CASE REPORT

Joubert syndrome is a rare autosomal recessive, congenital central nervous system anomaly. In 85% of cases the diagnostic MRI finding of Molar tooth sign is seen which is characterised by

1. Deep interpenduncular fossa
2. Thick elongated superior cerebellar peduncles
3. Cerebellar vermis hypoplasia

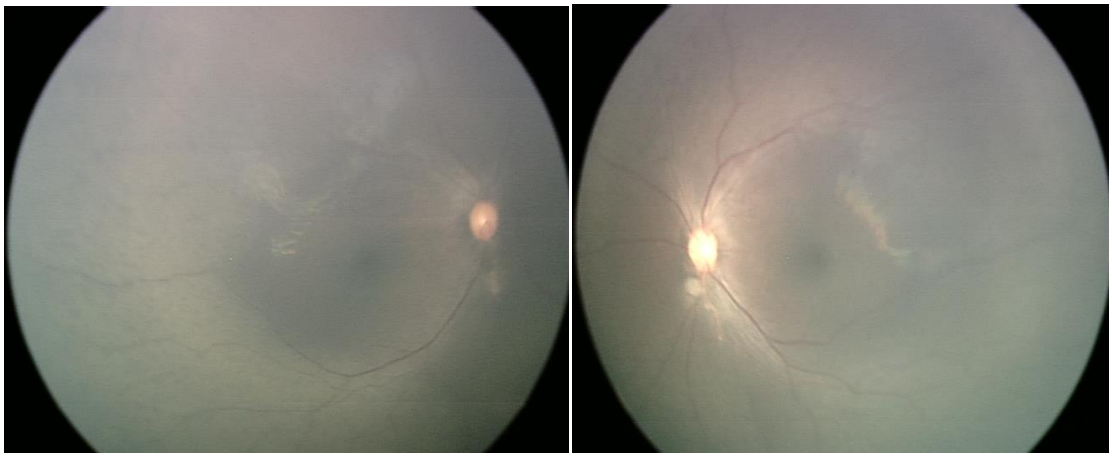
The significance of identifying this rare entity lies in the fact that potential systemic complications could be anticipated thus avoiding life threatening events.

Case Report

A 6 month old child was referred to our institute with complaints of abnormal movements of eyes, inability of the child to identify faces and delayed developmental milestones

Examination:

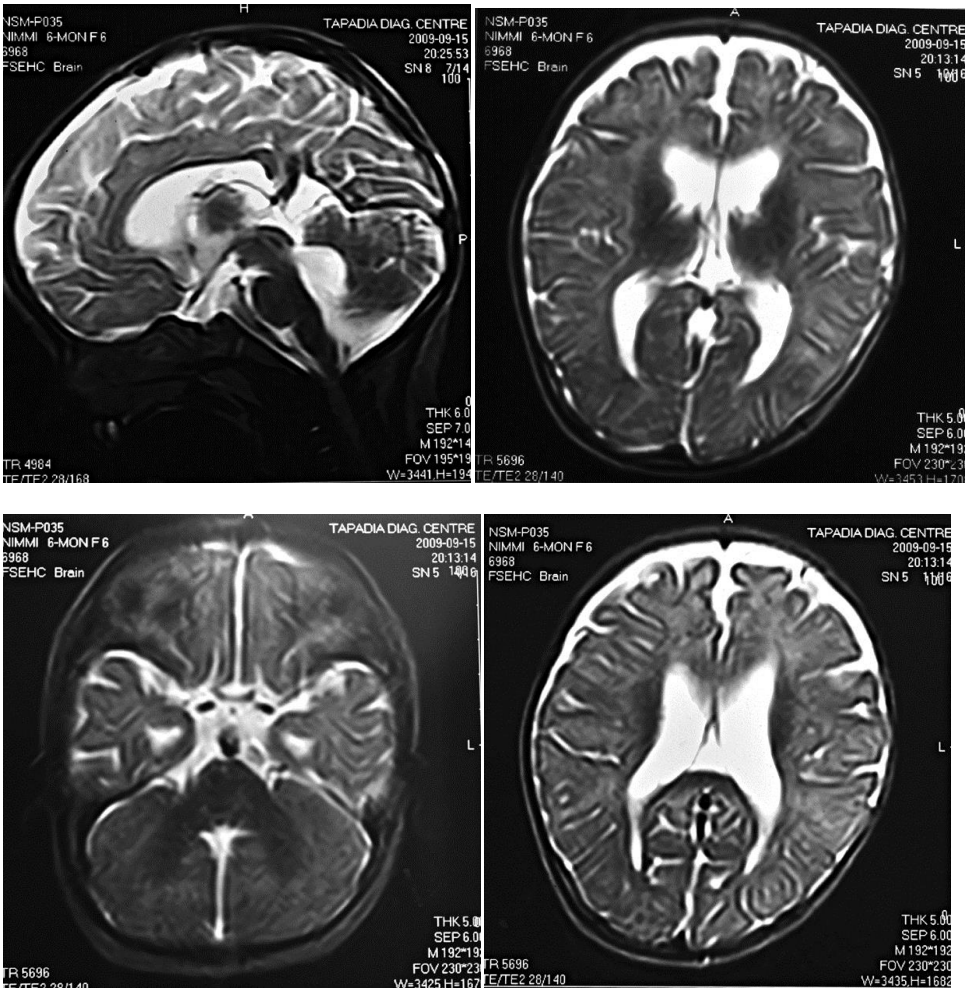
On physical examination the child was unable to fix or follow light, horizontal nystagmus increasing on lateral gaze, pigmentary changes in mid peripheral retina, developmental delay and hypotonia was noted. A clinical diagnosis of Jouberts syndrome was made and neuro imaging was sorted.



Imaging

MR imaging of T1, T2 flair axial and T2 sagittal revealed

1. Prominent cisterns, sulcal spaces and ventricular system
2. Dysgenesis of superior cerebellar vermis and thinning of superior cerebellar peduncles – resulting in Molar tooth sign
3. Thinning of corpus callosum



Discussion

Joubert syndrome was first described in 1969 by Dr. Marie Joubert et al in four siblings with agenesis of the cerebellar vermis, episodic hyperpnoea, abnormal eye movements, ataxia and mental retardation and in one child, the presence of associated occipital meningo encephalocele. Bolshauser and Isler reported three children with same phenotype 8 years late, and coined the term "Joubert Syndrome"

The cardinal neuroradiological hallmark of Joubert Syndrome is "Molar tooth sign" resulting from constellation of cerebellar vermis hypo/aplasia, horizontally oriented and thickened superior cerebellar peduncles and deepened interpenduncular fossa. This is caused by lack of normal decussation of the superior cerebellar peduncle fibres leading to enlargement of peduncles making them follow a horizontal course extending perpendicular to brainstem between the midbrain and cerebellum. Other diagnostic criteria include intellectual disability, hypotonia, abnormal respiratory pattern and / or abnormal eye movements. In addition anomalies like retinal dystrophy, ocular coloboma, oral frenulae and tongue tumors, polydactyly, cystic renal disease and congenital hepatic fibrosis are also seen contributing to the wide spectrum of the phenotypic presentation of the disease.

Opthalmic findings include abnormalities of ocular motility characterized by nystagmus which can be horizontal, vertical and/or torsional and oculomotor apraxia characterized by difficulty in smooth pursuit movements, dysconjugate eye movements and head thrusting.

Other ocular anomalies include strabismus, amblyopia, ptosis, IIIrd nerve palsy, optic disc drusen etc. Involvement of retina follows two basic types, one which mimics Lebers congenital amaurosis with congenital blindness with flat ERG and the other with pigmentary retinal dystrophy. Coloboma of choroid and retina are also seen which are associated with hepatic fibrosis in majority of the cases resulting in COACH syndrome.

The treatment of such patients is by multidisciplinary team approach and by providing supportive medical care.

Conclusion

Joubert syndrome is a rare congenital anomaly and the clinical history and the classic MRI finding of Molar tooth sign aid in diagnosis of this rare entity.