Disc case studies – Part 2

Tilted disc syndrome and visual field defect

In the second of these practice-based studies, Kirit Patel considers how life rarely follows the descriptions of textbooks.

IN PRACTICE WHAT we observe and expect to see does not always match up to reality. Using the example of an optic disc coloboma syndrome, I will discuss a visual field defect measured which was not as we would have expected it to be.

CASE HISTORY

A 68-year-old patient came in for a routine eye examination. His previous history included annual examination by an eye specialist for a congenital condition and was discharged 10 years ago. He took an anti-cholesterol medication.

Spectacle prescription

R PLANO / -1.00DC x 90 VA 6/6 ADD +2.00DS N5
L +1.00DS VA 6/6 ADD + 2.00DS N5

IOPs

R 14mmHg
L 15mmHg (Perkins 9am).

Fundus examination

Right eye (Figure 1) revealed:

- Tilted optic disc
- The disc appears to have rotated 25 degrees along the horizontal
- Pronounced crescent on the nasal aspect together with thinning of the retinal pigment epithelium.

Left eye (Figure 2) revealed a normal circular optic disc with a C:D ratio of 0.5.

VISUAL FIELDS AND GDx PLOT

The right eye visual field should have revealed a superior temporal field defect corresponding to the inferior nasal coloboma.

Surprisingly, there was a nasal step and a corresponding loss of nerve fibres in the superior temporal region revealed by the GDx plot (Figure 3). The loss of nerve fibres and the nasal step would immediately lead one to suspect normal-tension glaucoma or possibly a non-arteric anterior ischaemic optic neuropathy.

The left visual field plot revealed no visual field defect and the GDx plot showed a lovely 'hour glass' appearance with no loss of nerve fibres.

The patient did not want to see an eye specialist at this stage since he had had extensive tests carried out in the past. We therefore decided to monitor the patient over a three-month period to assess any more loss of nerve fibres or an increase in the visual field defect.

TILTED DISC SYNDROME

Tilted disc syndrome is a rare congenital condition where the optic disc is rotated upon its axis.

It represents a classical optic nerve coloboma and, while there is no actual rotation of the optic disc, the disc appears more heaped up in the superior aspect due to the hollowing out of the inferior disc, following incomplete closure of the embryonic ocular fissure at six weeks' gestation. Tilted disc syndrome is bilateral in 80 per cent of patients. Visual acuity is rarely affected and often there is myopic astigmatism, typically with oblique axis. There will always be a pronounced crescent of the inferior nasal aspect of the nerve and peripapillary area, known as the conus and sometimes it can extend to involve the entire inferior aspect of the nerve. There can also be moderate to severe thinning of the retinal pigment epithelium, choroid and the sclera and this can result in choroidal neo-vascularisation and haemorrhaging.

Visual field loss is typically a superior temporal defect as the coloboma affects the inferior nasal aspect of the optic disc. Figure 4 shows a patient with bilateral retinal coloboma. If tilted disc syndrome is bilateral, then expect to see a superior bi-temporal field defect similar to those caused by chiasmal compressive lesion – namely, pituitary adenoma, suprasellar masses.
However, the field defect in tilted optic disc syndrome will cross the vertical midline due to the oblique insertion of the papilla into the globe, whereas a chiasmal field defect will not. In our patient there is a vertical crossing and an inferior-nasal field defect in one eye only, therefore, eliminating a chiasmal lesion.

Several studies have described an association between tilted disc syndrome and CNS disease-like tumours such as craniopharyngioma, craniosynostosis and Ehlers-Danlos disease, so CT and/or an MRI scan would be useful.

In our patient there was an inferior-nasal step field defect which did not tally with the inferior nasal coloboma. It is rare to see a nasal step in isolation and scotoma greater than 5-10 degrees should alert the optometrist to glaucoma.

In fact, 40 per cent of patients with glaucoma exhibit a nasal step. Our patient’s unaffected left eye had a deeper cupping but no visual field defect and no nerve fibre deficit.

The GDx laser polarimeter allows practitioners to map the nerve fibre and so the next logical step was to perform this test.

Miraculously, the GDx revealed a deficit in the superior temporal nerve fibre layer which matches the inferior nasal field defect.

Could this mean this patient has primary normal-tension glaucoma? It was unwise to reach such a simple conclusion and with the patient’s agreement we will be analysing the visual fields and nerve fibre layer every three to six months to monitor any further loss of nerve fibres.

Not having any previous notes meant not knowing whether the patient had this visual field deficit since birth and whether any scans were taken.

Further reading
Kline LB. Optic Nerve Disorders, 1996; 37-53.

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