

Bilateral Buried Optic Nerve Head Drusen : A Case Report In An Adolescent Male With Keratoconus

Madhuri Upadhyaya¹ , Geetha Ganesan² , Greesha Raveendran³,

Abstract

Optic nerve head drusen (ONHD) are uncommon, usually asymptomatic acellular calcific deposits in the optic nerve head. They are thought to occur in genetically predisposed individuals as by-products of impaired axonal metabolism due to a narrow scleral canal. They are commonly incidentally detected on routine ophthalmic examination and are generally non progressive. Here we present a case report of a 16 year old male who came to us with diminution of vision in both eyes since 1 year and was diagnosed to have keratoconus. Fundus examination showed blurring and elevation of both optic disc margins. Suspecting papilledema, MRI brain was done and found to be normal. Further tests confirmed the diagnosis of bilateral optic nerve head drusen.

Key words: Optic disc drusen, blurring of disc, B-scan, fundus autofluorescence, papilledema

Date of Submission: 20-11-2023

Date of Acceptance: 30-11-2023

I. Introduction

Optic nerve head drusen (ONHD) was first described by Muller in 1858 [1]. It is an uncommon condition where there are deposits of acellular calcified substances like hyaline, amino acids, nucleic acids and mucopolysaccharides in the prelaminar part of the optic nerve head [2]. The exact pathophysiology is not known but mechanisms like abnormal axonal metabolism, axonal disruption, extrusion of mitochondria into the extracellular space and continuous calcium deposition is hypothesized [3].

II. Case Report

A 16 year old male came to our hospital with complaints of gradually progressive painless diminution of vision in both eyes since about a year. Visual acuity was 6/18 in the right eye and 6/36 in the left eye. The best corrected visual acuity (BCVA) was 6/9 in the right eye with -1.50 DS -3.25DC x 90 whereas it was 6/18 in the left eye with -3.50 DS -3.50DC x 100. Corneal pentacam revealed both eye keratoconus, he was planned for corneal collagen cross linking and sent to retina OPD for retina evaluation. Dilated fundus examination showed blurring and elevation of both disc margins without any other associated features (Figure 1 and Figure 2). Colour vision was normal, visual fields showed generalized constriction and fundus autofluorescence revealed no hyperfluorescence over the optic disc. Suspecting intracranial pathology, MRI brain was done which turned out to be normal. On the following visit, ultrasound B-scan was done which revealed hyperechoic lesions in the posterior pole, posterior to the optic nerve head in both eyes which also showed high amplitude spikes on A-scan consistent with the diagnosis of buried optic nerve drusen (Figure 3 and Figure 4). Patient and his family were counselled regarding the condition and advised regular follow up.



Figure 1



Figure 2



Figure 3



Figure 4

III. Discussion

Optic disc drusen, though an uncommon condition, can cause a significant diagnostic dilemma since it can mimic disc edema.

They can be of two types, visible drusen, which protrude from the disc margin and can easily be visualized as prominent, refractile, rounded, pale deposits and buried drusen, which cause elevation of the disc with blurred margins, often mimicking disc edema. Retinal vessel anomalies like increased tortuosity, branching or looping can also be associated.

They are usually asymptomatic and an incidental finding on routine ophthalmic examination. However they can be associated with visual field defects like enlargement of the blind spot, nerve fiber bundle defects, generalized constriction and also acute vision loss due to anterior ischemic optic neuropathy, vascular occlusion or choroidal neovascularization [4]. Buried ONHD are more prevalent in children, and studies suggest that either they remain stationary (more commonly) or undergo a transition phase in adolescence, where they become more prominent due to increase in size, number, anterior migration or thinning of the overlying nerve fibre layer [5].

Accurate diagnosis can be a challenge in cases of buried drusen, which can mimic papilledema. B-scan ultrasonography is a simple and reliable diagnostic modality which is more sensitive compared to fundus autofluorescence and CT scan. With the advancement in optical coherence tomography (OCT) like the enhanced depth imaging (EDI-OCT) and swept source (SS-OCT), the diagnosis has become more streamlined. There is no known effective treatment for this condition but a routine follow up is advisable to detect the development or progression of visual field changes or any other complications, which can be managed symptomatically.

IV. Conclusion

Optic disc drusen, especially buried drusen, can mimic papilloedema, which can result in very elaborate and unnecessary investigations like CT scan, MRI to rule out causes of increased intracranial tension. But with the evolution of newer and more sophisticated diagnostic modalities, an accurate and early diagnosis is possible in a much more cost effective manner. However we need to keep in mind that there can also be a possibility of patients having superimposed true papilledema because of idiopathic intracranial hypertension (IIH) or an intracranial space occupying lesion (ISOL), which can sometimes be life threatening. In addition, optic disc drusen in small optic discs may mask glaucomatous cupping or they may be more susceptible to glaucomatous damage (6)

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