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Joubert Syndrome- A Rare Congenital Anomaly

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Abstract
Joubert syndrome is an inherited autosomal recessive trait that affects many parts of the body. However, in some cases Joubert's syndrome appears to be sporadic. The classic clinical presentation is a child with developmental delay, ataxia, and oculomotor and respiratory abnormalities. Neonates may exhibit nystagmus, alternating apnea and hyperpnea, and seizures.
Molar tooth sign and complete or almost complete aplasia of the vermis are neuro imaging hallmarks of Joubert's syndrome.
We present you a case of Joubert's syndrome in 8year old who came with the complaint of intractable seizure and ataxia and who on examination had nystagmus and hypotonia.
Keywords: *jouberts syndrome, sporadic, congenital anomaly, neuroimaging.*

Introduction

In 1969, Joubert described a syndrome a syndrome of episodic hyperpnea, abnormal eye movements, ataxia and mental retardation in five children[1]. The abnormal eye movements relate to abnormalities of supranuclear controls and nystagmnus. It is inherited as an autosomal recessive trait, and there is a male preponderance. Ten causative genes have been identified to date. All these genes encode for proteins of the primary cilium, including JSRD in the group of "ciliopathies". Some but not all affected patients have mutations or chromosome 9q34.3.[6]

The most characteristic finding is complete or almost complete aplasia of the vermis. There is midline clefting, aplasia and heterotopia of cerebellar nuclei and abnormalities of pyramidal decussations, structure of the inferior olivary-nuclei, descending trigeminal tract, solitary fascicle and dorsal column nuclei. Some patients have ocular anomalies.[2]

Clinical Issues

The estimated incidence of JSRD is 1:80,000-100,000 live births. There is no gender predilection.

JSRD typically presents in infancy and childhood. The classic clinical presentation is a child with developmental delay, ataxia, and oculomotor and respiratory abnormalities. Neonates may exhibit nystagmus, alternating apnea and hyperpnea, and seizures.[3][6]

Case Report

A 3 year old female child presented to us with the history of seizures since 2 years. The seizures consists of generalized tonic clonic activity lasting approximately 5-10 minutes. The seizures were progressively increasing in frequency and severity despite of antiepileptic treatment. The also complains of difficulty in walking.

On clinical examination, the child was temperamentally stubborn. The cognition was impaired. There was apparent paraparesis (atonia) and nystagmus. In this patient, scalp electroencephalography (EEG) revealed generalized and focal epileptiform activity findings. The patient first got a CT scan done which revealed the typical molar tooth sign, vermian aplasia, open umbrella sign and bat wing appearance. MRI confirmed the diagnosis and revealed foreshortened midbrain, narrow isthmus, deep interpeduncular fossa, and thickened superior cerebellar peduncles surrounding an oblong or diamond-shaped fourth ventricle festigial shifting of the rostrum and also ruled out no involvement of the corpus callosum. After the diagnosis was made on imaging history for familial consanguinity was not present suggesting most likely this was a case of sporadic Jouberts syndrome.

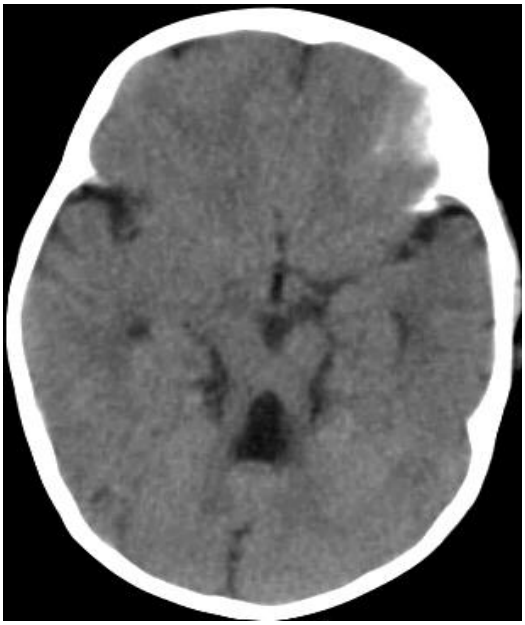

Figure No-1: intial CT showing typical molar tooth sign

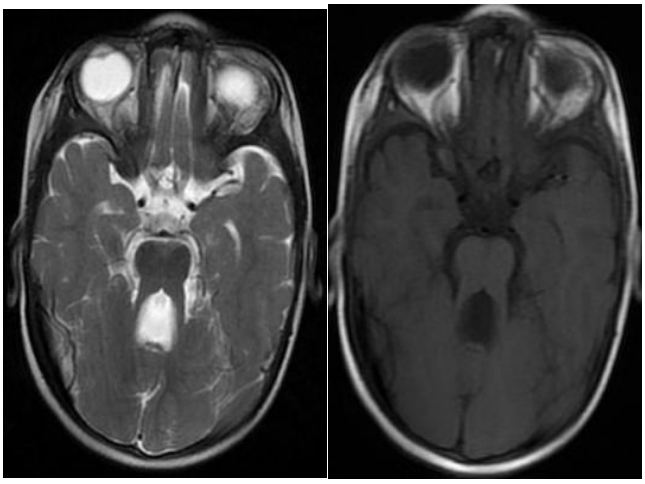

Figure No-2: T1WI& T2WI showing typical molar tooth sign due to vermian hypoplasia with thickened superior cerebellar peduncles

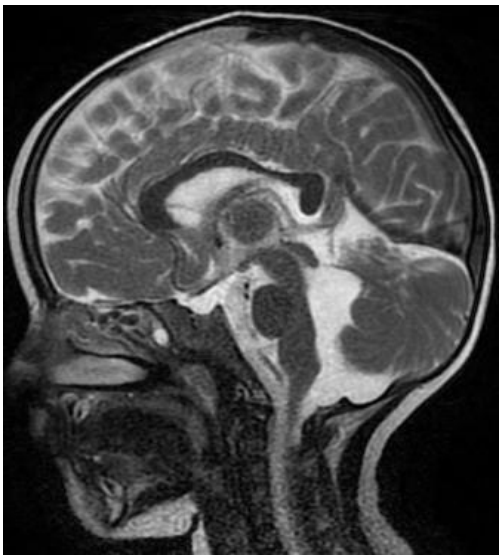

Figure No-3: Sagittal T2WI shows superior displacement of tectal plate

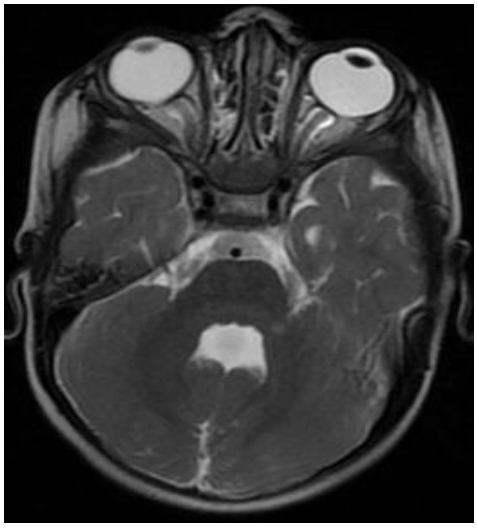

Figure No-4 : T2WI showing typical open umbrella sign and bat wing appearance of fourth ventricle

Discussion

MRI is the imaging tool of choice. The most characteristic finding is complete or almost complete aplasia of the vermis, thickened and abnormal orientation of the superior cerebellar peduncles, thinning of the isthmic portion of the brainstem with a deep interpeduncular cistern producing the pathognomonic Molar tooth sign. There is midline clefting, aplasia and heterotopia of cerebellar nuclei and abnormalities of pyramidal decussations, structure of the inferior olivarynuclei, descending trigeminal tract, solitary fascicle and dorsal column nuclei. Some patients have ocular anomalies.[4]

The molar tooth appearance is also contributed by failure of normal decussation of the superior cerebellar peduncularfibres during embryonic development resulting in thickening of peduncles that follow a more horizontal course extending perpendicular to the brainstem between the midbrain and cerebellum. The absence of crossing fibres also leads to decreased anteroposterior diameter of the brainstem at the level of midbrain and deep interpeduncular cistern. Combination of these findings demonstrates the classical molar tooth configuration on axial and coronal MR images of the midbrain. Additional MR findings include absence of vermis resulting in a midline cleft between the two cerebellar hemispheres resulting in batwing appearance of the fourth ventricle on axial images and open umbrella sign.[4]

Anomalies of the kidneys, eyes, extremities, liver, and bile ducts are common in the JSRD spectrum. Six major JSRD phenotypic subgroups are recognized: Pure JS, JS with ocular defect, JS with renal defect, JS with oculo-renal defects, JS with hepatic defect, and JS with oro-facio-digital defects.[3][5]

Classic JS is the "pure" syndrome. The oculo-renal form is termed CORS (**c**erebello-**o**culo-**r**enal **s**yndrome). COACH syndrome consists of **c**erebellar vermis hypoplasia, oligophrenia, ataxia, ocular **c**oloboma, and hepatic fibrosis.[3]

With appropriate clinical presentation, detailed physical evaluation and the typical MR finding of the molar tooth sign, diagnosis of Joubert syndrome can be made.

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