Pediatric Oncall Journal (January-March 2020)
Volume 17, Issue 1: 28
DOI : https://doi.org/10.7199/ped.oncall.2020.6

PEDIATRIC ONCALL
CHILD HEALTH CARE

IMAGES IN CLINICAL PRACTICE

MOLAR TOOTH SIGN

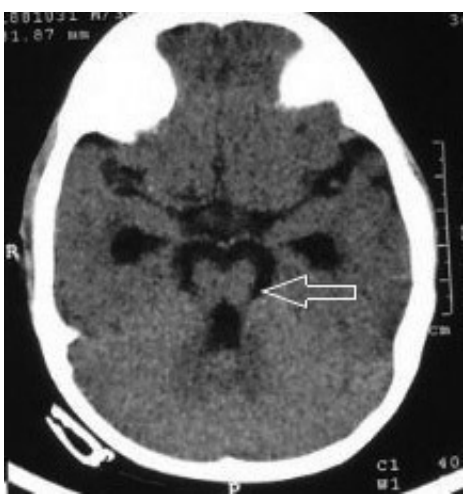
Rajani H S, Narayanappa D.

Department of Pediatrics, JSS Medical College, JSS Academy of Higher Education and Research, Mysore, India.

ARTICLE HISTORY
Received 27 Jun 2019
Accepted 22 Jul 2019

A 3 year old male child, first born of a consanguineous marriage diagnosed previously as cerebral palsy with epilepsy due to birth asphyxia presented to us with breakthrough seizures. He had global developmental delay, microcephaly, irregular breathing with intermittent hyperpnoea, horizontal nystagmus, strabismus and oculomotor apraxia. Fundus examination was normal. CT brain showed partial vermian hypoplasia with elongated superior cerebellar peduncles giving a 'Molar Tooth Sign' in the midbrain, hypoplastic corpus callosum and thinned out brain stem (Figure 1). Ultrasound abdomen was normal.

Figure 1. CT brain shows partial vermian hypoplasia with elongated superior cerebellar peduncles giving a 'Molar Tooth Sign' in the midbrain.



What is the diagnosis?

Joubert Syndrome (JS). It is a rare predominantly autosomal-recessive disorder characterized by agenesis of cerebellar vermis, abnormal eye movements with nystagmus, episodes of hyperpnoea and apnea, delayed generalized development, retinal coloboma and dystrophy and sometimes, multi-cystic kidney disease.[1-3] It is due to defective malfunctioning cilia in the retina, renal tubule and neural cell migration producing heterogeneous syndrome complexes known as ciliopathies.[1] The combination of hypoplasia of the cerebellar peduncles seen as molar tooth sign and hypoplasia of the vermis resulting in bat-wing appearance of the fourth ventricle in the CT or MRI are highly suggestive of JS.[1-3] Molar tooth sign, though a hallmark of JS, may be seen in other ciliopathies like Varadi-Papp syndrome, Malta syndrome, Senior-Loken syndrome, COACH syndrome, nephronophthisis and Cogan's syndrome.[1,3,4] JS is genetically heterogeneous with 34 causative genes identified to date, of which 33 genes are autosomal recessive and one gene (OFD1) is X-linked. Most of the genes encode proteins known to be involved in the function of the primary cilium or basal body.[5] The treatment for JS is supportive. Annual screening is recommended for liver, kidney and retinal abnormalities.[6] The prognosis for infants with JS depends on whether or not the cerebellar vermis is partially developed or entirely absent, as well as on the extent and severity of other organ involvement, such as the kidneys and liver.[7] Manifestations may be in mild form with minimal motor disability and good mental development or severe motor disability, moderate impaired mental development, and multi-organ impairments.[7]

CONTACT Rajani H S

Email: drrajanihs@jssuni.edu.in

Address for Correspondence: Dr. Rajani H S, Assistant Professor, Department of Pediatrics, JSS Hospital, Mahatma Gandhi Road, Mysore 570004, India.

Compliance with Ethical Standards

Funding: None

Conflict of Interest: None

References :

1. Nag C, Ghosh M, Das K, Ghosh TN. Joubert Syndrome: The Molar Tooth Sign of the Mid-Brain. Ann Med Health Sci Res. 2013; 3: 291-294
2. ZHU Zhen, PA Mier, ZHU Jie-ming, QIAN Bin. CT and MRI diagnosis of Joubert syndrome. Chinese J Radiol. 2005; 39: 1256-1259.
3. Choh SA, Choh NA, Bhat SA, Jehangir M. MRI findings in Joubert syndrome. Indian J Pediatr. 2009; 76:231-5.
4. Brancati F, Dallapiccola B, Valente EM. Joubert Syndrome and related disorders. Orphanet J Rare Dis. 2010;5:20
5. Xiang J, Zhang L, Jiang W, Zhang Q, Wang T, Li H, Li H. Prenatal Diagnosis and Genetic Analysis of a Fetus with Joubert Syndrome. Biomed Res Int. 2018: 2018; 7202168
6. Parisi M, Glass I. Joubert Syndrome. 2003 Jul 9 [Updated 2017 Jun 29]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1325/
7. Poretti A, Snow J, Summers AC, Tekes A, Huisman TA, Aygun N, et al. Joubert syndrome: neuroimaging findings in 110 patients in correlation with cognitive function and genetic cause. J Med Genet. 2017; 54:521-9.