IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 21, Issue 11 Ser.6 (November. 2022), PP 45-47
www.iosrjournals.org

"A bat wing and a molar tooth in brain"

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Abstract:-
Joubert syndrome (JS) is a very rare autosomal-recessive condition and is characterised by agenesis of cetrebellar vermis, abnormal eye moments with nystagmus, episodes of hyperapnea and apnea, delayed generalised motor development, retinal coloboma and dystrophy and multi cystic kidney disease. Prenatal diagnosis by ultrasonography and antenatal magnetic resonance imaging (MRI) we had diagnosed a case of JS in a 11 year old male child.

Date of Submission: 10-11-2022 Date of Acceptance: 25-11-2022

I. Background:-

Joubert Syndrome is a rare genetic development disorder, is named after french neurologist Marie Joubertwho described the condition in 1969.It is characterized by cerebellar vermis and brainstem are underdeveloped and malformed.This can lead to neonatal breathing dysregulation developmental delay, intellectual disability, hypotonia, ataxia and nystagmus.

II. Case report:-

A eleven - year-old male child presented to neurology department with history of delayed milestones, mental retardation, generalised hypotonia, abnormal movements of limbs, nystagmus, abnormal breathing pattern, seizures and recurrent chest infections.

He is a product of consanguineous marriage and normal vaginal delivery at term in the hospital and history of birth asphyxia. In early life he had feeding difficulties and abnormal moments of eyes and limbs were noted by his mother. Rapid and abnormal breathing pattern in the respiration was noticed by his father and there is a history of recurrent chest infections. There is an episode of abnormal breathing with opening of mouth and protrusion of tongue is noted. His cry was weak. Hearing was also not present i,e, no response to sound. Similar complaints were noticed after few months and are increasing. He has delayed motor and mental development / milestones. Immunization was complete and no other complaints were noted in the siblings in the family.

On physical examination the patient showed hypotonia, with abnormal and rapid breathing moments, nystagmus. No morphological abnormalities noted. No organomegaly noted. Ultrasound abdomen was normal and biochemical tests and complete urine examination were within normal limits.

MRI brain examination showed Axial T1- and T2 – weighted magnetic resonance imaging (MRI) images showed clefted vermis, tickened superior cerebellar peduncles (molar tooth sign),elongated midbrain and bat wing appearance of fourth ventricle. No other associated brain anamolies such as cortical dysplasias, greymatter heterotopias, ventriculomegaly and corpus callosum agenesis were noted.

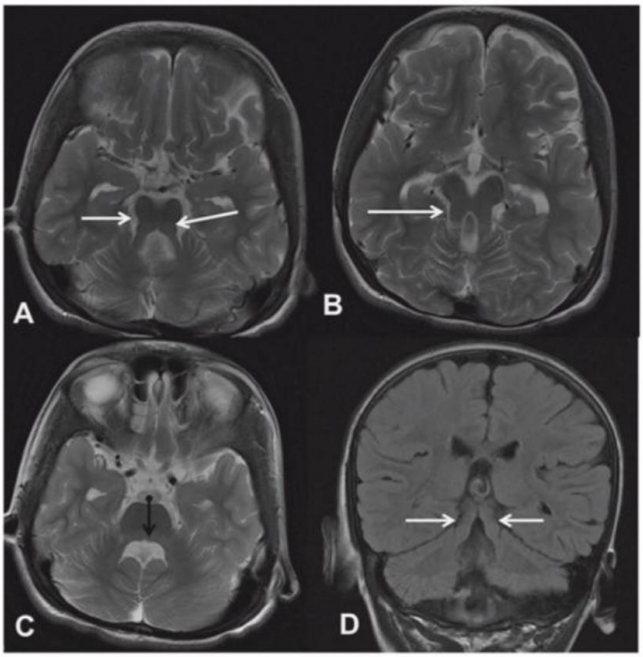

Fig.1. Molar tooth sign and Bat wing appearance of fourth ventricle.

Our patient also had a posterior fossa cyst which is a Dandy-walker variant (Figure 2).

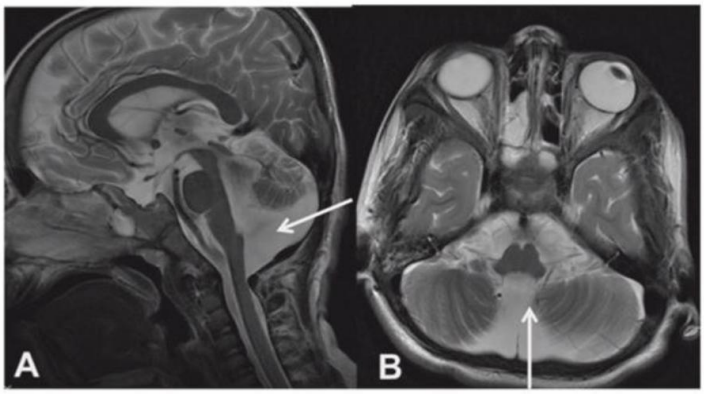

Fig. 2. Posterior fossa cyst.(Dandy Walker variant).

III. Discussion:-

Joubert Syndrome is a rare congenital syndrome associated with varying degrees of vermian hypoplasia and failure of fibre decussation in superior cerebellar peduncles and pyramidal tracts which causes thickened superior cerebellar peduncles to have more horizontal course between the brainstem and cerebellum [1].The incidence ranges between 1/80000 to 1/100000 live births[2]. Joubert Syndrome – related disorders(JSRD) were defined based on associated multi-organ involvement (retinal dystrophy, nephronophtisis, hepatic fibrosis, polydactyly).JSRD has six phenotypic subtypes: Pure JS, JS with occular defect, JS with renaldefect, JS with occulorenal defects, JS with hepatic defects, JS with orofacial digital defects. Pure JS is classified into two types depending on presence or absence of retinal dystrophy. Type 2 is associated with retinal dysplasia and therefore has a poor prognosis.

The clinical features include episodic hyperapnea and apnea in neonatal period, occular abnormalities, hypotonia, truncal ataxia, developmental delay, intellectual impairment and abnormal facies [3,4].

The diagnosis based on characteristic imaging features in computed tomograpgy (CT) or magnetic resonance imaging (MRI) which include molar tooth sign and varying degrees of vermian hypoplasia causing batwing appearance of the fourth ventricle. The molar tooth sign is produced by a thinned ponto-mesencephalic junction and deep inter peduncular fossa due to dysgenesis of the isthmus and thickening of superior cerebellar peduncles which are oriented horizontally[5,1]. Other associated brain anamolies include cortical dysplasias, greymatter heterotopias, ventriculomegaly and corpus callosum agenesis[5,6].

In diagnosed cases of JS, further investigations should be done to exclude any systemic abnormalities.The protocol should include occular investigations (Visual acuity, occular motility,ERG), kidney, and liver function tests, urine analysis and abdominal ultrasound abdomen to identify hepatic fibrosis and multi cystic kidney disease[2].

Management is supportive and requires a multidisciplinary approach. In particular, special care should be taken in children with breathing abnormalities in infants. Medication like opioids should be used with caution as these patients are sensitive to respiratory depressents and anaesthetic agents like nitrous oxide should be avoided. Cognitive and behavioral abnormalities should be dealt with adequate neuropsychological support and rehabilitation. Prognosis depends upon the extent and severity of breathing dysregulation and systemic abnormalities[5,2].

IV. Conclusion:-

Joubert syndrome should be ruled out in all patients presenting with hypotonia, ataxia, nystagmus, breathing abnormalities and developmental delay. Its hallmarks include molar tooth sign and batwing shaped fourth ventricle. As JS is associated with multiorgan involvement, these patients should undergo a diagnostic protocol to acesses systemic abnormalities. Caution should be taken while administering drugs in these patients as they are prone to respiratory depression.

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