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CASE REPORTS

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Multimodal Imaging Characteristics of a Rare Co-occurrence of Optic Nerve Head Drusen and Peripapillary Myelinated Retinal Nerve Fibres

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ABSTRACT

Although isolated optic nerve head drusen (ONHD) and peripapillary myelinated retinal nerve fibres (MRNF) are described in the literature many times as far as we could reach, the combination of the two has been reported in only a single case without multimodal imaging. Here, we present multimodal imaging of a 47-year-old healthy woman with MRNF who had blind spot enlargement and a visual field defect in the left eye ascribed to the ONHD. ONHD may accompany MRNF which can hide the disc drusen. Clinicians, therefore, should be aware of this rare coexistence to explain possible complications such as visual field defects associated with drusen in patients with MRNF.

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Myelinated retinal nerve fibres; optic nerve head drusen; visual field defect

Introduction

Optic nerve head drusen (ONHD) are made up of calcified aggregates of extracellular materials including hyaline, calcium, and other proteins at the optic disc. Retinal ganglion cell nerve fibre degeneration or alterations in axoplasmic flow have been associated with their pathogenesis. ONHD, whose incidence ranges from 0.4% to 2.4%, are bilateral in 75% to 86% of cases.[1–4] Though it is mostly benign and asymptomatic, they can sometimes be associated with several complications including ischaemic optic neuropathy, central retinal artery, and vein occlusion.[5–7] In addition, visual field defects including nasal step defect, peripheral depression, arcuate defect, and enlarged blind spot can also be detected.[3] Visual field defects arising from disc drusen are considered to be the result of the pressure caused by the calcific material on the retinal nerve fibre layer.[8] In the present case, we demonstrate multimodal imaging of a 47-year -old healthy woman who had visual field defects associated with the disc drusen hidden by peripapillary myelinated retinal nerve fibres (MRNF).

Case report

A 47-year-old housewife was referred to our clinic with a preliminary diagnosis of papilloedema. Her past medical history revealed a throbbing headache for 2 months. The patient did not have any known chronic systemic disease, a history of surgery, or long-term drug usage or smoking. Her general physical and neurological examinations were unremarkable. On neuro-ophthalmological assessment, visual acuity without any correction was 10/10 in each eye on a Snellen chart. Colour vision using Ishihara pseudoisochromatic plate was 15/15 in each eye. Pupillary light and near reflexes were normal. Both pupils were isochoric, and there was no relative afferent pupillary defect. Ocular movements were full. Slit-lamp examination and intraocular pressure were normal in each eye. Fundus examination revealed blurred borders of both optic discs resulting from peripapillary MRNF with feathered borders (Figure 1). However, spontaneous venous pulsation was present and both optic discs were pink. Fundus autofluorescence imaging revealed superficial drusen on the nasal side of both discs with a "hyperautofluorescence" appearance (Figure 2). On red-free fundus imaging, the bilateral white appearance of MRNF was observed at the peripapillary area (Figure 3). Optical coherence tomography (OCT) passing through both optic nerve heads demonstrated thickened retinal nerve fibre layer (RNFL) and lumpy-bumpy internal contour (Figure 4). The visual field was normal in the right eye, and there was blind spot enlargement and a nasal step defect in the left eye (Figure 5). The patient was diagnosed as having both

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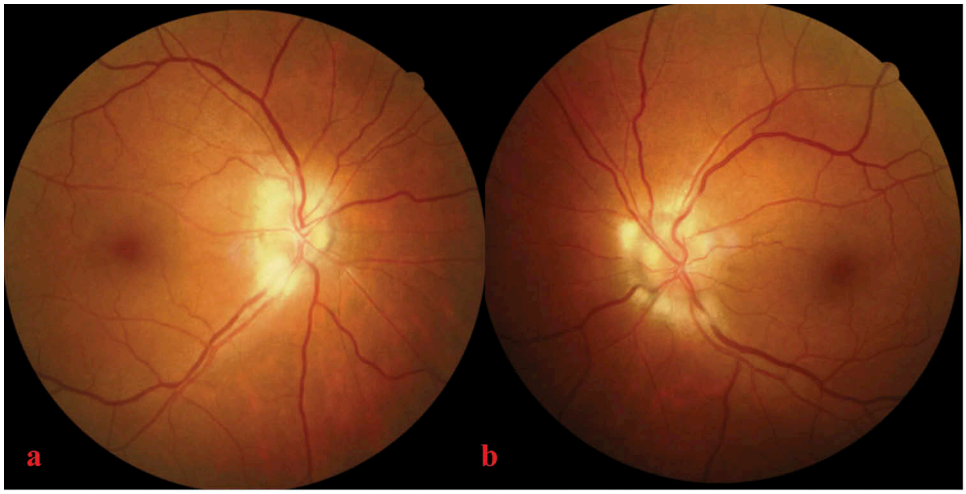


Figure 1. Colour fundus picture of right (a) and left (b) eyes depicting bilateral blurred border of the optic disc because of peripapillary myelinated retinal nerve fibres with feathered borders.

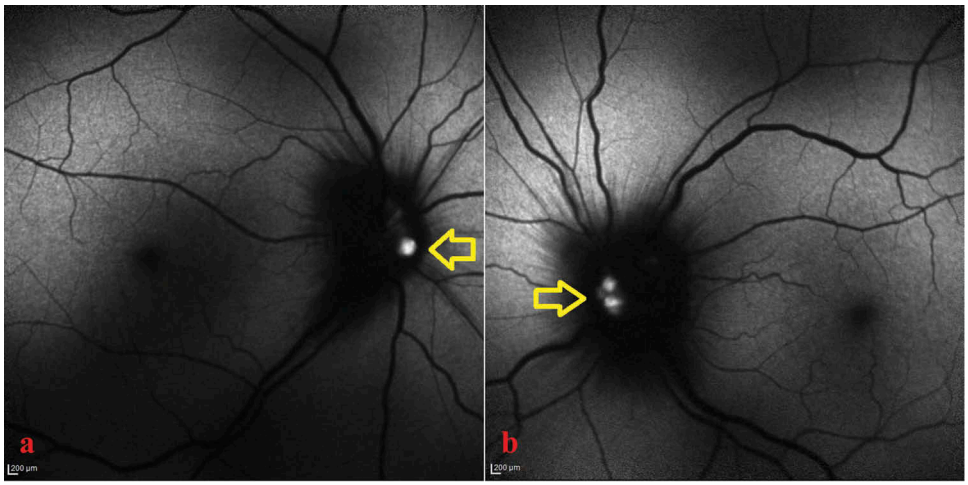


Figure 2. Fundus autofluorescence images of right (a) and left (b) eyes showing hyperautofluorescence appearance, corresponding to drusen in the nasal side of both discs (yellow arrows).

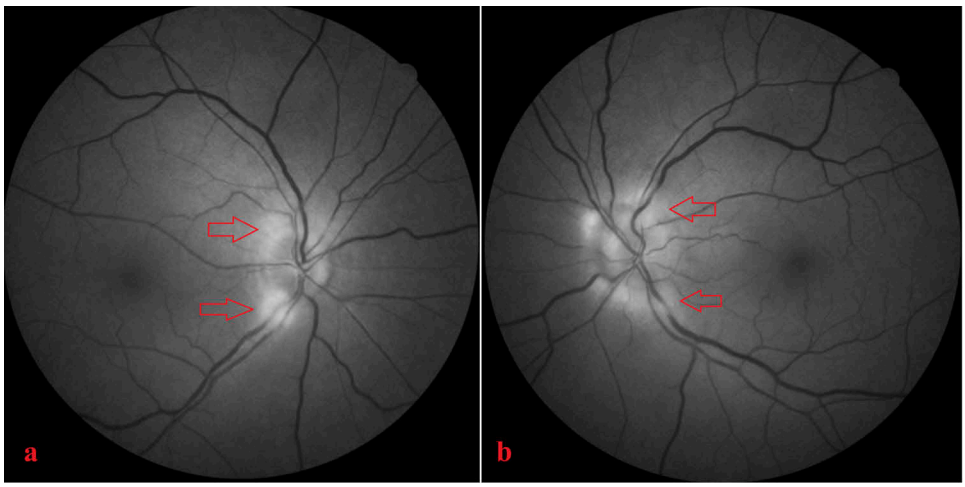


Figure 3. Red-free images of right (a) and left (b) eyes showing peripapillary white appearance because of the high lipid content of myelinated retinal nerve fibres (red arrows).

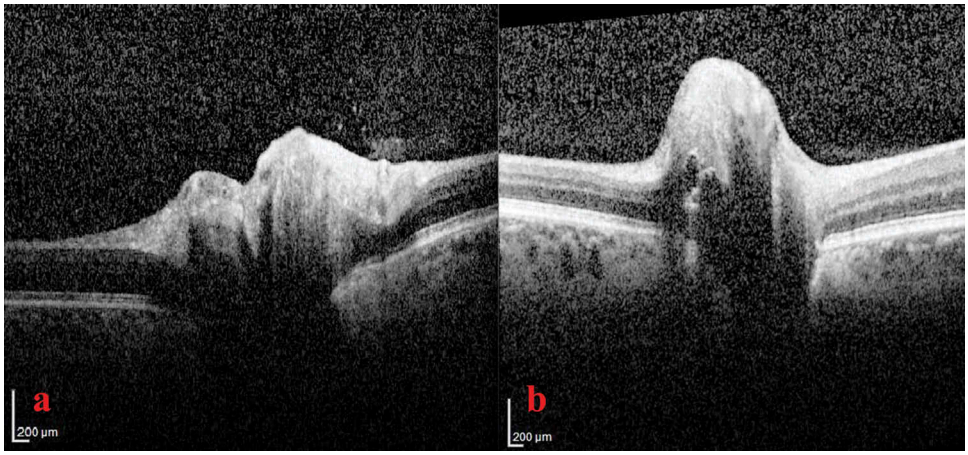


Figure 4. Optical coherence tomography scans of right (a) and left (b) eyes passing through both optic nerve heads demonstrating a "lumpy-bumpy" internal contour appearance.

ONHD and peripapillary MRNF after clinical examination and multimodal fundus imaging. The visual field defects were ascribed to the disc drusen. She had already been evaluated radiologically because of the misdiagnosis of papilloedema revealing normal results. A lumbar puncture had been performed with an opening pressure of 16 cmCSF and normal constituents 16 cm H_2O and normal constituents. A diagnosis of migraine was made. She was given prophylactic treatment, and periodic follow-up was recommended.

Discussion

MRNFs are rare congenital anomalies that may be located at the disc, peripapillary area, and/or elsewhere on the retina.[9–11] Their prevalence are 0.54% and bilaterality has been reported as 7.7% of cases.[10] Although MRNF is usually MRNFs are usually sporadic, although they may be associated with many systemic syndromes such as Down, Turner, Goltz-Gorlin, and GAPO (growth retardation, alopecia, pseudoanodontia, optic atrophy).[11] Current investigations have suggested that oligodendrocyte progenitor cells arising from neuroepithelial cells and ectopically located in the retina have been responsible for the pathogenesis. These fan-shaped lesions, which are consistent with the distribution of the RNFL settle into the outermost surface of the retina. They appear as grey-white well-demarcated opaque patches with feathery edges and may obscure the details of the underlying retina and its vascular structures.[9–11] Many studies have reported that MRNF are more common in patients with anisometropia, strabismus, and amblyopia.[10–12]

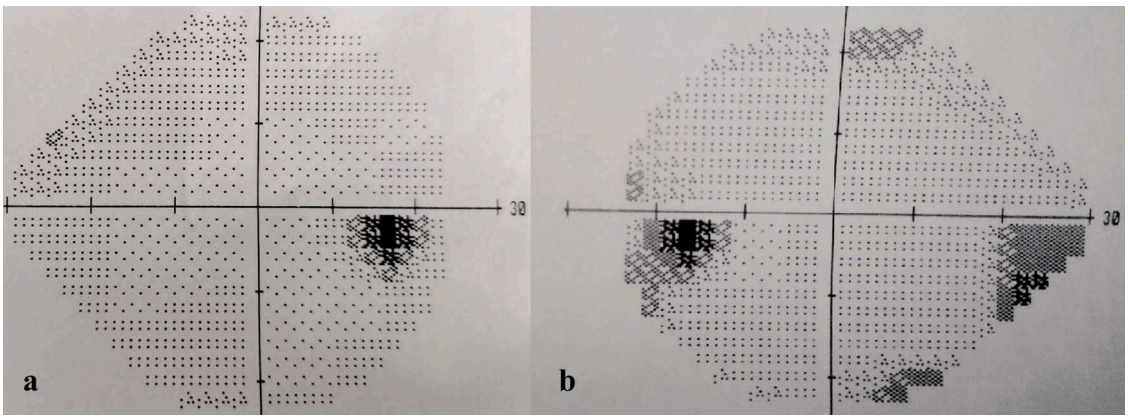


Figure 5. Visual field test showing normal appearance in the right eye (a) and nasal step defect and blind spot enlargement in the left eye (b).

In this case report, we present a rare coexistence of ONHD and MRNF. Although many patients with ONHD or MRNF have been reported in the literature up to date, the coexistent of these entities has been reported in only a 7-year-old male previously. The authors stated that the MRNF progressed during the adolescent period and could be associated with ONHD. However, although the authors demonstrated this association, they could not visualise the disc drusen in their report.[13] In this context, our case is the first to visualise this association with multimodal imaging.

In conclusion, ONHD and MRNF may rarely coexist. The frequency and clinical significance of this combination remain unknown. Since the MRNF may obscure the disc details, drusen can be easily overlooked and so possible complications associated with drusen, including visual field defects and/or vision loss may be attributed to other disorders, leading to misdiagnoses. Clinicians, therefore, should keep in mind that the drusen may accompany MRNF. For this reason, when the clinicians observe peripapillary MRNF, they should perform disc examination closely and carefully, and if necessary, they should utilise ancillary tests such as fundus autofluorescence and/or OCT to examine the buried or superficial drusen.

Declaration of interest statement

All authors contributed significantly to the creation of this manuscript and each fulfilled criterion as established by the ICMJE. No potential conflict of interest was reported by the authors. Patient consent was obtained to write this case report.

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