CASE REPORT

Joubert Syndrome - A Case Report

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Abstract:

Joubert syndrome is a very rare malformation. 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.

Key words: Joubert Syndrome, JSRD, Agenesis of Cerebellar Vermis, Molar Tooth Sign

Introduction:

Joubert syndrome is a rare, autosomal recessive condition, first described by Joubert in 1969. 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 [1]. Joubert Syndrome can be identified on axial magnetic resonance imaging as the molar tooth sign (MTS); other criteria include intellectual disability, hypotonia, and often, abnormal respiratory pattern and/or abnormal eye movements. The clinical course can be variable, but most children with this condition survive infancy to reach adulthood. We report a case of Joubert syndrome having abnormal breathing pattern, hypotonia and Molar Tooth Sign (MTS) on C.T. Head.

Case report:

2 months old male child brought to Paediatrics ward of Dr. V. M. Govt. Medical College, Solapur with breathlessness of 15 days duration. On examination the baby was afebrile, had abnormal breathing pattern - hyperpnoea followed by apnea, cyanosis and hypotonia. Reflexes were absent. Cardiovascular and respiratory system examinations were normal. Gastrointestinal and genitourinary examination revealed no abnormality. Baby had convulsion on 2nd day of admission. C.T. head showed 'Molar tooth appearance' and 'bat wing appearance. Ophthalmic examination and USG abdomen were normal. Complete haemogram, serum electrolytes and blood sugar were normal. Kidney function test and X-ray were normal. Patient improved with symptomatic treatment given in the form of oxygen, injection gardenal and gavage feeding. CT Head / MRI is sufficient to confirm the diagnosis on detection of 'Molar tooth sign.' Baby discharged and advised to come for follow up for further metabolic and chromosomal study.

Discussion:

Joubert syndrome (JBTS; OMIM 213300) is a rare, autosomal recessive disorder characterized by a specific congenital malformation of the hindbrain and a broad spectrum of other phenotypic findings [1]. JSRD are classified in six phenotypic subgroups: Pure JS; JS with ocu-

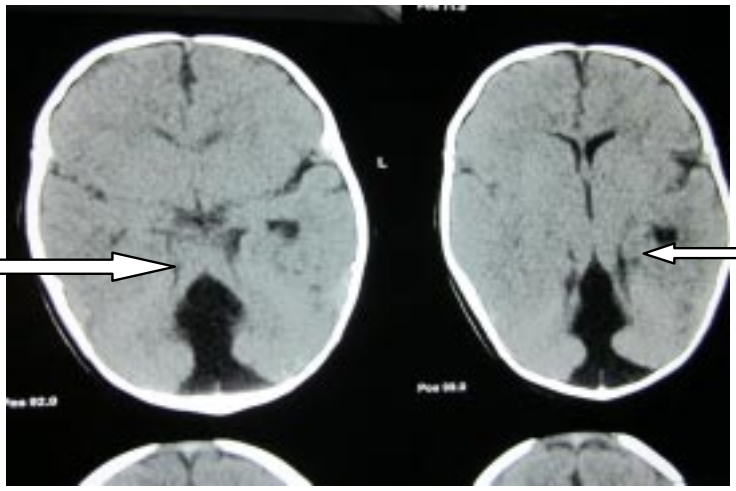

Fig. 1 : Molar Tooth Sign on MRI

lar defect; JS with renal defect; JS with oculorenal defects; JS with hepatic defect; JS with orofaciodigital defects [2].

JSRD (Joubert Syndrome related disorder) presents with episodic hyperpnoea, abnormal eye movements, ataxia and intellectual disability [3]. The term 'JSRD' includes all conditions sharing MTS and this neuroradiolagical sign now represent the mandatory criterion to diagnose JSRD [4]. There can be many associated systemic features including progressive retinal dysplasia, coloboma, congenital heart disease, microcystic kidney disease, liver fibrosis, polyldactyly and soft tissue tumors of tongue [5].

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" [2].

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%.

The neurological features are hypotonia, ataxia and developmental delay, intellectual disability, altered respiratory pattern in neonatal period and abnormal ocular movement. Other findings are like retinal dystrophy, renal disease in 25% cases as NPHP (Nephronophthisis), congenital hepatic fibrosis and polyldactyly in 8-16% cases [1].

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 [4].

Managing respiratory and feeding problems related to breathing abnormalities or hypotonia is the priority. Prolonged apnea is life threatening. Sometimes respiratory abnormalities resolve spontaneously. Prognosis depends on severity of breathing dysfunction, renal and hepatic complications. After the first months of life, global prognosis varies considerably among JSRD subgroups, depending on the extent and severity of organ involvement [2].

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