CLINICAL SCIENCE

Optical coherence tomography of superior segmental optic hypoplasia

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Aim: To describe optical coherence tomography (OCT) images of superior segmental optic hypoplasia (SSOH).

Methods: Five patients (two men and three women, ages 10–45 years) presented with ophthalmoscopic features and visual field defects of SSOH. All affected eyes had good visual acuity and inferior altitudinal or inferonasal visual field loss. The mothers of three patients had type 1 diabetes mellitus. OCT (Humphrey Instrument, CA, USA) was used to evaluate tomographically the optic disc and peripapillary retina of both eyes of each patient. Control data on retinal nerve fibre layer (RNFL) thickness were obtained from 13 normal eyes, one eye each from 13 normal subjects.

Results: Seven of 10 eyes in patients had SSOH. Scans in the vertical meridian through the affected optic discs showed a superior defect of the optic disc associated with decreased RNFL thickness and, in some cases, an abnormal extension of a complex of retinal pigment epithelium and choroid over the edge of the lamina cribrosa. Circular scans around the seven optic discs revealed various decreases of peripapillary RNFL thickness in the superior quadrants. Vertical scans through the fovea also showed superior thinning of RNFL. Quantitative assessment of the peripapillary RNFL thickness revealed significantly decreased values in the superior quadrants compared to normal eyes.

Conclusions: OCT provides a new tool for quantitative evaluation of optic nerve hypoplasia as exemplified in this study of SSOH. It can reveal minimal degrees of segmental hypoplasia previously undetected.

Superior segmental optic hypoplasia (SSOH), also termed “topless optic disc” by Landau *et al*, is a developmental disorder characterised by relative superior entry of the central retinal artery, superior retinal nerve fibre deficiency, superior scleral halo, and superior disc pallor.¹ Patients with SSOH have good visual acuity and inferior altitudinal or sector-like visual field defects.^{1–7} The pathogenesis of SSOH remains undefined. Several reports point to an association with maternal type 1 diabetes mellitus.^{1–3,5} This report describes optical coherence tomography (OCT) images obtained from five patients with SSOH.

PATIENTS AND METHODS

Patients

We identified five patients with SSOH while evaluating them for incidentally discovered inferior visual field defects (Table

1). There were two men and three women. The mean age at examination was 33 years (range 10–45 years). Routine examinations included visual acuity assessment, cycloplegic refraction, visual field testing using the Goldmann perimeter, direct ophthalmoscopy, and fundus photography. Scanning laser ophthalmoscopy, colour vision tests, and electroretinography were performed in some cases.

None of the patients had neurological disease. Family history in two patients (patients 1 and 2) was not contributory. Patient 4 was the mother of patient 5. Type 1 diabetes mellitus was present in the mothers of patients 3, 4, and 5.

Methods

OCT (Humphrey Instrument, CA, USA) images were obtained from both eyes of each patient, by the methods described by

Table 1 Clinical findings of five patients with superior segmental optic hypoplasia

	Patient 1		Patient 2		Patient 3		Patient 4		Patient 5*	
Age (years), sex	24, male		24, male		45, female		44, female		10, female	
Family history	—		—		—		+		+	
Neurological disease	—		—		—		—		—	
Maternal diabetes mellitus	—		—		+		+		+	
Laterality	unilateral		unilaeteral		bilateral		unilaeteral		bilateral	
Visual symptoms	—		—		—		—		—	
	R	L	R	L	R	L	R	L	R	L
Visual acuity	20/20	20/20	20/20	20/20	20/20	20/20	20/20	20/20	20/20	20/20
Refractive errors	−3.0 D	−1.5 D	−6.0 D	−6.0 D	−1.5 D	−1.75 D	+0.5 D	+0.5 D	+0.5 D	+1.0 D
Visual field	IAD	normal	normal	IAD	ITD	ITD	normal	ITD	IAD	IAD
Ophthalmoscopic findings										
Superior entrance of CRA	+	—	—	+	+	+	—	+	+	+
Loss of superior RNFL	+	—	—	+	+	+	—	+	+	+
Superior disc pallor	+	—	—	+	+	+	—	+	+	+
Superior scleral halo	+	—	—	—	+	—	—	—	—	—

*Daughter of patient 4.

IAD = inferior altitudinal defect; ITD = inferotemporal defect; CRA = central retinal artery; RNFL = retinal nerve fibre layer.

Figure 1 Patient 1. Left panels: right eye with SSOH; right panels: normal left eye. (A) Visual fields. The affected eye has an inferior altitudinal defect. (B) Optic discs. The affected eye shows superior entry of the central retinal artery and vein. (C) Left image is a vertical OCT scan through the optic disc. It shows shrunken superior portion of the optic disc with thinned nerve fibre layer and extension of a complex of retinal pigment epithelium and choroid towards the centre of the optic disc. Right, the unaffected eye with a normal configuration of optic nerve head and peripapillary retina. Asterisks represent termination of a complex of retinal pigment epithelium and choroid at the level of lamina cribrosa. (D) Left image represents a circular scan at a 1.74 mm radius from the centre of the optic nerve head. (Top) OCT images. (Bottom) Quantitative assessment of thickness of peripapillary retinal nerve fibre layer (RNFL) for twelve 30 degree subdivisions from the temporal to the inferior segment. The affected eye shows decrease in RNFL thickness. The RNFL thickness map has a single hump with a peak in the inferior quadrant. The normal fellow eye shows a double hump map with peaks in the superior and inferior quadrants. Thick bar = 250 μ m. (E) Left side image represents a scan in the vertical meridian through the fovea. The affected eye demonstrates a normal foveal configuration and decreased RNFL thickness superior to the fovea. The unaffected eye is normal in RNFL thickness. Explanation of symbols is the same in the following figures.

Uchino *et al*⁸ for obtaining high resolution cross sectional tomograms of the optic nerve head and the posterior fundus. Our routine examination consisted of obtaining (1) scans in the vertical meridian through the optic nerve head (Figs 1–5C), (2) circular scans around the optic disc at the 1.74 mm radius from the centre of the optic disc (Figs 1–5D), and (3) vertical scans through the fovea (Figs 1–5E). An additional session was performed in selected patients to take horizontal and vertical scans in various posterior retinal areas (Fig 6). The configuration of the optic nerve head and the thickness of retinal substructures were evaluated qualitatively with reference to the false colour coded images.

The thickness of the peripapillary retinal nerve fibre layer (RNFL) was assessed quantitatively using commercially

Figure 2 Patient 2. Left panels: right eye, normal. Right panels: left eye with SSOH. (A) Visual fields. The affected eye has an inferior altitudinal defect. (B) Optic discs. The affected eye shows superior entry of the central retinal artery and vein. Both eyes have temporal conus. (C) Vertical scans through the optic disc. The affected eye shows a defective configuration of the optic disc due to shrinkage (hypoplasia) of the upper “half.” (D) Circular scan of the peripapillary area. The affected eye shows absence of the RNFL in the superior half, with a single hump pattern of RNFL thickness map. (E) Vertical scan through the macula. The affected eye shows absence of RNFL superiorly but not inferiorly.

available OCT software in twelve 30 degree segments and four 90 degree quadrants. The mean RNFL thickness was determined from three circular scans in each eye of each patient. In addition, control data of peripapillary RNFL thickness were obtained from 13 eyes of 13 healthy subjects (eight men and five women, aged 38–60 years with the mean of 47 years).

Statistical analysis was performed using the unpaired Student's *t* test for the peripapillary RNFL thickness between SSOH eyes and normal eyes.

RESULTS

All patients were asymptomatic and had good visual acuities with correction of mild to moderate myopia. Goldmann perimetry revealed inferior altitudinal visual field defects in four eyes and inferonasal defects in three eyes (Figs 1–5A). Results of other functional examinations including pupillary responses, colour vision, and electroretinography were unremarkable. The intraocular pressures of each patient were normal.

Superior segmental optic hypoplasia was unilateral in three patients (patients 1, 2, and 4) and bilateral in two patients (patients 3 and 5). Ophthalmoscopy and optic disc photography of the seven affected eyes showed various features of

Figure 3 Patient 3. Left panels: right eye; Right panels: left eye. Both eyes have SSOH. (A) Visual fields. An inferonasal visual field defect is demonstrated in both eyes, being much more prominent in the right eye. (B) Optic discs. The right optic disc shows superior entry of the central retinal artery. The left eye has relatively mild anomaly, which shows a thinner neuroglial rim superiorly. (C) Vertical scan through the disc. Both eyes show thinning of the RNFL in the superior portion of the optic disc. (D) Circular scan of the peripapillary area. Both eyes show loss of superior RNFL in the RNFL thickness map. The peripapillary RNFL defect appears less severe in the left eye. (E) Vertical scan through the fovea. Right eye shows RNFL defect in areas superior to the fovea.

SSOH. Four eyes of four patients demonstrated the relative superior entrance of the central retinal artery, superior RNFL and superior disc pallor. Although obviously abnormal in OCT examinations, ophthalmoscopy and optic disc photography in three eyes of three patients showed nearly normal findings (Figs 1–5B). Scanning laser ophthalmoscopy performed in selected patients confirmed retinal nerve fibre loss in the superior peripapillary region but not inferiorly.

OCT scans in the vertical meridian through the optic disc disclosed that eyes with SSOH had an asymmetric tomographic configuration due to structural changes in the superior portion, distinct from the symmetric configuration in normals. In the superior segments of affected optic discs, the RNFL contiguous with the optic nerve was thinned, and in some eyes the complex of retinal pigment epithelium and choroid extended more towards the centre of the optic disc cup, terminating over the edge of the lamina cribrosa and suggesting superior narrowing of the scleral canal (Figs 1–5C). Circular OCT scans of the peripapillary retina with a 1.74 mm radius from the centre of the optic disc showed a decrease of the RNFL thickness in the superior quadrants of the affected eyes (Figs 1–5D). Scans in the vertical meridian through the fovea revealed decrease of the RNFL thickness superior to the fovea (Figs 1–3E). Notably, three eyes with minimal fundus features of SSOH (left eyes of

Figure 4 Patient 4. Left panels: right eye, normal. Right panels: left eye with minimal SSOH. (A) Visual fields. The affected eye shows a mild inferonasal defect. (B) Optic discs. The affected eye shows almost normal appearance. (C) Vertical scan through the disc. The affected eye shows slightly thinned RNFL in superior portion. (D) Circular scan of peripapillary region. The affected eye shows a mild loss in the superior segment of the RNFL.

patients 3, 4, and 5) demonstrated recognisable thinning of the RNFL superiorly.

Scans elsewhere in the superior retinal quadrants also revealed a decrease of RNFL thickness (Fig 6A). Scans in the horizontal meridian through the fovea showed normal RNFL thickness nasally and temporally (Fig 6B).

Three unaffected fellow eyes of five patients demonstrated, as expected, normal optic nerve head configuration and normal RNFL thickness in all quadrants (Figs 1–5C).

Quantitative assessment of peripapillary RNFL thickness was performed using the OCT software. In normal eyes, the peripapillary RNFL thickness varied in a “double hump” pattern in the superior and inferior quadrants (Figs 7 and 8); the mean thickness value of 13 normal eyes was 88.3 μm (SD 15.8) in the temporal quadrants, 127.3 (15.8) in the superior, 81.7 (14.6) in the nasal, and 137.3 (15.8) in the inferior. In eyes with SSOH, the RNFL thickness varied in a “single hump” pattern in the inferior quadrant; the mean thickness value of seven affected eyes was 74.7 (SD, 11.6) in the temporal quadrants, 56.7 (25.9) in the superior, 47.2 (24.0) in the nasal, and 124.8 (19.2) in the inferior. A statistical comparison between the affected eye group and normal eye group revealed that for twelve 30 degree subdivided segments the affected group had significantly decreased RNFL thickness in four segments from the superotemporal to superonasal segments ($p < 0.001$) (Fig 8) and that for the four 90 degree quadrants the affected group had significantly decreased RNFL thickness in the superior quadrants ($p < 0.001$) (Fig 7). The normal fellow eyes of three patients showed RNFL thickness within the normal range.

Figure 5 Patient 5. Left panels: right eye; Right panels: left eye. Both eyes have SSOH. (A) Visual fields. An inferior altitudinal defect is demonstrated in right eye. An inferonasal defect is shown in left eye. (B) Optic discs. Right eye shows superior entry of the central retinal artery and vein. Left eye shows normal appearance without thinning RNFL. (C) Vertical scan through the disc. Right eye shows abnormal configuration of the superior disc, decrease of the RNLF thickness in the superior portion of the optic disc, and extension of a complex of retinal pigment epithelium and choroid over the region of lamina cribrosa. Left eye appears to have thinned RNLF in the superior portion of the optic disc. (D) Circular scan around the disc. Both eyes demonstrate a loss of RNFL in the superior quadrant, with a single hump RNFL thickness map.

DISCUSSION

OCT images of SSOH presented here provide corollaries for what has been reported previously about these disc anomalies based upon their ophthalmoscopic and fundus photographic appearance. In vertical tomographic images OCT shows the disproportion between the normal lower part of the disc under the central retinal artery entrance and the hypoplastic portion above the central retinal artery entrance. Likewise, vertical OCT sections show the differences in the RNFL thickness below the disc and above and provide unique quantitative information about this thickness. Also, in some cases, OCT shows an overlap, or intrusion, of the superior peripapillary pigment epithelium and choroid over the edge of the lamina cribrosa (see Figs 1, 2, and 5). This fact may contribute to the appearance of the superior peripapillary halo, or crescent, previously described in SSOH.⁵

Horizontal OCT sections through the disc and fovea show that the papillomacular RNFL is intact—that is, normally thick, a finding that correlates with the normal visual acuity and colour vision in eyes affected with SSOH.

Our OCT study of eyes with partial inferior field defects confirms quantitatively what Okazaki and coworkers and Namba and coworkers reported clinically.^{9,10} Segmental hypoplasia can be so mild that it easily can be overlooked ophthalmoscopically (see cases 3, 4, and 5 in our report). This fact, developed from our study, indicates that all previous ophthal-

Figure 6 (Top) Scans in the vertical and horizontal meridians through various retinal areas in the affected left eye of patient 2, displayed in tomographic scanning direction. The RNFL is thinned in areas above the horizontal meridian. S = superior; I = inferior; F = fovea; N = nasal. (Bottom) Scans in the horizontal meridian through the fovea. Left panel, normal eye; Right panel, affected eye (patient 2). The affected eye shows no involvement of RNFL in areas nasal and temporal to the fovea.

moscopic surveys of the incidence of SSOH have underestimated its frequency of occurrence. OCT will be a powerful clinical tool in future studies of this problem.

The anatomical findings from our OCT study of SSOH do not contribute to the mystery of its embryogenesis. SSOH frequently, but not always, occurs in offspring of type 1 diabetic mothers.³⁻⁶ It has been reported in identical twins born of a mother with type 1 diabetes.¹¹ Three of our five patients had

Figure 7 The peripapillary RNFL thickness (μm) assessed from the 1.74 mm radius circular scan is shown for each quadrant around the optic disc. Open squares represent mean values from 13 eyes of 13 normal subjects, and solid squares represent mean values from seven eyes with SSOH. Error bars indicate 1 SD from the mean. Asterisks indicate a significant defect of the RNFL thickness in affected eyes ($p < 0.001$). T = temporal quadrant; S = superior; N = nasal; I = inferior.

Figure 8 The peripapillary RNFL thickness (μm) assessed from the 1.74 mm radius circular scan is shown for 30 degree segment around the optic disc. Open squares represent mean values from 13 eyes of 13 normal subjects, and solid squares from seven eyes with SSOH. Error bars indicate 1 SD from the mean. Asterisks indicate a significant defect of the RNFL thickness in affected eyes ($p < 0.001$). T = temporal quadrant; S = superior; N = nasal; I = inferior.

diabetic mothers. We believe ours is the first report of SSOH in a type 1 diabetic mother, herself an offspring of a diabetic mother, and in her non-diabetic daughter (cases 4 and 5). Clearly, genetic factors can play a part.

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