CASE REPORT

JOUBERTS SYNDROME

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ABSTRACT: Joubert syndrome is a rare autosomal recessive disorder with key finding of cerebellar vermis hypoplasia with a complex brainstem malformation that appears as 'molar tooth sign' on axial magnetic resonance images. This syndrome is difficult to diagnose clinically because of its variable phenotype. The exact diagnosis is often not made for several years after birth. We present a two year old male toddler brought to the pediatric outpatient clinic with global developmental delay, polydactyly and abnormal eye movements. MRI showed molar tooth appearance of superior cerebellar peduncles, the fourth ventricle shaped like a bat wing and hypoplasia of the vermis which is suggestive of jouberts syndrome.

KEYWORDS: Jouberts syndrome, 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.

CASE REPORT: A two year old male child, third born to third degree consanguineous parents brought to Pediatric OPD with history of delayed milestones, hypotonia, extra fingers. Child born by normal vaginal delivery of birth weight 2.75kg. History of birth asphyxia present for which child was admitted in NICU. History of recurrent apnea and seizures present during admission. Child was discharged on day 12 of life with anti-epileptic drug. On examination, child had polydactyly in all 4 limbs, hypotonia, oculomotor apraxia, nystagmus, global developmental delay. Other systemic examination was normal. MRI brain showed 'Molar tooth sign' which is pathognomonic of Jouberts syndrome.

DISCUSSION: Dr. Marie Joubert and coworkers in 1969, for the first time described four siblings with ataxia, cognitive impairment, eye movement abnormalities, cerebellar vermis agenesis, and episodic tachypnea in a French- Canadian family.⁽²⁾

Joubert syndrome and related disorders (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 orofaciocaudal defects.⁽³⁾ JSRD presents with episodic hyperpnea, abnormal eye movements, ataxia and intellectual disability.⁽²⁾ The term JSRD includes all conditions sharing molar tooth sign and this neuroradiological sign now represent the mandatory criterion to diagnose JSRD.⁽⁴⁾ There can be many associated systemic features including progressive retinal dysplasia, coloboma, congenital

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heart disease, microcystic kidney disease, liver fibrosis, polydactyly 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 in 10%. Homozygous deletion of NPHP1 gene is in 1-2%.

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 Nephronophthisis, congenital hepatic fibrosis and polydactyly in 8-16% cases.⁽¹⁾ Typical facial features for this syndrome include high round eyebrows, broad nasal bridge, anteverted nostrils, and low-set ears. Ocular findings include colobomas, oculomotor apraxia, nystagmus, strabismus, ptosis, and abnormal retinal pigmentation.⁽⁶⁾ 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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