Case Report

Joubert Syndrome: A Case Report

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ABSTRACT

Joubert Syndrome (JS) is a very rare autosomal recessive neurodevelopmental disorder. It is characterized by cerebellar vermis hypoplasia, hypotonia and abnormal breathing pattern or abnormal eye movements. We report a case of 13 years old male child with seizures, abnormal eye movements, developmental delay and ataxia. On radiological evaluation MRI of brain revealed "molar tooth sign" and "bat wing appearance" of fourth ventricle.

Keywords: Joubert syndrome, vermian hypoplasia, bat wing appearance, molar tooth sign

INTRODUCTION

Joubert syndrome was first described in 1969, by Dr Marie Joubert and colleagues in four siblings with cognitive impairment, ataxia, episodic tachypnea, eye abnormalities and cerebellar vermis agenesis in a large French-Canadian family with consanguinity traced upto 11 generations to a common ancestor.[1] They usually have slowed generalized motor activity because of psychomotor retardation. Respiratory irregularities like rapid respirations may occur during infancy. The incidence of Joubert syndrome has been estimated between 1 in 80,000 to 1 in 100,000 live births.[2] Joubert Syndrome is a rare autosomal recessive neurodevelopmental disorder.[3] characterized by absence of dysmorphic signs and variability of clinical presentations can sometime delay the diagnosis. Joubert syndrome may also have involvement of the central nervous system (occipital encephalocele, corpus callosal agenesis), eyes (coloboma, retinal dystrophy, oculomotor apraxia), kidneys (nephrocalcinosis, cystic dysplasia) and liver (hepatic fibrosis).

CASE REPORT

A 13 year old male child with seizures since one week was admitted in Chalmeda Anand Rao Institute of Medical Sciences which is a teritiary care hospital in North Telangana. There is a history of four episodes of generalized seizures and inability to fix gaze, weakness and ataxia. There is no history of head injury, ear discharge and tuberculous contact. The child was product of 3rd degree consanguineous marriage. Birth history was uneventful. It was a full term, normal vaginal delivery, cried immediately after birth, birth weight of 3kgs. He was on breast feeding and complementary feeding was started around 8-9 months of age. The developmental history suggestive of there is a global developmental delay. The boy was attained sitting, walking and speech milestones at one, two and four years

Figure 1: Child with ataxic gait

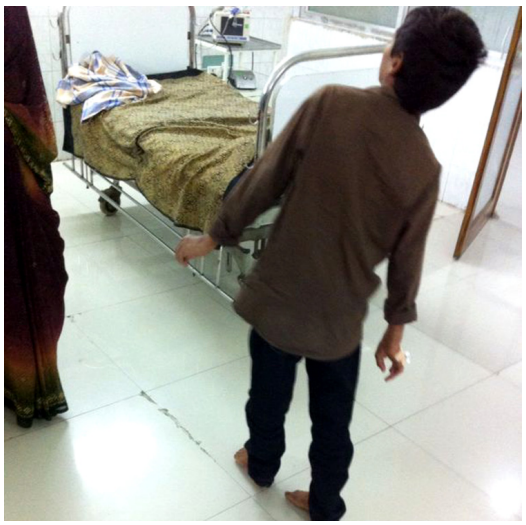

Figure 2: Child unable to maintain balance with closed eyes

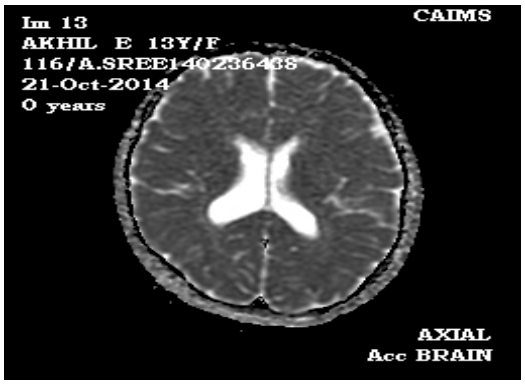

Figure 3: MRI scan of brain showing molar tooth sign

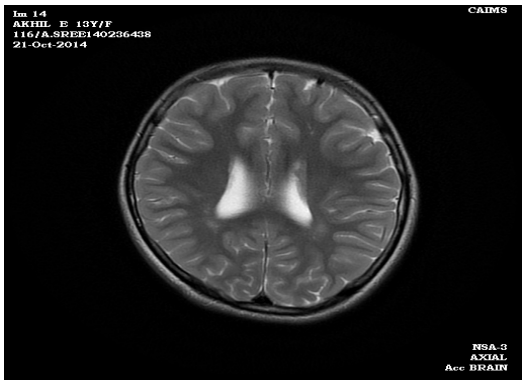

Figure 4: MRI scan of brain showing bat wing appearance

respectively. He has abnormal eye movements since few months old. On general physical examination child was malnourished. Vitals are stable. child was conscious, able to speak few words. The child also has dysarthria. Cranial nerves examination was normal, except rotatory nystagmus. Motor system examination revealed that child is having hypotonia, decreased tendon reflexes. He was ataxic in his gait could not maintain his balance with closed eyes (Figure 1, 2). Hematological and biochemical investigations are normal. Electro encephalogram revealed generalized seizure disorder. On radiological examination MRI scan of brain was showing brain stem thinning and atrophy of posterior half of midbrain i.e "molar tooth sign", and bilateral superior and middle cerebellar peduncles with prominent bilateral lateral ventricles i.e "Bat wing appearance".suggestive of Joubert Syndrome (Fig-3, 4). The seizures were controlled with phenytoin sodium and child was discharged on maintenance dose of phenytoin sodium. Advised to come for follow up.

DISCUSSION

Joubert syndrome is a rare genetic disorder with autosomal recessive inheritance characterized by (a) Hypoplasia to total aplasia of cerebellar vermis and

dilatation of fourth ventricle in typical Bat wing configuration (b) Intellectual impairment or developmental delay of variable degree (c) Hypotonia in infancy (d) Irregular breathing pattern in infancy or abnormal eye movements.[4] Central nervous system malformations include hydrocephalus, cystic enlargement of posterior fossa, abnormalities of corpus callosum, white matter cyst and absence of pituitary gland, abnormal migration defects mainly periventricular nodular heterotopia in polymicrogyria.[5]

The term "Joubert Syndrome and Related Disorders" (JSRD) refers to those individuals with Joubert Syndrome also having additional clinical findings.[6] JSRD are categorized into six phenotypic subgroups (i). Pure JS , (ii). JS with ocular defects, (iii). JS with renal defects, (iv). JS with occulo-renal defects, (v). JS with hepatic defects and (vi). JS with oro-facio-digital defects.[7]

The hallmark imaging features of Joubert Syndrome are dysgenesis of the isthmus (part of brain stem between pons and inferior colliculus), thickening of superior cerebellar peduncles, hypoplasia of the vermis characterized by enlarged fourth ventricle, incomplete fusion of the halves of the vermis on axial or coronal MRI planes. Hypogenesis of the vermis results in a triangular shaped mid fourth ventricle and Bat wing shaped fouth ventricle.[8] Initiation of periodic comprehensive assessments and a program of interventions including special education, physical , occupational and speech therapy have shown significant benefits in attainment of developmental milestones in children with JSRD.[4]

CONCLUSION

JS is a rare autosomal recessive disorder involving the cerebellum. Most of the cases are present with abnormal eye movements, irregular respiration, developmental delay, hypotonia and gait disorders.

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