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Atretic Occipital Encephalocele in A Case of Joubert's Syndrome

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Abstract: Joubert syndrome is a very rare malformation. It is an uncommon autosomal recessive neurodevelopmental disorder involving cerebellar vermis and brain stem. It is estimated to affect between 1 in 80,000 and 1 in 100,000 newborns. Joubert syndrome is an autosomal recessive disorder marked by agenesis of cerebellar vermis, ataxia, hypotonia, oculomotor apraxia, neonatal breathing problems and mental retardation. We report a 12 month-old girl who presented with developmental delay, failure to thrive, decrease vision and abnormal eye moments. On examination decreased muscle tone, nystagmus, and gait ataxia is present.
Keywords: Joubert syndrome, cerebellar vermis, nystagmus, gait ataxia

INTRODUCTION

In 1969, Dr. Marie Joubert and colleagues first described four siblings with cognitive impairment, ataxia, episodic tachypnea, eye movement abnormalities, and cerebellar vermis agenesis in a large French-Canadian family with consanguinity traced 11 generations to a common ancestor [1]. It is characterized by congenital malformation of the hindbrain and a broad spectrum of other phenotypic findings which are now known to be caused by defects in the structure and/or function of the primary cilium. Many children with Joubert syndrome exhibit dysmorphic facial features that include broad forehead, arched eyebrows, eyelid ptosis, wide-spaced eyes, open mouth configuration, and facial hypotonia[2]. Joubert syndrome also have other clinical manifestations involving the CNS (occipital encephalocele, corpus callosal agenesis), eyes (coloboma, retinal dystrophy, nystagmus, oculomotor apraxia), kidneys (nephronophthisis, cystic dysplasia), liver (hepatic fibrosis), and limbs (polydactyly)[3].

CASE REPORT

A 12 month old male child was brought to our hospital by her father with the chief complaints of delayed milestones, abnormal eye moments and abnormal respirations. Boy had abnormal eye moments being noticed by parents since age of 9 months in form of upward rolling of his eyes with head tilt and poor fixation. Boy hadn't achieved milestones like sitting and waliking by 12 months of age. There is no history of consanguineous marriage. The birth history consisted of delivery at full term gestation by vaginal route at district hospital. Child did not cry immediately after birth, cried after rescuitation. Child was admitted in NICU for irregular respiration. During hospitalization the pediatrician's notes stated that the child is hypotonic with attacks of apnea and hyperpnoea. According to history from mother the child was sleepy for the first 1 1/2 months, with no crying, no limb movement, and very poor sucking. Clinical system examination revealed nystagmus, decreased muscle tone, and gait ataxia. Routine hematological, urine examination, 2d ECHO, and thyroid profile were unremarkable. In view of suspicious intracranial pathology, MRI brain was advised and performed after properly sedating the child. MRI revealed ventriculomegaly, partial vermian agenesis, atretic occipital encephalocele with atretic tissue continuum with right cerebellar hemisphere, molar tooth sign and enlaged subarachnoid spaces in frontal lobes. Screening of abdomen revealed multicystic renal dysplasia. Spinal cord revealed normal morphology and signal intensity.

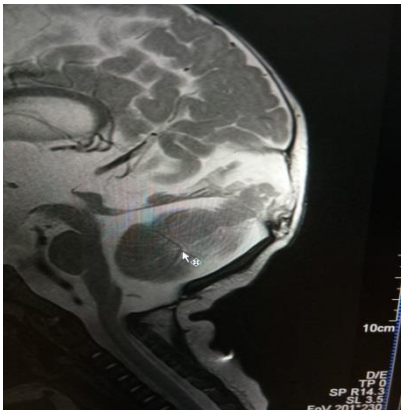

Fig-1: Sagittal T2 weighted images revealing atrertic occipital encephalocele

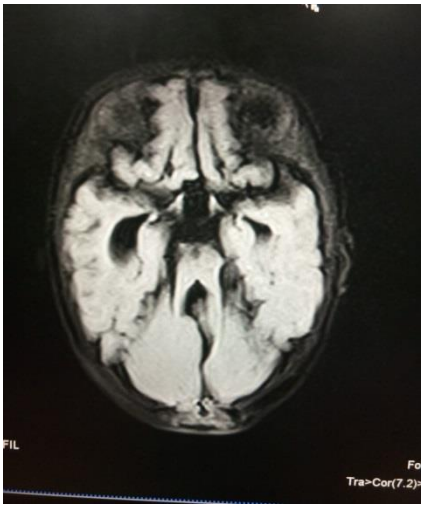

Fig-2: Axial FLAIR images revealing Molar Tooth sign with atretic occipital encephalocele with vermian dysgenesis

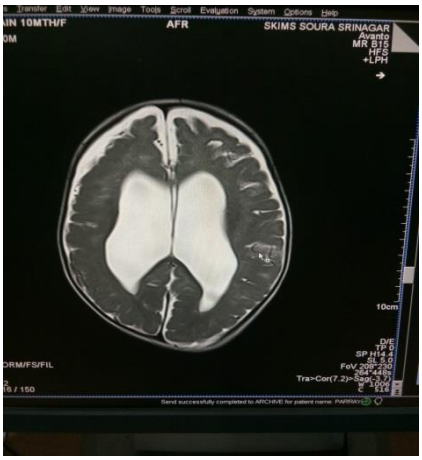

Fig-3: Axial T2 weighted images revealing ventriculomegaly with enlarged frontal subarachnoid spaces

DISCUSSION

Joubert syndrome (JBTS) is a rare, autosomal recessive disorder characterized by a specific congenital malformation of the hindbrain and a broad spectrum of other phenotypic findings [2]. JSRD are classified in six phenotypic subgroups: Pure JS; JS with ocular defect; JS with renal defect; JS with oculorenal defects; JS with hepatic defect; JS with orofaciodigital defects [4]. JSRD (Joubert Syndrome related disorder) presents with episodic hyperpnoea, abnormal eye movements, ataxia and intellectual disability [5]. The term JSRD includes all conditions sharing MTS and this neuroradiolagical sign now represent the mandatory criterion to diagnose JSRD [6].Molar tooth sign refers to the appearance of mid brain in axial section in which the elongated superior cerebellar peduncles give the midbrain an appearance reminiscent of a molar tooth. It was initially described in jouberts sundrome but can occur in other

few conditions e.g Cogan's syndrome, nephronophthisis and hepatic fibrosis.it results fromlack of normal decussation of superior cerebellar fiber tracts which in turn enlargement of peduncles.this leads to reduction in anterioposterior diameter of midbrain and deepening of interpeduncular cistern[7].

The dentate nuclei, the major source of cerebellar output to the cerebral cortex, are fragmented into islands. Malformation of various pontine and medullary structures, including the basis pontis, reticular formation, inferior olivary, dorsal column and solitary tract nuclei, have been reported, which may explain the respiratory defects in JS[4]. Developmental abilities, in particular language and motor skills, are delayed in all JSRD patients, with variable degrees of severity. (CNS) malformations include hydrocephalus, cystic enlargement of the posterior fossa, abnormalities of the corpus callosum, white matter cysts, and absence of the pituitary gland. Abnormal migration defects, mainly periventricular nodular heterotopia, and polymicrogyria [4]. Atretic occipital encephalocele refers to small subscalp lesions that consist of dura, fibrous tissue and dysplastic brain tissue. Most common site being the parietal region followed by occipital region.

Satran D, *et al*. mentioned that early death sometimes present in JS. Other associated clinical features than the major findings could be seen in JS with varying frequencies are as follows [8,9]:

- Polydactyly
- Renal cyst
- Occipital meningocele
- Retinal dysplasia and chorioretinal coloboma
- Ptosis
- Ocular fibrosis
- Liver anomalies
- Congenital heart defects
- Duodenal atresia and choanal atresia
- Soft tissue tumors of the tongue
- Mental retardation and early death
- Micropenis*

JSRD is transmitted in autosomal recessive fashion and some X linked recessive pattern. It is diagnosed by chorionic villus sampling. Fetal USG may be useful. Fetal MRI is the diagnostic method of choice [6]. Ten causative genes have been identified to date, all encoding for proteins of the primary cilium or the centrosome, making JSRD part of an expanding group of diseases called ciliopathies [4]. Mutations in the AHI1 gene are cause in 10- 15% cases. Mutation in the CEP 290 (NPHP6) gene is in10%. Homozygous deletion of NPHP1 gene is in1-2%.

Initiation of periodic, comprehensive developmental assessments and a program of interventions including special education, physical, occupational, and speech therapy, with adaptive equipment as needed, have shown significant benefits in attainment of developmental milestones for many children with JSRD [2].

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