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RESEARCH ARTICLE

IMAGING FINDINGS IN JOUBERT SYNDROME

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ABSTRACT

Joubert syndrome is a rare autosomal recessive disorder characterized by abnormal respiratory pattern and eye movements, hypotonia, ataxia, developmental retardation with neuropathologic abnormalities of cerebellum and brainstem including inherited hypoplasia or aplasia of vermis. The reported prevalence is less than 1 in 100,000.

Cross-sectional axial imaging demonstrates isolated abnormality of the cerebellum, consisting of:

- small dysplastic or aplastic cerebellar vermis
- absence of fibre decussation in the superior cerebellar peduncles and pyramidal tracts , which can be assessed by diffusion tensor imaging
- abnormal inferior olivary nucleus
- dysplasia and heterotopia of cerebellar nuclei

The posterior fossa typically shows a bat wing 4th ventricle and prominent thickened elongated superior cerebellar peduncles giving characteristic molar tooth sign like appearance.

In a minority of cases minor lateral ventriculomegaly (6 - 20% of cases), and corpuscallosal dysgenesis (6-10% of cases) is also present.

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INTRODUCTION

In 1969, Dr. Marie Joubert and colleagues first described four siblings with cognitive impairment, ataxia, episodic tachypnea, eye movement abnormalities, and cerebellar vermis agenesis in a large French-Canadian family with consanguinity traced 11 generations to a common ancestor (Marie Joubert *et al.,* 1969). The incidence of Jouberts syndrome has been estimated as between 1/80,000 and 1/100,000 live births (Harjinder Gill *et al.,* 2011). Joberts syndrome is an autosomal recessive neurodevelopmental disorder (Edwin *et al.,* 2000). Many children with Joubert syndrome exhibit dysmorphic facial features that include broad forehead, arched eyebrows, eyelid ptosis, wide-spaced eyes, open mouth configuration, and facial hypotonia (Melissa and Parisi, 2009). Joubert syndrome also have other clinical manifestations involving the CNS (occipital encephalocele, corpus callosal agenesis), eyes (coloboma, retinal dystrophy, nystagmus, oculomotor apraxia), kidneys (nephronophthisis, cystic dysplasia), liver (hepatic fibrosis), and limbs (polydactyly) (Harjinder Gill *et al.,* 2011).

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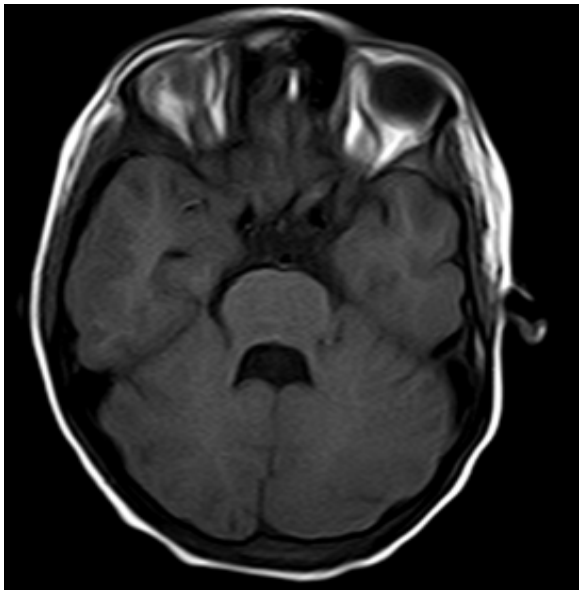
MATERIAL AND METHODS

This is a retrospective study. MRI scan of the patient included in this study was performed using Philips Achieva 1.5 T MRI machine at department of radiology Shri B.M Patil Medical College, Hospital and Research Centre, Bijapur. The standard MRI brain protocol at our institution includes. T1W axial and sagittal images, T2W coronal and axial images, T2 FLAIR axial images, DWI and gradient sequence.

CASE REPORT

A 6 year old male child was brought to our hospital by his father with the chief complaints of delayed milestones, abnormal eye moments and abnormal respirations. Boy attained sitting, walking, and speech milestones at 1, 2, and 4 years respectively and they are delayed. The birth history consisted of delivery at full term gestation by vaginal route at tertiary care hospital. Child did not cry immediately after birth, cried after rescuitation. Child was admitted in NICU for irregular respiration. Routine hematological, urine examination, 2d ECHO, and thyroid profile were unremarkable.



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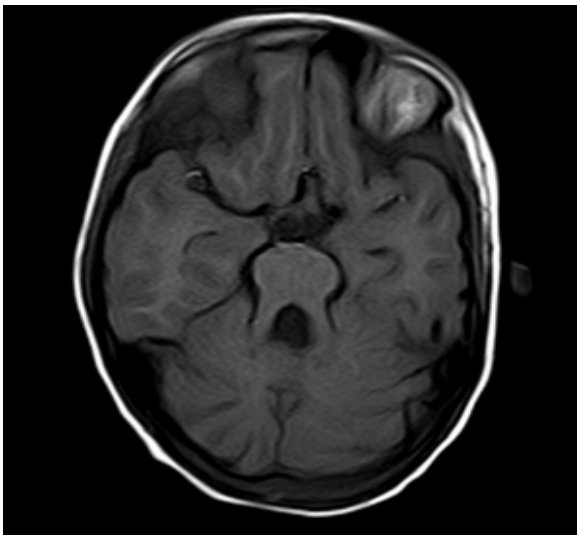

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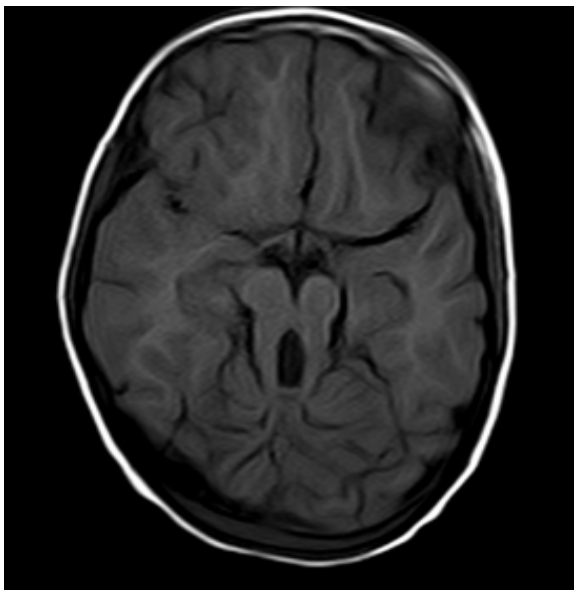

C

Images A, B, C (axial T1WI) show aplasia of vermis with molar tooth appearance and batwing appearance of IV ventricle

MRI scan done revealed aplasia of vermis, thickened and elongated bilateral superior cerebellar peduncles which showed a more horizontal course between the midbrain and cerebellum with decreased antero-posterior diameter of midbrain and deeper interpeduncular cistern - molar tooth appearance. Batwing appearance of IV ventricle was also seen.

DISCUSSION

JS was originally described in 4 siblings with vermian agenesis presenting with episodic hyperpnoea, oculomotor apraxia, ataxia, and cognitive impairment (Joubert *et al.,* 1969). The primary MR imaging features of Joubert syndrome are (1) thining of the isthmus with widened interpeduncular fossa; (2) thickened superior cerebellar peduncles; (3) hypoplasia of the vermis with fourth ventricular deformity; (4) rostral shift of fastigium and (5) sagittal vermian cleft due to incomplete fusion of the two halves of vermis. These findings are present in almost all cases. In one study,the molar tooth sign was identified in 85% of patients with Joubert syndrome, and it has been reported to be pathognomonic of this disorder. The absence of a normal vermis creates a midline cleft between the two normal appearing cerebellar hemispheres, which results in a characteristic bat wing (open umbrella) appearance of the fourth ventricle on transverse CT and MR images (Patel and Barkovich, 2002). A classification into two groups has been proposed on the basis of the presence or absence of retinal dystrophy (Saraiva and Baraitser, 1992). In the group with retinal dystrophy, there is a high prevalence of multicystic renal disease, and there appears to be a worse prognosis in terms of survival than in the group without retinal dystrophy 3 The exact location of the defective gene has not been established, although both the X chromosome and the nephronophthisis 1 region on chromosome 2 have been excluded (Saraiva and Baraitser, 1992; Hildebrandt *et al.,* 1998).

The importance of recognizing Joubert syndrome is related to the outcome, its autosomal recessive trait, and the potential complications that may develop. A follow-up study in 19 children with Joubert syndrome showed that three children died before 3 years of age, whereas the remaining children showed neuromotor developmental retardation and various levels of reduced cognitive development (King *et al.,* 1984). Retinal dysplasia is correlated highly with renal cystic disease and seems to carry a worse prognosis in terms of survival (Saraiva and Baraitser, 1992; Steinlin *et al.,* 1997). Retinal dysplasia is often difficult to diagnose early; hence, regular ocular screening should be performed. In patients with retinal anomalies, it also is advisable to monitor renal function and perform ultrasonography (US) of the kidneys to detect cystic renal disease (Habre *et al.,* 1997). The diagnosis is important for future procedures that require anesthesia (Habre *et al.,* 1997). Patients with Joubert syndrome are extremely sensitive to the respiratory depressant effects of anesthetic agents, such as opioids, and nitrous oxide. Therefore, these agents should be avoided and close perioperative respiratory monitoring is essential (Habre *et al.,* 1997).

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