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SHORT COMMUNICATION

Case report

Duplication of the optic disc: True or pseudo? A coloboma or not a coloboma?

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PURPOSE. *To present an unusual case of optic disc pseudoduplication with colobomata.*
METHODS. *Clinical evaluation, fundus photography and literature review.*
Results. Optic disc duplication is a rare clinical entity.
CONCLUSIONS. *We report what we believe to be the first case of pseudoduplication of the optic disc with coexistent bilateral optic disc colobomata. (Eur J Ophthalmol 2004; 14: 163-5)*

KEY WORDS. *Double disc, Pseudoduplication, Coloboma*

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Case report

An 11 year old boy presented to our eye clinic following referral from an optician who had noted "peculiar optic discs" during a routine examination. The patient had no visual complaints. He was fit and well and was not taking any medications. He had been a premature baby, born at 34 weeks by casarean section following a breech presentation. His birth weight was 2200 gms. At birth, it was felt by the pediatricians that he had slightly dysmorphic facies with a degree of brachycephaly and a thin upper lip with a prominent philtrum. Therefore, chromosomal analysis was performed but was normal. TORCH titres were also negative. There was no history of maternal drug or alcohol abuse during pregnancy. There was no family history of ocular problems.

On examination, his unaided visual acuities were 6/5 right eye, 6/4 left eye. Pupils were equal and reactive. Colour vision was normal in both eyes. Anterior segment examination was unremarkable. Intraocular pressures were 14 mmHg bilaterally. Posterior segment examination of the right eye revealed what appeared to be two optic discs of approximately equal size (Fig. 1a). The superior "normal" optic disc was pink in colour with an inferior notch. The inferior disc-like lesion was markedly excavated with heavily pigmented margins. Tissue of uniform colour and consistency bridged the discs and appeared to merge with their edges. The inferotemporal branches of the retinal blood vessels emerging from the superior disc dipped into the inferior excavation before re-emerging to supply the inferior retina. Left eye fundoscopy also revealed an anomalous optic disc which was large in size with a marked inferonasal notch in keeping with a coloboma of the optic nerve head (Fig. 1b).

β-scan ultrasonography of the right orbit confirmed the presence of only one optic nerve. Visual field testing showed bilateral, absolute, superior altitudinal visual field defects corresponding to the inferior disc notches (Fig. 2).

A diagnosis of unilateral pseudoduplication of the right optic disc with coexistent bilateral optic disc colobomata was made. Baseline photographs were taken and a copy given to the patient's parents for reference during further follow-up by the optician.

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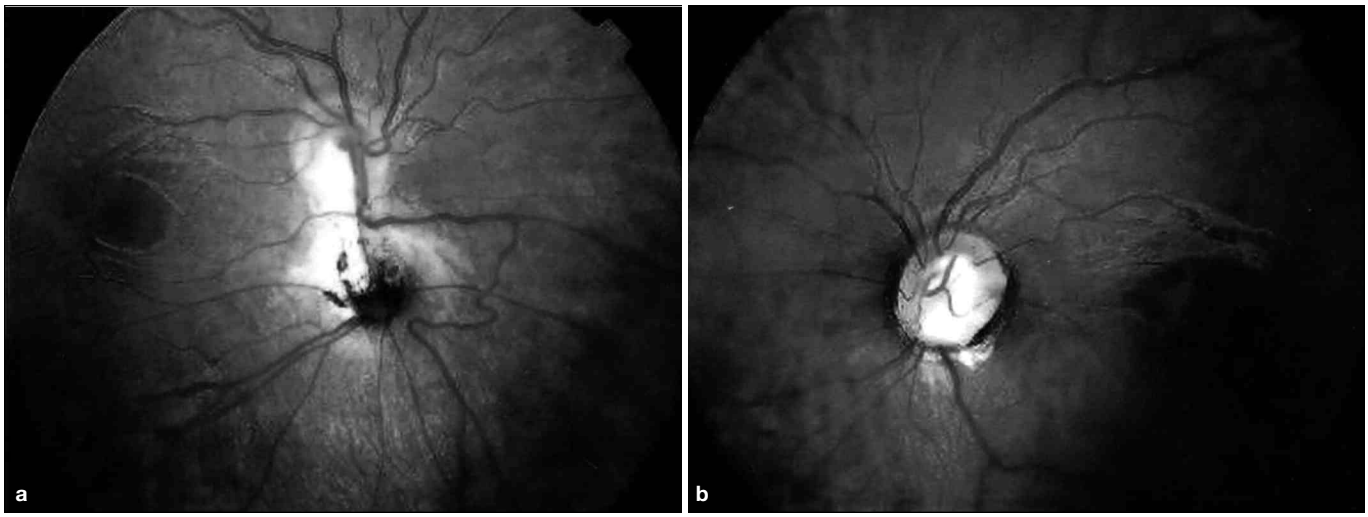

Fig. 1 - a) *Pseudoduplication of the right optic disc with coexistent disc coloboma.* **b)** *Left optic disc coloboma.*

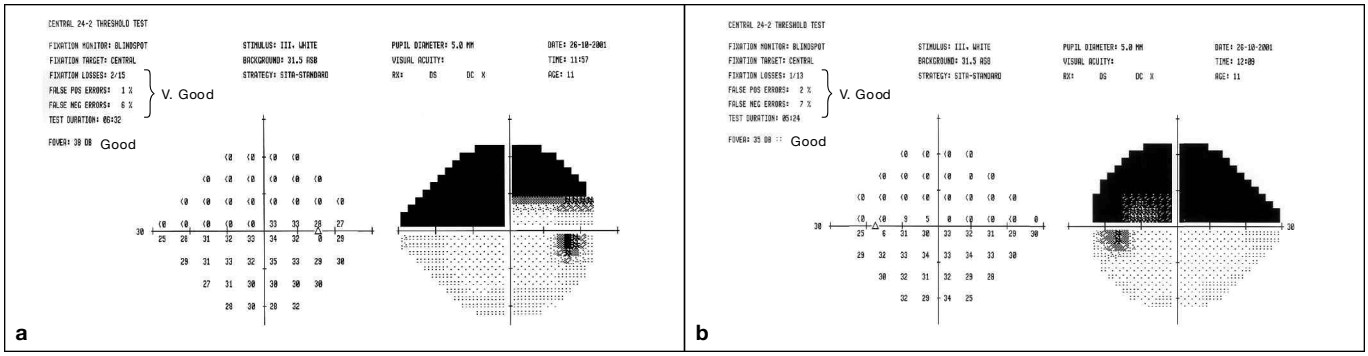

Fig. 2 - *Humphreys 24-2 threshold visual fields.* **a)** *Right superior absolute scotoma.* **b)** *Left superior absolute scotoma.*

DISCUSSION

True duplication of the optic disc occurs with separation of the optic nerve into two or more strands. Although rare in humans, it is common in some lower vertebrates, such as teleostei (fish with an ossified skeleton) (1). Pseudoduplication is also extremely rare and arises when a well circumscribed disc-like lesion with apparent cupping, associated vasculature and surrounding chorioretinal atrophy occurs adjacent to the normal optic disc. This entity is readily distinguishable from other retinal lesions such as staphyloma, inflammatory foci and choroidal rupture.

Two optic foramina in the same orbit on X-ray, dual blind spots in the same visual field or the presence of synchronous pulsations of each major disc artery all indicate the existence of double optic nerves. β-scan ultrasonography or CT/MRI scanning can confirm if true duplication of the optic nerves and discs exists. In our patient the presence of only one optic nerve on ultrasonography established the diagnosis of pseudoduplication of the optic disc.

Optic nerve head and chorioretinal colobomata occur due to closure defects in the proximal embryonic fissure at six weeks of gestation and may occur in conjunction. An optic disc coloboma typically presents as an inferior excavation of the optic nerve head. It may be unilateral or bilateral, as in this case, and may be asymmetrical. It is usually an isolated finding; systemic associations are infrequent e.g. trisomy 13 syndrome, thalidomide or LSD embryopathy.

To the best of our knowledge, this is the first re-

ported case of bilateral optic disc colobomata and unilateral pseudoduplication of the optic disc. There have been a limited number of documented cases of pseudodoubling of the optic disc in one eye with a normal fellow eye (2, 3, 4). Interestingly, Konrad and Harrison (5) described a case of bilateral optic disc pits and unilateral pseudodoubling of the optic disc. The authors surmised that pseudoduplication of the optic disc is a chorioretinal coloboma, which coincidentally is of similar size and shape to the normal optic disc, with sufficient optic disc involvement to create a vascular communication between the excavated second 'disc' and the central retinal vessels. They proposed that the coexistence of bilateral optic nerve pits was consistent with this theory. In our patient, the presence of marked bilateral inferior optic disc colobomata as well as unilateral pseudoduplication of the optic disc is strong evidence that this is indeed the case.

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