Case Report

Molar Tooth Sign in Joubert's Syndrome: A Case Report

Radiology Section

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ABSTRACT

Joubert's syndrome is a rare cause of hypotonia and developmental delay in infancy and childhood [1]. Radiologically, the syndrome is characterized by malformations of the hind brain in the form of 'Molar tooth sign' and enlargement of the fourth ventricle with a bat wing configuration.

Key Words: Joubert's Syndrome, Molar tooth, Hypotonia

INTRODUCTION

Joubert's syndrome is a rare cause of hypotonia and developmental delay in infancy and childhood [1]. It is a very rare autosomal recessive condition which was first described by the French neurologist, Marie Joubert in 1969 [2]. Joubert et al described four French sibs, with most cases of the Joubert's syndrome being sporadic. Radiologically, the syndrome is characterized by specific congenital malformations of the hind brain and a broad spectrum of phenotypic findings such as intellectual disability, hypotonia, abnormal respiratory movements and eye movements [1].

CASE REPORT

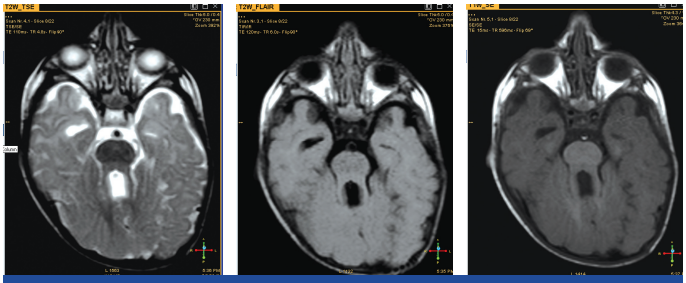
A 7 months old female patient was referred to the Radiology Department for MRI of the brain to rule out developmental brain malformation. The girl was referred from the Paediatric Department for the evaluation of hypotonia and developmental delay. This was the first child who was born to her mother at a government hospital by normal vaginal delivery. Her neonatal and early infantile period was uneventful. Later, as the baby grew, her mother noticed developmental delay with decreased crying and decreased limb movements.

On examination, the paediatricians found general hypotonia in the child. The patient could not follow light or moving objects. With this clinical history, the patient was referred to the Radiodiagnosis Department for MRI of the brain.

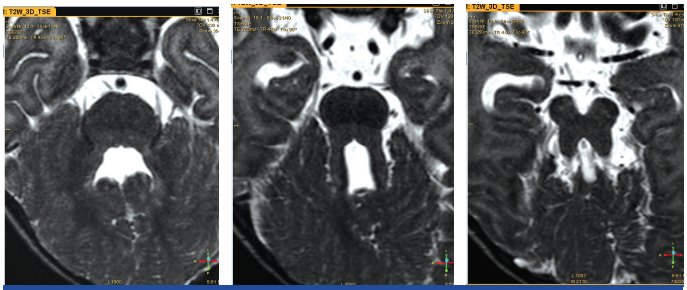
The MRI was done in a 1.5 Tesla Philips Achieva machine with multiplanar T1, T2 and FLAIR sequences. Diffusion weighted images were also done.

FINDINGS ON MRI

There was deepening of the interpeduncular cistern with elongation and thinning of the isthmus. The superior cerebellar peduncles were thick, elongated and abnormally oriented perpendicular to the dorsum of the pons. This was along with a deep cisterna interpeduncularis which formed a 'Molar tooth sign' [Table/Fig-1 & 2]. Moderate dilatation of the 4th ventricle was noted, with a bat wing configuration of the rostral fourth ventricle (Table/Fig-2). The cerebellar vermis was aplastic. The cerebral hemispheres were normal. The rest of the ventricular system was normal.



[Table/Fig-1]: Axial T2, FLAIR & T1 weighted images showing thick & elongated superior cerebellar peduncles abnormally oriented perpendicular to the pons.



[Table/Fig-2]: Axial CISS 3D sequences showing bat wing configuration of fourth ventricle, thick & elongated superior cerebellar peduncles abnormally oriented perpendicular to the pons and deep interpeduncular cistern.

DISCUSSION

Joubert's syndrome is an autosomal recessive disorder which is characterized by hypotonia, imbalance, developmental delay and respiratory abnormalities which are characterized by tachypnoea, followed by apnoea [3].

Joubert's syndrome related disorders (JSRD) are included in the rapidly expanding group of disorders which are called 'Ciliopathies', because all the six gene products (NPHP1, AHI1, CEP290, RPGRIP1L, TMEM611 and ARL13B) are implicated in the JSRD function in the primary cilium/basal body organelle [4,5].

Quisling et al reported the findings from the MRI which was performed on 44 patients with the clinical diagnosis of Joubert's syndrome [6]. The following features are noted in Joubert's syndrome.

Mesencepahlon: Deepening of the interpeduncular fossa and elongation and thinning of the isthmus was noted. The interpeduncular fossa was elongated.

Cerebellum: The superior cerebellar peduncles were thick and were abnormally oriented perpendicular to the dorsum of the pons. The appearance of the mesencepahlon and the superior cerebellar peduncles together formed the 'Molar tooth sign'. This sign is also seen in other disorders like Pontine Tegmental cap dysplasia. There was a variable degree of vermian hypoplasia, the volume of the inferior vermis was roughly equivalent to that of the superior vermis. The fastigium was shifted rostrally. The floculonodular lobe was hypoplastic but it was always present. The cerebellar hemispheres demonstrated normal volume. However, they may have been oddly oriented secondary to the vermian hypoplasia.

There was mild to marked fourth ventricular dilatation enlargement which appeared to be secondary to the vermian hypoplasia. The rostral fourth ventricle was often bat shaped. The brain stem and the supratentorial findings were less common, like a mild prominence of the subarachnoid spaces, mild lateral ventriculomegaly and a mild delay in myelination.

Cerebellar vermian anomalies have been reported with other disorders, such as the Dandy-Walker syndrome and rhomboencephalosynapsis. These disorders can usually be easily distinguished from Joubert's syndrome. In the Dandy-Walker syndrome, a large cystic abnormality of the posterior fossa is seen and it is often accompanied by posterior fossa enlargement. In rhomboencephalosynapsis, the cerebellar hemispheres are fused and unlike in Joubert's syndrome, a midline cerebellar cleft is not present [7, 8].

Haug et al [8] showed overlapping of the clinical features of Joubert's syndrome and the orofacial digital syndromes type 1 and type 3. However radiological imaging helps to differentiate these two pathologies. Apart from the central nervous system changes, there may be retinal pigmentary changes and renal cysts [1].

A similar case report was reported by Wajih Abdallat et al [9] in an 8 months old female patient with similar clinical and radiological features. The treatment is usually symptomatic and the prognosis of this disease is usually poor with hypotonia and severe developmental delay [10].

CONCLUSION

Increased awareness of the possibility of this syndrome which occurs in patients who exhibit developmental delay, hypotonia and respiratory problems with classical radiological findings in an MRI study will lead to earlier diagnosis, appropriate counseling and proper rehabilitation.

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