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CLINICAL INVESTIGATION

Superior Segmental Optic Hypoplasia Found in Tajimi Eye Health Care Project Participants

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Abstract

Purpose: To investigate the prevalence and characteristics of superior segmental optic hypoplasia in Japanese.

Methods: We studied 14779 subjects, aged 40 years or older, who underwent IMAGEnet fundus photography as part of a large-scale eye disease screening project conducted in Tajimi, Japan. A single researcher reviewed all of the photographs for the presence of ocular abnormality in the optic nerve head and retina, paying special attention to the presence of superior segmental optic hypoplasia.

Results: Fundus photographs of 14431 cases (28396 eyes) were successfully reviewed. We found superior segmental optic hypoplasia in 37 cases (54 eyes; 0.3% of the cases and 0.2% of the eyes). Of the 37 cases, 23 (62%) showed the corresponding visual field defect in at least one eye.

Conclusion: The prevalence of superior segmental optic hypoplasia is about 0.3% in the Japanese population. **Jpn J Ophthalmol** 2004;48:578–583 © Japanese Ophthalmological Society 2004

Key Words: glaucomatous optic neuropathy, optic disc anomaly, prevalence, superior segmental optic hypoplasia

Introduction

Superior segmental optic hypoplasia is a congenital anomaly affecting the optic nerve head and the retina. This condition is ophthalmoscopically characterized by a relatively superior entrance of the central retinal artery, pallor of the superior optic disc, a superior peripapillary scleral halo, and thinning of the superior peripapillary nerve fiber layer.[1–8] Perimetry reveals an inferior altitudinal defect or an inferior sector defect connecting to the blind spot. Visual acuity is not affected in most cases. In a study of this condition, Unoki et al.[1] suggested that optical coherence tomography (OCT) is useful for detecting mild cases of superior segmental optic hypoplasia.

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Superior segmental optic hypoplasia was originally reported as a kind of optic nerve hypoplasia. The term superior segmental optic hypoplasia became generally accepted after Kim et al.[2] investigated the association of this condition with maternal type I diabetes mellitus. Hoyt coined the term "topless optic disc"[3] to emphasize the appearance of the optic disc. Although an association with maternal diabetes is suspected, the etiology of this congenital disorder is not well understood. Unoki et al.[1] reported familial cases of superior segmental optic hypoplasia and suggested the involvement of a genetic factor in this condition.

Because thinning of the neuroretinal rim of the optic nerve head occurs in superior segmental optic hypoplasia, predominantly in the nasal superior region, differentiation of this condition from glaucomatous optic neuropathy is of clinical importance. In particular, normal-tension glaucoma resembles superior segmental optic hypoplasia by its localized rim thinning and lack of elevated intraocular pressure.

To investigate the basic features of superior segmental optic hypoplasia, especially its prevalence, we based the present study on the statistics reported as part of a large-scale eye disease screening project in Japan.

Materials and Methods

The data used in the present study were obtained by the Eye Health Care Project in Tajimi City, Gifu Prefecture, between September 2000 and October 2001, and were provided to us by the Japan Glaucoma Society, which conducted this large-scale eye disease screening project. Our usage of the data was approved by the Japan Glaucoma Society, which has contracted with Tajimi City so that the society can use the data strictly for scientific purposes as long as subject anonymity is preserved. Moreover, informed consent was obtained from all participants before the data from the eye disease screening project were used, and all participants gave the Japan Glaucoma Society permission to use the individual data for scientific research on condition of anonymity. The details of the Eye Health Care Project in Tajimi have been published elsewhere.[9]

The project comprised two study populations: one consisted of 4000 randomly selected citizens aged 40 or older who underwent a detailed ophthalmological check-up, and the other consisted of members of the general population aged 40 or older, who received a general screening. The following ophthalmological examinations were conducted for both eyes of the latter population: visual acuity testing, refractometry with a refractometer (KP-8100PA, Topcon, Tokyo, Japan), corneal thickness measurement with an SP-2000P (Topcon, Tokyo, Japan), perimetry with a frequency doubling technology (FDT) screener (Humphrey Instruments, San Leandro, CA, USA) using the C-20-1 screening program, 45° fundus photography with an IMAGEnet 6S (Topcon, Tokyo, Japan), applanation tonometry with a Goldmann tonometer, slit-lamp biomicroscopy, and van Herick testing. In addition, the participants were requested to fill out a questionnaire on their medical history, and their systemic blood pressure, body weight, and height were also measured. Since most of the participants were unfamiliar with FDT testing, the test was carried out a second time after repeating the full explanations of the testing; the second result was adopted if the first result was abnormal or unreliable. While there was no question regarding the subject's ethnicity in the questionnaire, practically speaking, all of the participants were Japanese. The general screening targeted 50165 citizens aged 40 or older. Of these, 14779 participated in the screening, yielding a response rate of 29.5%. Additionally, we offered a second, voluntary definitive examination on another day to participants who were suspected of having any ocular diseases. At the definitive examination, visual field testing with a Humphrey Field Analyzer (HFA) (Humphrey-Zeiss, Dublin, CA, USA) using the 30-2 SITA Fast program was conducted for subjects with suspected optic disc abnormalities of any type.

In the present study, all of the 45° IMAGEnet fundus photographs of the above-mentioned 14779 participants were reviewed for the presence of ocular abnormality in the optic nerve head and the retina of the posterior pole by one of the authors (TY), who was also one of the chief investigators for the Eye Health Care Project in Tajimi. The reviewer paid special attention to the presence of superior segmental optic hypoplasia. Subject information, except for the bilateral fundus photographs, was masked to the reviewer. Superior segmental optic hypoplasia in the present study was defined as rim thinning of the optic nerve head most prominent in the superior nasal region with corresponding nerve fiber layer defects in the superior nasal region in at least one eye. In cases with superior segmental optic hypoplasia, visual function, that is, visual acuity, the FDT screening result, and the HFA result, if available, were reviewed after reading the fundus photographs. In addition, demographic and other ophthalmological data were reviewed. An FDT abnormality was defined as the presence of test points with a P value less than 5% in the probability map. An HFA abnormality was defined according to criteria proposed by Anderson and Patella[10]: when the pattern standard deviation probability plot showed a cluster of three or more contiguous nonedge points having sensitivity with a probability of less than 5%, with one of these having a probability of less than 1%, in one hemifield, then the hemifield was rated as abnormal. If a corresponding inferior defect in the FDT screening or the HFA testing was present, the eye was rated as definite superior segmental optic hypoplasia; if not, it was rated as suspect superior segmental optic hypoplasia.

The data were analyzed using StatView version 5.0 (SAS Institute, Cary, NC, USA) on a personal computer. Differences among the groups were evaluated using a χ-squared test when applicable.

Results

Of the 14779 participants in the screening project, 45° IMAGEnet fundus photographs of 28396 eyes of 14431 cases were successfully reviewed. A total of 1162 eyes of 814 cases were not used, because the fundus photos were unavailable for 312 eyes and because the photos were of poor quality for 850 eyes. The age and sex distribution of the study population is shown in Tables 1A and B. Of the 14431 cases reviewed, we found 54 eyes of 37 cases with superior segmental optic hypoplasia (about 0.2% of the eyes and about 0.3% of the cases): 23 cases (0.2%) had definite superior segmental optic hypoplasia in at least one eye, and the remaining 14 cases (0.1%) were suspect. Of the 23 definite cases, 5 were definite bilaterally, 5 were definite in one eye and suspect in the other eye, and 13 were unilateral. Of the 14 suspect cases, 7 were bilateral and 7 unilateral. The age and sex distribution of cases with superior segmental optic hypoplasia found in the present study are shown in Tables 2A and B (Table 2A for the overall cases and Table 2B for the definite cases). There was a statistically significant

Table 1A. The age and sex distribution of the study population

Age (years)	Male	Female	Total
40–49	996/7024	2024/7143	3020/14167
50–59	1531/7931	3131/7880	4662/15811
60–69	1895/5287	2486/5279	4381/10566
≥70	1162/3822	1554/5799	2716/9621
Total	5584/24064	9195/26101	14779/50165

Values are population screened / population targeted.

Table 1B. The population whose fundus photos were examined and included in the present study

Age (years)	Male	Female	Total
40–49	985	2009	2994
50–59	1513	3101	4614
60–69	1861	2427	4288
≥70	1094	1441	2535
Total	5453	8978	14431

Table 2A. The age and sex distribution of superior segmental optic hypoplasia, all cases

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)

Values are number of cases / population studied (prevalence in percent).
$P = 0.0103$ versus age and $P = 0.1765$ versus sex (χ-squared test).

Table 2B. The age and sex distribution of superior segmental optic hypoplasia, definite cases only

Age (years)	Male	Female	Total
40–49	3/985 (0.3)	7/2009 (0.3)	10/2994 (0.3)
50–59	3/1513 (0.2)	4/3101 (0.1)	7/4614 (0.2)
60–69	1/1861 (0.1)	2/2427 (0.1)	3/4288 (0.1)
≥70	2/1094 (0.2)	1/1441 (0.1)	3/2535 (0.1)
Total	9/5453 (0.2)	14/8978 (0.2)	23/14431 (0.2)

Values are number of cases / population studied (prevalence in percent).
$P = 0.0425$ versus age and $P = 0.8942$ versus sex (χ-squared test).

difference in prevalence among age groups ($P = 0.0103$ and $P = 0.0425$, for overall and definite cases, respectively). No significant sex difference was found ($P = 0.1765$ and $P = 0.8942$, for overall and definite cases, respectively).

The data for the 37 superior segmental optic hypoplasia cases found in the present study are summarized in Table 3, and example fundus photos are shown in Fig. 1. Of the 37 superior segmental optic hypoplasia patients, 10 were men and the remaining 27 were women. Their ages ranged from 40 to 76 years (mean ± SD: 53.1 ± 10.3 years). The visual acuity was better than or equal to 20/20 in 40 eyes (74%) and worse than 20/25 in 2 eyes (4%) of the 54 eyes with superior segmental optic hypoplasia. When the left eyes were selected in bilateral cases and the affected eyes were selected in unilateral cases, the refractive error as spherical equivalent was −1.54 ± 2.72 D (mean ± SD) and ranged from −8.25 to +1.88 D; the intraocular pressure was 14.2 ± 2.5 mmHg (mean ± SD) and ranged from 9 to 19 mmHg. Maternal diabetes was not identified because the medical questionnaire did not address this issue. One patient with superior segmental optic hypoplasia was reported to have diabetes mellitus but the remaining 36 patients did not.

Table 3. Data on superior segmental optic hypoplasia cases found in the present study

				Visual acuity		FDT defects (number)		HFA defects		Refractive error (D)		IOP (mmHg)		Type of SSOH	
Case	Sex	Age (years)	DM	RE	LE	RE	LE	RE	LE	RE	LE	RE	LE	RE	LE
1	f	58	−	20/20	20/20	I (1), S (2)	I (1)	NA	NA	−0.25	−0.38	13	13	Def	Def
2	f	65	−	20/15	20/15	0	0	NA	NA	1.63	1.75	13	13	Susp	Susp
3	m	73	−	20/30	20/20	0	I (2)	NA	NA	0.00	0.25	12	12	—	Def
4	f	54	−	20/15	20/15	0	0	ITD	Normal	−1.00	−1.00	14	15	Def	—
5	m	58	−	20/25	20/15	0	0	NA	NA	0.88	0.63	17	17	Susp	Susp
6	m	56	+	20/15	20/15	I (1)	0	NA	NA	0.13	−0.38	15	15	Def	—
7	f	51	−	20/40	20/50	0	0	NA	NA	−6.63	−9.00	17	17	Susp	—
8	f	68	−	20/20	20/25	0	0	NA	NA	−7.38	−8.25	15	15	Susp	Susp
9	f	50	−	20/15	20/15	0	0	NA	NA	−0.75	−0.63	12	12	—	Susp
10	f	42	−	20/20	20/15	0	0	NA	NA	−2.38	−2.50	11	12	Susp	—
11	f	64	−	20/20	20/20	0	0	NA	NA	1.88	3.00	18	17	Susp	—
12	f	47	−	20/15	20/15	0	I (1)	NA	NA	−3.25	−3.00	15	15	Susp	Def
13	f	50	−	20/20	20/15	I (2)	S (1)	ITD	Normal	−3.50	−4.00	16	16	Def	—
14	f	42	−	20/20	20/20	0	0	NA	NA	−4.50	−5.50	15	15	Susp	Susp
15	m	46	−	20/15	20/20	0	0	ITD	Normal	−1.50	−1.38	13	14	Def	—
16	f	42	−	20/20	20/25	0	0	NA	NA	−3.25	−2.50	13	13	—	Susp
17	f	42	−	20/20	20/20	I (1)	0	NA	NA	−3.63	−3.25	16	14	Def	Susp
18	m	42	−	20/15	20/15	0	I (1)	NA	NA	−0.13	−0.50	14	14	Susp	Def
19	f	40	−	20/15	20/15	0	I (3)	NA	NA	−1.38	−0.50	15	15	—	Def
20	f	51	−	20/15	20/15	0	0	ITD	Normal	−0.38	−0.13	13	12	Def	—
21	f	76	−	20/40	20/50	I (4)	0	IAD	Normal	1.88	1.75	14	14	Def	—
22	f	47	−	20/20	20/20	I (1)	0	NA	NA	−1.00	−2.00	18	18	Def	—
23	f	66	−	20/25	20/25	I (1)	0	ITD	ITD	1.13	0.50	12	12	Def	Def
24	f	47	−	20/20	20/20	0	0	NA	NA	−2.63	−3.13	18	18	Susp	Susp
25	m	45	−	20/15	20/20	0	0	NA	NA	0.38	0.38	16	16	Susp	Susp
26	f	40	−	20/20	20/20	0	I (2)	Normal	IAD	−6.63	−7.00	12	12	—	Def
27	f	52	−	20/15	20/15	0	S (4)	NA	NA	0.00	0.88	11	10	—	Susp
28	m	57	−	20/15	20/15	I (1)	I (4), S (2)	GD	IAD	−5.38	−6.63	16	18	—	Def
29	f	50	−	20/15	20/15	0	0	NA	NA	0.00	0.38	13	13	—	Susp
30	f	56	−	20/25	20/25	0	0	NA	NA	−4.69	−5.24	15	14	Susp	Susp
31	m	74	−	20/25	20/25	0	I (2)	NA	NA	0.63	1.00	14	14	Susp	Def
32	f	68	−	20/25	20/20	0	0	ITD	ITD	1.38	1.25	10	9	Def	Def
33	f	43	−	20/15	20/15	0	0	Normal	ITD	0.13	0.25	14	14	—	Def
34	m	61	−	20/20	20/25	I (1)	0	NA	NA	0.50	0.75	13	11	Def	Susp
35	f	46	−	20/15	20/20	0	S (2)	ITD	ITD	−1.50	−0.75	19	19	Def	Def
36	f	50	−	20/25	20/20	I (1), S (1)	S (2)	NA	NA	−1.75	−1.50	17	17	Def	—
37	m	47	−	20/15	20/15	0	I (2)	ITD	IAD	−5.13	−3.63	11	11	Def	Def

Refractive error is expressed as spherical equivalent. The right eye of case 28 was rated as optic hypoplasia rather than superior segmental optic hypoplasia.

FDT, frequency doubling technology screener; HFA, Humphrey Field Analyzer; D, diopter; SSOH, superior segmental optic hypoplasia; IOP, intraocular pressure; DM, history of diabetes mellitus; RE, right eye; LE, left eye; f, female; m, male; I, inferior defects; S, superior defects; ITD, inferior temporal defect; IAD, inferior altitudinal defect; GD, generalized depression; NA, not available; Def, definite; Susp, suspect.

Discussion

The definition of superior segmental optic hypoplasia employed in the present study differs in part from previous definitions. Originally, this anomaly was characterized by a relatively superior entrance of the central retinal artery, pallor of the superior optic disc, a superior peripapillary scleral halo, and thinning of the superior peripapillary nerve fiber layer.[1–8] However, optic disc pallor and a superior peripapillary scleral halo are not necessarily visible in all patients, at least not in Japanese patients. According to Unoki et al.[1] a superior scleral halo was not seen in five of seven affected eyes in Japanese. In a study of four Japanese cases by Hashimoto et al.,[7] only one eye showed a superior scleral halo and disc pallor. Additionally, optic disc pallor was not found in Japanese patients highly suspect of having superior segmental optic hypoplasia.[11,12]

The present study revealed that the prevalence of superior segmental optic hypoplasia is about 0.3% in Japanese. This is the first report on the prevalence of superior segmental optic hypoplasia based on a large-scale screening for eye disease. About two-thirds of the patients had a corresponding visual field defect. Because a diagnosis of superior segmental optic hypoplasia is based on the appearance of the optic disc and the corresponding visual field, diagnostic measures including IMAGEnet fundus photography and an FDT screener seem to be appropriate for the detection of this disease. It should be emphasized that, in contradistinction to an epidemiological study, the present study was based on a large-scale screening; thus, the 0.3% prevalence

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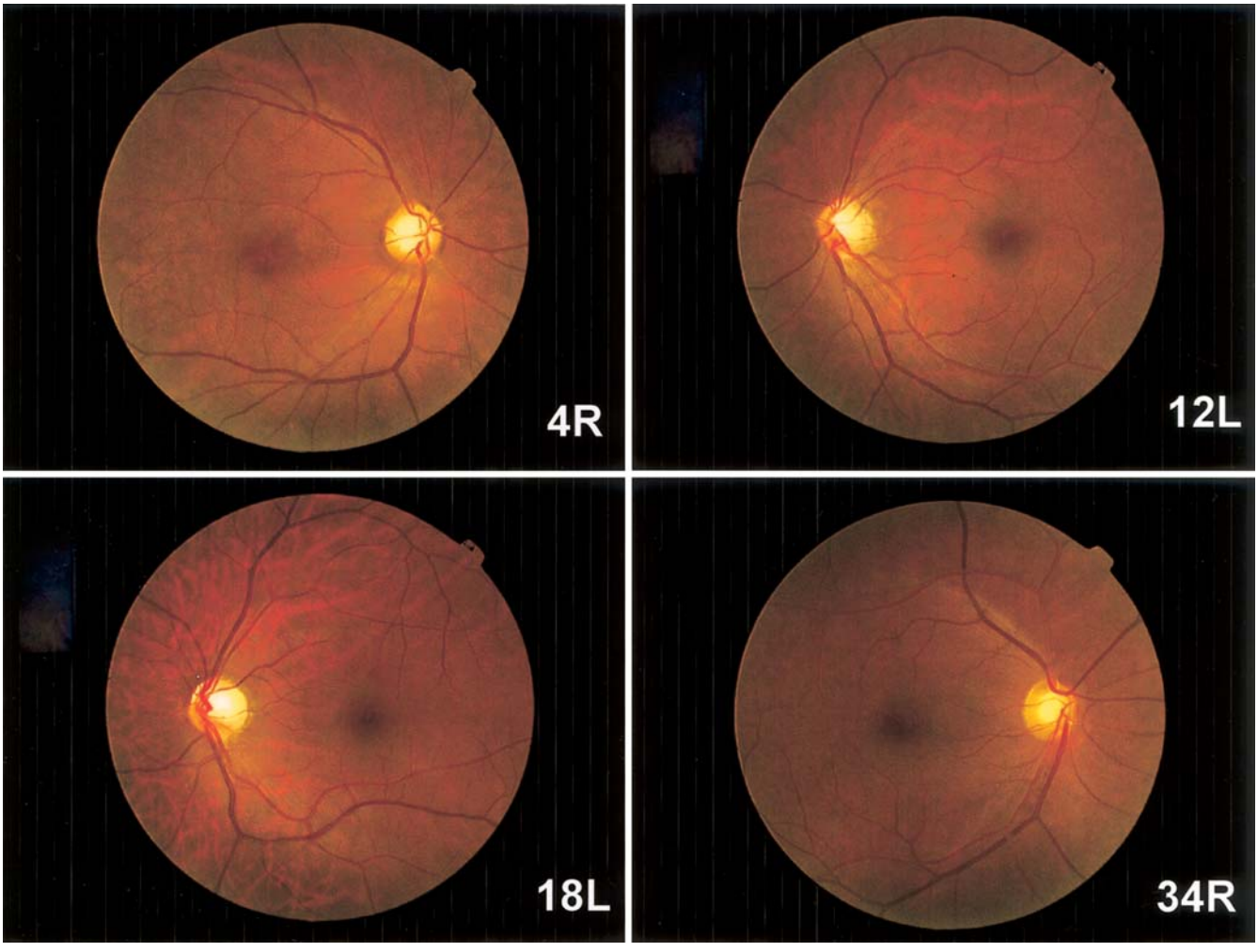

Figure 1. Representative fundus photographs of superior segmental optic hypoplasia found in participants in the large-scale eye disease screening project in Tajimi, Japan, on which this study was based. *Upper left*: the right eye of case 4; *upper right*: the left eye of case 12; *lower left*: the left eye of case 18; *lower right*: the right eye of case 34. The affected eyes show rim thinning in the superior nasal region with corresponding nerve fiber layer defects, but lack pallor of the superior optic disc and a superior peripapillary scleral halo.

rate needs further validation. Additionally, the reason for the age difference found in the present study should be sought; we cannot explain it at this moment.

Superior segmental optic hypoplasia resembles glaucomatous optic neuropathy in that localized rim thinning is seen when the optic disc shows cupping. In glaucoma cases with normal intraocular pressure, that is, normal-tension glaucoma, differentiation from superior segmental optic hypoplasia is important because both conditions lack elevated intraocular pressure. The key means of differentiation is the localization of the rim thinning and the characteristic visual field changes: an inferior altitudinal defect or inferior sector defect connecting to the blind spot. Since the 0.3% prevalence of superior segmental optic hypoplasia is substantial and represents about one-tenth of that of normal-tension glaucoma in Japanese,[9,13] awareness of this condition should increase in clinical practice.

In conclusion, we found the prevalence of superior segmental optic hypoplasia is 0.3% in Japanese based on data from a large-scale screening for eye disease.

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