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Findings of optical coherence tomography angiography of nasal optic disc hypoplasia

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ABSTRACT

Purpose: This study aimed to present a case of a woman diagnosed with congenital nasal optic disc hypoplasia (NOH) in the right eye.

Observations: A woman in her 20s presented with a small optic disc, irregular optic disc margins with a double-ring sign, and wedge-shaped defects temporally extending from Mariotte's blind spot in the right eye. The radial peripapillary capillary (RPC) density around the optic nerve disc was examined using optical coherence tomography angiography (OCTA). The best-corrected visual acuity was 1.2 OU, and intraocular pressure was 12 mmHg OU. Optical coherence tomography C-scan detected thinning of the circumpapillary retinal nerve fiber layer, and OCTA revealed that a decrease in RPC density on the nasal side of the optic disc in the right eye was markedly lower than that in the other eye areas. Furthermore, it was lower than the RPC density on the nasal side of the left eye.

Conclusions and importance: OCTA may help detect the clinical features and pathogenesis of NOH.

1. Claims of priority statement

We conducted a literature search on May 23, 2024, utilizing PubMed and Google Scholar databased with the key words "nasal optic disc hypoplasia" and "optical coherence tomography angiography." No prior reports were found on the findings of OCTA in cases of NOH.

2. Introduction

Developmental abnormalities of the optic nerve or head during the embryonic period cause congenital optic disc hypoplasia. Several patterns of optic disc hypoplasia exist, with varying degrees of severity.[1] Although superior segmental optic disc hypoplasia (SSOH) is the most well-known,[2] nasal optic disc hypoplasia (NOH) has also been reported. NOH is characterized by (i) a small optic nerve disc, (ii) pallor of the nasal optic disc or irregularity of the optic disc margins, and (iii) wedge-shaped defects temporally extending from Mariotte's blind spots.[3]

In SSOH, optical coherence tomography (OCT) reveals thinning of the circumpapillary retinal nerve fiber layer (cpRNFL) thickness superior to the optic nerve disc. Laser speckle flowgraphy (LSFG) demonstrates decreased blood flow velocity in the optic nerve disc in association with this area.[4] Meanwhile, OCT angiography (OCTA) reveals a decrease in the density of the radial peripapillary capillaries (RPC) of the optic nerve disc.[5] In contrast, the cpRNFL thickness on the nasal side of the optic disc is reduced in NOH.[6–8] In addition, blood flow velocity in the optic disc is reduced in LSFG, which is consistent with that area.[8] However, studies on RPC density using OCTA in NOH are lacking. Therefore, this study aimed to examine the RPC density around the optic nerve disc in eyes with NOH using OCTA. We conducted a literature search on May 23, 2024, utilizing PubMed and Google Scholar databased with the key words "nasal optic disc hypoplasia" and "optical coherence tomography angiography." No prior reports were found on the findings of OCTA in cases of NOH.

3. Case report

A woman in her 20s with myopia and no other ophthalmologic symptoms or medical and family histories presented with unremarkable symptoms. The best-corrected visual acuity using the Japanese standard Landolt visual acuity chart was 1.2 OU. Intraocular pressure (IOP) was within normal ranges, recorded at 12.0 mmHg OD and 12.7 mmHg OS.

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Fundus examination revealed a small optic disc and irregular optic disc margins with a double-ring sign in both eyes. In addition, the distance from the center of the disc to the macula/disc diameter (DM/DD) ratio was 3.46 in both eyes, indicating a small disc head (Figs. 1a and 2a). Additionally, the C-scan of OCT revealed thinning of the cpRNFL in the nasal quadrants of both eyes (Figs. 1b and 2b). Given that NOH was suspected based on these fundus findings, a visual field examination was performed using a Humphrey field Analyzer (24-2 Swedish Interactive Threshold Algorithm standard) and Goldmann perimeter. A visual field abnormality was detected, showing a wedge-shaped defect temporally extending from Mariotte's blind spot in the right eye (Fig. 1c and d). The left eye did not exhibit any visual field abnormalities (Fig. 2c and d).

4. Investigations

OCTA (RS-3000 Advance 2; Nidek Co., Ltd., Gamagori, Japan) was used to quantify the RPC density of the optic disc. Nidek's RS-3000 Advance 2 OCT system and AngioScan (updated; version 1.10.0) were used to evaluate the OCTA images. OCTA scans of the optic disc were conducted with a 4.5 mm × 4.5 mm diameter and a 256 B-scan composition. The RPC density slab thickness for the RS-3000 Advance 2 system encompasses the entire NFL layer, from the internal limiting membrane to NFL/ganglion cell layer. The RPC density can be separately quantified and evaluated for up to four sectors. On OCTA, the RPC density on the nasal side of the optic disc in the right eye was markedly lower than that in the other areas of the eye (Fig. 1e and Table 1). These results were consistent with the areas of visual field abnormalities and decreased cpRNFL thickness. Furthermore, it was lower than the RPC density on the nasal side of the left eye (Table 1).

5. Differential diagnosis

In this case, we diagnosed the small size of the optic disc, irregular margins with a double-ring sign, abnormality in the wedge-shaped visual field extending to physiologic blind spot on visual field examination, as well as thinning of the cpRNFL coinciding with the area of the visual field abnormality and NOH in the right eye. The left eye demonstrated fundus and OCT findings similar to those of the right eye;

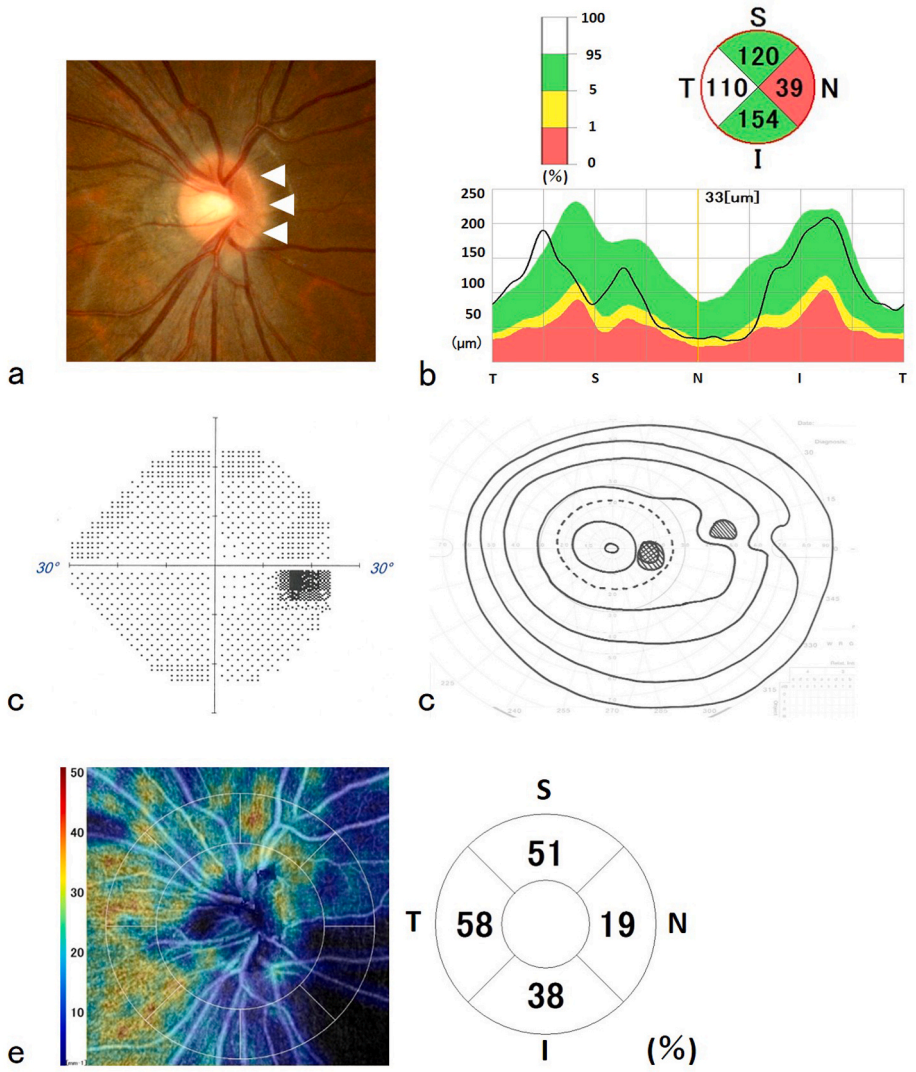

Fig. 1. Images of the right eye with nasal optic disc hypoplasia (NOH). a. Fundus photograph illustrating the double-ring appearance of the nasal optic nerve head (arrowheads). b. Thickness of the circumpapillary retinal nerve fiber layer (cpRNFL) for each quadrant (S, superior; N, nasal; I, inferior; and T, temporal) of the optic disc using optical coherence tomography and thinning of the nasal cpRNFL. c. Humphrey 24-2 Swedish interactive threshold algorithm (SITA) standard test and d. Goldmann perimetry revealing a wedge-shaped temporal visual field defect. e. Radial peripapillary capillary (RPC) density color map, aligned with the temporal-superior-nasal-inferior-temporal (TSNIT) graph, showing markedly reduced RPC density on the nasal side in the NOH eye. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

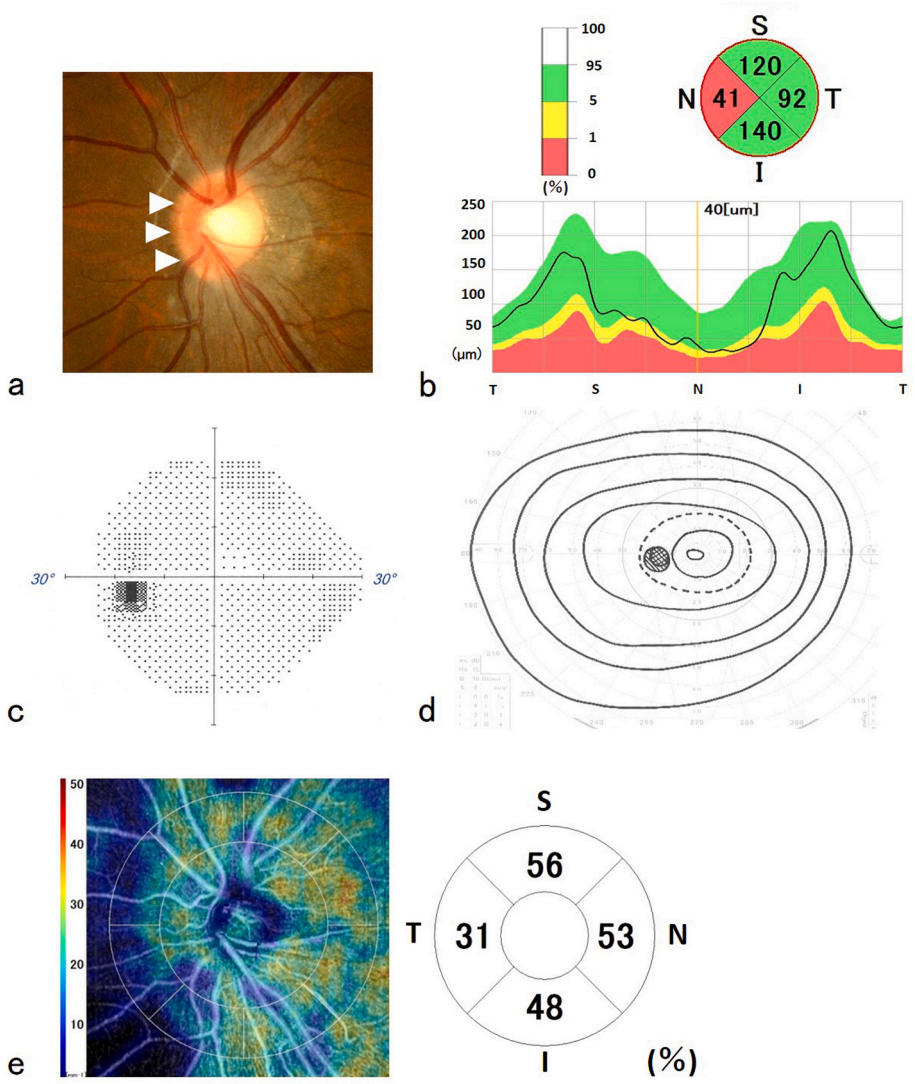

Fig. 2. Images of the normal left eye. a. Fundus photograph illustrating the double-ring appearance of the nasal optic nerve head (arrowheads). b. Thickness of the circumpapillary retinal nerve fiber layer (cpRNFL) for each quadrant (S, superior; N, nasal; I, inferior; and T, temporal) of the optic disc using optical coherence tomography and thinning of the nasal cpRNFL. c. Humphrey 24-2 Swedish interactive threshold algorithm (SITA) standard test and d. Goldmann perimetry did not exhibit any visual field abnormalities. e. Radial peripapillary capillaries (RPC) density color map corresponding to the temporal-superior-nasal-inferior-temporal (TSNIT) chart showing reduced RPC density on the nasal side. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

Table 1
Characteristics of both eyes at baseline and after 6 months in this case.

Eye	Right		Left	
	Baseline	After 6 months	Baseline	After 6 months
SE (D)	−2.75	−2.75	−3.37	−3.37
IOP (mmHg)	12.0	11.3	12.7	12.0
MD (dB)	−0.45	−0.28	−0.10	0.20
cpRNFL thickness (μm)				
Superior	120	113	120	120
Temporal	110	119	92	98
Inferior	154	151	140	134
Nasal	39	34	41	41
RPC density (%)				
Superior	51	49	56	57
Temporal	58	57	53	58
Inferior	38	47	48	53
Nasal	19	19	31	35

SE, spherical equivalent; D, diopter; IOP, intraocular pressure; MD, mean deviation; cpRNFL, circumpapillary retinal fiber layer thickness; RPC, radial peripapillary capillaries.

however, the absence of visual field abnormalities did not support a NOH diagnosis, as defined by Buchanan and Hoyt. Glaucoma is a differential diagnosis because it causes visual field abnormalities similar to those observed in NOH. However, only 2.5 %–3.0 % of patients with glaucoma present with temporal visual field abnormalities,[6,9] such as NOH. Additionally, the patient's IOP was within the normal range. Since the condition is currently non-progressive, glaucoma has been ruled out. During the 6 months of follow-up, all test results remained within the margin of error, and no progression of the condition was observed (Table 1). In the future, regular examinations should be performed to monitor for any changes in ocular findings and potential glaucoma conditions. Furthermore, no genetic testing was performed in this case. Although systemic complications are not currently evident, comprehensive systemic and genetic testing will be pursued if deemed clinically necessary in the future.

6. Outcome and follow-up

Given that it is non-progressive, NOH, a congenital condition, does

not require treatment. Thus, the patient should be carefully followed up regularly in the future to ensure that the findings do not progress and are not complicated by glaucoma.

7. Discussion

In this case, the RPC density on the nasal side of the optic disc in the NOH eye was markedly lower than that in the other areas and on the nasal side of the contralateral eye. Furthermore, areas of visual field abnormalities and decreased cpRNFL thickness coincided with the areas of reduced RPC density.

In SSOH, which is a partial optic nerve hypoplasia, as well as NOH, thinning of the cpRNFL of the superior optic disc on OCT C-scans with visual field abnormalities are characterized by inferior arcuate visual field defect with physiologic blind spot at the apex.[2,10] Furthermore, LSFG exhibits a reduction in the optic disc microcirculation, consistent with the thinning areas of the cpRNFL.[4] Additionally, decreased RPC density in the optic disc using OCTA has been demonstrated, suggesting the usefulness of OCTA in SSOH.[5]

In NOH, the nasal cpRNFL thickness is reduced.[6,7] Additionally, the nasal cpRNFL thickness of the NOH eyes substantially decreases compared to that of the normal eyes, and no considerable differences are detected in the other cpRNFL thicknesses between the NOH and normal eyes.[8] Similarly, the velocity ratio of the optic disc nasal blood flow is substantially lower in the NOH eyes than in normal eyes.[8] Furthermore, a substantial correlation has been detected between cpRNFL thickness and microcirculation in the nasal area.[8] However, no previous studies have been conducted on RPC density using OCTA in NOH.

8. Conclusions

In the present case, these results reveal that even when NOH was suspected based on the findings of the fundus and OCT C-scan images, differences existed in the presence of visual field abnormalities. Furthermore, the density of the RPC may be involved in these differences. Thus, OCTA may be useful in detecting the NOH clinical features and pathogenesis.

CRediT authorship contribution statement

Miki Yoshimura: Visualization, Validation, Resources, Methodology, Investigation, Formal analysis, Data curation. **Yuki Hashimoto:** Writing – original draft, Visualization, Validation, Resources, Project administration, Methodology, Investigation, Funding acquisition, Formal analysis, Data curation, Conceptualization. **Arisu Hatanaka:** Investigation, Data curation. **Takeshi Yoshitomi:** Writing – review & editing, Supervision, Conceptualization.

Patient consent

Written informed consent was obtained from the patient after the nature and possible consequences of the study had been explained.

Conflicts of interest

None.

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Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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