Photo Essay

A Child with Joubert Syndrome: Clinical and Imaging Features

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PRESENTATION

Joubert syndrome is a rare autosomal recessive congenital syndrome that was first described by Joubert in 1969.[1] Diagnosis is based upon clinical features such as hypotonia, developmental delay, and progressive ataxia.[2,3]

The syndrome can have variable ophthalmic features such as ptosis; ocular motor signs, including nystagmus, horizontal strabismus, "wheel-rolling" torsional eye movements, periodic alternating gaze deviation, elevation of the abducting eye on lateral gaze (with bilateral superior rectus and superior oblique muscle over-action), and skew deviation; and also fundus disorders such as optic nerve drusen, cellophane maculopathy, and pigmentary change at the periphery of the retina.[4-6]

In Joubert syndrome, abnormality of the cerebellum is seen on cross-sectional axial brain MRI. The posterior fossa typically exhibits a bat wing 4th ventricle and hypoplastic cerebellar vermis with hypoplasia of the superior cerebellar peduncle resembling the "molar tooth sign".[7]

Our patient was a 2-year-old boy who was referred to the neuro-ophthalmology clinic for evaluation by a pediatrician due to abnormal eye movements, especially during the past year. His medical history and general examination indicated that he had hypotonia with slow limb movements and developmental delay. However, he did not show any signs of specific organ involvement associated with hepatic, renal or skeletal disease except for polydactyly. He had no significant family history and his parents were not consanguineous.

Upon ocular examination, the patient had poor fixation and could not follow moving objects. He had bilateral ptosis, high rounded eyebrows, and a broad nasal bridge [Figure 1]. The patient exhibited primary position nystagmus (see-saw pattern) in conjunction with saccadic dysfunction. Anterior segment examinations were normal. Fundus examination showed a dystrophic retinal appearance in conjunction with optic disc coloboma of the right eye [Figure 2a] and optic disc drusen of the left eye with mottled pigmentation at the periphery of the retina in both eyes [Figure 2b], that was confirmed using fluorescein angiography [Figure 3a and b]. Brain MRI showed hypoplasia of the cerebellar vermis with straight and thickened superior cerebellar peduncles

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suggestive of the "molar tooth sign" [Figure 4a]. Brain MRI revealed apposed cerebral hemispheres and absence of the vermis [Figure 4b].

DISCUSSION

Joubert syndrome is characterized by the following criteria: cerebellar vermis hypoplasia, developmental delay, hypotonia, and abnormal eye movements or breathing pattern.[3]

One of the differential diagnoses for Joubert syndrome is COACH syndrome, which entails cerebellar vermis hypoplasia, oligophrenia, congenital ataxia, coloboma, and hepatic fibrocirrhosis.[8] However, our patient did not exhibit these features.

Khan et al in a case series study, reported the ophthalmic features of patients suffering from Joubert syndrome as saccadic dysfunction, compensatory head turns, primary position nystagmus (see-saw pattern), dystrophic retinal appearance, and ptosis.[6]

In our study, the patient was found to have hypotonia and global developmental delay. His ocular features included bilateral ptosis, primary position nystagmus with a see-saw pattern, saccadic dysfunction, optic nerve coloboma (right eye), optic nerve drusen (left eye), and pigmentary change at the periphery of retina in both eyes. Few cases of optic disc drusen have been reported in patients with Joubert syndrome.[9]

In conclusion, primary position nystagmus in children with developmental delay, which indicates subsequent exhaustive fundal examination and neuroimaging.

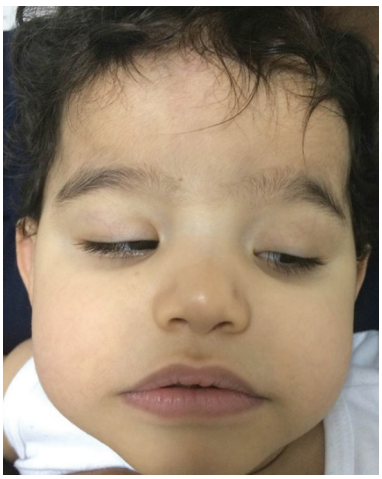

Figure 1. Bilateral ptosis, high rounded eyebrows, and broad nasal bridge.

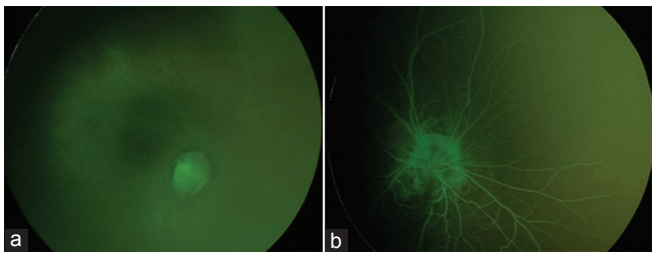

Figure 3. (a) Fluorescein angiography of the right eye: The blood vessels transiting the coloboma to reach the normal retina. (b) Fluorescein angiography of the left eye. Staining of the optic disc borders without optic disc leakage.

Declaration of Patient Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patients parents have given their consent for his images and other clinical information to be reported in the journal. They understand that the childs name initials name and initials will not be published and due efforts will be

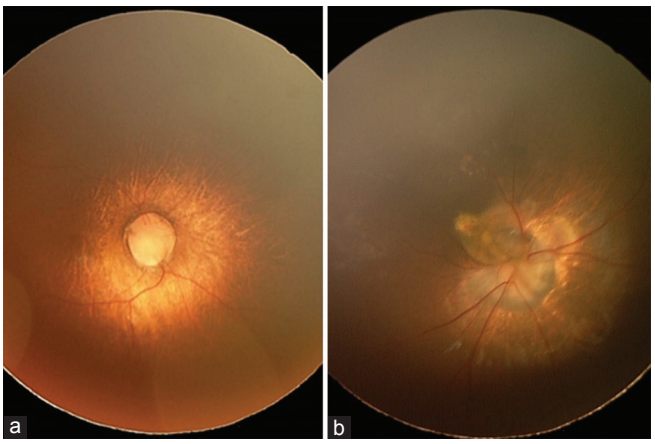

Figure 2. (a) Right eye: Optic nerve coloboma with the absence of physiologic cup and central retinal artery and vein. (b) Left eye: Optic disc drusen mimicking optic disc swelling. Also note peripheral pigmentary changes of the retina are evident.

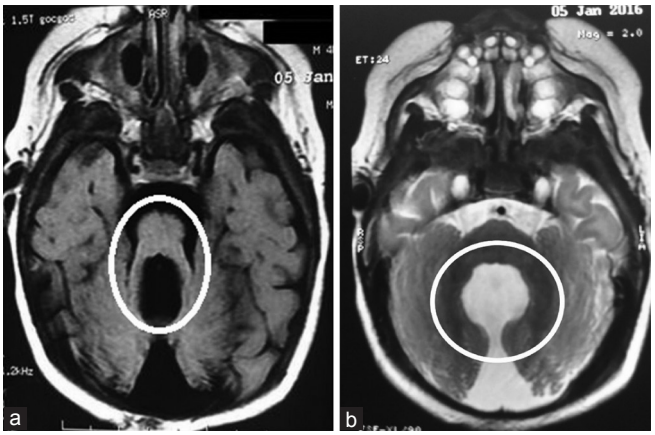

Figure 4. (a) Brain MRI, T1-weighted, an axial cut without contrast: Cerebellar vermis hypoplasia, thickening of superior cerebellar peduncles and prominent cisterna magna, referred to as the "molar tooth sign". (b) Brain MRI, T1-weighted, an axial cut without contrast. Absence of the vermis with apposed cerebellar hemispheres.

made to conceal his identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of Interest

There are no conflicts of interest.

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