

# Nanophthalmos with Optic Nerve Head Drusen Complicated by Central Serous Chorioretinopathy - A Case Report

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**Abstract:** We report a case of 45-year-old man with complaints of blurring of vision. Fundus examination showed optic disc drusen with neurosensory detachment. Ultrasound examination revealed short axial length and optic disc drusen. Fundus fluorescein angiography revealed pigment epithelial detachment. Nanophthalmos is a clinical spectrum of disorder with phenotypically normal but structurally small eye usually presenting due to angle closure glaucoma and spontaneous choroidal effusion. We report the first case presenting with simultaneous occurrence of optic drusen and central serous chorioretinopathy in nanophthalmos.

**Keywords:** Nanophthalmos, optic disc drusen, neurosensory detachment, pigment epithelial detachment and central serous chorioretinopathy

## 1. Introduction

Nanophthalmos is a developmental disorder of the eye characterized by shortening of both the anterior and the posterior segments with thick sclera, choroid and high lens to eye volume ratio not associated with any other ocular congenital malformations.<sup>1,2</sup> It may be associated with optic nerve head drusen,<sup>1</sup> which are hyaline calcificated deposits of mitochondria formed at the prelaminar optic nerve head due to altered axonal metabolism and axonal disruption.<sup>3,6</sup> Nanophthalmos can be complicated by angle closure glaucoma, uveal effusion syndrome and exudative retinal detachment.<sup>1</sup> This is the first case report presenting the simultaneous occurrence of nanophthalmos with optic nerve head drusen complicated by central serous chorioretinopathy (CSCR).

## 2. Case Report

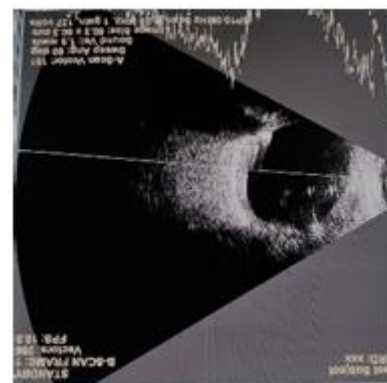
A one-eyed 45 year old male reported to our outpatient department with complaints of blurring of vision in the left eye past one month. The patient had a history of penetrating trauma to the right eye and it had undergone phthisis. He gave history of use of glasses with lensometer reading of +8.00DS in right eye (just for weight balance) and +8.00DS/+1.50DC × 25° in left eye. Systemic and family history was unremarkable. His best corrected visual acuity was 6/24 in the left eye. Further, anterior segment examination of the left eye revealed nystagmus, microcornea measuring 9.5×9.7 mm, shallow anterior chamber (Van Herrick grading II) and narrow vertical palpebral aperture (7 mm) in the primary gaze. The pupil was briskly reacting to light. The extraocular movements were normal in all gazes. The intraocular pressure of the left eye was 20mm Hg as measured by applanation tonometry. Fundus examination showed blurring of the disc margins and multiple, round, multilobulated, semi-translucent yellowish deposits at the optic disc suggestive of optic disc drusen. In addition, a well-circumscribed elevation at the macula with fine

xanthophyll pigment deposits pointed towards a neurosensory detachment (Figure 1A).



**Figure 1 (A):** Fundus photograph showing optic disc drusen and neurosensory detachment

The ultrasound B-scan showed hyperechoic round structures with acoustic shadowing at the optic nerve head region suggestive of drusen (Figure 1B).

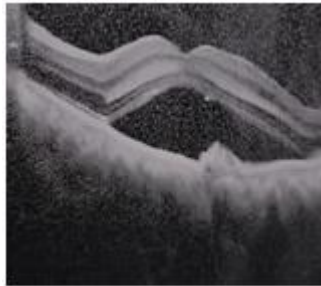


**Figure 1 (B):** USG B-scan showing hyperechoic structures at the optic nerve head region – optic disc drusen

The ultrasound A-scan measurements of anterior chamber depth was 3.30 mm, lens thickness was 4.15 mm and the axial length was 18.67 mm. Further, the ultrasound B-scan

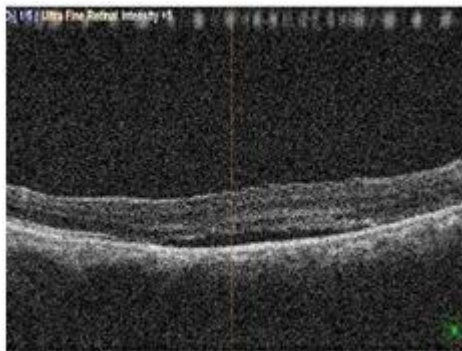
revealed an increased retinochoroidal thickness of 3.11 mm. All these parameters suggested that the left eye is nanophthalmic.

Optical coherence tomography (OCT) scan taken through the fovea 1 month ago at the onset of symptoms (outside) revealed an optically clear area in the sub-retinal space suggestive of neurosensory detachment along with a pigment epithelial detachment (Figure 2A).



**Figure 2 (A):** OCT scan showing neurosensory detachment with pigment epithelial detachment

The current OCT through the fovea showed only a shallow neurosensory detachment (Figure 2B) with a central macular thickness of 532  $\mu\text{m}$ .



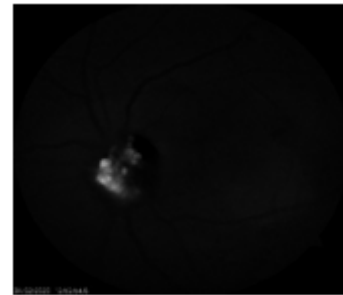
**Figure 2 (B):** OCT scan after one month showing shallow neurosensory detachment

Fundus fluorescein angiography showed a hyperfluorescent spot in the superotemporal region of the macula in the early phase increasing in intensity in the late phase suggestive of pigment epithelial detachment (Figure 3).



**Figure 3:** Fundus fluorescein angiography showing pigment epithelial detachment with absence of active leak

There was no active leakage. Fundus autofluorescence revealed hypoautofluorescence at the macula and multiple hyperautofluorescent lesions at the optic disc consistent with the finding of optic disc drusen (Figure 4).



**Figure 4:** Fundus autofluorescence showing optic disc drusen

Based on these findings, a diagnosis of nanophthalmos with high hypermetropia, optic nerve head drusen and central serous chorioretinopathy of the left eye was made. The patient has been kept under observation and is planned to be followed up monthly to monitor for progression or spontaneous resolution.

### 3. Discussion

Nanophthalmos is a clinical spectrum of disorders with phenotypically small but structurally normal eye with pachychoroid.<sup>1,2</sup> Nanophthalmos is a subtype of microphthalmia with a global reduction in the size of the eye not associated with any other ocular anomaly, with an axial length  $<21\text{ mm}$ , a shallow anterior chamber, high lens to eye volume, high hyperopia, thick sclera and choroid.<sup>2</sup> They can present as an isolated anomaly or with other malformations like coloboma, anterior segment dysgenesis, lens abnormalities, and posterior segment anomalies.<sup>1,2</sup> Nanophthalmic eyes have abnormal collagen fibrils in sclera,<sup>1,5</sup> which impairs the vortex venous drainage. Nanophthalmos can also present with irregular astigmatism, optic disc drusen,<sup>1,11</sup> chorioretinal folds, retinal cysts, foveal hypoplasia, increased subfoveal and choroidal thickness, retinitis pigmentosa<sup>1,11</sup> and retinoschisis<sup>1</sup>.

Patients with optic nerve head drusen usually have well preserved visual acuity,<sup>6,7</sup> but can rarely present with anterior ischemic optic neuropathy, central retinal artery or venous occlusion, peripapillary choroidal neovascularization, and central serous papillopathy.<sup>6,8</sup> Drusen alters the normal barrier between the prelaminar optic nerve and the peripapillary retina, causing focal choroidal hyperpermeability. Central serous chorioretinopathy (CSCR) is a disease with an altered retinal pigment epithelium, characterized by localized serous neurosensory detachment of the macula and focal retinal pigment epithelial detachments.<sup>9</sup> In our case, the abnormal pachychoroid and the inelastic sclera of the nanophthalmic eye along with drusen seems to have led to choroidal hyperpermeability with vessel engorgement, increased choroidal hydrostatic pressure and a breach of the retinal pigment epithelium causing subretinal fluid accumulation.

Kumar et al have described the occurrence of serous macular detachment in the setting of nanophthalmos, similar to CSCR.<sup>1</sup> Suelves et al have described the association of optic disc drusen with central serous papillopathy.<sup>6</sup> Buys et al have reported the occurrence of retinitis pigmentosa, optic disc drusen and nanophthalmos.<sup>11</sup> We report the first case in

literature with simultaneous occurrence of nanophthalmos with optic nerve head drusen (ONHD) as a potential cause of central serous chorioretinopathy. So, it is important to recognize the association of nanophthalmos with optic disc drusen and CSCR. A dilated fundus examination must be done in all cases of nanophthalmos to look for these possible causes of visual deterioration.

## References

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