Photo Essay

Tuberous sclerosis complex with Best's vitelliform macular dystrophy: A combined presentation

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Key words: Autosomal disorder, Best's dystrophy, tuberous sclerosis complex

An 11-year-old male presented with gradual diminution of vision in the left eye since 2 years. BCVA was 6/6, N6 in the right and 6/36 and N8 in the left eye. Fundus evaluation of both eyes showed well-defined circular lesions at the macula with atrophic changes, vitelliform deposits at the lower edge, and fibrosis at the fovea in the left eye. A translucent white lesion was seen in the superficial retina, without calcification and no evidence of dilated vessels surrounding it measuring 1 DD in size in superotemporal quadrant of the right eye and a vertically elongated translucent lesion with white specks suggestive of calcification above the disc in the left eye [Fig. 1]. His father [Fig. 2] and paternal grandmother had a history of dry macular degeneration. General examination revealed facial angiofibromas, shagreen patch [Fig. 3], and multiple hypomelanotic macules over back and legs. Abdominal scans showed multiple renal angiomyolipomas. Patient was on carbamazepine 300 mg/day for epilepsy and was advised regular follow-ups for both the systemic and ocular conditions.

Swept source optical coherence tomography (SSOCT) shows a hyperreflective prominent Verhoeff's membrane (white arrow) and surrounding subretinal fluid and hypertrophic retinal pigment epithelium (white star) at the fovea suggestive of Stage 4 of BVMD [Fig. 4a and b].[1,2] SSOCT through the astrocytoma showed an inner retinal mass lesion compressing on the outer retina with central full-thickness involvement having a moth-eaten appearance [Fig. 4c and d].[3] Magnetic resonance imaging brain showed cortical and subependymal tubers and ventricular bands [Fig. 5a]. EOG showed an abnormal Arden's ratio (left eye: 1.151; right eye: 1.141) [Fig. 5b].

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Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/ijo.IJO_693_17

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Manuscript received: 13.08.17; Revision accepted: 03.10.17

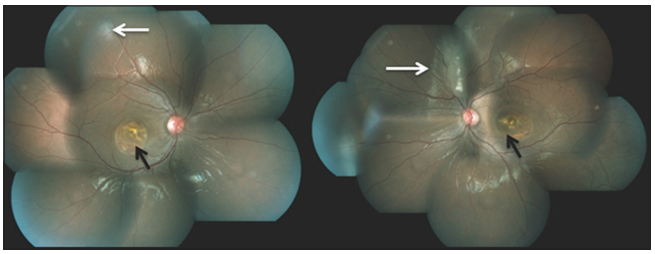

Figure 1: Fundus photographs showing the well-circumscribed vitelliform lesions at the macula (black arrows) and translucent astrocytomas (white arrows)

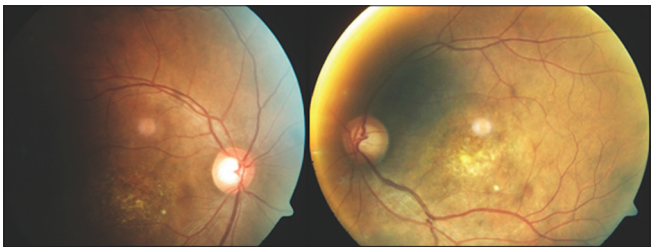

Figure 2: Fundus photograph of the father showing atrophic changes at the macula

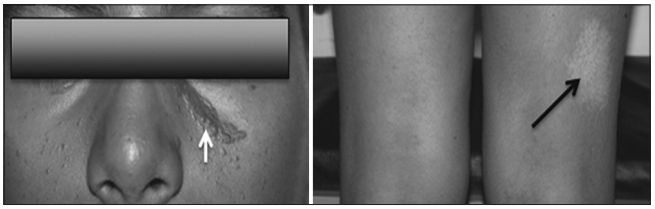

Figure 3: The external photograph shows facial angiofibromas (white arrow) and shagreen patch on the left eye thigh (black arrow)

Discussion

Tuberous sclerosis has an incidence of about 1/10,000 population,[4] whereas Best's dystrophy has a reported incidence of 1–9/100,000.[1] The diagnosis of BVMD was

Cite this article as: Dhami A, Devi PV, Bhende M. Tuberous sclerosis complex with Best's vitelliform macular dystrophy: A combined presentation. Indian J Ophthalmol 2018;66:295-6.

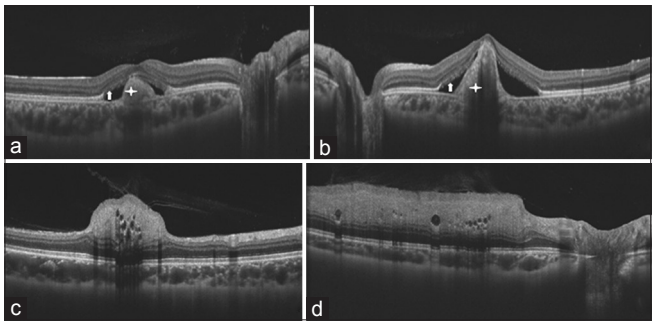

Figure 4: (a) Swept source optical coherence tomography through the macula of the right eye, (b) left eye showing a prominent Verhoeff's membrane (white arrow) and subfoveal hypertrophic lesion (white star), and (c and d) swept source optical coherence tomography through the astrocytoma shows inner retinal mass lesion involving the inner retinal layers having a moth-eaten appearance

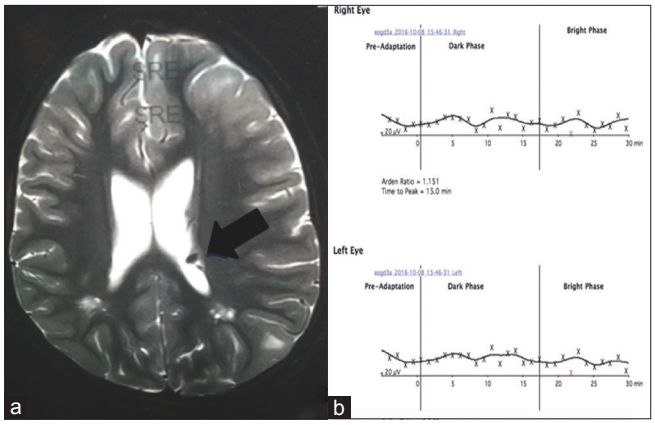

Figure 5: (a) The magnetic resonance imaging of brain revealed cortical tubers, subependymal nodules, and radial bands in the ventricles (black arrow); (b) the electrooculogram showing an abnormal Arden's ratio (right eye: 1.151, left eye: 1.141)

established clinically and confirmed by abnormal Arden's ratio on EOG and the presence of macular lesions in the father and grandmother.[3] A diagnosis of retinal astrocytic hamartoma with TSC was established due to the presence of the major criteria as described by Northrup and Krueger.[5]

Conclusion

The report presents an unusual combined presentation of both autosomal dominant disorders in a patient with clinical features classical of both TSC and Best's dystrophy.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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