Case Report

Superior Segmental Optic Nerve Hypoplasia: A Rare Mimicker of Normal-Tension Glaucoma–A Case Series from Türkiye

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Abstract

This retrospective case series presents the characteristic features of superior segmental optic nerve hypoplasia (SSONH) in four patients who were initially misdiagnosed with normal-tension glaucoma (NTG), aiming to raise awareness of this rare condition in Türkiye. Four patients (two females and two males) with a mean age of 38 years were included. All were initially diagnosed with NTG and treated with brimonidine drops for three years. Comprehensive ophthalmological examinations were performed, including optic disc photography, optical coherence tomography retinal nerve fiber layer (RNFL) analysis, and visual field testing, with follow-up evaluations conducted over at least one year. Bilateral involvement was observed in two cases, and unilateral involvement in the other two. History of maternal diabetes was noted in 50% of the patients. During medication-free follow-up, all patients demonstrated stable structural and functional parameters, supporting the diagnosis of SSONH. These findings suggest that SSONH should be considered in young patients presenting with superior RNFL thinning and corresponding inferior visual field defects. The non-progressive nature of the condition helps differentiate it from glaucomatous optic neuropathy. Recognizing unilateral cases is essential for avoiding misdiagnosis.

Keywords: Superior segmental optic nerve hypoplasia, normal tension glaucoma, Türkiye

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Introduction

Superior segmental optic nerve hypoplasia (SSONH) is a rare congenital developmental anomaly first described by Petersen and Walton[1] in 1977 as a distinct form of optic nerve hypoplasia. Unlike generalized optic nerve hypoplasia, SSONH is characterized by preserved visual acuity with sectoral visual field defects and has gained attention as a crucial differential diagnosis for glaucoma, particularly normal-tension glaucoma (NTG).

The four key diagnostic features of SSONH are relative superior entrance of the central retinal artery, pallor of the superior optic disc, superior peripapillary halo, and thinning of the superior peripapillary nerve fiber layer.[2] However, not all patients exhibit all of these characteristics, and the presence of at least two features with documented non-progressive disease course may be sufficient for diagnosis.[3] Figure 1 demonstrates characteristic fundoscopic appearances of SSONH, showing both unilateral and bilateral presentations with the classic "topless disc" appearance described by Landau et al.[4]

Maternal diabetes has been recognized as a significant risk factor, though recent studies suggest other factors may contribute to SSONH development.[3,5] The condition predominantly affects females and remains stable over time without progression, distinguishing it from glaucomatous optic neuropathy.[6]

Despite being well-documented in East Asian countries, SSONH remains underrecognized in Türkiye, where it is often misdiagnosed as glaucoma, leading to unnecessary treatments and interventions. To address this gap in awareness, we present a retrospective case series of four patients who were initially misdiagnosed with NTG but later identified as having SSONH.

Case Reports

All patients were evaluated at our glaucoma clinic and underwent comprehensive ophthalmological examinations including optic disc photography (Visucam 500; Carl Zeiss Meditec AG, Jena, Germany), retinal nerve fiber layer (RNFL) analysis (Cirrus HD-OCT 4000; Carl Zeiss Meditec, Inc., Dublin, CA, USA), and visual field testing (HFA-II 750; Carl Zeiss Meditec, Inc., Dublin, CA, USA). Detailed medical histories were reviewed, and follow-up assessments were carried out over a minimum of one year to evaluate the stability of structural and functional parameters. Table 1 summarizes the sociodemographic and clinical features of all four patients. The cohort included two females and two males with a mean age of 38 years (range 30-55). All patients had been treated with brimonidine drops for suspected NTG for at least three years before correct diagnosis.

Case 1

A 30-year-old female patient presented for glaucoma follow-up after relocating. Visual acuity was 20/20 bilaterally with intraocular pressure (IOP) of 16 mmHg. Figure 2 shows bilateral superior RNFL thinning on optical coherence tomography (OCT) analysis. Maternal type 1 diabetes was documented. No progression was observed during three years of external follow-up. Following SSONH diagnosis, brimonidine therapy was discontinued, and two-year medication-free follow-up showed stable parameters (IOP 16-18 mmHg) with resolution of ocular surface complaints.

Case 2

A 32-year-old female patient sought a tertiary opinion after receiving NTG diagnoses from two centers. Visual acuity was 20/20 in the right eye and 20/30 in the left (due to amblyopia from childhood ankyloblepharon surgery). Guided progression analysis showed stable RNFL parameters over time in right-sided unilateral SSONH (Figure 3). Maternal insulin-dependent diabetes

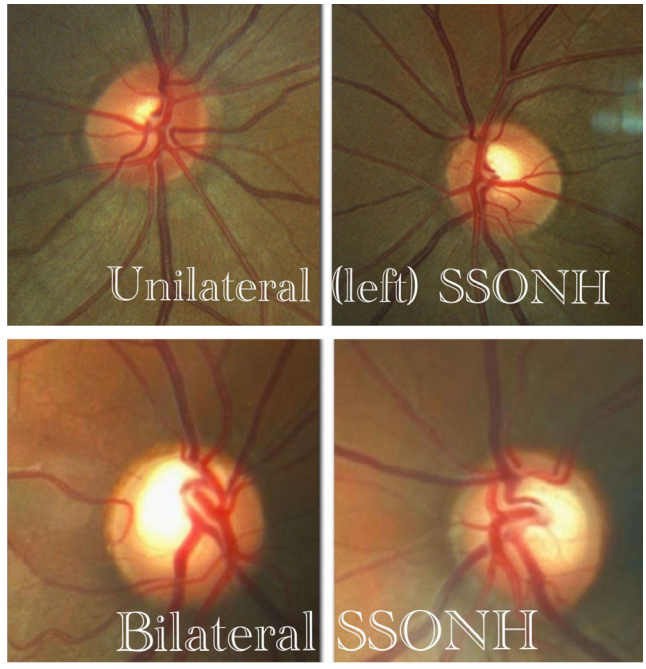

Figure 1. Appearance of the right and left optic disc in unilateral and bilateral cases of superior segmental optic nerve hypoplasia (SSONH)

Table 1. Sociodemographics and clinical features of the patients

	Case 1	Case 2	Case 3	Case 4
Age (years)	30	32	55	35
Sex	Female	Female	Male	Male
BCVA (Snellen feet) (R, L)	20/20, 20/20	20/20, 20/32	20/20, 20/20	20/20, 20/20
IOP (mmHg) (R, L)	16, 16	12, 13	17, 17	15, 16
CCT (µm) (R, L)	585, 580	530, 540	612, 618	585, 588
Average RNFL thickness (µm) (R, L)	75, 77	74, 89	76, 63	64, 82
Eye involvement	Bilateral	Unilateral	Unilateral	Bilateral
Maternal diabetes	Yes	Yes	Unknown	No
Systemic comorbidity	None	None	None	None
Ocular comorbidity	None	Unilateral congenital ankyloblepharon	Presbyopia	None
Mean follow-up in SSONH (months)	24	27	13	18

BCVA: Best corrected visual acuity, R: Right eye, L: Left eye, IOP: Intraocular pressure, CCT: Central corneal thickness, RNFL: Retinal nerve fiber layer, SSONH: Superior segmental optic nerve hypoplasia

was documented. Two-year follow-up without treatment confirmed stable IOP (11-14 mmHg) and no structural progression.

Case 3

A 55-year-old male patient initially presented with presbyopic complaints and was diagnosed with glaucoma. Despite irregular medication compliance, no progression was observed over two years. Visual acuity was 20/20 bilaterally with IOP of 17 mmHg. Maternal diabetes history was unavailable due to early maternal loss. The patient was diagnosed as having left unilateral SSONH and showed stable parameters (IOP 14-18 mmHg) over 13-month medication-free follow-up.

Case 4

A 35-year-old male patient sought a second opinion three years after receiving a diagnosis of asymptomatic NTG. Maternal gestational glucose intolerance was suspected. Visual field testing demonstrated characteristic

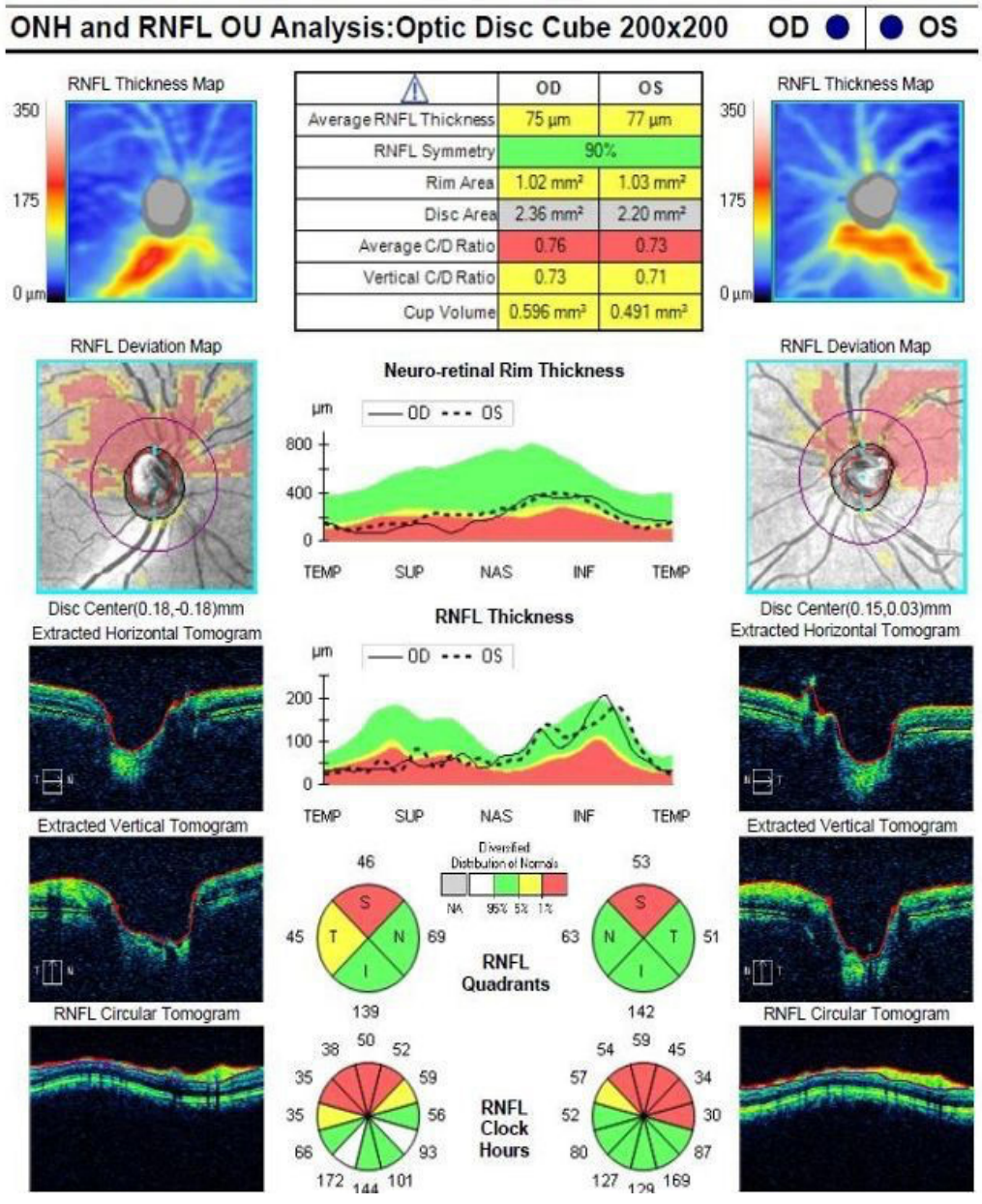

Figure 2. Representative optical coherence tomography (OCT) retinal nerve fiber layer (RNFL) findings in superior segmental optic nerve hypoplasia (SSONH)

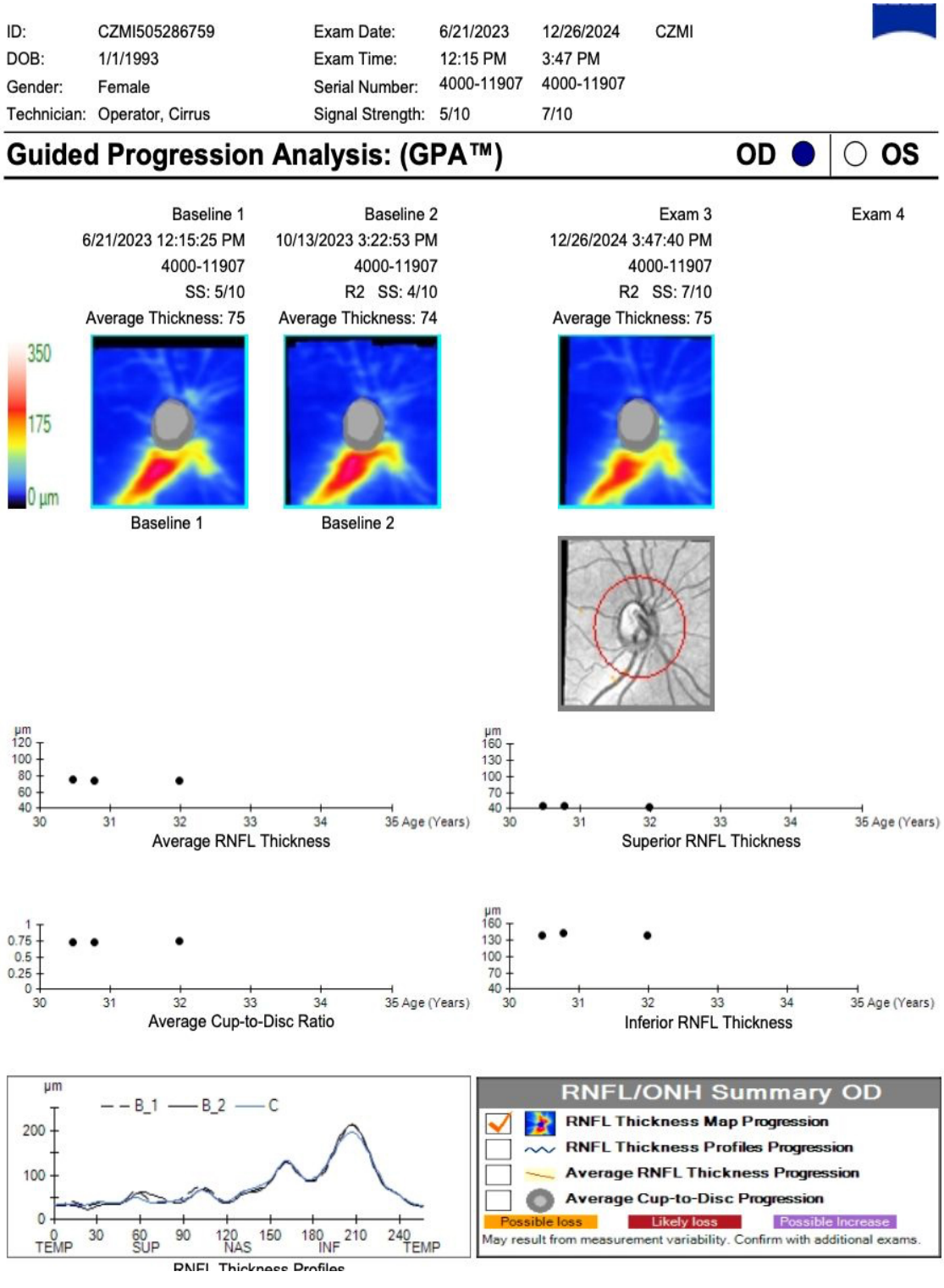

Figure 3. Longitudinal retinal nerve fiber layer (RNFL) imaging in superior segmental optic nerve hypoplasia (SSONH) demonstrating structural stability over time

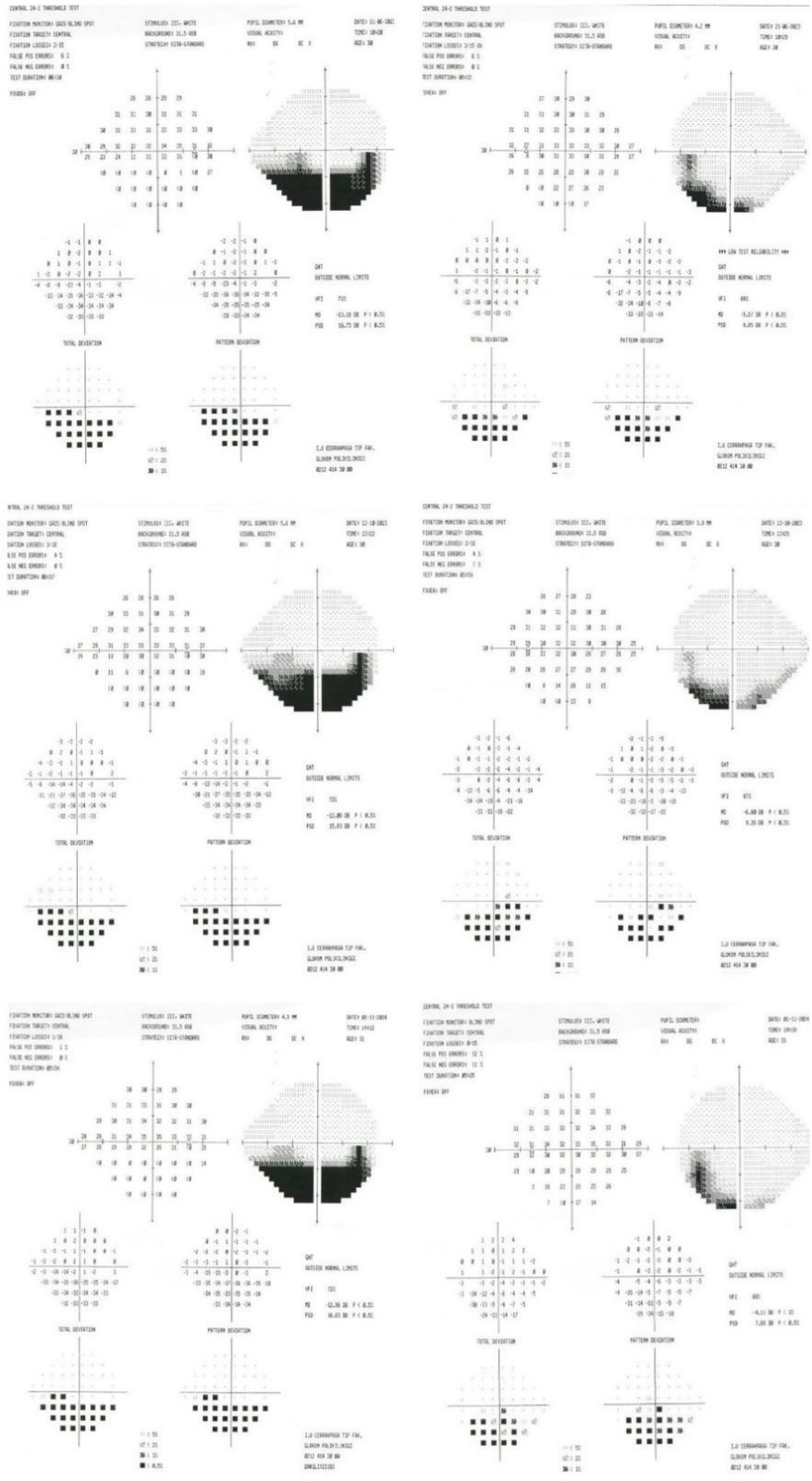

Figure 4. Long-term visual field stability in superior segmental optic nerve hypoplasia (SSONH)

inferior arcuate scotomas (Figure 4). The patient exhibited bilateral SSONH with more prominent findings in the right eye. Lack of structural or functional progression over time (18 months) supported a diagnosis of SSONH over glaucomatous optic neuropathy.

Discussion

This case series highlights the diagnostic challenges of SSONH, particularly its differentiation from NTG. All four patients were initially misdiagnosed and received unnecessary treatment, emphasizing the importance of awareness among ophthalmologists.

The demographic profile shows interesting patterns. While previous studies suggested female predominance,[3,4] our cohort included two males (50%), reinforcing that SSONH affects both genders.[7] Notably, two patients (50%) presented with unilateral involvement, which contrasts with typical bilateral presentations reported in the literature.[1,3] Unilateral SSONH, though less common, has been documented and should be recognized as a valid presentation pattern.[8] This finding is particularly important, as unilateral cases may be more easily misdiagnosed as glaucoma due to asymmetric presentation.

Maternal diabetes was documented in two cases (50%), consistent with established associations.[1,2] However, the fourth patient lacked a history of maternal diabetes, supporting recent findings that other factors may contribute to SSONH development beyond maternal diabetes.[9]

OCT RNFL analysis revealed characteristic superior RNFL thinning in all cases, differing from the inferotemporal thinning commonly observed in glaucoma. This pattern provides a valuable diagnostic distinction. Visual field defects predominantly affected inferior regions, further supporting the SSONH diagnosis.

The most significant diagnostic feature was stability of structural and functional parameters during long-term follow-up without treatment. This non-progressive nature distinguishes SSONH from glaucomatous optic neuropathy, which typically shows progression if untreated. Our cases demonstrated stable parameters over mean 20.5 months of medication-free follow-up.

Several conditions should be considered in the differential diagnosis of SSONH. NTG is the most common diagnostic challenge, as both disorders may present with superior RNFL thinning and inferior visual field defects. However, SSONH is a congenital and usually non-progressive anomaly observed in younger patients, whereas NTG typically appears later in life, often when systemic vascular disease is more prevalent.[10,11] Although our patients were younger than the typical age group for NTG, all underwent systemic internal medicine and neurology consultations, as well as magnetic resonance imaging (MRI), and no significant pathological findings were detected. Considering the age group in which SSONH is typically diagnosed, periventricular leukomalacia (PVL) should also be included in the differential diagnosis, as it may mimic both SSONH and NTG by demonstrating optic disc cupping and superior RNFL loss with corresponding inferior field defects.[12] Yet, PVL is usually associated with a history of prematurity and characteristic periventricular or subcortical white matter changes on MRI, which help to distinguish it from SSONH.[13] In addition, split RNFL variations, most often observed superiorly, may resemble SSONH in asymptomatic adults with IOP below 21 mmHg, but these represent benign anatomical variants without underlying congenital optic nerve anomalies.[14]

An important clinical implication is the unnecessary treatment burden. All patients had received brimonidine drops for 3 years, causing financial burden and, in one case, ocular surface complications. This underscores the importance of accurate differential diagnosis to prevent unnecessary interventions.

Limitations of this study include the retrospective nature and small sample size. However, given SSONH's rarity (prevalence <1%), case series remain valuable for understanding this condition. Recent advances in OCT angiography may provide additional diagnostic tools through peripapillary vessel density measurements, though this was not evaluated in our cases.[15]

SSONH should be considered in young patients with suspicious optic nerve appearance, superior RNFL thinning, and inferior visual field defects, especially with maternal diabetes history. However, the absence of maternal diabetes should not exclude the diagnosis. Importantly, both bilateral and unilateral presentations should be recognized, as unilateral cases may be more prone to misdiagnosis as glaucoma. The stability of structural and functional parameters over time, even without treatment, remains the key diagnostic feature distinguishing SSONH from NTG. Long-term follow-up is essential for accurate diagnosis. Raising awareness among Turkish ophthalmologists regarding this rare congenital anomaly is crucial for preventing misdiagnosis and unnecessary treatments, thereby improving patient care and reducing the healthcare burden.

Ethics

Informed Consent: Written informed consent was obtained from all patients.

Declarations

Authorship Contributions

Surgical and Medical Practices: C.Y.E., Ö.O., Concept: C.Y.E., İ.K.S., Design: C.Y.E., İ.K.S., Data Collection or Processing: C.Y.E., İ.K.S., Analysis or Interpretation: C.Y.E., İ.K.S., Literature Search: C.Y.E., İ.K.S., Ö.O., Writing: C.Y.E., İ.K.S.

Conflict of Interest: No conflict of interest was declared by the authors.

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