

## Optic Disc Drusen, Important Detail in the Differential Diagnosis of Optic Disc Edema

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### Authors' contributions

This work was carried out in collaboration between all authors. Author MCS designed the case study, conducted diagnostic procedure and wrote the first draft of the manuscript. Authors RD and EÇ conducted a diagnostic procedures and managed the literature searches. Authors RD and Sİ conducted diagnosis and treatment. All authors read and approved the final manuscript.

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Case Report

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### ABSTRACT

Pseudopapilledema is used for pathologies that mimicking the optic disc edema. Among these, pseudo papilledema is commonly produced by optic disc drusen (ODD). A 17-year-old female patient was referred to the ophthalmology department from child neurology department with papilledema diagnosis because of bilateral elevated and blurred optic disc. Following ophthalmologic evaluation with fundus examination, B-scan ultrasound, red-free fundus photography with revealed autofluorescence the diagnosis of papilledema was excluded in the case and optic disc drusen was diagnosed. Swelling of optic disk and blurs of the optic disk margins is not equal papilledema. It is important to keep up pseudopapilledema in the differential diagnosis of papilledema.

**Keywords:** Optic disc drusen; papilledema; pseudopapilledema; children; pediatric; diagnosis.

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## 1. INTRODUCTION

Pseudopapilledema is used for pathologies that mimicking the optic disc edema. Among these, pseudopapilledema is commonly produced by optic disc drusen (ODD). ODD are globular, calcified hyaline bodies which contain mucoprotein and mucopolysaccharides, at the head of the optic nerve. ODD may result from abnormal axonal metabolism or may form secondarily from extracellular preliminary deposits. ODD usually don't produce any visual symptoms and detect incidentally. Myelinated nerve fibers, peripapillary glial tissue, tilted disc, compact disc due to hyperopia are other pathologies that should come to mind in the differential diagnosis. In this report, we report a case of the bilateral ODD in a 17-year-old female patient mimicking papilledema.

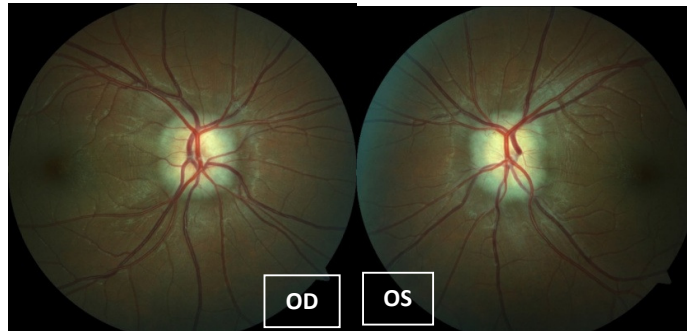
## 2. CASE REPORT

A 17-year-old female patient was referred to the ophthalmology department from child neurology department with papilledema diagnosis because of bilateral elevated and blurred optic disc. During the ophthalmological examination,

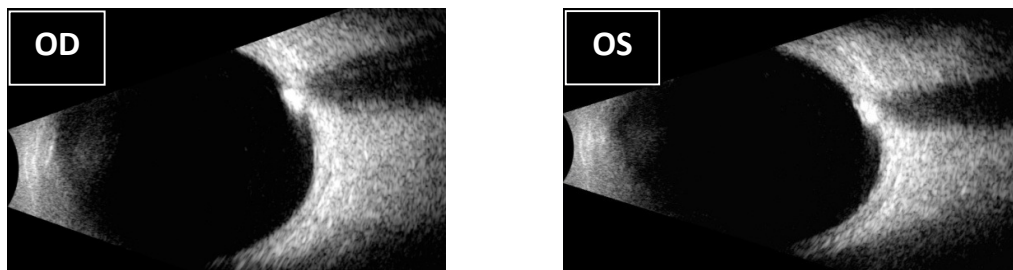
corrected visual acuity was 20/20 OD and 20/20 OS. The patient was orthophoric with no restriction of extraocular motility. Anterior segment examination in both eyes was unremarkable and pupils were briskly reactive with no relative afferent papillary defect. Color vision tested by Ishihara isochromatic plates was 21/21 in both eyes. Bilateral optic disk elevation with blurred disk margins was observed on fundus examination (Fig. 1). B-scan ultrasound revealed bilateral hyperechogenic lesions at the junction of the retina and optic nerve (Fig. 2). Red-free photography revealed autofluorescence of the drusen without the administration of fluorescein (Fig. 3). Retinal nerve fiber layer thickness was normal (Figs. 4a and 4b). The diagnosis of bilateral pseudopapilledema was made and the patient and the clinician in pediatric neurology department informed. Any therapy or interventional procedures not recommended. We obtained signed statement from the patient's parents.

## 3. DISCUSSION

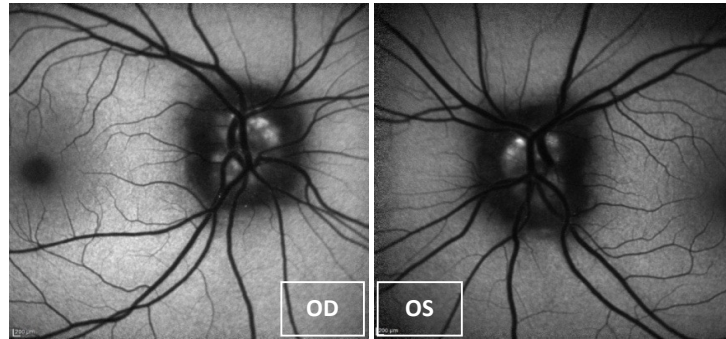
Optic disc drusen affects 2% of the population, however, those with a family history was found to be 3.4%. It is clinically apparent in only about



**Fig. 1. Optic disc photos of a 17 year old girl with bilateral buried optic disc drusen. The disc margins are blurred, but there are no hemorrhages or exudates**



**Fig. 2. B-scan ultrasonography of case showing a hyperechoic region over the optic nerve head (OD and OS B-scan)**



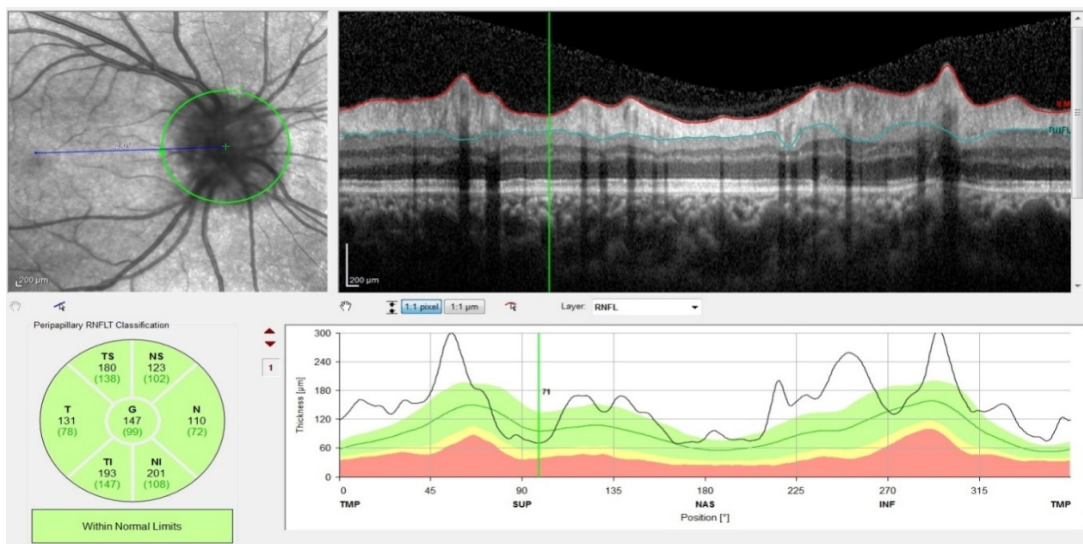
**Fig. 3. The red-free fundus photographs of case; revealing autofluorescence of the optic discs**

0.35% of individuals. Men and women are affected equally. Hereditary ODD development is associated with small scleral foramina and resistance of the normal axoplasmic flow [1-2].

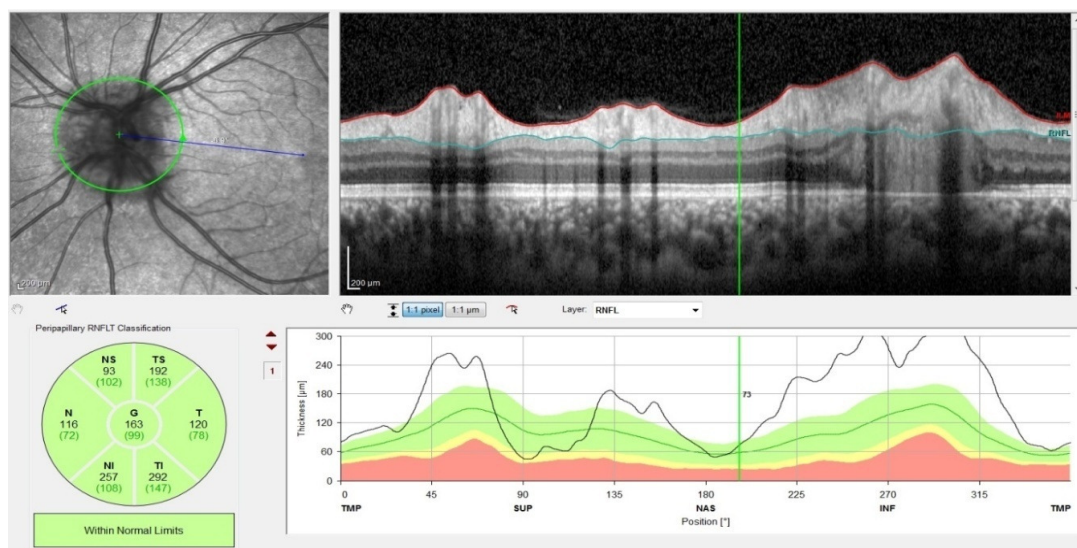
Most patients of ODD are asymptomatic, however, episodic blurred vision can be seen in some patients, due to probably transient ischemia caused by compression effects and occur bilaterally. Optic disc drusen can be superficial or buried. Buried optic disc drusen are often seen in younger patients and may become visible by moving forward over time [3]. We found asymptomatic bilateral buried ODD in our 17-year-old female patient. Small buried drusen may cause only elevation of the disc but especially superficial drusen may often seen white nodules. Especially in papilledema with buried drusen are particularly vulnerable to misdiagnosis and unnecessary initiatives such as

lumbar puncture. B-scan ultrasound and red-free photography without the administration of fluorescein may be helpful in identifying ODD and to rule out true papilledema [4]. Thickening of the retinal nerve fiber layer is often seen [2]. Nasal RNFL thickness has the highest diagnostic ability to differentiate true papilledema from pseudopapilledema [5].

Visual field defects have been reported in 76% of eyes with visible drusen and in 46% of those without visible drusen but with the appearance of pseudopapilledema [6]. Novel et al reported 32% inferior nasal, 21% nonspecific, 21% generalize constriction, 18% enlargement of the blind spot visual field defects in 15 children with buried and superficial ODD [7]. We detect small nasal visual field defect only on right eye. There is no established treatment for visual field defects related with ODD [2]. Optic disc drusen may



**Fig. 4a. OCT of right optic nerves showing elevated optic nerve heads and peripapillary RNFL thickening**



**Fig. 4b. OCT of left optic nerves showing elevated optic nerve heads and peripapillary RNFL thickening**

associated with retinitis pigmentosa, pseudoxanthoma elasticum, angioid streaks and Alagille syndrome (90% unilateral, 50% bilateral) [8-9]. Juxtapapillary choroidal neovascularization, vitreous hemorrhage, vascular occlusions such as partial anterior ischemic optic neuropathy, rarely may occur with ODD [2].

#### 4. CONCLUSION

Swelling of optic disk and blurs of the optic disk margins is not equal papilledema. It is important to keep up pseudopapilledema in differential diagnosis of papilledema. It is necessary to remind the pseudopapilledema reasons such as ODD which is not required treatment in the case of papilledema and it will avoid unnecessary invasive procedures.

#### CONSENT

Written informed consent was obtained from the patient and her parent for publication of this case report and any accompanying images.

#### ETHICAL APPROVAL

All authors hereby declare that all experiments have been examined and approved by the appropriate ethics committee and have therefore been performed in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki.

#### COMPETING INTERESTS

Authors have declared that no competing interests exist.

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