CASE REPORT

Unusual association of keratoconus with situs inversus and micronystagmus

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SUMMARY

Keratoconus is a bilateral non-inflammatory ectasia of the cornea and the presentation is usually as an isolated condition. A few ocular and a number of systemic associations have been described. Situs inversus of retinal vessels is a rare developmental anomaly due to anomalous insertion of optic stalk into the optic vesicle. In this case report, we describe an unusual association of keratoconus with situs inversus and micronystagmus in a young man. An additional feature, a blue dot cataract, was also present. To the best of our knowledge, such a conglomeration of multiple ocular features has not been described previously.

BACKGROUND

Keratoconus is a bilateral non-inflammatory ectasia of the cornea. The involvement of both eyes may be symmetric or asymmetric. The disease is common, with reported incidence varying from 50 to 230/100 000 of the general population.¹ Six to eight per cent of patients may have a positive family history.² It is usually an isolated condition. Lately, a wide range of ocular associations have also been described with mechanical trauma being implicated as an important risk factor, in other words, vernal keratoconjunctivitis³ and floppy eyelid syndrome.⁴ Other ocular associations described in the literature are Leber's congenital amaurosis,⁵ retinitis pigmentosa,⁶ Fuchs' corneal dystrophy,⁷ lattice dystrophy,⁸ granular dystrophy,⁹ cataract¹⁰ etc.

Situs inversus of retinal vessels is a rare developmental anomaly due to anomalous insertion of optic stalk into the optic vesicle; the central retinal vessels emerge from the nasal aspect of the optic nerve head instead of from the temporal aspect, and course nasally before turning temporally. The condition is bilateral in 80% of cases and has been described in myopia,¹¹ tilted optic disc,¹² optic disc pit,¹³ optic disc coloboma,¹⁴ Ehlers Danlos syndrome,¹⁵ familial dextrocardia¹⁶ and optic neuritis.¹⁷ Other associated anomalies reported are cilioretinal arteries¹⁸ and ectopia of macula.¹⁸

We report an unusual coexistence of bilateral keratoconus and situs inversus of the optic disc in a young man. Micronystagmus and congenital blue dot cataracts were also present in both eyes. To the best of our knowledge, bilateral keratoconus with bilateral situs inversus of the optic disc has not been reported before.

CASE PRESENTATION

A 25-year-old man presented to us with a 2-year history of progressive diminution of vision for

distance in both eyes. There was no significant ocular or medical history. His family history was not significant.

On ocular examination, his uncorrected distance visual acuity was 6/60 in the right eye and 6/24 in the left, on Snellen's chart. The best corrected visual acuity (BCVA) was 6/18 (right eye) with -4.00 DS/ -5.00 DC at 45° and 6/12 (left eye) with -3.00 DS/ -6.00 DC at 165° . The near visual acuity was N6 in both eyes. On slit lamp biomicroscopy, corneal thinning just inferior to the pupil was seen in both the eyes (figure 1A). Fleischer ring was present in the inferior part of the cornea (figure 1B) and, on diffuse examination, enlarged corneal nerves could also be seen. Multiple small whitish coloured lenticular opacities were seen in the central and peripheral parts of the lens suggestive of blue dot cataract (figure 1C).

Fundus examination of both eyes revealed vessels emerging from the nasal aspect of the optic disc and choroidal sclerosis. Also present were myelinated nerve fibres superior to the disc in the left eye (figure 2). A diagnosis of situs inversus of the disc was performed. Jerky nystagmus with fast and slow phases of small amplitude was also seen on fundus evaluation. The fast phase was towards the right and the slow phase towards the left. The patient was not aware of its presence and thus we referred to it as micronystagmus.

INVESTIGATIONS

On Pentacam (Oculus, Wetzlar, Germany), K1 and K2 were 45.6 D and 50.1 D @ 137.6° in the right eye, and 45.7 D and 50.7 D @ 75.4° in the left eye, respectively. The thinnest pachymetry was 528 μ in the right eye and 525 μ in the left eye. The maximum elevation was +19 (more than 15) on the anterior elevation map and +43 (more than 20) on the posterior elevation map in the right eye (figure 3), and +20 on the anterior elevation map and +44 on the posterior elevation maps in the left eye (figure 4) in the central 3 mm zone, suggestive of central cone in both eyes. The Belin maps confirmed the diagnosis of keratoconus. The average progression index was also more than 1.2 in both eyes.

DISCUSSION

Keratoconus typically has its onset at puberty and is progressive up to the age of 30–40 years. It seldom occurs in children and rarely may be congenital.¹ The aetiology of the disease is not well understood with both genetic and environment factors being implicated in the causative role. In the past few years, although a number of candidate genes have

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Figure 1 Slit lamp image showing (A) corneal thinning, (B) Fleischer ring, (C) blue dot cataract.

been identified that have increased the understanding in the molecular aetiology of the disease, a large number of genetic factors are yet to be elucidated.¹⁹

An anomalous insertion of the primitive optic stalk into the optic vesicle generally results in congenital crescents or conus, but in some cases, this anomalous insertion is of a less dramatic degree resulting in tilting of nerve head (dysversion of optic disc).¹⁸ In dysversion of the optic disc, the central retinal vessels usually emerge from the nasal instead of temporal side (being rotated through 180°), which is defined as situs inversus of the disc, but, rarely, the distortion may be in a vertical direction resulting in an oblique tilting of the disc.¹⁸ The condition may be associated with bitemporal or altitudinal field defects.¹⁸ The presence of keratoconus in a patient with unilateral situs inversus due to tilted disc has been reported.²⁰ Our patient had bilateral situs inversus with no tilting of the disc and had medullated nerve fibre in the left eye, denoting a malfunction in optic nerve development. Although situs inversus of the disc alone is usually visually innocuous, a case report of its association with reduced binocularity and stereopsis has been described.²¹

The association of increased corneal astigmatism and tilted disc syndrome with situs inversus has been described in the

literature,²² but no genetic association has been ascribed. As the presence of keratoconus is not uncommon, its presence in our patient, as an independent coexistent disease or a manifestation of an underlying genetic defect, cannot be precluded.

The association of nystagmus and keratoconus has been reported mostly in the context of a syndrome having other systemic clinical features as well. Our patient had jerky microneystagmus, which was apparent only on fundus examination, and no other clinical features suggestive of any syndrome. Since microneystagmus has very small amplitude with fast and slow phases within parafoveal areas,²³ it is not known to affect vision. The aetiopathogenesis of microneystagmus is not well described. Although considered physiological, it has been reported in patients with cone dystrophy²⁴ and oculopalatal disorders.²⁵ The presence of microneystagmus in our patient as an incidental finding or as a sign of visual impairment due to situs inversus cannot be ruled out. There was also presence of an additional feature, blue dot cataract. To the best of our knowledge, this conglomeration of multiple ocular features in the form of situs inversus of the optic disc with microneystagmus and blue dot cataract, in association with keratoconus, has not been reported before.

Figure 2 Fundus photograph: Bilateral situs inversus of the disc and myelinated nerve fibre above the disc in the left eye.

Figure 3 Showing Quad map of right eye on pentacam.

Figure 4 Showing Quad map of left eye on pentacam.

Patient's perspective

I have been told about my eye condition by the concerned doctors and I give my consent to report on it for publication as it will increase the understanding of my disease in future.

Learning points

- ▶ Situs inversus has usually been described in association with tilted disc syndrome, but it can also be present as an isolated condition.
- ▶ Although situs inversus is usually visually innocuous, it can also affect visual development.
- ▶ Keratoconus is usually an isolated condition. Its association with other ocular anomalies of the eye points towards some common pathogenesis.

Contributors PJ and RA identified the case and drafted the report. RA and JG helped in formulating the final version submitted.

Competing interests None declared.

Patient consent Obtained.

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