


Research Article

An Illustrative Case Report of Anterior Migration of Optic Disc Drusen in A Child

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Abstract

Optic disc drusen (ODD) are acellular calcified deposits located both intracellularly and extracellularly within the optic nerve. Their appearance typically change over the first two decades as they migrate anteriorly towards the optic nerve head, from an initial buried location. They can pose a diagnostic challenge as can simulate the appearance of optic disc swelling. Distinguishing ODD from true optic disc swelling is of paramount importance to avoid delayed diagnosis of potentially life threatening neurologic disease and on the other hand prevent invasive and unnecessary work-up for elevated intracranial pressure, if not clinically indicated. A number of imaging modalities are available to assist in the diagnosis and are discussed below.

Keywords: Optic disc drusen (ODD), Ophthalmology, EDI-OCT, OCT RNFL

Introduction

Optic disc drusen (ODD) are acellular calcified deposits located both intracellularly and extracellularly within the optic nerve. Their appearance typically change over the first two decades as they migrate anteriorly towards the optic nerve head, from an initial buried location. They can pose a diagnostic challenge as can simulate the appearance of optic disc swelling. Distinguishing ODD from true optic disc swelling is of paramount importance to avoid delayed diagnosis of potentially life threatening neurologic disease and on the other hand prevent invasive and unnecessary work-up for elevated intracranial pressure, if not clinically indicated. A number of imaging modalities are available to assist in the diagnosis and are discussed below.

Methods

We present an illustrative case of the change in appearance of ODD due to anterior migration over a 3-year period, in a young child.

Results / Case report

A young female aged 3 years was referred by her optician to the Ophthalmology department for urgent review and evaluation of blurred disc margins. Her parents had taken her to the optician regarding a recent onset squint, which was first noticed 3 weeks previously. She had been well up until the day of her review, during which she was feeling nauseous and vomited on four occasions. She had no significant past medical history. She was born by elective caesarean section at full term, weighing 4.28kg. She was on no regular medications. There was a positive family history of strabismus.

On examination, her logMAR visual acuities were 0.175 and 0.275 in the

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right and left respectively. Orthoptic examination showed a small constant left convergent squint, measuring 16-18 prism diopters base out for near and 14 prism diopters base out for distance. Her extraocular movements appeared full. Her pupils were equal and reactive to light. There was no relative afferent pupillary defect present. Cycloplegic refraction revealed a hyperopia, with measurements of +3.50 dioptres sphere in each eye. On funduscopy, the optic disc margins were poorly defined in both eyes, more marked in the right eye (figure 1A) compared to the left (figure 1B). There were no haemorrhages, exudates or vessel obscurations. OCT of the optic nerve heads revealed a non-specific elevation, but no optic disc drusen were seen (figure 1A and 1B). Fundus autofluorescence was normal. B-scan was unable to be performed, as she was feeling too unwell at the time. Given her history of sudden onset esotropia, symptoms of nausea and vomiting and possible swollen discs, she was admitted under the Pediatric team for further investigation. A full neurological examination revealed no focal neurology. An urgent MRI head showed no intracranial pathology. Her symptoms of nausea subsequently settled. Her observations remained stable and she was discharged the following day.

Follow-up at the Ophthalmology clinic a few weeks later confirmed a fully accommodative esotropia and she commenced full-time wear of her glasses. B-scan ultrasonography of the eyes identified bilateral hyper-reflective lesions in the optic nerve head, consistent with bilateral optic disc drusen. Orthoptic review continued, but she was discharged from the regular Pediatric Ophthalmology clinic.

Three years later, she was again referred by her optician for urgent evaluation of her optic discs, which were reportedly more elevated and more blurred than the previous images. She reported a two-week history of frontal headaches, associated with nausea. She had no history of transient visual obscurations and no tinnitus. On examination, visual acuities were 0.1 and 0.0 in the right and left eyes respectively.



Figure 1: Comparison of optic disc photos and EDI-OCT of optic nerve head of the right eye (1A) and left eye (1B) on initial presentation. Repeat optic disc photos and EDI-OCT of optic nerve head of the right eye (2A) and left eye (2B) on subsequent presentation 3 years later. Optic disc photos show increased blurring of the optic disc margins in both eyes. EDI-OCT shows marked increase in elevation of optic nerve head: maximum elevations increased from 743 microns to 886 microns (right eye), 579 microns to 818 microns (left eye).



Figure 2: B-scan ultrasonography of the right eye showing a hyperechoic lesion in the optic nerve head, consistent with optic disc drusen.

Color vision was normal on Ishihara testing. Her extraocular movements appeared full. Her pupils were equal and reactive to light. There was no relative afferent pupillary defect present. Funduscopy confirmed a change in appearance of the optic discs, with increased blurring of the margins compared to her previous review and EDI-OCT showed a pronounced elevation of the ONH in both eyes (Figure 2A and 2B). Fundus autofluorescence was again normal. B-scan ultrasonography of the eyes identified bilateral hyper-reflective lesions in the optic nerve head, consistent with previously diagnosed bilateral optic disc drusen (figure 2). Given the constellation of her recent onset symptoms and documented change in appearance of her optic discs, investigations to out-rule secondary pathology were arranged. MRI head was normal. Lumbar puncture was performed. Opening pressure was within the normal range and the cerebrospinal fluid analysis was normal. Her headaches settled spontaneously and she developed no further symptoms. The change in optic disc appearance was attributed to migration of her optic disc drusen.

Discussion

Optic disc drusen (ODD) are acellular calcified deposits located both intracellularly and extracellularly first described by Muller in 1858 [1]. They typically begin ‘buried’ in the substance of the optic nerve in early years, migrate forward with age and become visible around the second decade [2]. When visible, optic disc drusen look like yellow crystals within the substance of the optic nerve head. Initially, the optic disc appears indistinct, simulating swelling and is often cupless because the optic disc drusen are buried [2]. Distinguishing ODD from true optic disc swelling is of paramount importance to avoid delayed diagnosis of potentially life threatening neurologic disease and on the other hand prevent invasive and unnecessary work-up for elevated intracranial pressure, if not clinically indicated. It is

important to consider optic disc drusen in the differential for papilledema, as 50 to 55% of children initially diagnosed with papilledema have optic disc drusen as their final diagnosis [3]. Features in favour of papilloedema include obscuration of peripapillary vessels caused by nerve fibre layer oedema involving the peripapillary retina, hyperemia, hemorrhages, cotton wool spots, Paton lines, and exudates around the optic disc.

ODD are commonly buried when occurring in children. They can therefore be particularly difficult to distinguish from true optic nerve swelling. Few reports have demonstrated the evolution of ODD in children over time [4-7], progressing from anomalous disc appearance to ophthalmoscopically visible optic nerve drusen. However, there is a paucity of photographic documentation of the natural history of changes in the optic disc appearance over time, particularly in the earlier stages whilst the ODD remain buried. Opportunities to document the full panorama of drusen evolution in a particular patient are limited. Our case demonstrates a marked increase in elevation of both optic discs, attributable to drusen migration, without the drusen being ophthalmoscopically visible.

Determining a definitive diagnosis based on clinical examination alone can be challenging for even the most distinguished ophthalmologists. In these cases, a variety of imaging modalities may be employed to evaluate for the presence of ODD. B-scan ultrasonography has long been the preferred method for diagnosis of ODD [8]. Drusen characteristically appear hyperechoic with posterior shadowing on ultrasonography. Leon and colleagues reported on the cost-effectiveness of performing ultrasonography prior to neuroimaging in patients with ODD, with an overall reduction in the number of other investigations undertaken [9]. However, drusen and raised intracranial pressure can coexist [10, 11] and a diagnosis of drusen does not preclude the need for investigations for papilledema. In patients with signs or symptoms suspicious for another disorder, further evaluation may be necessary even if optic disc drusen are detected. Mosajee et al found the prevalence of coexisting ODD and raised intracranial pressure in children to be 10% in their pediatric practice [12].

Mosajee et al [12] also highlighted the importance of using ultrasonography not only to detect drusen but also to measure optic nerve sheath diameter, which if large is indicative of papilloedema. Carter et al [13]. Described a method of using ultrasound to identify fluid around the optic nerve (crescent sign) as a reliable sign of optic disc swelling. A-scan mode is used to determine the optic nerve sheath width in the primary gaze. If the optic nerve sheath was found to be wide (≥ 3.3 mm), a 30° test was then performed to differentiate fluid from solid thickening of the optic nerve sheath. The patient is instructed to re-fixate at least 30° away from the primary gaze

toward the ultrasound probe, at which point the optic nerve sheath was measured again. If fluid is present, the measured width of the optic nerve is expected to decrease, based on the assumption that when the eye is turned the optic nerve sheaths become stretched and the fluid is redistributed over a greater area. A reduction in width $>10\%$ is considered positive for the presence of fluid.

In more recent years, there has been considerable interest in the use of OCT in the diagnosis of ODD. Spectral-domain OCT (SD-OCT) offers a limited depth of imaging and is considered unreliable at distinguishing between buried optic disc drusen and true optic disc edema in children [14]. The introduction of enhanced depth imaging optical coherence tomography (EDI-OCT) has improved the visualization of more deeply buried ODD [15]. EDI-OCT allows for evaluation of the entire nerve head up to the lamina cribrosa which is hypothesized to be the most posterior extent where ODD may be found. The Optic Disc Drusen Studies Consortium recommends obtaining EDI-OCT (for identification of ONHD and measurement of scleral canal size), OCT RNFL, OCT Macula, and fundus autofluorescence [16]. Autofluorescence, however, does not reliably detect buried drusen [17]. Therefore, its usefulness in identifying optic disc drusen in children, who are more likely to harbour non-calcified buried drusen, has not been conclusively determined.

Learning points

This case illustrates the migration of optic disc drusen over a 3-year period in a child, where the margins of the discs over this time become more elevated and indistinct. EDI-OCT show a significant increase in the elevation of the optic nerve heads bilaterally, whereby it is difficult to outrule concurrent papilloedema. As discussed, ODD and papilloedema can coexist and it is therefore pertinent to take the full clinical picture into account when evaluating similar patients. The changes documented in this case were attributed to ODD migration, following full investigation, including neuroimaging and CSF sampling, as this was considered clinically indicated.

Conflict of interest

The authors report no conflict of interest.

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None to declare

Abbreviations

ODD: optic disc drusen, ONH: optic nerve head, OCT: optical coherence tomography, SD-OCT: spectral-domain optical coherence tomography, EDI-OCT: enhanced depth imaging optical coherence tomography

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