ejpmr, 2016,3(12), 317-319

SJIF Impact Factor 3.628

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Case Report

ISSN 2394-3211

EJPMR

JOUBERT SYNDROME –A RARE CASE OF CEREBELLAR ATAXIA

Sajeeth Kumar K. G.[1] and Santhosh Narayanan*[2]

[1]Professor, Department of Medicine Govt. Medical College, Kozhikode, Kerala, India.
[2]Senior Resident, Department of Medicine Govt. Medical College, Kozhikode, Kerala, India.

***Corresponding Author: Sajeeth Kumar K. G.**

Department of Medicine Govt. Medical College, Kozhikode, Kerala, India.

Article Received on 02/10/2016 **Article Revised on 22/10/2016** **Article Accepted on 14/11/2016**

ABSTRACT

Joubert syndrome is an uncommon autosomal recessive neurodevelopmental disorder involving cerebellar vermis and brain stem. We report a case of 14 year old male boy who presented with ataxia, abnormal movements and intention tremor. Magnetic resonance imaging (MRI) revealed characteristic Molar tooth sign and bat wing appearence of fourth ventricle.

KEYWORDS: Magnetic resonance imaging.

INTRODUCTION

Joubert syndrome is a rare congenital malformation of brain characterised by either absence of cerebellar vermis or its underdevelopment.Vermis is responsible for balance, co ordination and integration of oculomotor functions.[1] Clinically joubert syndrome manifests as ataxia,oculomotor apraxia,hypotonia and low intellect. Other associated features include polydactyly, hepatic and renal anomalies. Treatment is symptomatic and supportive.[2] Prognosis depends on the severity of vermian involvement.

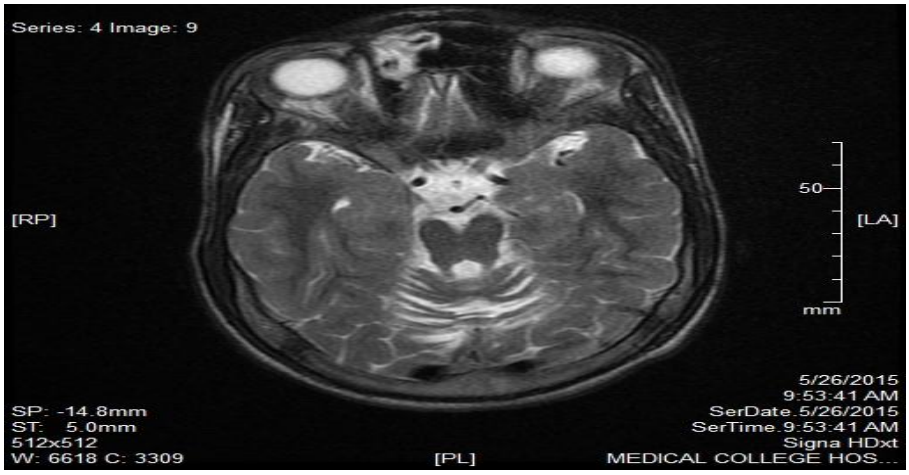
CASE REPORT

14 year old boy presented to us with complaints of swaying while walking, tremors on holding objects/during activity. His symptoms were present from childhood but did not come to medical attention.His mother told that he had delayed developmental milestones and his scholastic performance was poor. There was no history suggestive of cranial nerve dysfunction, weakness of limbs, sensory abnormalities. No h/o vertigo, tinnitus, or exanthematous fever in the past.T here is no history of consanguineous marriage. The birth history consisted of delivery at full term gestation by vaginal route at tertiary care hospital.He had no comorbid illness like hypertension,diabetes mellitus,tuberculosis.He had no addictions.There is no history of similar illness in the family.

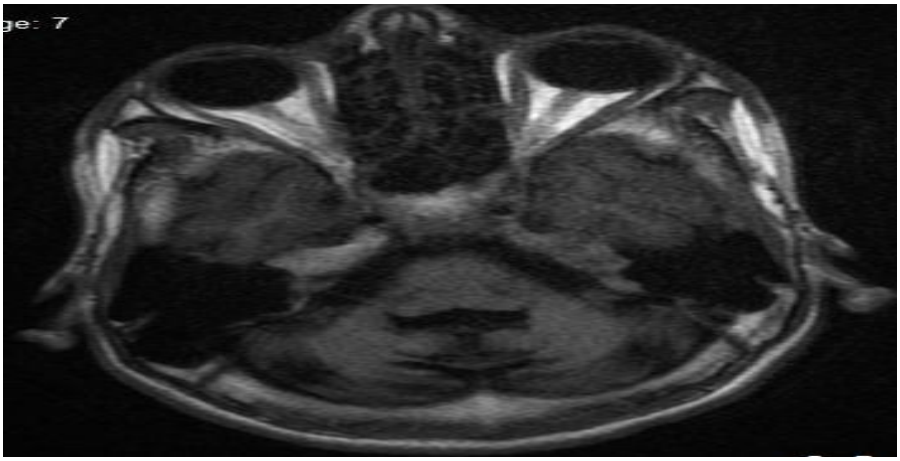
On examination he was conscious,oriented to time,place and person.His vitals were stable.Nervous system examination revealed a scanning type of speech,anisocoria possibly congenital. No nystagmus present. His eye movements were abnormal with difficulty in fixing gaze,difficulty in initiating horizontal saccades,and defective convergence reflex.There was hypotonia of all four limbs.Gait ataxia was present.There was dysdiadochokinesia,intention tremors,but finger nose test,heel knee test was normal.Other systems were within normal limits.

Investigations revealed normal hemogram,serum electrolytes,blood sugars,renal,liver function tests.

CT head showed cerebellar hypoplasia.MRI Brain revealed “Molar tooth sign” and “Bat wing appearance of fourth ventricle” characteristic of joubert syndrome.There was no associated retinal or renal abnormalities.Ultrasonography of abdomen was within normal limits.



MRI BRAIN SHOWING MOLAR TOOTH SIGN



MRI BRAIN SHOWING "bat wing sign of fourth ventricle"

DISCUSSION

The features necessary for a diagnosis of classic Joubert syndrome include the following (a) The molar tooth sign on axial views from cranial MRI studies comprised of these 3 findings: cerebellar vermis hypoplasia (CVH), deepened interpeduncular fossa, and thick, elongated superior cerebellar peduncles (b) intellectual impairment/developmental delay, of variable degree; (c) hypotonia in infancy; (d)one or both of the following (not required but supportive of the diagnosis): irregular breathing pattern in infancy (episodic apnea and/or tachypnea, sometimes alternating) and abnormal eye movements (nystagmus and/or oculomotor apraxia (OMA)) 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[3]. 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.

The hallmark imaging features of JS are: dysgenesis of the isthmus (part of the brainstem between the pons and inferior colliculus), which is seen as elongation and thinning of the pontomesencephalic junction, and deep interpeduncular fossa; thickening of the superior cerebellar peduncles; hypoplasia of the vermis characterized by incomplete lobulation and enlarged fourth ventricles; and incomplete fusion of the halves of the vermis, creating a sagittal vermis cleft seen on axial or coronal MRI planes[4]. Combination of the first three features produce the characteristic MTS on axial MRI. Hypogenesis of the vermis results in a triangular- shaped midfourth ventricle and a bat-wingshaped superior fourth ventricle.[5] 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.

CONCLUSION

Jouberts syndrome is a rare autosomal recessive disorder involving cerebellum. Most of the cases present with

abnormal eye moments, irregular respiration and developmental delays. Examination will reveal hypotonia and gait disorders. This syndrome has characteristic molar tooth appearance on MRI imaging.

REFERENCES

1. Marie Joubert, Jean-Jacques Eisenring, J. Preston, et al. Familial agenesis of the cerebellar vermis : A syndrome of episodic hyperpnea, abnormal eye movements, ataxia, and retardation. Neurology 1969; 19; 813.
2. Melissa A. Parisi. Clinical and molecular features of Joubert syndrome and related disorders. Am J Med Genet C Semin Med Genet. 2009; 151C(4): 326–340.
3. Joubert M, Eisenring JJ, Robb JP, Andermann F. Familial agenesis of the cerebellar vermis. A syndrome of episodic hyperpnea, abnormal eye movements, ataxia and retardation. Neurology. 1969; 19: 813–25.
4. Kendall B, Kingsley D, Lambert SR, Taylor D, Finn P. Joubert syndrome: A clinico-radiological study. Neuroradiology. 1990; 31: 502–6.
5. Egger J, Bellman MH, Ross EM, Baraitser M. Joubert-Boltshauser syndrome with polydactyly in siblings. J Neurol Neurosurg Psychiatry. 1982; 45: 737–9.