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CASE REPORT OF JOUBERT SYNDROME.

Radiodiagnosis

Dr Danquale Vance Kynshikhar Resident, Dept. of Radiodiagnosis, IGMC, Shimla, HP, India

Dr Sanjay Kumar* Resident, Dept. of Radiodiagnosis, IGMC, Shimla, HP, India. *Corresponding Author

Dr Suresh Thakur Associate Professor, Dept. of Radiodiagnosis, IGMC, Shimla, HP, India.

Dr Anupam Jhobta Professor, Dept. of Radiodiagnosis, IGMC, Shimla, HP, India.

ABSTRACT

Joubert Syndrome is a rare autosomal recessive neurodevelopment disorder characterized by malformation of the mid and the hindbrain. It is characterized mainly by hypotonia, ataxia, abnormal eye movements including ocular apraxia and nystagmus, abnormal breathing patterns. Joubert Syndrome and Related Disorders(JSRD) is a wider spectrum of Joubert Syndrome based on the additional involvement of kidneys, liver and eyes. We report a 14 years old female patient who presented with global development delay, difficulty in walking, abnormal eye movement. Neurological examination shows hypotonia, ataxia and nystagmus. Magnetic Resonance Imaging revealed the characteristic molar tooth sign and batwing appearance of the fourth ventricle.

KEYWORDS

Joubert Syndrome(JS), Joubert Syndrome-Related Disorders(JSRD), Molar Tooth Sign(MTS).

INTRODUCTION:

Joubert syndrome (JS) is an autosomal recessive neurological disorder named after Marie Joubert in 1969[1]. It is characterized by brain stem and cerebellar malformations and different degree of involvement of other systems other than the nervous systems[2]. Clinically patients usually presents with oculomotor apraxia, hypotonia, ataxia, episodic hyperpnea with apnea, and developmental delay[3,4,5]. Classic Joubert syndrome is characterized by brainstem and cerebellar malformation called the molar tooth sign (MTS) on imaging, hypotonia, developmental delay, and one of the following: abnormal breathing pattern or abnormal eye movement[1,2]. JS may be associated with multiorgan involvement including the eye, renal, liver and polydactyly, and the term Joubert syndrome-related disorders (JSRD) was used. The term JSRD refer to those conditions having Molar Tooth Sign on MRI imaging, but also involving other distinctive organ systems[1,2]. The Molar Tooth Sign is now considered as an essential criteria for the diagnosis of JSRD[1].

CASE REPORT

A 4 year old boy presented to the paediatric out patient department with complaints of development delay, abnormal eye movements, difficulty in walking. There was also history of episodes of abnormal breathing during his first few months of life. He was delivered via normal vaginal delivery with birth weight of 2.5kg. There was no history of any consanguinity in the family or any similar cases in the family. On physical examination of the patient revealed that the patient was having bilateral nystagmus with abnormal cerebellar functions. Laboratory investigations including hemogram, blood sugar, liver function tests, renal function tests and urine examination were all within normal limits. Magnetic resonance imaging (MRI) of the brain of the patient was done. On MRI images, the interpeduncular fossa was deep and the superior cerebellar peduncles were thick and elongated giving 'Molar Tooth Sign' appearance. The cerebellar vermis was dysplastic. The fourth ventricle was dilated with a deformed roof and floor with a midline sagittal cleft separating the two cerebellar hemispheres giving the Batwing Sign/Umbrella Sign. On coronal images, the cerebellar hemispheres appeared in close approximation in the midline separated by a sagittal cleft due to agenesis of the cerebellar vermis giving the 'Buttock Sign'. Based on the clinical and the MRI findings, a diagnosis of Joubert syndrome was given.

Figure 1. MRI T1 WI axial section showing deep interpeduncular fossa and narrow isthmus (yellow arrow) with thick and elongated superior cerebellar peduncles (red arrows) giving 'Molar Tooth Sign' appearance.

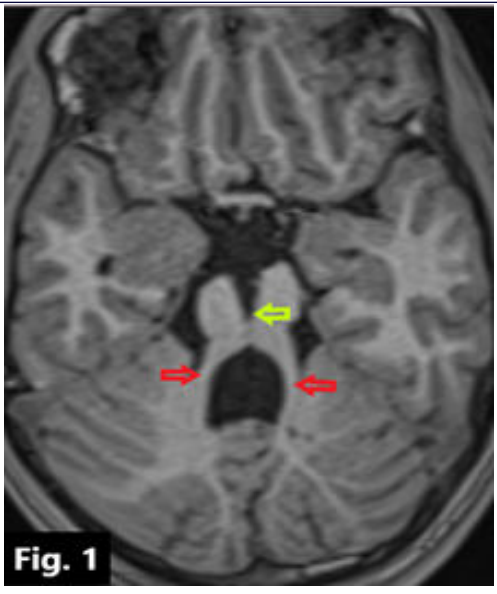

Figure 2(a). MRI T2 WI axial section at the level of the pons showing dilatation of the fourth ventricle (red asterisk) with a midline sagittal cleft (red arrow) separating the two cerebellar hemispheres giving the 'Umbrella Sign'.

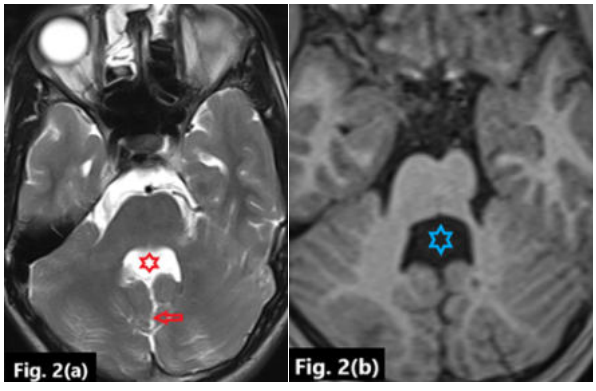

Figure 2(b). MRI T1 WI axial section showing dilatation of the fourth ventricle with deformed roof and floor giving the shape of a batwing, 'Batwing Sign'.

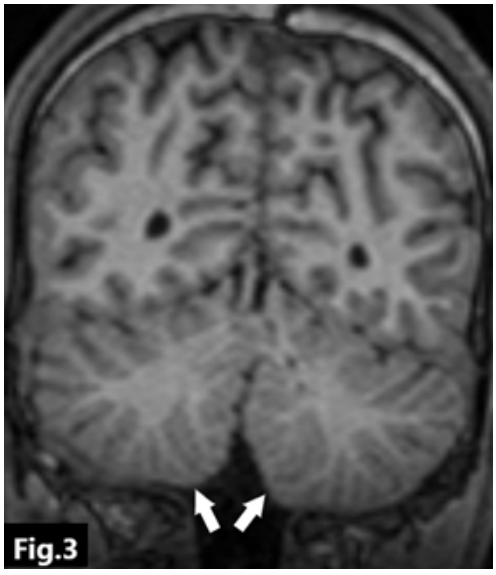

Figure 3. MRI T1 WI coronal section showing the cerebellar hemispheres (white arrows) are in close approximation in the midline separated by a sagittal cleft due to agenesis of the cerebellar vermis giving the 'Buttock Sign'.

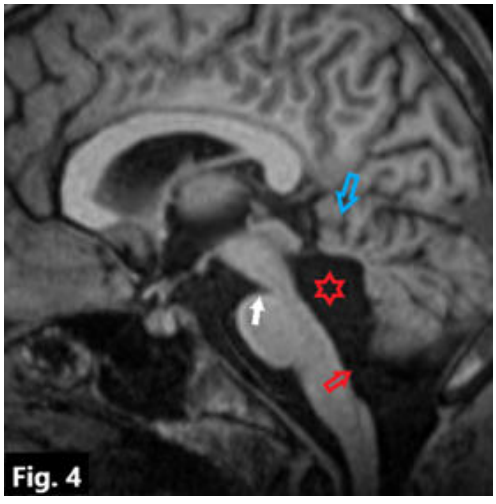

Figure 4. MRI T1 WI sagittal section showing narrow ponto-mesencephalic junction(white arrow) with dilated fourth ventricle and a deformed roof (red asterisk). Cerebellar vermis appears dysplastic (blue arrow). Foramen of Magendie is wide opening into the cisterna magna (red arrow).

DISCUSSION:

Joubert Syndrome was first described in the year 1969 in four siblings with clinical features of episodic rapid breathing, abnormal eye movements, ataxia and mental retardation with malformations of the cerebellar vermis[6]. Joubert Syndrome is an autosomal recessive disorder and has an estimated incidence of 1 in 100000 live birth[7]. By 2009, approximately 200 cases have been reported worldwide[8].

The clinical manifestations usually start in the neonatal period, however most cases are often diagnosed months and years after birth[9]. Patients usually presents with abnormal breathing patterns, abnormal eye movement, ataxia and mental retardation. The essential clinical features for the diagnosis of Joubert syndrome are hypotonia, congenital ataxia, developmental delay and at least one of the following: neonatal respiratory abnormalities and abnormal ocular movements including nystagmus and oculomotor apraxia[10,11]. In our case the patient was 4 years old and presented with complaints of development delay, abnormal eye movements, difficulty in walking and history of episodes of abnormal breathing during his first few months of life.

The term 'JS and related disorders' (JSRD) refers to conditions having Molar Tooth Sign on MRI imaging but also involving other distinctive organ systems other than the central nervous system. JSRD are classified into 6 sub-groups based on organs involvement and genotype-phenotype correlation. These includes i)pure JS, ii)JS with ocular defect, iii)JS with renal defect, iv)JS with oculorenal defect, v)JS with hepatic defect, vi)JS with oro-facio-digital defects[1,2].

MRI of the brain plays a very important role in the diagnosis of Joubert Syndrome and JSRD. The Molar Tooth Sign on CT Scan and MRI brain is an important finding in the diagnosis of Joubert Syndrome[1,2].

It is important to differentiate between Joubert Syndrome and rhombencephalosynapsis as there are few similarities clinically and radiologically. However, correct diagnosis can be achieved confidently on the basis of the findings on CT/MRI[12].

There is lack of decussation of fibres of superior cerebellar peduncle leading to decrease antero-posterior diameter of the midbrain and dysplastic ponto-mesencephalic junction. The Molar Tooth Sign results from a combination of hypoplasia of the cerebellar vermis, deep interpeduncular fossa, enlargement of the superior cerebellar peduncles and distortion of the fourth ventricle[10,13]. Another important sign for the diagnosis of Joubert Syndrome is Batwing sign or the umbrella sign. This sign results due to agenesis/hypogenesis of the cerebellar vermis resulting to dilatation of the fourth ventricle and appearance of a cleft in the midline between the two cerebellar hemispheres. This results in Bat Wing appearance or Umbrella Sign on axial images on MRI and CT scan images[5,6]. Buttock sign is also seen on coronal images due to the absence of the vermian lobe resulting in separation of the cerebellar hemispheres by a midline cleft[5]. Another sign which was reported in the year 2014 is the shepherd's crook sign[14]. On sagittal images of the posterior fossa, the Shepherd's Crook sign is seen where the brain stem and the pons form the shaft and the abnormal superior cerebellar peduncles and the cerebellar hemisphere form the arc of the crook[14]. Other findings including corpus callosum dysgenesis, absent septum pellucidum and mild enlargement of the lateral ventricles are usually associated with Joubert Syndrome [15].

Joubert Syndrome usually have poor prognosis[12]. Symptomatic treatment and supportive care of patients of Joubert Syndrome is important due to cognitive disability and breathing difficulty in these patients. The degree of brainstem involvement and other organs involvement are important prognostic factors[16]. Hence patients with Joubert Syndrome should be evaluated carefully for any other system involvement and should be treated accordingly.

CONCLUSION

Joubert Syndrome should be considered as a possible diagnosis in all patients with developmental delay, breathing abnormalities, nystagmus and hypotonia. Certain important signs on imaging for the diagnosis of Joubert Syndrome include Molar Tooth Sign, Umbrella sign/Batwing shaped fourth ventricle and the Buttock sign. Early diagnosis of Joubert Syndrome is possible considering the clinical presentations with the imaging findings and appropriate rehabilitation and supportive care can be started as soon as possible.

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