Fundus findings in a case of Joubert syndrome

Sir,

A 9-month-old male child born out of nonconsanguineous marriage diagnosed as having Joubert syndrome (JS) was referred for an ophthalmic examination as parents noticed that the child was unable to follow objects. The diagnosis was based on classical findings of JS seen on brain magnetic resonance imaging (MRI). The MRI brain of the child showed prominent, thickened, and elongated superior cerebellar peduncles ("molar tooth sign") [Fig. 1a, arrow] and "Batwing" fourth ventricle [Fig. 1b, arrow].

Parents gave history of developmental delay and the absence of neck holding/sitting balance. On examination, facial dysmorphism (prominent forehead, open mouth, and low set ears) was seen. Horizontal nystagmus was quite obvious. Ocular examination revealed normal anterior segment. Fundus examination revealed clear media, normal optic disc, and macula. However, midperipheral fundus of both eyes revealed symmetrical pigmentary changes which consisted of uniformly distributed hypopigmented discrete lesions similar to those seen in retinitis albipuncatata. In addition, there was relatively coarse intraretinal pigmentation along the retinal venules. Arterioles showed generalized narrowing. Placoid, yellowish hypopigmented lesions were also evident in the posterior retina, in proximity to the optic disc [Fig. 2]. These lesions were not as excavated as typical colobomas. Furthermore, the inferior branch of the central retinal vein was seen to follow a normal course over these lesions in the left eye. In a coloboma, there is some alteration of vasculature in the overlying atrophic retina. However, these still could be small colobomas or lacunar lesions as seen in Aicardi syndrome. Only histopathological studies would reveal their true nature. The fundus images were captured using RETCAM 3 (Clarity Medical Systems, USA).

The child had a hypermetropia of +6 D spherical in both eyes when examined under cycloplegia (atropine 1%). Visually evoked potential (VEP) and electroretinogram showed no significant response in either eye. The brain evoked response audiometry was normal. Genetic analysis is not done as MRI was very typical of JS.

JS is a rare, autosomal recessive congenital malformation of the brainstem and cerebellar vermis. This syndrome was first described by Joubert *et al*. in 1969, who reported four patients with episodic tachypnea, developmental delay, ataxia, abnormal eye movements, and absence of the cerebellum.[1] Diagnostic criteria in JS include hypotonia, ataxia, global developmental delay, and the neuroradiological finding of "molar tooth sign".[2] The term "Joubert syndrome and related disorders" (JSRD) is sometimes used when associated findings suggest a unique distinct syndrome.[3]

JSRD are clinically heterogeneous presenting with neurological signs with variable multiorgan involvement, mainly of retina, kidneys, liver, and skeleton. Ten genes have been identified to date, all of which encode for proteins of the primary cilium, which are known to play a key role in the development and functioning of several cell types, include retinal photoreceptors, neurons, kidney tubules, and bile ducts.[4]

Saccadic function (typically with head thrusts or turns), primary position nystagmus (usually see-saw), pursuit abnormality, and retinal findings suggestive of dystrophy are the most common ophthalmic manifestations of JS.[5] Retina is one of the organs most frequently involved in JSRD, in the form of retinal dystrophy, due to progressive degeneration of photoreceptor cells. Severity ranges from congenital retinal blindness to retinal dystrophy characterized by a progressive course and variably conserved vision.[6] Coloboma involving the retinal pigment epithelium, neurosensory retina, and choroid are known associations of this disorder.[7] Asymmetric flash VEPs and see-saw nystagmus suggest an abnormality in optic nerve decussation.

Retinal pigment epithelial changes are there but not like those seen in typical retinitis pigmentosa (RP). Furthermore, the small placoid hypopigmented lesions inferior to disc as documented by us have not been described before. The

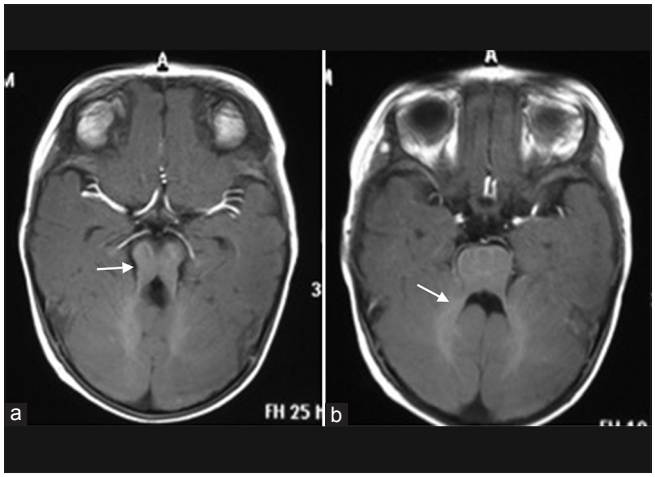

Figure 1: Axial T1-weighted postcontrast image of the brain at the level of midbrain shows prominent thickened and elongated superior cerebellar peduncle giving characteristic "molar tooth appearance" (a, arrow) and "Bat wing" fourth ventricle (b, arrow)

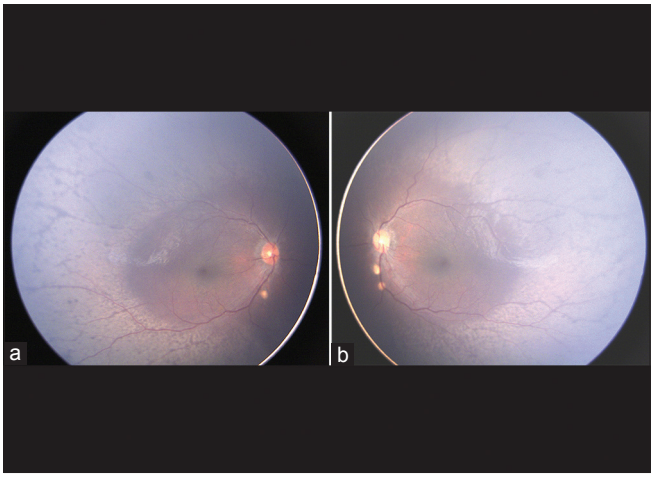

Figure 2: Fundus photograph of right (a) and left eye (b) showing symmetrical pigmentary changes. Placoid, yellowish hypopigmented lesions are also evident in the posterior retina, in proximity to the optic disc

photodocumentation of fundus findings of JS has not been done so far in literature and to the best of our knowledge there are no published reports of fundus findings in JS from India.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/ijo.IJO_441_16

Cite this article as: Chawla R, Bypareddy R, Vekaria L, Venkatesh P, Ananthashayana VH. Fundus findings in a case of Joubert syndrome. Indian J Ophthalmol 2017;65:329-30.