Brief Communication

Congenital Optic Disc Drusen Misdiagnosed As Benign Idiopathic Intracranial Hypertension

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ABSTRACT

A 16 year old female, was referred by the neuro-physician for the evaluation and management of decrease visual acuity in the left eye and papilledema, she was diagnosed Benign Idiopathic Intracranial Hypertension. She was under treatment for 4 years for papilledema. Visual acuity was 6/6 in the right eye and 1/60 in the left eye. Slit lamp examination was normal. Fundoscopy showed lumpy appearances of the optic nerve heads with vessels and macula normal, in both eyes. Her B scan ultrasonography and Computerized Tomography scan were carried out that revealed buried optic disc drusen a cause of pseudo-papilledema. On General examination she had typical Cushingoid features that she developed due to excess steroid therapy she was prescribed.This casehighlights the importance of identifying buried optic disc drusen, being misdiagnosed and treated as true papilledema secondary to Benign Idiopathic Intracranial Hypertension.

Key Words: Optic disc drusen, pseudopapilledema, Benign Idiopathic Intracranial Hypertension, cushingoid features.

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INTRODUCTION

Bilateral optic disc swelling can be due to papilledema or pseudopapilledema. The former is represented as true disc edema and warrants immediate magnetic resonance imaging (MRI), lumbar punctures, diuretics and steroid administration for evaluation and management. Latter is a group of congenital optic disc etiologies including hypoplastic, dysplastic, or tilted optic disc, persistent hyaloid remnants and optic disc Drusen (ODD).[1] Pseudopapilledema is most commonly observed in ODD. Drusen are extracellular, proteinaceous deposits of calcium and hyaline caused by impaired axoplasmic flow resulting in extrusion of drusen or degeneration of the retinal ganglion cell nerve fiber axons. With time, drusen causes elevation of the optic nerve head. There are two types of drusen, superficial and buried. Buried drusen mimics true disc edema on fundoscopy and may lead to misdiagnosis.[2]

According to a study, drusen occur in 0.3 – 2% of the general population, are bilateral in 70%, with no sex predilection.[3] ODD are inherited in an irregular dominant fashion. The close relatives of the patients can be screened for the final diagnosis. There are several options for managing ODD including lowering the intraocular pressure and vasoactive therapy. To date, there is no treatment established to modify the clinical development of optic nerve head drusen (ONHD). However, lowering the intraocular pressure can aid in neuro-protection, yet it is still indefinite whether or not an elevated intraocular pressure is damaging to the optic disc in ONHD.[4]

Papilledema can present as a medical emergency, presenting with nausea, vomiting, headache and profound loss of vision. It is crucial to exclude the causes of optic disc swelling. The initial differential diagnosis includes etiologies from raised intracranial pressure resulting from a space-occupying lesion (e.g. tumor, abscess), hydrocephalus, consequence of

trauma as subdural hematoma or cerebral edema, meningitis. Optic disc swelling can cause papilledema from optic neuritis, uveitis, scleritis. Subsequent treatment depends on the cause.[5] Benign Idiopathic Intracranial Hypertension is an important cause of papilledema and is the diagnosis of exclusion.

True edema presents with dilation and telangectasia of the disc capillaries and may demonstrate hemorrhages on or adjacent to the disc. In pseudopapilledema, disc vessels are normal and disc margins are clearly visible.[6]

This case highlights the importance of identifying buried optic disc drusen, being misdiagnosed and treated as true papilledema secondary to Benign Idiopathic Intracranial Hypertension.

Case Presentation

16 year old female presented to the outpatient department of Jinnah Post Graduate Medical Centre with the complaint of headache and decrease vision in the left eye for the past five years. She was a known case of Benign Idiopathic Intracranial Hypertension and was admitted in the neuro-medicine department in a renowned hospital and was referred for visual assessment and papilledema evaluation. She was under treatment of a neuro-physician for last four years and was diagnosed papilledema. Her neurological examination was normal. She underwent multiple lumbar punctures with normal composition of cerebrospinal fluid and serial MRIs that were normal. She was prescribedoral steroids and acetazolamideon and off for the past three years. She had consulted various ophthalmologists in her vicinity where she was found to have bilateral optic disc edema and referred to neuro-physician. We performed her ophthalmologic examination that revealed her visual acuity was 6/6 in the right eye using Snellen chart, and left eye was 1/60, whereas her best corrected visual acuity was 6/60 after +2.50DS. On slit lamp examination her anterior segment was normal in both the eyes; there was relative afferent papillary defect in the left eye. Intraocular pressures were 14mm of Hg in both eyes and extraocular movements were normal. Color vision was normal in the right eye and impaired in the left eye. Confrontation tests revealed constricted peripheries and fundoscopy of both eyes (Figure 1) showed lumpy disc appearance with indistinct margins and macula was normal.B scan ultrasonography of both eyes (Figure 2) was done that showed elevation of the optic nerve head, with hyper-echoicreflection at the centre of the posterior globe in contrast to papilledema where signal intensity is reduced due to edema. Computerized Tomography (CT) scan of the orbit and brain (Figure 3) revealed focal calcification involving the junction of posterior globe and optic disc suggestive of bilateral optic disc drusen.Red-free fundus photography (Figure 4) showed autofluorescence of the optic discs. Perimetry (Figure 5) showed profound field losses in both eyes. She had no neurological signs and was diagnosed with bilateral ODD. On detailed inquiry she denied any nausea, vomiting and she complained of weight gain of 10 kgs in the past 3 years, with puffiness of the face, generalized weakness, muscles and bone pains,

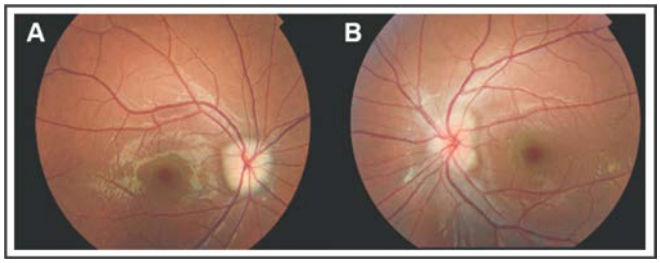

Figure 1: Fundus Photos of the Right (a) and Left eye (b) Respectively showing lumpy disc appearances.

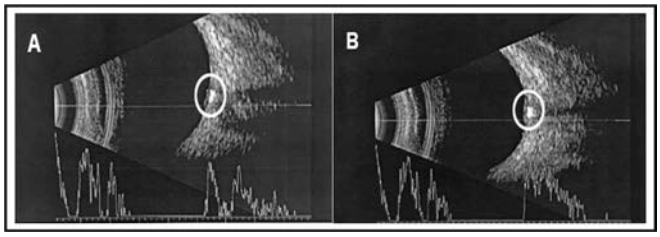

Figure 2: B Scans of Right (A) and Left eye (B) respectively showing hyper echoic reflection in the posterior globe depicted by circles.

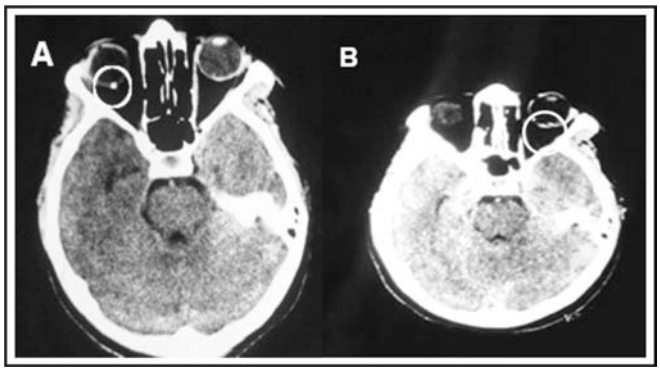

Figure 3: Axial CT Scan (Brain and Orbit) showing focal calcification (round circles) in the posterior globe of left (A) and right (B) eyes respectively.

menstrual disturbances and depression since the treatment continued. On general physical examination she had typical cushingoid appearance secondary to excess steroid therapy. The diagnosis was explained and the patient was referred to endocrinologist and medicine department for evaluation and management. Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

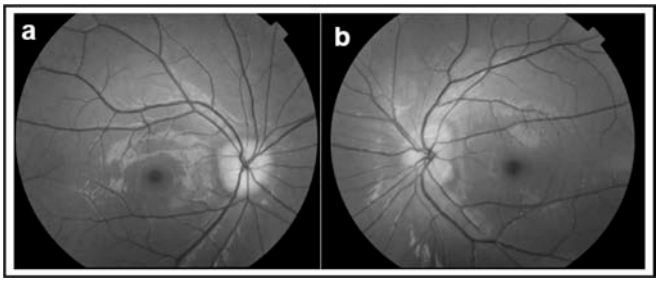

Figure 4: Fundus Autoflourescence of Right (a) and Left eye (b) respectively revealed autoflourescence.

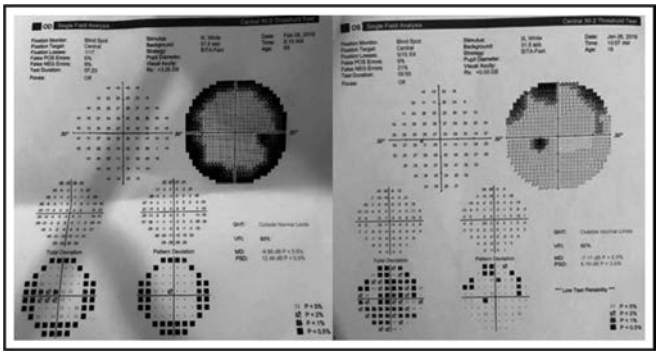

Figure 5: Showing profound visual field defects in right (left picture) and left (right picture) eyes respectively.

DISCUSSION

When doctors examine the fundus and find bilaterally elevated optic nerve heads, they may face a diagnostic challenge. Differentiating congenital etiologies of optic disc elevation (known as pseudo-papilledema) from true papilledema is overbearing. According to a case serial study, approximately 75% of population diagnosed as papilledema turned out to have pseudo-papilledema or anomalous discs. True papilledema usually presents with neurological signs and symptoms like vertigo, nausea, vomiting or tinnitus, whereas 33% of patients are found to be asymptomatic.[7] ODD, which are laminated hyaline bodies within the optic nerve head, could lead to hard amorphous optic disc elevations. Whereas, true papilledema is a medical emergency and needs immediate attention therefore, diagnosis of ODD is very important to prevent an extensive, invasive, expensive and unnecessary neuro-ophthalmologic work-ups.[8]

The use of systemic corticosteroidsfor Benign Idiopathic Intracranial Hypertension has always been controversial sincetheir mechanism of action isuncertain. Review of literature says that patients presenting with visual loss who are prescribed steroids show initial improvement but recurrence of visual loss is evidenced on tapering of steroids, followed by recurrence of papilledema and visual loss. So use of long-term steroids to treat papilledema has largely been restricted outweighing their adverse side effects.[9]

ODD are typically bilateral and more observed to be buried in younger patients than in adults. Thus, they may be difficult to distinguish from true papilledema, that commands need for investigation of the underlying cause. Ancillary testing such as B scan, fundus auto flourescecnce and CT scan should be performed in every case to rule out pseudo-papilledema, because an initial misdiagnosis of papilledema can lead to serious and invasive procedures in patients increasing their suffering.

CONCLUSION

As papilledema mimics pseudopapilledema it can easily be misdiagnosed, and when no apparent cause is ruled out for papilledema it is labelled as Benign Idiopathic Intracranial Hypertension. It is pivotal for primary care physicians as well as specialist care ophthalmologists to be aware of anomalous disc appearancesin order to have an accurate diagnosis along with proper, timely treatment.

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Conflict of Interest

Authors declared no conflict of interest.

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Authors' Designation and Contribution

Nida Shamim; Consultant Ophthalmologist: *Concepts, Design, Literature search, Data acquisition, Data analysis, Statistical analysis, Manuscript preparation, Manuscript editing.*

Muhammad Ali Tahir; Consultant Ophthalmologist: *Data acquisition, Manuscript editing, Manuscript review.*

Alyscia Cheema; Professor: *Manuscript editing, Manuscript review.*

Anum Javed; Resident: *Design, Literature search, Statistical analysis, Manuscript preparation, Manuscript editing.*