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Case Report

Familial Joubert syndrome in two siblings

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ABSTRACT

Joubert syndrome is a rare autosomal recessive neurological disorder with episodic hyperpnoea, developmental delay, abnormal eye movements and gait abnormalities. We describe a case of 6 months old baby boy who presented with hyperpnoea and developmental delay. MRI was performed which showed the characteristic molar tooth sign. (Rawal Med J 2012;37:56-57).

Keywords

Familial Joubert syndrome, molar tooth sign, MRI.

INTRODUCTION

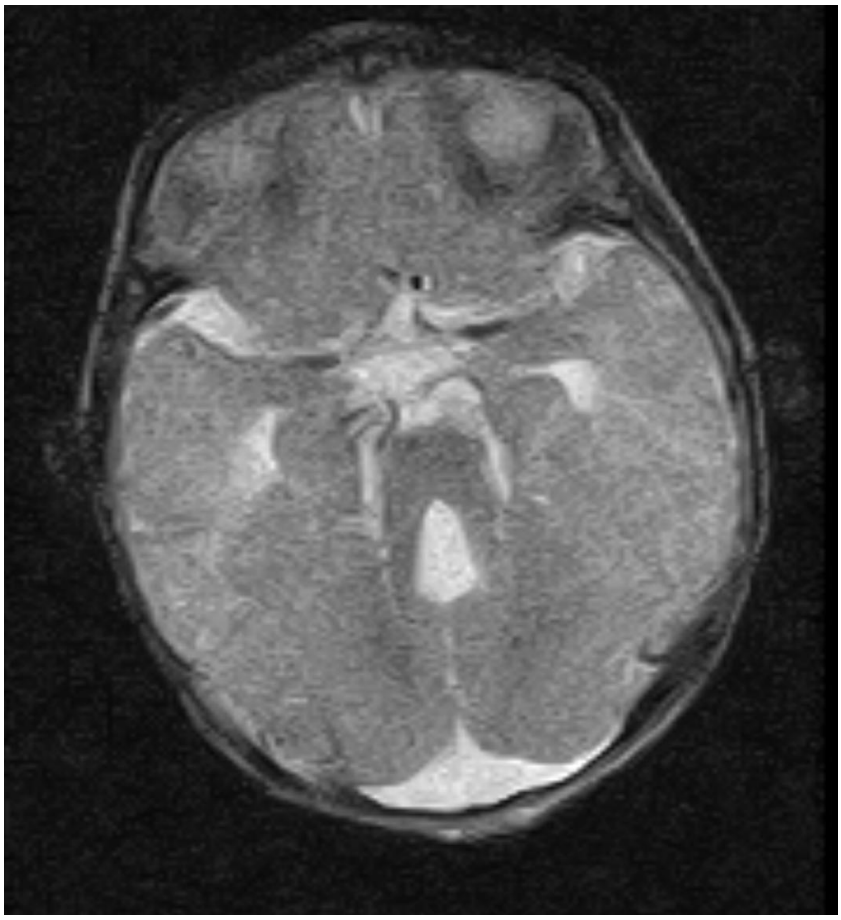
Joubert syndrome (JS) is a rare neuroradiological disorder with characteristic clinical and radiological findings. Magnetic resonance imaging findings include hypoplastic or aplastic cerebellar vermis, thickened and maloriented superior cerebellar peduncles, deep interpeduncular fossa and batwing shaped fourth ventricle.[1] These findings usually give a characteristic appearance of a molar tooth on axial MR images which in combination

with clinical findings of abnormal respiratory pattern and developmental delay are diagnostic for Joubert syndrome.

CASE PRESENTATION

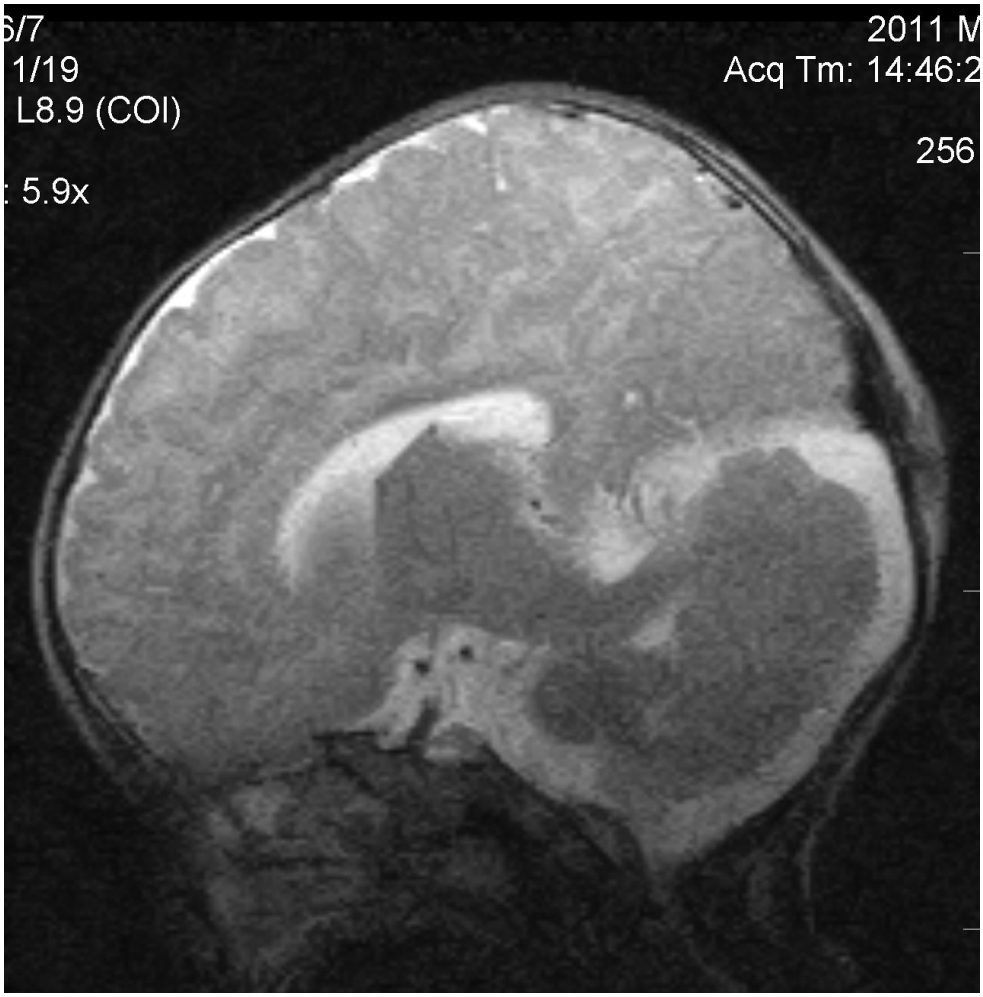
A six months old baby boy presented to our Department with history of a small swelling on the back of his head, abnormal rapid and deep breathing with inability to hold the neck. He was advised to have MRI brain by the neurosurgeon which showed thickened superior cerebral peduncle giving molar tooth appearance (Fig 1) with hypoplastic cerebellar vermis (Fig 2). Additional findings of mega cisterna magna was also present.

Fig 1. Axial T2 weighted image showing the characteristic molar tooth appearance.



Based on the clinical presentation as well as MRI findings he was diagnosed as having JS. Further history revealed that baby has four siblings, all from consanguinous marriage.

Fig 2. Sagittal T2 weighted image showing abnormally thickened and horizontally placed superior cerebellar peduncle.



One of his elder sisters who is around three years old was completely bed bound and could just turn to right or left while lying. She also had an MRI at the age of six months at our institution. When we followed her records, we found that she also presented with developmental delay and abnormal eye movements at the time of MRI. She was also

diagnosed as having JS based on her clinical presentation and MRI findings of hypoplastic vermis, deep interpeduncular fossa and molar tooth appearance of superior cerebellar peduncles. Rest of the siblings were normal.

DISCUSSION

Joubert syndrome also known as Joubert Bolthauser syndrome is a rare familial syndrome characterized by complex mid and hind brain malformations as well as clinical findings of rapid breathing, abnormal jerky movements of eye, developmental delay and dysmorphic facies. It is equally represented in both genders. It is transmitted as an autosomal recessive trait with mutations reported in many genes. A French neurologist named Marie Joubert in 1969 reported this syndrome in a family of four siblings who presented with ataxia, abnormal eye movements, mental retardation and episodic hyperventilation with cerebellar vermis agenesis.[2] Name of Joubert syndrome was given many years later and Raynes et al in 1999 reported Joubert syndrome in three sisters with two of them being monozygotic twins.[3]

MRI is the imaging of choice and findings usually include pathognomonic molar tooth sign. The absence of a normal appearing vermis gives rise to a midline cleft which gives rise to characteristic appearance of bat wing shaped fourth ventricle. Diffusion tensor imaging and fiber tractography show elongated and thickened superior cerebellar peduncles with a horizontal configuration.[1] Histopathological findings include dysplasia of olivary and paraolivary nuclei with reduction of neurons of pons. Distortion of dentate nuclei with anomalies of dorsal column nuclei and an absence of decussation of pyramids and superior cerebellar peduncles have been reported.[4]

Joubert syndrome and its related disorders all show the characteristic molar tooth sign on MRI. These have been given a new classification based on presence or absence of retinal, renal and hepatic findings in addition to characteristic findings of abnormal breathing pattern and eye movements, developmental delay and ataxia. In pure Joubert syndrome, there is no retinal, renal or liver involvement. Other types include JS with retinal defects, JS with renal defects, JS with oculorenal defects, JS with hepatic defects and JS with oro-facio-digital defects.[5]

Prenatal diagnosis can be done through chorionic villous sampling at around 11 weeks and antenatal ultrasound at 20 weeks of gestation to detect vermian hypoplasia and enlargement of 4th ventricle and by Molar tooth sign in fetal brain by MRI.[5] Genetic counseling is important once a diagnosis has been made in one neonate as this is an autosomal recessive disorder. Diagnosing JS as early as possible is important as these patients are highly sensitive to anaesthetics such as nitrous oxide and opioids which are respiratory depressants. Treatment of Joubert syndrome is mainly supportive and includes physical, occupational and speech therapy. In summary, JS is a rare familial disorder in which active search should be performed for this disease in siblings of patient as it has significant social and medical implications. MRI is considered the gold standard for diagnosis.

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