The Potpourri of Clinical Signs in Tuberous Sclerosis Complex

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Clinical Description

A 52-year-old male, known case of hypertension and diabetes mellitus type 2, presented with complaints of diminution of vision (right eye) for 03 weeks, which was painless, sudden onset, and gradually progressive. Best-corrected visual acuity was OD 20/60 and OS 20/20. Anterior segment of both eyes was unremarkable. Right eye fundus showed optic disc pallor, and the left eye revealed hyperemic optic disc with raised and blurred margins and yellowish, raised, a retinal lesion with indistinct margins, superior to fovea, approximately 1/5th of disc diameter with no overlying vitritis [Figure 1a]. Ancillary investigations revealed optic atrophy (right eye) secondary to nonarteritic anterior ischemic optic neuropathy (NAION), and recent onset (NAION) (left eye). Autofluorescence and fundus fluorescein angiography revealed retinal astrocytoma in the left eye. The patient was also assessed by a rheumatologist and a cardiologist to rule out secondary causes of NAION and a neurologist for tuberous sclerosis. Systemically patient was diagnosed to have adenoma sebaceum [Figure 1c], shagreen patches [Figure 1d]. Magnetic resonance imaging (MRI) brain showed cortical tubers [Figure 1b]. Although our patient presented with decreased vision in the right eye, which was secondary to NAION, however, in literature, we could not find any association between tuberous sclerosis and NAION.

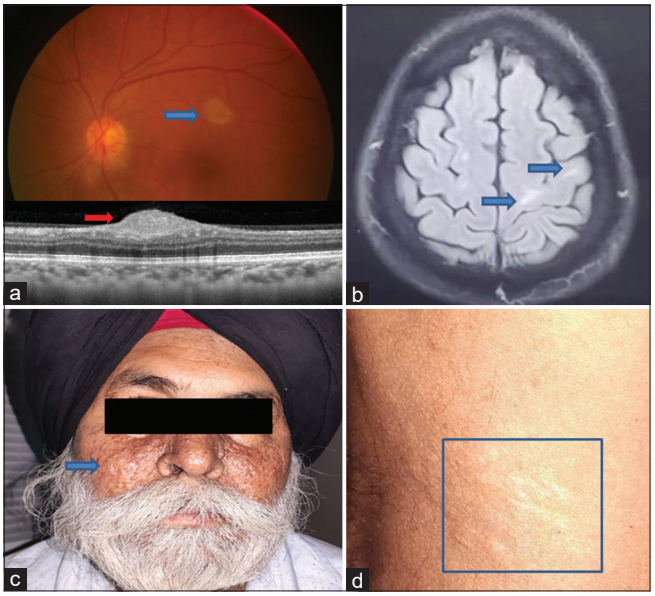

Figure 1: (a) Fundus picture of the left eye showing retinal astrocytoma (blue arrow), hyperemic disc, and corresponding swept-source optical coherence tomography B scan showing dome-shaped lesion arising from the inner retina (red arrow). (b) Axial section of MRI brain in fluid-attenuated inversion recovery sequence showing subcortical linear hyperintense signals along the left precentral gyrus of the frontal lobe likely to represent cortical tubers (blue arrows), (c) showing facial angiofibromas/adenoma sebaceum (blue arrow), and (d) showing shagreen patch on the back (blue square)

Discussion

We present a spectrum of manifestations in the tuberous sclerosis complex (TSC). In tuberous sclerosis complex, the most common ocular finding is retinal astrocytoma, which is seen in approximately 50% of the patients, and in approximately 50% of these patients, retinal astrocytoma occurs bilaterally.[1] Retinal astrocytoma in TSC usually remains stable, and generally does not affect vision; thus, no treatment is required. However, few retinal astrocytomas progress, and few may develop later which may be a matter of concern, and may require treatment.[2] Thus, precise diagnosis and follow up of retinal astrocytoma is essential not only for supporting the diagnosis of TSC but also to keep an eye for their growth and conversion to aggressive hamartomas so that timely intervention can be taken to prevent the worsening of visual acuity.

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.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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