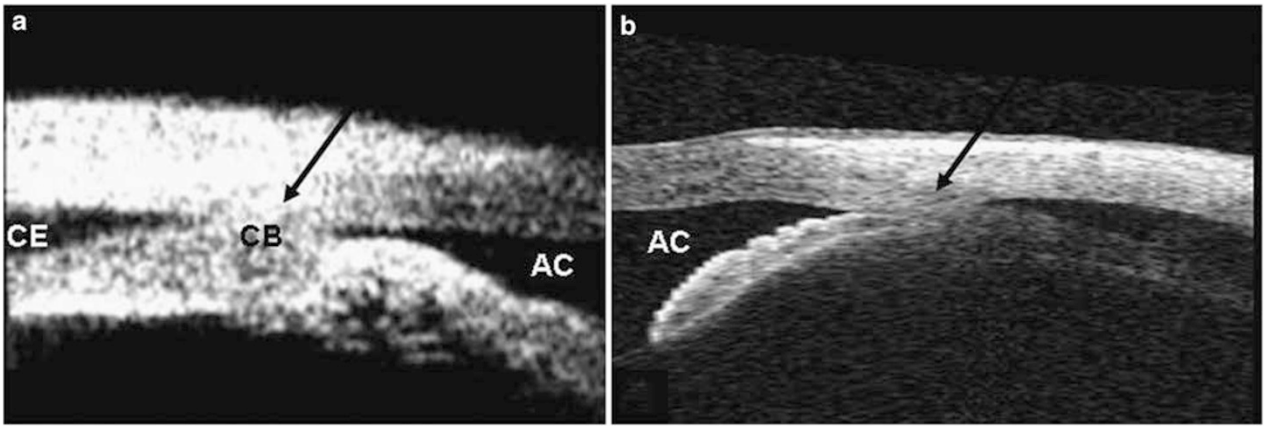

Figure 2 (a) UBM image showing choroidal effusion with no ciliary body detachment at 6-o'clock position (arrow). (b) SL-OCT image showing choroidal effusion with no ciliary body detachment at the same position (arrow). AC = anterior chamber; CB = ciliary body; CE = choroidal effusion.

intervention.[1] UBM has been used for diagnosis, when gonioscopy is not possible due to a shallow anterior chamber or hazy media.[4] Injection of viscoelastic material into the anterior chamber may facilitate gonioscopic visualization when other methods fail.[5] Gonioscopy and UBM may be difficult to perform in younger children. SL-OCT is atraumatic, rapid, and slit-lamp-adapted. Although the SL-OCT provides images with a higher axial resolution, a major limitation compared to the UBM is its inability to visualize structures posterior to the iris.

In our patient, the cleft was well defined and easily located by SL-OCT, producing images similar in quality to those produced by UBM, without risk of further ocular injury or patient discomfort. Testing can be repeated frequently, with minimal difficulty, to follow the ocular response to intervention. Lastly, given that SL-OCT is easily performed in the upright position (compared with majority of UBM devices performed in the supine position), the findings may more likely reflect gonioscopic and slit-lamp biomicroscopic clinical findings.

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Sir,

Novel mutation in *PAX3* gene in Waardenburg syndrome accompanied by unilateral macular degeneration

Waardenburg syndrome (WS) is a congenital pigmentary anomaly that affects the eye, hair, and skin. It is accompanied by facial abnormalities and deafness.[1] WS is clinically and genetically heterogeneous, and WS type 1 (WS1) is characterized by dystopia canthorum. WS1 results from mutations in the *PAX3* gene.[2] We report a patient with WS1 who presented with unilateral vision decrease and a novel mutation in the *PAX3* gene.

Case report

A 54-year-old woman with heterochromia of the right iris noticed a decrease in her vision. She had dystopia canthorum, hypopigmentation of her eyelashes and skin, and unilateral hearing impairment. A diagnosis of WS1 was made. Her best corrected visual acuities were 0.09 OD and 1.5 OS with refractive errors of −16.0 diopters (D) OD and −5.5 D OS. Intraocular pressure was 11 mmHg OD and 12 mmHg OS. The right fundus appeared albinotic over the entire retina accompanied by chorioretinal atrophy in the posterior pole probably due to the high myopia (Figure 1). A B-scan ultrasonogram showed an elongated axial length of 28.1 mm in the right eye, and a normal appearing left eye with an axial length of 25.0 mm. Posterior staphyloma-like changes were not detected in both eyes. Optical coherence tomography revealed a thickened pigment epithelial layer under the macula, suggestive of a myopic macular scar from a possible choroidal neovascularization (Figure 2).

After an informed consent was obtained, a search for mutations in the *PAX3* gene was made from the DNA extracted from the peripheral blood. A novel A→C transversion was identified in exon 5 resulting in a tyrosine→serine change at codon 243 (Figure 3). This codon is located in a highly conserved homeodomain. The change was not observed in 191 healthy controls, and this codon is conserved in different species.

Comment

Most of the symptoms and signs of our patient were consistent with those reported for patients with WS1. However, even with more than 50 mutations in the *PAX3* gene reported, none of the patients has been reported to have a myopic macular degeneration as seen in our patient. In WS, the refractive errors vary considerably from hyperopia to myopia.[3] The *PAX3* gene is a transcription factor that is expressed during embryonic development and is critically involved in the development of melanocytes.[4] The similarity in the phenotypic expression of different point mutations in the *PAX3* gene indicates that these mutations cause a complete loss of function. However, a recent study suggested that different point mutations tend to exhibit independent properties of DNA binding.[5]

The novel mutation found in this study is predicted to alter highly conserved tyrosine at codon 243, which plays a role in homeodomain DNA binding through a phosphate contact.[5] Although further studies are required, it is possible that, owing to the different property of deficient melanocytes, this patient was vulnerable to choroidal neovascularization in association with the high myopia. In any case, ophthalmologists should be aware that patients with WS1 may also have myopic macular degeneration.

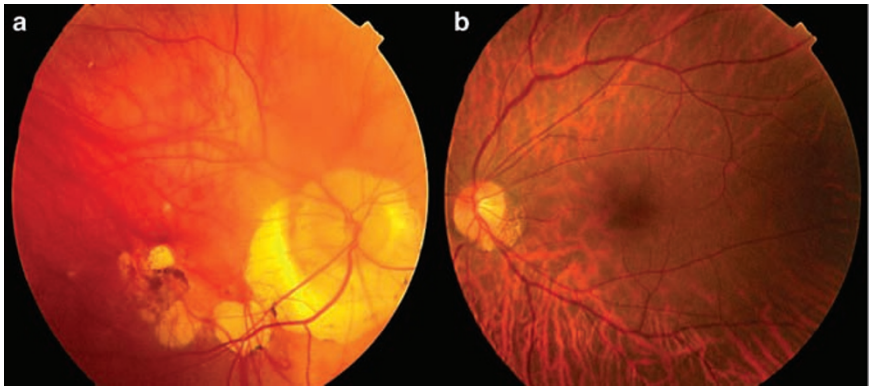

Figure 1 Fundus photographs of a patient with Waardenburg syndrome. (a) Photograph of the right eye showing albinotic appearance and a chorioretinal atrophy in the posterior pole due to high myopia. (b) The left eye shows a conus temporal to the disc with normal appearing macula.

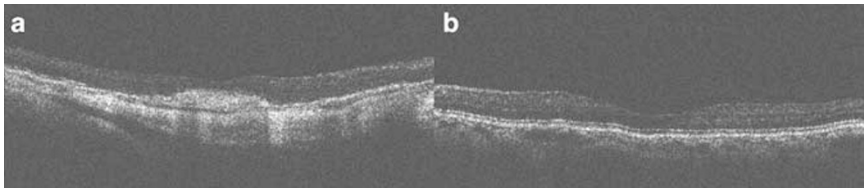

Figure 2 Optical coherence tomographic images of the right and left eye. (a) Optical coherence tomographic image of the right eye shows a thickened retinal pigment epithelial layer under the macula. (b) Optical coherence tomographic image of the left eye showing normal findings.

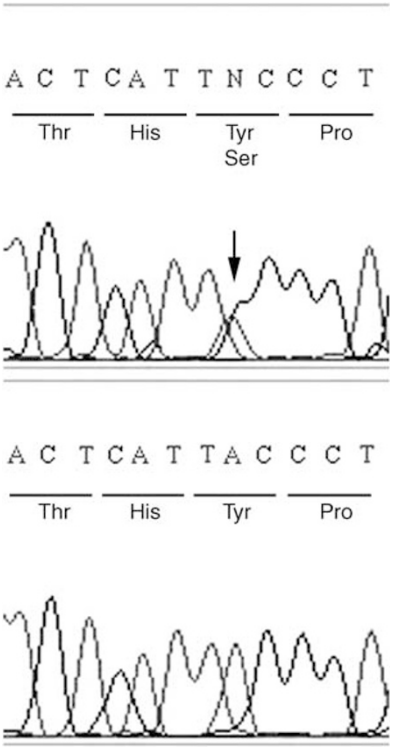

Figure 3 Sequencing results of the patient (upper) and a normal control (lower). A heterozygous A→C transversion can be seen in exon 5 resulting in a tyrosine-to-serine change at codon 243.

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Sir,

Haemorrhagic pituitary tumour presenting with unilateral paracentral visual disturbance

Case report

A 49-year-old postmenopausal woman presented with a 2-month history of 'smeary and patchy' vision in her left eye. Visual acuities were 6/6 bilaterally; Ishihara testing was normal (13/13) on the right but slightly impaired (12/13) on the left. Pupillary reflexes were normal. Slit lamp biomicroscopy, including dilated fundoscopy, was normal. On Amsler grid testing, she described patchy loss of vision paracentrally in the left eye; visual fields to confrontation demonstrated normal blind spot and peripheral fields. Three weeks later her symptoms persisted. Fluorescein angiography and repeat examination were normal. Maculopathy was suspected and electrophysiological examination was requested.

VEPs (Figure 1) showed P100 component delay from both eyes, left worse than right. Both eyes showed greater abnormality in the traces from the contralateral hemisphere than the ipsilateral, suggestive of chiasmal dysfunction. PERG showed no macular dysfunction. Urgent brain MRI revealed a 3×2.3 cm pituitary mass with internal haemorrhage extending superiorly from the pituitary fossa to abut the optic chiasm (Figure 2). Subsequent endocrine tests identified the tumour to be nonfunctioning.

Following urgent neurosurgical assessment, she reported sudden deterioration in vision, suggestive of apoplexy. Humphrey perimetry then identified a small right temporal hemianopia, more marked inferiorly, with more generalized field loss inferotemporally on the left. She underwent emergency transsphenoidal pituitary resection. Histology confirmed a nonfunctioning adenoma.

Comment

This case illustrates two important points. First, the presentation with central visual disturbance reinforces that the classical bitemporal hemianopia may not occur in chiasmal compression, present in one series in only 12 of 34 patients.[1] Second, electrophysiology can detect and localize chiasmal dysfunction even when visual acuity and visual fields are normal, being more sensitive than perimetry or acuity.[2–4] The key diagnostic feature is that potentials generated in the hemisphere contralateral to the stimulated eye show the maximum abnormality.[5] The functional localization by VEPs can direct MRI evaluation, and may assist surgical planning.[1]