

Evaluation of Optical Coherence Tomography Findings in Regards of Nanophthalmos Cases

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ABSTRACT

Nanophthalmos is a subtype of microphthalmia developed as a result of halt of growth in the globe and early closure of the embryonic fissure. Our purpose is to evaluate optical coherence tomography (OCT) findings and macular images in nanophthalmos cases and to distinguish from posterior microphthalmia. We evaluated 6 eyes of 3 patients diagnosed as nanophthalmos with an ocular axial length less than 20 mm. Axial length, anterior segment depth, corneal thickness, mean macular thickness and retinal nerve fiber layer thickness were measured using biometry, sonography and spectral domain OCT. The mean axial length was 15.19 mm in 6 eyes, ranging from 15 mm to 15.45 mm. The mean macular thickness of the was 550 µm in 3 eyes, which was higher than normal ranged reported in the literature. In our cases, the mean RNFL thickness was 145.5 µm, which was also higher than normal ranged reported in the literature. The mean anterior chamber depth was measured as 2.52 mm, indicating anterior chamber narrowing when compared to those reported in the literature. anterior chamber narrowing was detected in cases compared to normal values in the literature. Fundus examination revealed a small optical disc small with ill-defined margins o (crowded disc) and a low cup: disc ratio. No papillomacular fold was observed but intraretinal cysts and loss of foveal pitting were observed on OCT imaging. Although it was reported that that papillomacular folds may occur in nanophthalmos eyes on OCT imaging, we did not observe in our cases.

Keywords: Nanophthalmos, optical coherence tomography, posterior microphthalmia.

INTRODUCTION

Nanophthalmos is a subtype of microphthalmia resulting from halt of globe growth and premature closure of embryonic fissure. Although there is microphthalmia, the structural organization and functions are preserved in the eye. In clinical practice, it is presented with secondary angle-closure glaucoma, spontaneous choroidal effusion and perioperative complications during cataract and retina surgeries. It may be autosomal dominant or autosomal recessive as well as sporadic. Five distinct genes including MFRP, TMEM98, PRSS56, BEST1 and CRB1 have been defined¹⁻².

It is characterized by small orbit, deep localization of small globe, high hypermetropia due to shorter axial length, small cornea, narrow anterior chamber and palpebral fissure. The disrupted choroidal development with decreased permeability to proteins leads to choroidal effusion and non-rhegmatogenous retinal detachment; followed by

decreased vision. In ocular examination, small optic disc with appearance of crowded disc, bone spicule mimicking retinitis pigmentosa in retina, choroidal effusion, non-rhegmatogenous retinal detachment and macular folds can be seen^{3,4}.

Microphthalmia is a developmental disorder resulting from halt of ocular development. It is characterized by total axial length which is below at least 2 standard deviation than corresponding age group⁵. Posterior microphthalmia is a subtype that is characterized by decreased total axial length and normal cornea diameter and can be associated with high hypermetropia and papillomacular retinal folds⁶. Nanophthalmos is distinguished from posterior microphthalmia by micro-cornea and uveal effusion; however, presence of retinal fold on OCT may lead confusion. Here, we will present 3 nanophthalmos cases together with OCT findings.

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CASE REPORT

Case 1

A 24-years old man presented to our clinic with decreased vision. The visual acuity at presentation was as follows: OD, 0.5 and OS, 0.4 (correction: +12,75+0,75X5° / +14,25+0,75X180°). Intraocular pressure (IOP) was measured by 21/17 mmHg by applanation tonometry. There was no consanguinity between parents. There was no additional disease in the history.

On biomicroscopy, narrow anterior chamber was detected in both eyes. No other abnormality was detected in anterior segment. Color vision was 8/12 in the right eye and 12/12 in the left eye. In fundus examination, optic was small (as being more prominent at right side) with ill-defined borders and low cup: disc ratio. Bilateral macular folds were observed (Figure 1a).

On OCT, macular thickness was 508/476 μm and retinal nerve fiber layer thickness was 164/182 μm (Figure 1b). Minimal intraretinal fluid was observed at right side (Figure 1a). Choroidal thickness was measured as 222/248 (Figure 1a). The anterior chamber dept was measured as 2,42 / 2,41 mm by anterior segment OCT.

Axial length was measured as 15,22/15,25 mm by pachymetry. Cornea thickness was 581 μm in right eye and 586 μm in left eye by pachymetry. Keratometry results were as follows: right, K1:49,91D X28° K2:50,58 DX128°; left, K1:50,38 Dx89° K2:51,88 Dx179°.

Case 2

A 19-years old woman, the sibling of case 1, presented to our clinic with decreased vision. The visual acuity at presentation was as follows: OD, 0.5 and OS, 0.6 (correction: +14 / +12 Intraocular pressure (IOP) was measured by 12/15 mmHg by applanation tonometry. There was no consanguinity between parents. There was no additional disease in the history.

On biomicroscopy, narrow anterior chamber was detected in both eyes. No other abnormality was detected in anterior segment. Color vision was 1/12 in the right eye and 12/12 in the left eye. In fundus examination, ill-defined optic nerve margins were observed in both eyes.

On OCT, macular thickness was 470/453 μm and retinal nerve fiber layer thickness was 150/154 μm (Figure 2b). Choroidal thickness was measured as 274/290 (Figure 2a). The anterior chamber dept was measured as 2,65 / 2,67 mm by anterior segment OCT.

Axial length was measured as 15,25/15,45 mm by pachymetry. Corneal thickness was 588 μm in right eye and 590 μm in left eye by pachymetry. Keratometry results were as follows: right, K1:50,30 DX171° K2:50,81 DX81°; left, K1:50,61 DX141° K2:50,61 DX51°

Case 3

A 5-years old children was presented with decreased vision by parents. The visual acuity at presentation was as

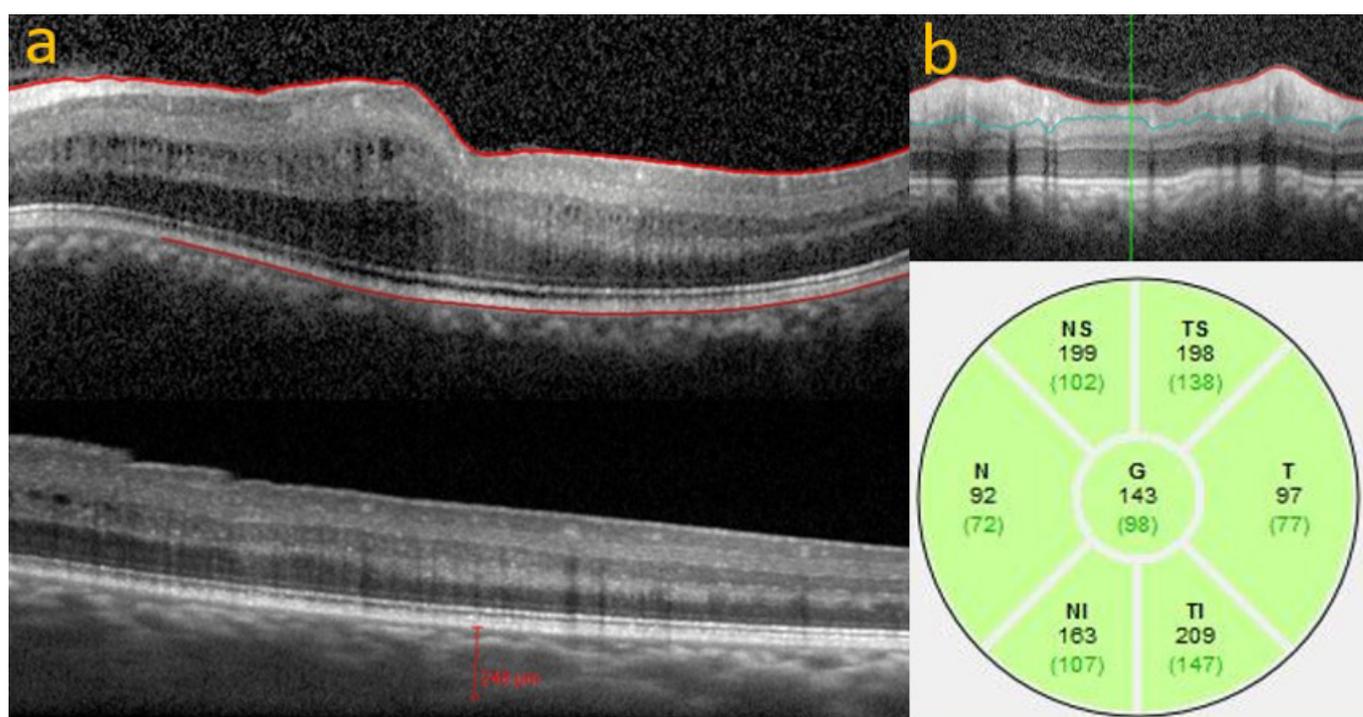


Figure 1: a) Macular and choroidal images on OCT in case 1; b) optic nerve section on OCT and RNFL analysis in case 1.

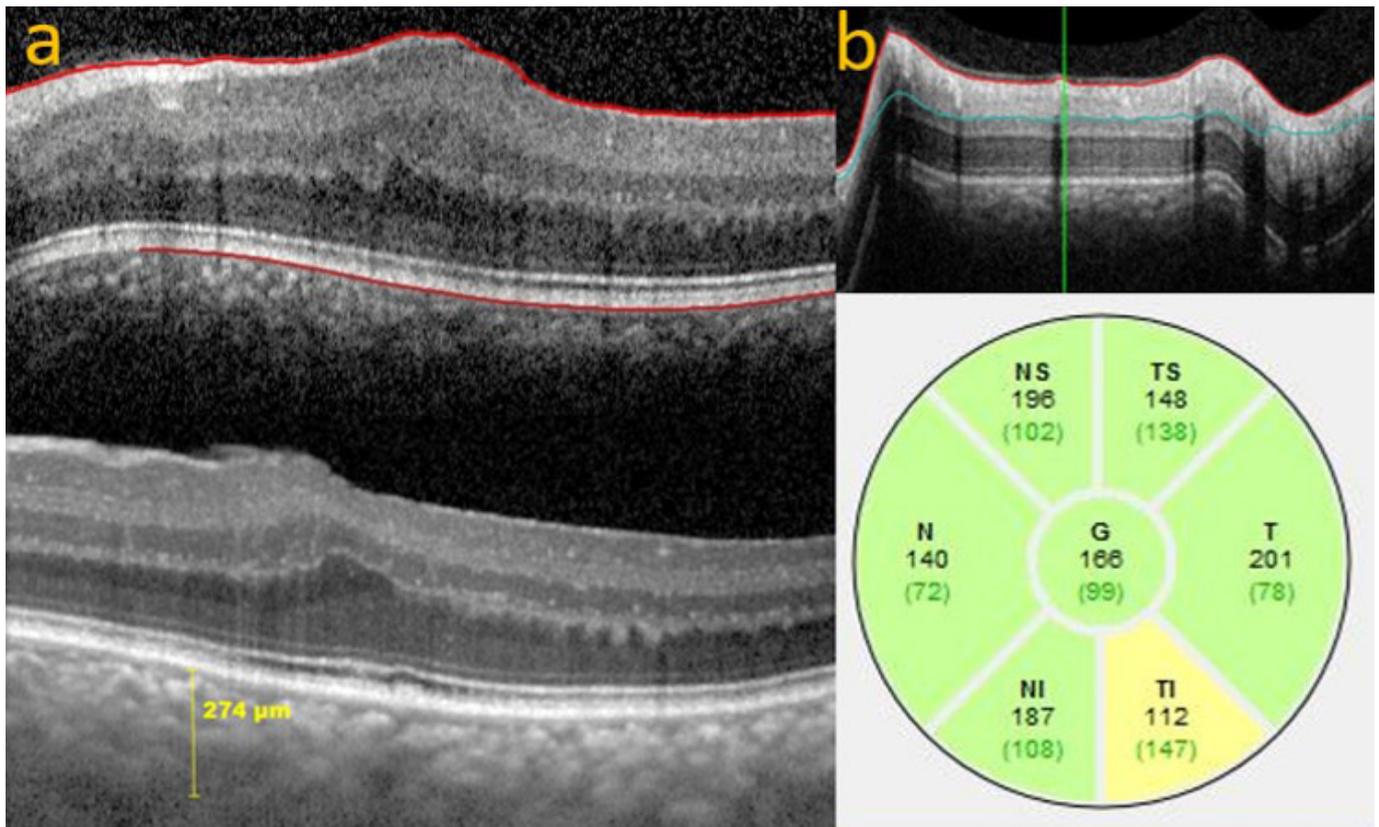


Figure 2: a) Macular and choroidal images on OCT in case 2; b) optic nerve section on OCT and RNFL analysis in case 2.

follows: OD, 0.3 and OS, 0.2. Intraocular pressure (IOP) was measured by 16/14 mmHg by applanation tonometry. There was no consanguinity between parents. There was no additional disease in the history.

On biomicroscopy, narrow anterior chamber was detected in both eyes. No other abnormality was detected in anterior segment. In fundus examination, ill-defined optic nerve margins were observed in both eyes.

On OCT, macular thickness was 683/712 μm (Figure 3a) and retinal nerve fiber layer thickness was 118/107 μm (Figure 2b). The anterior chamber depth was measured as 2,50 / 2,50 mm by Lenstar LS 900 (Haag Streit AG, Koeniz, Switzerland). Intraretinal cysts were observed on OCT (Figure 3a).

Axial length was measured as 15/15 mm by sonographic pachymetry. Corneal thickness was 560 μm in right eye and 564 μm in left eye by Sirius (CSO, Florence, Italy). Keratometry results were as follows: right, K1:50,01 DX116° K2:50,33 DX26° left, K1:49,95 DX30° K2:50,36 DX111°.

DISCUSSION

Nanophthalmos is a rare ocular disease. It is characterized by narrow palpebral space, deeply localized small globe, short axial length, high hypermetropia and normal or

decreased corneal diameter. In histopathological studies, scleral thickening and scleral collagen fiber abnormalities were shown⁷. High hypermetropia was also seen in our cases (mean: +14,50; min-max: +12,00 to +16,00).

There is limited epidemiological data regarding prevalence of nanophthalmos and microphthalmia. Its prevalence was found as 0.002-0.017% in a cohort study from United Kingdom and 0.0009% in a study from China^{8, 9}. In the literature, nanophthalmos appears with different definitions based on axial length. Weiis et al. defined upper limit of axial length as 18 mm for diagnosis nanophthalmos¹⁰ while it was defined as 20.5 mm in series reported by Singh, in and Anderson³. Based on up-to-date data, Day et al. defined axial length as 21 mm for microphthalmia and 20 mm for nanophthalmos¹¹. In our cases, axial length was shorter than 20 mm with a mean value of 15.1 mm. In the EPIC-Norfolk Eye Study from Europe (2013), the rate of individuals with axial length < 21 mm was 1.2% and those with axial length < 18 mm was 0.14%¹². Based on these data, it may be proposed that number of patients with microphthalmia and nanophthalmos is underestimated.

In the literature, mean anterior chamber depth has been reported as 3.11 mm which varies across ethnicities¹³. The lower limit of normal is reported as 2.91 mm and anterior chamber depth was lower than normal in our cases,

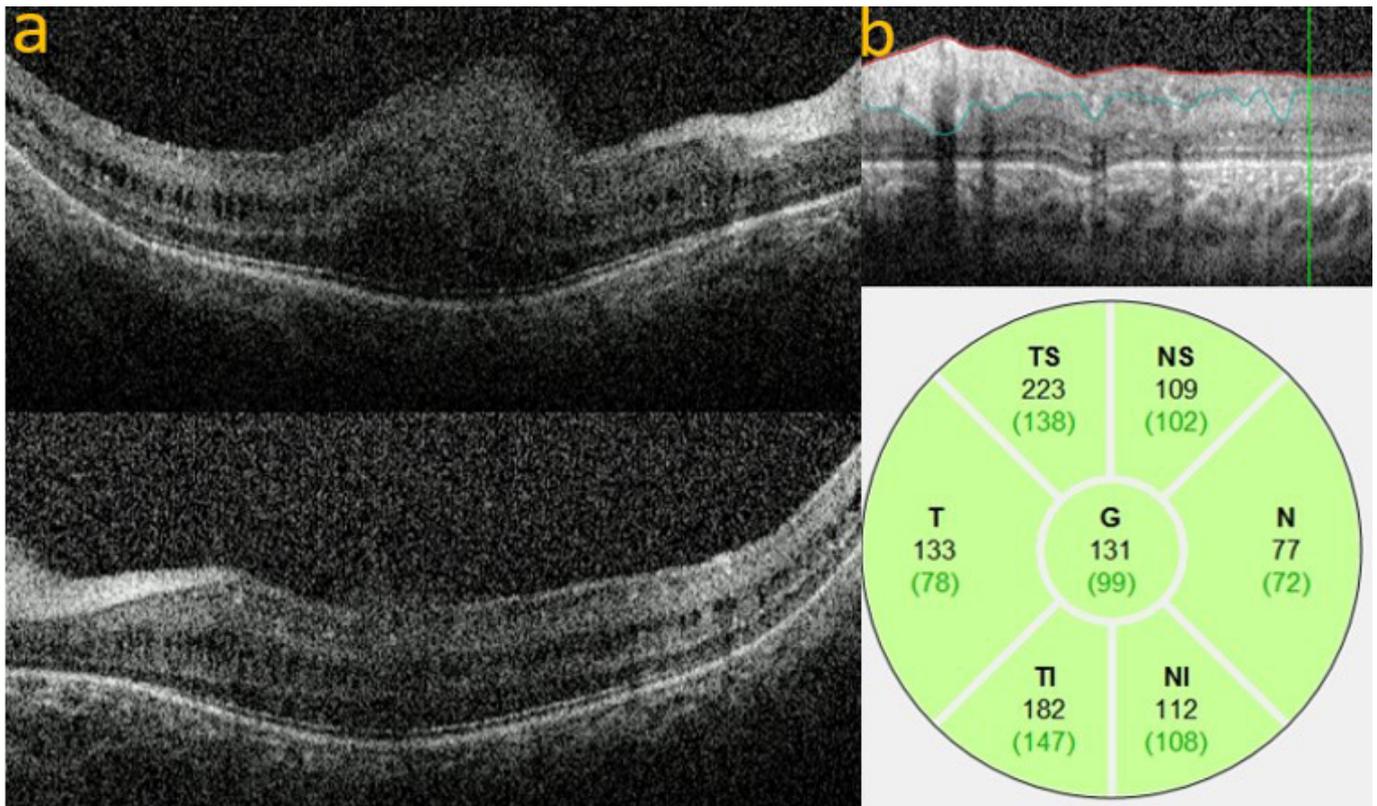


Figure 3: a) Macular and choroidal images on OCT in case 3; b) optic nerve section on OCT and RNFL analysis in case 3.

indicating narrowed anterior chamber depth. However, no glaucomatous pathological finding was observed.

In the literature, corneal thickness has been reported as 483-570 µm in normal population¹⁴. In our cases, near-normal corneal thicknesses were recorded. No reduction was observed in corneal thickness. Mean keratometry value was found as 50.32 (min-max: 49.91-50.81)..

In the literature, normal macular thickness has been reported as 212 ± 20 µm on OCT¹⁵. In three cases presented, macular thickness was found to be high. Normal value for retinal nerve fiber layer is 99.4±9.6 µm¹⁶. In our cases, RNFL values were found to be higher than normal value reported. In a previous study, normal value for subfoveal choroidal thickness was reported to be 363±84 µm on EDI-OCT¹⁷. In case, choroidal thickness was lower than normal in case 1 while it was comparable to normal values in case 2. The finding of thicker macula and thinner choroidal layer may explain poorer visual acuity in case 1. Given these values, it is likely that case 1 would face with nanophthalmos-related complications in the future.

In a nanophthalmos case by Yalcindag et al., papillomacular folds restricted to neurosensory retina were observed on OCT¹⁸. Authors suggested that nanophthalmos may be a subtype of posterior microphthalmia, considering

that papillomacular fold may be a finding of posterior microphthalmia. On the other hand, Timeoney et al. reported papillomacular fold produced by choroidal and retina in patients with Kenney-Caffey syndrome-related nanophthalmos¹⁹. No papillomacular fold was observed in our cases.

Ward et al. reported that there was macular hypoplasia and loss of foveal pitting on OCT in cases with nanophthalmos and that there was macular fold in only one of 15 patients. In the only case with papillomacular fold, anterior segment depth was found to be almost normal and authors reported that the case might be posterior microphthalmia²⁰. The filling of outer retinal layers in foveal pitting results in poor visual acuity. Loss of foveal pitting was observed in our cases in agreement with OCT finding reported by Ward et al. who revealed that true nanophthalmos and microphthalmia are distinct presentations based on lack of papillomacular folds in vast majority of the cases. Aras et al. defined papillomacular folds in posterior microphthalmos. Kim et al. reported that neurosensory retina fold not involving choroid and retinal pigment epithelium was found in posterior microphthalmos⁶. There are studies reporting that papillomacular fold is observed in posterior microphthalmos²²⁻²⁵. In a case report, it was suggested that papillomacular fold is more characteristic in posterior microphthalmia.

In a study including 19 patients with nanophthalmos, Xiao et al. observed macular hypoplasia, increased macular retinal thickness, enlarged outer retinal layer and loss of foveal pitting in all cases. Preretinal fold was observed in only two eyes. Authors explained loss of foveal pitting by abnormal localization of inner retinal layers²⁶. Although their findings are consistent to those found in our cases, no choroidal thickening was observed in our cases.

In all three cases, optic disc was small with ill-defined margins in fundus examination. Possible causes for poor visual acuity are considered as increased macular thickness, loss of foveal pitting and amblyopia.

In our case report, it is though finding papillomacular fold on OCT favors posterior microphthalmia in differential diagnosis of nanophthalmos and posterior microphthalmia. In addition, number of patients with nanophthalmos is limited in epidemiological studies, it is thought that its prevalence is underestimated. In advanced ages, it may manifest with secondary angle-closure glaucoma, spontaneous choroidal effusions and perioperative complications during cataract and retina surgeries. The cases with high hypermetropia (+10 to +20) should be assessed for microphthalmia or nanophthalmos. The diagnosis at younger ages will reduce potential complication in the future.

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