Bilateral Optic Disc Drusen in Hypermetropic Children of a Family

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Optic disc drusen are calcified, round, yellow nodules, with various sizes tend to be located within any part of the optic nerve head. According to most theories they are caused by impaired ganglion cell axonal transport, probably related to a small sclera canal and mechanical obstruction. We report a case of 12 year old male child and his 13 year old sister, both presented with decrease vision in our outpatient department. Fundus examination of both patients revealed bilateral optic disc swellings with no hyperemia. After investigations, it was proved that both patients suffered from bilateral buried optic disc drusen. Decrease vision was because of hypermetropia in both children.

CASE REPORT

12 year old male child and his 13 year old sister presented in our eye outpatient department with decrease vision in both eyes since last 5 year. Both were student and had no systemic illness. General physical examination and systemic examination were unremarkable.

Ophthalmologic examination of male child revealed best corrected visual acuity with correction of +19.5 DS in both eyes to be 6/60 (using Snellen’s visual acuity chart) with no pin hole improvement. Best corrected visual acuity of female patient with correction of +19.0DS in both eyes was 6/36 (using Snellen’s visual acuity chart) with no pin hole improvement. There was no deviation in any eye and extra ocular muscle movements were full in both eyes of both patients. Both pupils were round, regular and reacting to light and no relative afferent pupillary defect was noted. Biomicroscopic examination of both anterior segments was unremarkable. Fundoscopic examination showed bilateral blurred and swollen optic discs margins with no hyperemia and with obliteration of the physiological optic disc cupping (Fig 1A, 2A). Venous pulsations were present in both eyes in both patients. Visual field examination by confrontation showed grossly restricted peripheral
visual fields in both eyes but it was not possible to perform automatic Humphrey perimetry as the patients were young in age so were uncooperative and unable to understand commands for automatic humphery perimetry. A diagnosis of bilateral optic nerve head drusen was made, which was subsequently confirmed by B scan ocular ultrasonography (Fig B) and Optical Coherence Tomography (Fig C) in our ophthalmic outpatient department.

Additionally CT scan brain and orbit was done to see the optic nerve head calcification and to exclude any central nervous system pathology. Patients and their parents were informed about the condition and cause of decrease vision, which was hypermetropia and their parents were also informed that they and their other offspring should have an ophthalmic examination to exclude optic disc drusens.

**DISCUSSION**

Optic disc drusen are congenital and developmental anomalies of the optic nerve head seen frequently in clinical practice, often as an incidental finding during routine ophthalmic examination. Optic disc drusen can affect children as well as adults. It is important to consider optic nerve head drusen in the differential diagnosis of papilledema or optic nerve swelling in any age group. The primary pathology of optic disc drusen is an inherited dysplasia of the optic disc and its blood supply, which influence the formation of optic disc drusen. Optic disc drusen has a greater tendency to form in eyes with a small scleral canal; therefore hypermetropic eyes would have a higher rate of optic disc drusen relative to myopic eyes. The diagnosis of optic disc drusen can be made with
clinical findings combined with B scan ultrasound, Fundus fluorescein angiography, Ocular computed tomography and newer modalities using optical coherence tomography of optic nerve head, but ocular B-Scan ultrasonography, a non-invasive and cost-effective technique is the most sensitive and method of choice in the recognition of optic disc drusen. Optic disc drusen shows auto-fluorescence (ability of substance to emit yellow-green light when stimulated by blue light in the absence of fluorescein dye) in Fundus fluorescein angiography. Incidental asymptomatic orbital calcifications are frequently encountered on modern high-resolution CT scan images of the brain and orbit. Optical coherence tomography can differentiate optic disc drusen from papilledema and small optic disc without disc drusen as optic nerve head drusen typically elevate the disc surface and appear as an optically empty cavity, sometimes with a perceptible reflection from the posterior surface. The surface of the small optic nerves was slightly elevated, but with less anterior reflectance, therefore OCT of optic nerve head exposed unique and clinically helpful views of optic nerve drusen.

The aim of presenting this case is that we should consider optic disc drusen in any patient who presents with bilateral optic disc swelling, particularly when the patient is hypermetropic and asymptomatic.

CONCLUSION
In our case the unique feature is presence of bilateral optic disc drusen in hypermetropic two children of the same family. Optic disc drusen may be hereditary and have close association with small scleral canal.

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REFERENCE