



CASE REPORT

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Evaluation of a patient with buried optic disc drusen in the differential diagnosis of papilledema

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Abstract

Optic disc drusen (ODD) is hyaline-like calcified material accumulation that may occur at the optic nerve head. It is seen in approximately in 0.3% of the population and is often bilateral. Since bilateral cases might mimic papilledema, it is important to make the differential diagnosis. In this report; a patient followed in the neurology department with suspected papilledema and diagnosed with bilateral ODD will be assessed by means of perimetry, retinal nerve fiber layer (RNLF) analysis and fundus autofluorescence (FAF) imaging methods.

Keywords: Optic disc drusen, papilledema, diagnosis

Introduction

Optic disc drusen (ODD) are benign, protein like calcified extracellular deposits that accumulate in front of the lamina cribrosa due to the anomalies and degenerations of the axonal metabolism. They are seen more often bilaterally among people with narrow scleral canal with an incidence of 0.3% of the population. It is possible to classify it as buried and exposed types. Bilateral buried optic disc drusen cases are specially important since they can be confused with papilledema. In this report, a patient followed in the neurology department of our hospital with suspected papilledema and diagnosed with bilateral buried disc drusen will be discussed.

Case Report

A 25 year old male patient, with complaints of floaters and blackouts on both eyes, more pronounced on the left, approximately for 1 month, was initially referred to the neurology clinic. Afterwards the patient referred to our clinic for consultation with suspected papilledema. Following a detailed ophthalmologic examination, the patient was undergone retinal nerve fiber layer (RNLF) analysis (SD-OCT, OTI, Toronto, Kanada), optic disc topography, perimetry, B-mod ultrasonography (B-USG)

and fundus autofluorescence (FAF) (HRA2, Heidelberg Engineering, Germany).

The visual acuity in both eyes was 10/10, and color vision, intraocular pressure and anterior segment examination were normal. In the fundus examination, a mild to moderate elevation of the optic disc is remarkable and the disc margins are irregular and blurred on both eyes, more pronounced on the left (Figure 1). The results of RNFL examination were normal (Figure 2). On the 30-2 Threshold Humphrey Perimetry examination; in addition to periphic arcuate scotoma in the temporal quadrant, enlarged of the blind spot was observed on the left eye compared to the right eye (Figure 3). On the B-USG, a slight elevation on the disc heads and hyperechogenic lesion causing shadowing behind the disc were observed, more pronounced on the left eye again (Figure 4). In the FAF examination, a hyperautofluorescence disc head was seen on the left (Figure 5). Though it was not seen ophthalmoscopically, the patient was diagnosed with “Buried Optic Disc Drusen” by means of USG and FAF monitoring methods.

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Figure 1. Fundus photography of the patient. A mild to moderate elevation of the optic disc is remarkable and the disc margins are irregular and blurred on both eyes, more pronounced on the left.

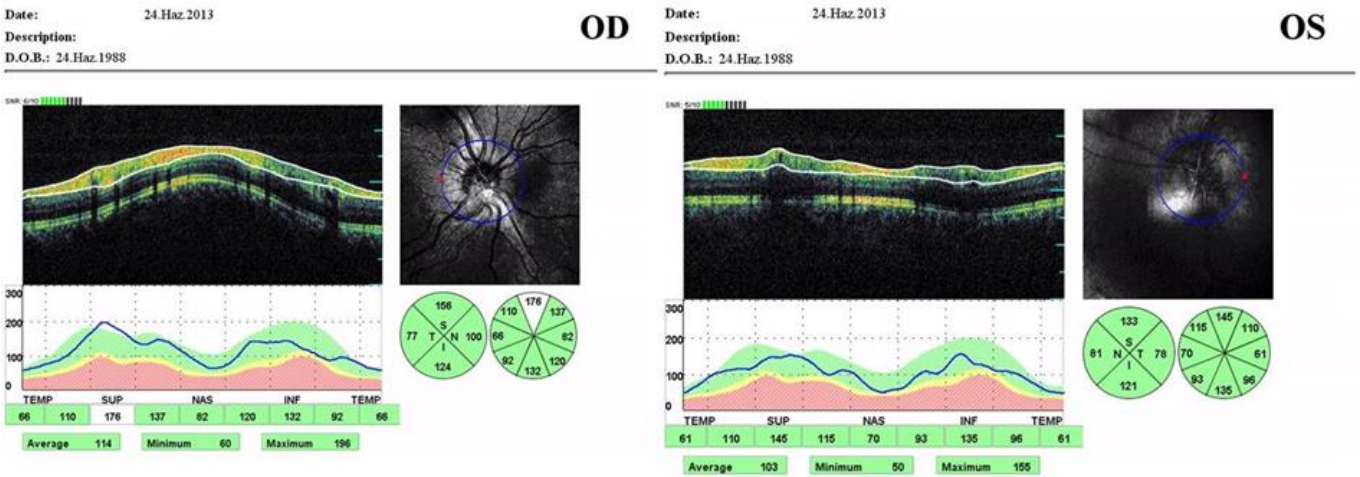


Figure 2. The RNLF analysis of both eyes are normal.

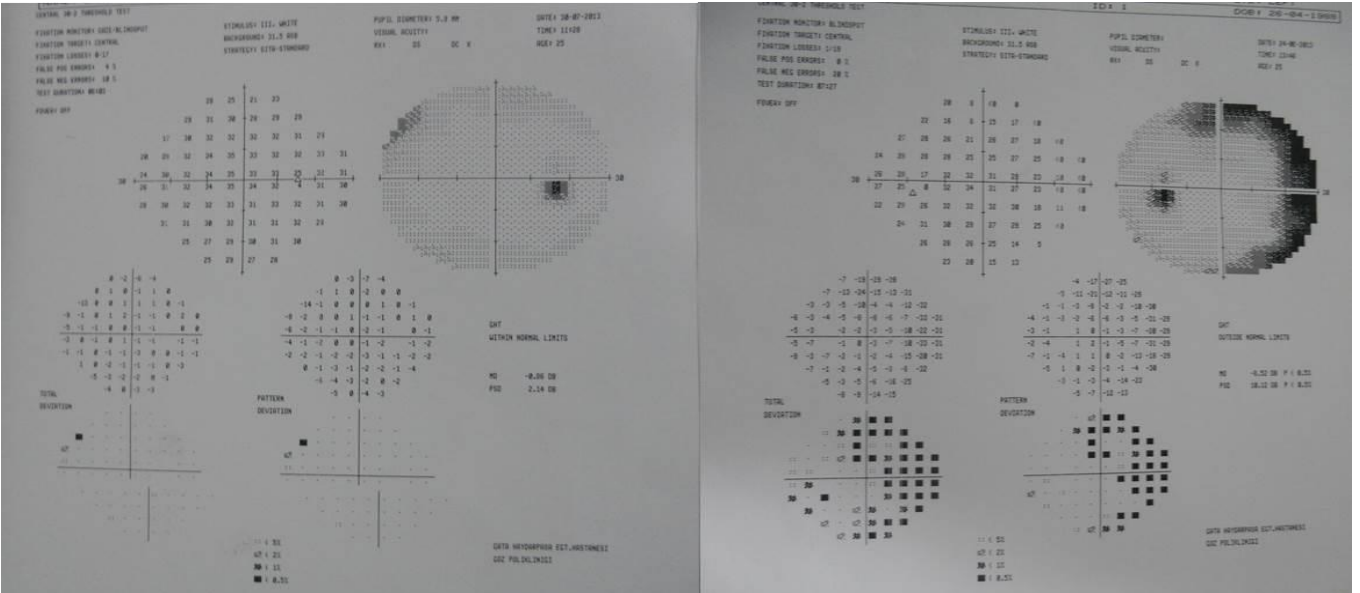


Figure 3. The 30-2 Threshold Humphrey Perimetry examination revealed a peripheral arcuate scotoma in the temporal quadrant and enlargement of the blind spot on the left eye.

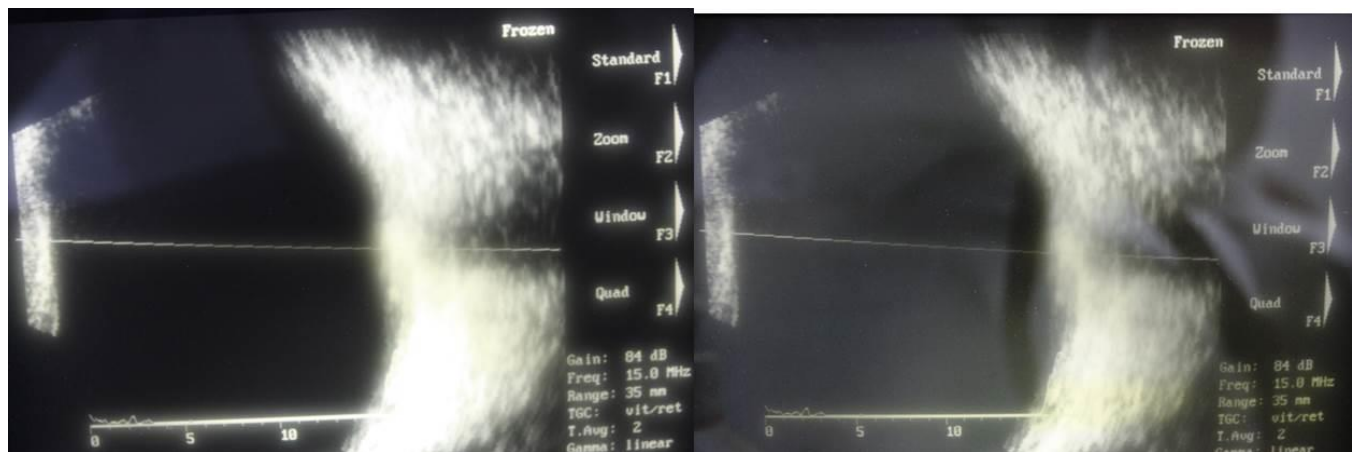


Figure 4. A slight elevation of the disc head and shadowing behind the disc due to the hyperechogenic lesion was observed in the B-scan ultrasonography.

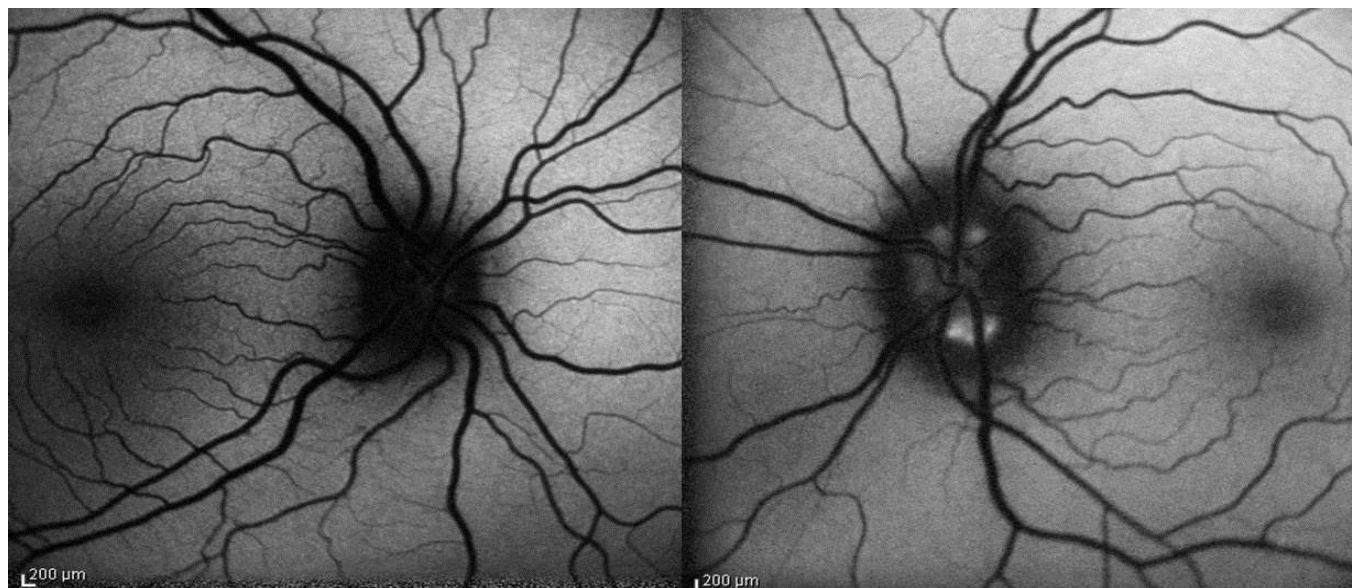


Figure 5. The characteristic sign of drusen in the FAF imaging can be seen in this figure. A hyperautofluorescence disc head was seen on the left eye of the patient.

Discussion

ODD is hyaline like calcified material accumulation that occur at the optic nerve head. It has an autosomal dominant inheritance pattern with incomplete penetrance. Actually, the narrow scleral canal and its vascular plexus are inherited, subsequently ODD developed. Elevation and blurriness of the disc head are basic findings of the ODD [1]. It is primarily associated with the disturbances in the axonal metabolism [1]. Secondly, it may also be developed due to the compression of prelaminar nerve fibers and capillaries from these deposits. Patients with ODD usually have normal visual acuities. However, visual fields defects may be so minimal that the abnormality never noticed or progress over time. It is found clinically in 0.3-2% of the population and seen 70% bilaterally without gender preference [2].

The ophthalmoscopic appearance of the ODD is related to the localization of drusen. While it is difficult to see a buried disc drusen on fundoscopic examination, exposed ones can easily be diagnosed. Buried disc drusen are usually seen in children and adolescents. It is important to differentiate them from papilledema, particularly in bilateral cases. Papilledema related to raised intracranial pressure is characterized with hyperemia of the disc, elevation of the peripapillary region and obstruction of the vessels around the disc margin. Soft exudates and peripapillary hemorrhages may be present. Spontaneous venous pulsations that are normally seen in funduscopy, are not seen in cases of true papilledema [1]. However, in ODD, there is not an obstruction in the peripapillary retinal vessels. The other sign in differential diagnosis of true papilledema from buried disc drusen is the absence of small vessels on the disc surface. Also staining and micro-

aneurysmal dilatation in the papillary vessels can be seen in the FFA [2]. There is published reports that stated papilledema and ODD could be seen at the same time. This relationship is mentioned for the first time in 1979. JD Rossiter et al. presented an 11-year-old girl who has bilaterally ODD concomitant with bilateral papilledema due to idiopathic intracranial hypertension in 2005 [3].

Hyaline like calcified materials are concentrated in the mitochondria of cells at first. As time goes by these deposits accumulate in the cells and then get out of cells where conventional extracellular accumulation formed. Most of the ODD cases are asymptomatic. They are generally detected incidentally during ophthalmologic examinations performed for other reasons. It has been reported that the presence of ocular vascular anomalies (abnormal tortuosity and branching, increased incidence of cilioretinal arteries and presence of retinochoroidal collaterals) with ODD concomitantly may result in complications such as anterior ischemic optic neuropathy, central retinal artery and vein occlusion or subretinal neovascularization [4].

In the differential diagnosis of ODD, especially for children, USG has a great importance by exhibiting hyperechoic lesion occurred due to deposition of calcium in optic disc. In initial stages, OCT measurements show an increase in RNFL thickness. As drusen progress and become superficial, the RNFL thickness decreases. The damage involves to especially the nasal nerve fibers resulting in temporal visual field defects [4]. In an controlled study designed by Roh et al., it has been found that RNFL thicknesses in ODD cases were lower than the control group [5].

In our case, it is important to observe visual field defects without any decrease in RNFL thickness. We think that this may be due to the inaccurate analysis of the instrument in RNFL measurements, particularly in the diseases which affect the optic nerve head morphology.

Mustonen [6] conducted a clinical trial on 200 patients with pseudopapilledema with and without visible optic disc drusen. Visual field defects were found in 38.9% of the pseudopapilloedema eyes without verified drusen, but in 73.4% of the verified drusen eyes. He stated that the largest field defects were found only in the eyes with drusen seen by oblique illumination or with superficial exposed drusen. These defects may disturb few patients so you can only detect the defects by perimetry.

ODD patients may have mild to moderate visual acuity loss. Almost 87% of the cases will have a progressive visual field loss in a period of their whole life. Calvo Gonzales and colleagues identified visual field defect patterns in the patients; and reported that visual field defect patterns are not always compatible with the location of

drusen. Authors have defined the patterns as well; 40% of inferior arcuate scotoma (half is the nasal scotoma), 40% of lower hemifield involvement (with mild upper hemifield involvement), 10% of superior arcuate and nasal scotoma and 10% of isolated nasal scotoma [7]. In our case, the patient had a peripheral arcuate scotoma in the lower and upper half of the temporal quadrant.

There is no effective treatment of ODD. Pfrrie M. et al. presented a 53 year old female patient with bilateral ODD who has 20/25 visual acuity in her right eye, and hand-motion in her left eye. They performed vitrectomy for the left eye and tried superficial excision of the drusen. But they ended the surgery because of the drusen's extensions into the optic disc and the central retinal vessels passing through [8]. In case of sudden vision loss in ODD patients; anterior ischemic optic neuropathy, central retinal artery or vein occlusion, peripapillary choroidal neovascularization and central serous papillopathy should be considered [9-11]. Color vision of ODD cases can be affected as well. Liutkeviciene and colleagues made a study in 2010 to explore this association. In this study 67 eyes of 37 ODD patients are compared with 200 eyes of 100 healthy participants for contrast sensitivity and tested with Farnsworth Munsell. The authors have reported better results in healthy eyes [12].

ODD should always be kept in mind to avoid unnecessary tests and interventions; in the differential diagnosis of pseudopapilledema. On the other hand, patients diagnosed with ODD should be informed for plausible visual field loss in the long term and must be followed up periodically.

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