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Superior segmental optic hypoplasia as a differential diagnosis of glaucoma

Tetsuya Yamamoto

Abstract:

Superior segmental optic hypoplasia (SSOH) is a congenital anomaly affecting the optic nerve head and retina. Although the conventional characterization of SSOH emphasizes the relatively superior entrance of the central retinal artery, the pallor of the superior optic disc, a superior peripapillary halo, and thinning of the superior nerve fiber layer, we encounter many cases with rim thinning in the superior nasal region that corresponds to a nerve fiber layer defect and an inferior wedge-shaped visual field defect connecting to the blind spot. However, among Asians, such cases usually lack pallor of the superior optic disc and more resemble glaucomatous optic neuropathy. We found the prevalence of SSOH to be 0.2%/eye and 0.3%/case among Japanese. We also noted that approximately half of all SSOH eyes show visual field changes. SSOH is an important differential diagnosis of glaucoma, especially normal-tension glaucoma, in Asian populations.

Keywords:

Glaucoma, nerve fiber layer defect, optic nerve head, superior segmental optic hypoplasia

Introduction

Superior segmental optic hypoplasia (SSOH) is a congenital anomaly affecting the optic nerve head and the retina. The conventional Western literature emphasizes its main features as relatively superior entrance of the central retinal artery, pallor of the superior optic disc, a superior peripapillary halo, and thinning of the superior nerve fiber layer. The optic anomaly was first reported in 1977 by Petersen and Walton.[1] Kim *et al*.[2] coined the term SSOH in 1989.

SSOH is considered to be a sign of maternal diabetes mellitus.[1-3] Petersen and Walton[1] reported 17 cases with optic hypoplasia that developed in children whose mothers had insulin-dependent diabetes mellitus. Landau *et al*.[3] identified three SSOH cases in a prospective study of 34 children whose mothers were patients with insulin-dependent diabetes mellitus.

Photographs of the optic nerve head from a previous study[2] showed apparent dissimilarity between SSOH and glaucoma. However, the author independently identified cases from glaucoma clinics having features similar to those of SSOH, which included rim thinning in the superior nasal region with a corresponding nerve fiber layer defect (NFLD) and inferior wedge-shaped visual field defect connecting to the mariotte blind spot [Figure 1a and b]. However, these cases lacked both the pallor of the superior optic disc and a positive family history of diabetes mellitus. In such cases, the following features are typically seen: an optic disc resembling glaucoma, normal intraocular pressure (IOP), a relatively young patient, normal visual acuity, and no complaint of visual disturbance. Further, visual field changes are nonprogressive in the majority of cases.

Here, the author presents the results of two previous studies[4,5] of "SSOH": A population-based study and a university hospital-based study.

Department of Ophthalmology, Gifu University Graduate School of Medicine, Gifu, Japan

Address for correspondence:
Prof. Tetsuya Yamamoto, Department of Ophthalmology, Gifu University Graduate School of Medicine, 1-1 Yanagido, Gifu-Shi 501-1194, Japan.
E-mail: mmc-gif@umin.net

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Outlines of Our Previous Studies

Study 1: Prevalence of superior segmental optic hypoplasia

We investigated the prevalence and characteristics of SSOH in 14,779 individuals, aged 40 years or older, who participated in a large-scale eye disease screening project conducted in Tajimi, Japan, between September 2000 and October 2001, designated Study 1.[4] The project was comprised of two study populations: one consisted of 4000 randomly selected citizens, aged 40 years or older, who underwent a detailed ophthalmological checkup as part of the well-known Tajimi study;[6] the other consisted of a screening of the general population. In the latter, the following ophthalmological examinations were conducted for both eyes: visual acuity testing, refractometry with a refractometer, corneal thickness measurement, frequency-doubling technology (FDT) perimetry, 45° fundus photography, Goldmann applanation tonometry, slit-lamp biomicroscopy, and van Herick testing. Of the 50,165 citizens invited, 14,779 actually participated in the screening, yielding a response rate of 29.5%. In addition, we offered a definitive examination on a separate day to participants who were suspected of having any ocular disease. At the definitive examination, visual field testing with a Humphrey Field Analyzer, using the Central 30-2 SITA Fast program, was conducted for individuals with suspected optic disc abnormalities of any type.

The author reviewed all of the photographs for the presence of ocular abnormality in the optic nerve head and retina, paying special attention to signs indicating the presence of SSOH. Fundus photographs of 28,396 eyes of 14,431 cases were successfully reviewed.

In this study, we defined SSOH as rim thinning of the optic disc most prominent in the superior nasal region with a corresponding NFLD in the superior nasal region. We then subdivided cases into definite type and suspect type according to the presence or absence of the corresponding visual field defect based on perimetric results from either the FDT or Humphrey Field Analyzer. We identified SSOH in 54 eyes of 37 cases (bilateral: 17, unilateral: 20; 0.2%/per eye and 0.3% per/case) in total [Table 1]. Owwf the 37 cases, 23 (0.2%) showed the corresponding visual field defect in at least one eye. There were 23 definite type cases (bilateral: 5, suspect in fellow eyes: 5, and unilateral: 13) and 14 suspect type cases (bilateral 7 and unilateral 7). There were 10 males and 27 females, aged 53.1 ± 10.3 (mean ± standard deviation, range: 40–76 years) years. The IOP by Goldmann applanation tonometry was 14.2 ± 2.5 mmHg (9–19 mmHg). The spherical equivalent was – 1.54 ± 2.72 D (–8.25 ± 1.88 D). Only one individual had a prior history of diabetes mellitus.

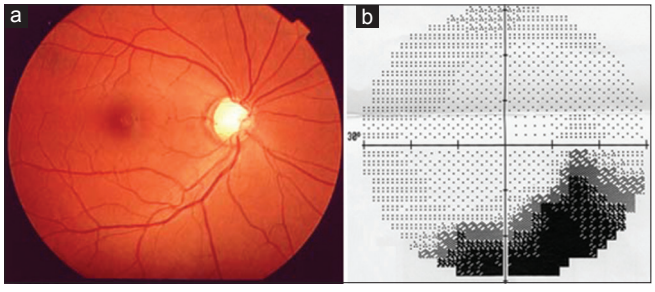

Figure 1: A case of superior segmental optic hypoplasia. A 43-year-old male with normal intraocular pressure (a) fundus photograph showing rim thinning in the superior nasal region and nerve fiber layer defect, (b) visual field indicating an inferior wedge-shaped visual field defect connecting to the blind spot

Study 2: Clinical features of superior segmental optic hypoplasia

We determined the clinical features of SSOH and compared nerve fiber layer thickness between SSOH and normal individuals quantitatively by spectral-domain optical coherence tomography (SD-OCT) in a retrospective fashion at the Department of Ophthalmology, Gifu University Hospital, Japan, in our study, designated Study 2.[6] We examined the medical charts detailing 106 eyes of 59 patients with SSOH [Figure 2]. All patients were examined between 2004 and 2015. We used the same diagnostic criteria for SSOH as in Study 1.[4] Eyes with SSOH were classified as being of the definite type or suspect type according to standard automated perimetry, as in Study 1. The findings of the SD-OCT images of 35 eyes with SSOH were compared with those of the 35 normal eyes.

All of the patients underwent a routine ophthalmological examination, including measurements of the best-corrected visual acuity, subjective refraction with a refractometer, slit-lamp biomicroscopy, IOP measurements with a Goldmann applanation tonometer, ophthalmoscopy, and optic nerve imaging with an SD-OCT. If glaucoma or optic nerve hypoplasia was suspected, perimetry with a Humphrey Field Analyzer was performed using the Central 30-2 SITA standard program.

Table 1: The age and gender distribution of superior segmental optic hypoplasia found

Age (years)	Male	Female	Total
40-49	4/985 (0.4)	11/2009 (0.5)	15/2994 (0.5)
50-59	3/1513 (0.2)	10/3101 (0.3)	13/4614 (0.3)
60-69	1/1861 (0.1)	5/2427 (0.2)	6/4288 (0.1)
70-	2/1094 (0.2)	1/1441 (0.1)	3/2535 (0.1)
Total	10/5453 (0.2)	27/8978 (0.3)	37/14431 (0.3)

Number of cases/population studied (prevalence as percent). Reprinted from reference 4, with permission

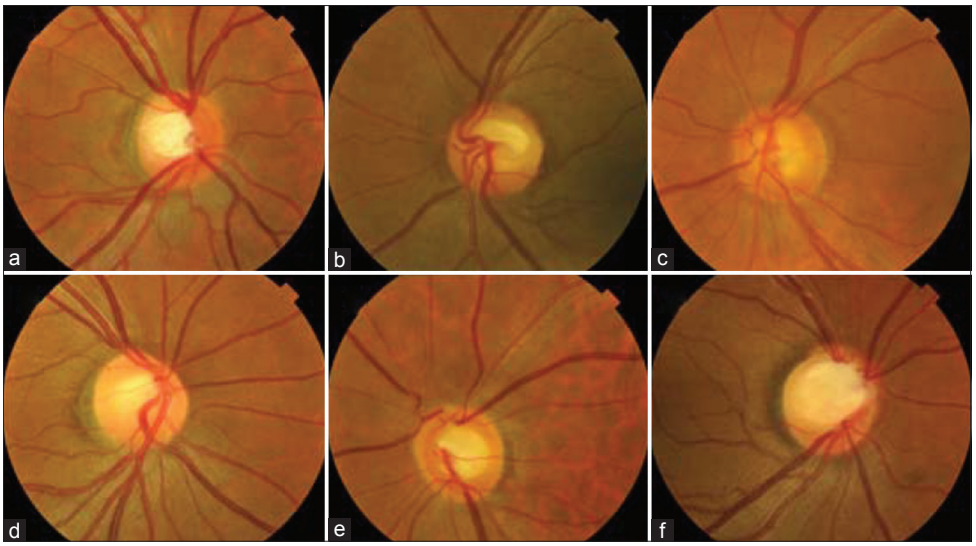

Figure 2: Examples of superior segmental optic hypoplasia cases found in Study 2. Reprinted from Yagasaki *et al*.,[6] with permission

Of the 106 eyes with SSOH, 56 (52.8%) were classified as the definite type and 50 (47.2%) were classified as the suspect type. Of the 56 definite type eyes, 14 had inferior visual field defects contiguous to the blind spot. The mean age of individuals was 35.7 ± 15.7 (mean ± standard deviation) years. The mean IOP was 14.9 ± 3.4 mmHg, the mean refraction error was −3.63 ± 3.38 D, and the average mean deviation was −2.51 ± 4.00 decibels.

SD-OCT showed that the average of the total nerve fiber layer thickness was significantly thinner in the SSOH group than in the normal group. Sectorial analysis demonstrated that the nerve fiber layer was thinner than that of controls in all quadrants. The comparison of the hourly sectors showed that the nerve fiber layer was thinner at the 10, 11, 12, 1, 2, 3, 5, and 6 o'clock sectors in the SSOH group than in the controls when left eyes were turned over and presented like right eyes.

Comments

A crucial feature of SSOH in the Asian population is its resemblance to glaucomatous optic neuropathy, in terms of rim thinning and the presence of NFLD. The prevalence of normal-tension glaucoma (NTG) is known to be 3.6% in Japanese, aged 40 years or older, and it accounts for 72% of all glaucoma.[5] As shown in our previous Study 1, the prevalence of SSOH is 0.3% in Japanese, which yields an SSOH to NTG ratio of approximately 1:12. Thus, when diagnosing NTG, differentiation from SSOH is important since both conditions lack elevated IOP. To succeed in this differentiation, we must carefully identify rim thinning dominantly in the superior nasal region and note characteristic visual field changes: an inferior altitudinal defect or inferior sector defect connecting to the blind spot. Increased awareness of these features as indicative of SSOH should be encouraged in clinical practice.

Our previous study (Study 1) showed that approximately one-half of the SSOH eyes were of the definite type (28 eyes, 51.9%) and one-half were of the suspect type (26 eyes, 48.1%). In Study 2, we found a similar distribution: definite type, 56 eyes (52.8%) and suspect type: 50 eyes (47.2%). Although the individuals in Study 1 were part of a large-scale eye disease screening study and all were at least 40 years of age, the proportion of each type of SSOH was nearly identical to that in Study 2. This suggests that SSOH develops in all age groups similarly and that most cases are diagnosed by chance, without apparent visual complaints, due to a lack or paucity of ocular symptoms in the majority of cases.

Previous studies strongly suggested that OCT measurement of nerve fiber layer thicknesses is useful for differentiating SSOH from normal eyes.[7-9] Our Study 2 indicated that not only the superior sectors but also the 3, 5, and 6 o'clock sectors were thinner in SSOH eyes. This is in agreement with the results from previous studies.

Unfortunately, SSOH is not widely recognized among ophthalmologists outside of Japan and Korea. Although four basic features, that is, (1) relatively superior entrance of the central retinal artery, (2) pallor of the superior optic disc, (3) a superior peripapillary halo, and (4) thinning of the superior nerve fiber layer, were previously reported in a Western population,[2] it is essential to pay additional attention to the signs of nerve fiber layer thinning in the superior nasal region as the main sign of SSOH among Japanese and probably in other Asian populations. It is well known that NTG is extremely prevalent in East Asian countries. NTG is normally associated with rim thinning in the inferior temporal or superior temporal

region in its early stages, while the IOP remains within normal limits. Although IOP is usually within normal limits in SSOH as well, the rim thinning is mostly in the superior nasal region with a corresponding NFLD at the same site. Thus, it is not difficult to differentiate between the two diseases, if careful attention is paid to the region of rim thinning and the location of the NFLD.

Conclusion

SSOH is a congenital anomaly affecting the optic nerve head and retina, and it remains an important but underappreciated differential diagnosis of glaucoma, especially in cases of NTG in Asians.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

T. Yamamoto, Grant Support (Alcon Japan, Alcon Pharma, Otsuka, Pfizer, Santen, Senju), Consultant/Advisor (Alcon Japan, Alcon Pharma, Astellas Pharma, Glaukos Japan, Inotek, Kowa, Otsuka, Pfizer, pH Pharma, Rohto, Santen, Seed, Senju), Lecture Fees (Alcon Japan, Alcon Pharma, AMO Japan, CREWT Medical Systems, Glaukos Japan, Johnson and Johnson, Kowa, Novartis, Otsuka, Pfizer, Santen, Senju).

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