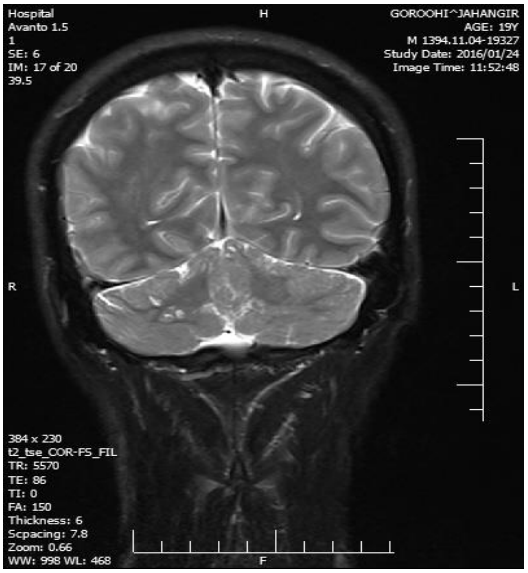

Figure 5. Vermian dysgenesis was more depisite on coronal T2 sequensis

Renal ultrasound showed no abnormality.

Conflict of Interests

Authors have no conflict of interests.

Acknowledgments

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