Case Report

Joubert Syndrome

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Abstract

Joubert syndrome is a very rare condition seen in our country. Herein, we report a case of Joubert syndrome in a one year four months old, male baby from a consanguineous marriage presenting with delayed developmental milestone, hypotonia, abnormal respiratory pattern and nystagmus . Cranial MRI shows ''Molar Tooth Sign''.

Keywords

joubert syndrome, vermis

Introduction

Joubert syndrome (JS) is a very rare, autosomal recessive condition, first described by Joubert in 1969. It is characterized by agenesis of cerebellar vermis, hypotonia and abnormal respiratory pattern and abnormal eye movement. Here, we present a case of Joubert Syndrome in a one year four months old male baby from Burdwan. This syndrome is never reported from eastern region of our country.

Case Report

A one year four month, male infant was admitted with abnormal limb and head movement. There was also history of nystagmoid eye movements, irregular breathing pattern with intermittent rapid breathing with opened mouth and protruded tongue. Mother also gives history of feeding difficulties and frequent chest infections.

In past history, the child delivered at term, birth weight 3 kg, had mild birth asphyxia. No convulsion ever seen. Mother also noticed weak cry and poor muscle tone. There is a gross motor delay in milestone. The symptoms were progressively increasing.

Prenatal history was uneventful. No similar illness in any siblings in the family. Immunization was incomplete.

On physical examination, anthropometric measurements including head circumference were within normal limits. No facial or limb deformity. Neurologically, the child had generalized hypotonia, reflexes were sluggish and plantar was equivocal. There was an abnormal head and limb movement. Mouth was opened with protruded tongue. Breathing pattern was abnormal with hyperapnea and tachypnea interspersed with normal pattern. Heart sounds were normal. No organomegaly seen.

Ocular examinations revealed bilateral divergent squint, restricted upward gaze and nystagmoid movement. Retinoscopy revealed refractory error of both eyes. Ophthalmoscopic examination was normal.

Investigation

Complete blood count, renal function test and Liver function test were normal.

Imaging Findings: MRI shows – hypoplastic cerebellar vermis with hypoplasia of the superior cerebellar peduncle resembling the ''Molar Tooth Sign'' in mid-brain (see **Fig. 1**). Hypoxia related periventricular white matter changes also seen.

Renal imaging showed no cystic abnormality.

Discussion

Joubert Syndrome is inherited as an autosomal recessive

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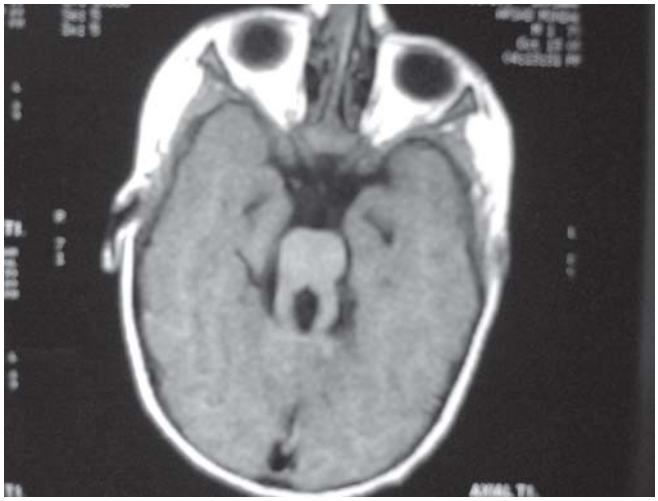

Fig. 1

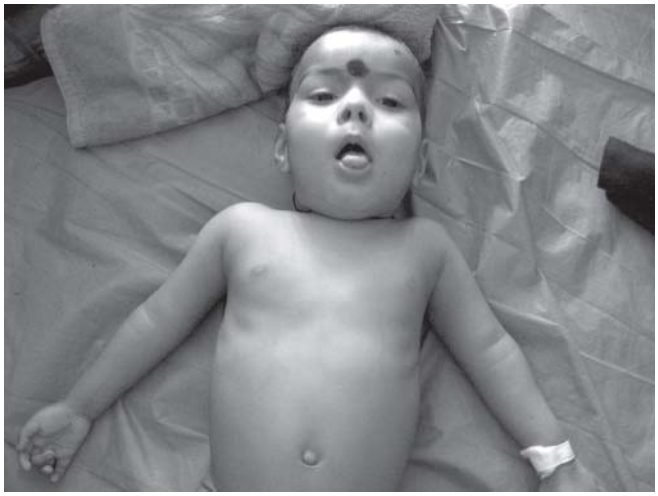

Fig. 2

condition. Genetic testing is not currently available but Pre-natal testing is possible for this condition. This syndrome was named after the French neurologist, Joubert, who first described the clinical findings[1–3]. The clinical features of Joubert syndrome consist of abnormal eye movements with nystagmus and inability of smooth pursuit of a moving object, episodes of hyperapnea and apnea, and delayed generalized motor development[4–7]. The syndrome is associated with retinal coloboma and retinal dystrophy in approximately 50%[3,5,6,8,9], tongue protrusion in more than 30%[3,5], multicystic kidney disease in 30% of patients with retinal dysplastic features[3,9], and polydactyly in up to 15% of patients[3–7].

In the group with retinal dystrophy, there is a high prevalence of multicystic renal disease, and there appears to be a worse prognosis[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[3,10].

The main imaging findings are partial or complete absence of the vermis, hypoplastic cerebellar peduncles, and fourth ventricular deformity[6,11]. The cerebellar hemispheres are usually normal. The cerebrum is usually not affected. Partial or complete absence of the cerebellar vermis has been described as an isolated anomaly or as part of Dandy-Walker syndrome, Down syndrome, or Joubert syndrome[12,13]. The combination of "molar tooth sign"[4] and severe hypoplasia of the vermis that gives a bat-wing appearance to the fourth ventricle[13] is highly suggestive of Joubert syndrome.

The importance of recognizing Joubert syndrome is related to the outcome, its autosomal recessive trait, and the potential complications that may develop. Retinal dysplasia is correlated highly with renal cystic disease and seems to carry a worse prognosis in terms of survival[3,7]. Furthermore, once a diagnosis of Joubert syndrome is made in one neonate, the diagnosis of Joubert syndrome can be made antenatally US during a subsequent pregnancy[13].

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