A white mulberry lesion in the retina

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ABSTRACT
A routine fundus examination after dilated retinoscopy in a 9-year-old child revealed a white mulberry-like lesion with adjacent dilated vessels. After suspicion, systemic investigations were done for the child which included ultrasound abdomen and magnetic resonance imaging brain, both of which showed the presence of abnormal growths. We describe a case of a rare phakomatosis diagnosed primarily due to a unique ophthalmic lesion.

Key words: Lesion, Mulberry, Retina, Sclerosis

DISCUSSION

Tuberous sclerosis is a multisystem involving pathology presenting to an ophthalmologist as a referral usually for ruling out ocular tumors [1]. Apart from the eyes, tumors can be present in the brain as cortical tubers and subependymal ependymomas and also in the kidney as renal angiomyolipomas. Tuberous sclerosis usually shows low penetrance and, thus, is a rare disease. Due to severe mental defects, it usually leads to an early death.

CASE REPORT
A 9-year-old male presented with non-specific complaints of itching in both the eyes for 1 month. On systemic examination, the child also had facial lesions (Fig. 1, Panel A) which were considered to be acne by parents. Axillary freckles, hypopigmented patch on the back, and a hyperpigmented patch on the right temporal region were also present. On examination, the visual acuity was 6/6 in both eyes. The anterior segment was grossly normal. The right eye retina showed a normal disc with an inferotemporal irregular flat white lesion <1/2 disc diameter. The left eye retina showed a normal disc with whitish mulberry-like growth abutting inferonasal disc margin extending up to 1.5 times the disc area with a tortuous feeder vessel (Fig. 1, Panel B and C). There was another transient flat white lesion in the inferonasal quadrant.

Ultrasound scan of the left eye revealed a hyperechoic mass with posterior shadowing suggestive of calcification. Ultrasound abdomen revealed bilateral renal growths. Magnetic resonance imaging of the brain revealed multiple outpouchings from the cortical matter and subependymal outgrowths (Fig. 1, Panel D). The patient was referred to pediatrics and dermatology for workup and prophylaxis for seizures. Siblings were screened. The patient was told to regularly follow-up on the eye outdoor patient department.
or oval tumors, gray in color, which have a smooth surface and are multiple. In general, stationary in its course, aggressive neovascular progression is a rare complication of this tumor.

Aggressive retinal astrocytoma, when encountered, shows a progressive enlargement of the tumor accompanied by total exudative retinal detachment and neovascular glaucoma [3]. Without adequate treatment, enucleation eventually becomes necessary due to blindness and ocular pain. Therefore, precise diagnosis and prompt treatment are important. Incidental diagnosis of tuberous sclerosis by an ophthalmologist, as in this case, is rare. Retinal lesions along with facial hamartomas in our case gave a clue to the diagnosis. The constellation of facial angiofibromas, subependymal nodules, and retinal astrocytic hamartomas is consistent with tuberous sclerosis [4]. Timely follow-up and ophthalmic examinations to look for neovascular proliferation in astrocytomas are imperative to save the patient from irreversible complications.

REFERENCES


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