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Clinical Image

Joubert's Syndrome-Prenatal Diagnosis with Magnetic Resonance Imaging

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Clinical History

A 36-year-old woman was referred for evaluation after abnormal (hypoplasia of cerebellar vermis, mild ventriculomegaly, enlarged cisterns magna) ultrasound findings. Fetal MRI performed at 22 weeks of gestation showed vermian hypoplasia, abnormal lobation and thickened cerebellar peduncles, setting the suspicion of Joubert Syndrome. Fetus had a normal karyotype. Parents refused termination of the pregnancy. The child gradually developed intellectual disability, hypotonia, ataxia, apneic episodes and abnormal eye movements. Magnetic resonance follow-up images of the child aged 2 years confirm prominence of the interpeduncular fossa and a deep cleft in the midbrain between thickened cerebellar peduncles comprising the molar tooth sign. Final diagnosis is Joubert Syndrome (JS).

Image Findings

Prenatal brain MRIs at 22 weeks' gestation (WG) reveal several brain abnormalities in a form of a thick elongated horizontal superior cerebellar peduncles and deformed fourth ventricle. Associated cerebellar hypoplasia with enlarged cisterna magna is also delineated.

Postnatal MRI study reveals enlarged fourth ventricle giving a characteristic bat wing appearance. At the level of mesencephalon, atypical appearance of both the superior cerebellar peduncles with deepening of interpeduncular cistern is seen exhibiting a characteristic "molar tooth appearance". Associated vermian dysgenesis is seen.

Discussion and Conclusion

Fetal MRI is the method of choice to portray posterior fossa malformations in a fetus as well as confirm sonographic findings and reveal additional abnormalities.[4] Therefore, all radiologists should be familiar with the expanding field of prenatal MRI. One of the main aims of this case report is, not only to present the image findings of prenatal and postnatal MRI of a JS, but also to raise awareness on the importance of prenatal MRI on the early and vital diagnosis of such abnormalities.

Joubert syndrome (JS) is an autosomal or X- linked recessive cerebellar and brain stem malformation.[2] It was first described by Marie Joubert, a French neurologist in 1969 and is estimated to affect between 1 in 80,000 and 1 in 100,000 newborns. Mutations in at least 27 genes have been shown to cause JS. [1]

Most infants with JS have hypotonia in infancy, which contributes to ataxia in early childhood.[3] Other characteristic features of the condition include episodes of hyperpnea or apnea in infancy, and abnormal eye movements (ocular motor apraxia). Most affected individuals have delayed development and intellectual disability, which can range from mild to severe. Distinctive facial features can also prevail in Joubert syndrome; these include a broad forehead, arched eyebrows,ptosis,hypertelorism, low-set ears, and a triangle-shaped mouth.

The hallmark feature of JS is a distinctive midbrain and hindbrain malformation known as the 'molar tooth sign' (MTS) on axial brain MRI or CT. This radiological feature reflects hypoplasia/aplasia of the vermis, thickening and horizontal reorientation of cerebellar peduncles and fourth ventricle deformity. The MTS is seen in 80-90% of patients with Joubert's syndrome.[2]

In association with the MTS, the absence of the vermis leads to a midline cleft between the two normal appearing cerebellar hemispheres, contributing to the 'bat wing' sign of the fourth ventricle. Moreover, cerebellar cortical dysplasia, abnormalities of the nuclei of the pons, cerebellum, and medulla can be found.

Joubert syndrome is sometimes associated with a broad range of additional signs and extracranial findings such as other eye abnormalities (retinal dystrophy, coloboma), kidney disease (multicystic kidney disease), liver disease (hepatic fibrosis), skeletal abnormalities (polydactyly), or congenital heart defects. A combination of the characteristic features of Joubert syndrome and one or more of these additional signs and symptoms are referred to as Joubert syndrome and related disorders (JSRD).

Differential Diagnosis

-Dandy Walker Syndrome

-Rhomboencephalosynapsis

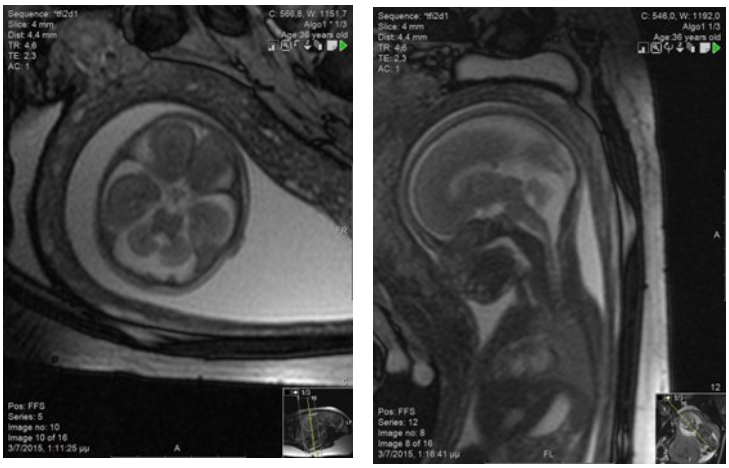

Figure 1: *MRI of a fetus brain at 22 WG using axial (a) T2-W and sagittal (b) sequence reveals hypoplastic cerebellar vermis and an enlarged fourth ventricle communicating with the cisterns magna (CM).*

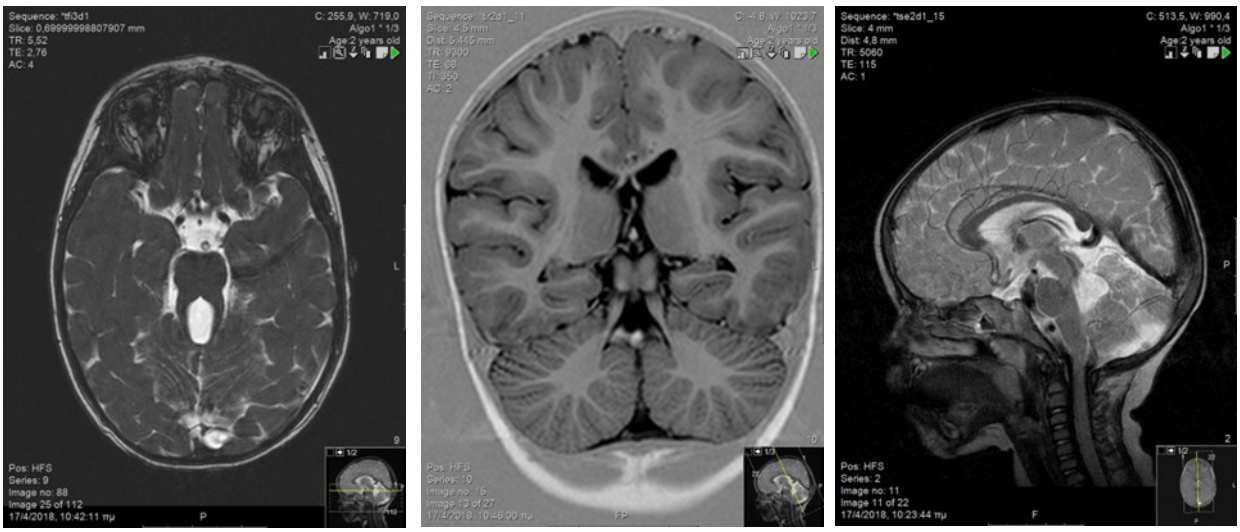

Figure 2: *Postnatal Brain MRI at age 2 using axial (a),coronal (b) and sagittal (c) TSE sequence unveils thick elongated horizontal superior cerebellar peduncles ,deformed fourth ventricle and cerebellar hypoplasia with enlarged communicating CM. The elongated superior cerebellar peduncles give the midbrain the appearance referred to as Molar Tooth Sign. There is also evidence of vermian agenesis and incomplete lobulation. Coronal (b) brain MRI images reveal at wing appearance of the fourth ventricle.*

Conflict of Interest

The authors declare no conflict of interest.

References

[1] Bin Dahman HA, Bin Mubaireek A-HM, Alhaddad ZH. (2016) Joubert syndrome in a neonate: case report with literature review.. Sudanese Journal of Paediatrics 2016;16(1):53-57.

[2] Joubert M, Eisenring JJ, Robb JP, Andermann F. (1969) Familial agenesis of the cerebellar vermis. A syndrome of episodic hyperpnea, abnormal eye movements, ataxia, and retardation. Neurology. Sep;19(9):813-25.

[3] Bosemani T, Orman G, Boltshauser E et al. (2015) Congenital abnormalities of the posterior fossa. Radiographics

[4] Saleem N. Sahar (2014) Fetal MRI :An approach to practice :A review .Journal of Advanced Research (2014)5,507-523

[5] Salem S.N.,Zaki M.S.(2010) Role of MR Imaging in Prenatal Diagnosis of Pregnancies at Risk for Joubert Syndrome and Related Cerebellar Disorders.AJNR AM J Neuroradiol 31:424-29 (Mar 2010)

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