Pseudoduplication of the optic nerve head

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Background: Pseudo-doubling of the optic nerve head is a spectacular clinical entity, in which a lesion resembling an optic disk appears adjacent to the true optic disk.

Case Report: A case of unilateral pseudo-doubling of the optic disk with bilateral optic nerve pits is presented.

Conclusions: The lesion is congenital, and represents a chorioretinal coloboma with optic disk involvement. Pseudo-doubling can be differentiated from true doubling of the optic nerve by the imaging techniques of ultrasonography, computerized tomography, and magnetic resonance imaging.

Key Words: Congenital optic disk anomaly, coloboma, doubling of the optic nerve, optic nerve pit, pseudo-doubling of the optic disk

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We present a case of pseudo-doubling of the optic disk—a spectacular and rare clinical presentation. Of additional interest is that this case includes bilateral optic nerve pits.

In this report, we discuss the differentiation between true and pseudo-doubling of the optic disk and the issues involved in classification of and differentiation from the family of congenital optic nerve head anomalies and other clinical presentations resembling this condition.

Case Report

An 84-year-old woman came to us with gradually decreasing vision in both eyes and difficulty adjusting to dimly lit environments. She was aware that the left eye was unusual, as her previous eye care practitioner had photographed "the back of the eye" and presented her as a case study at a local ophthalmology meeting some 50 years earlier.[1] She was taking anti-hypertensive medication, but was in otherwise good health.

With her refraction of +3.50/–0.50 × 90 and +4.75/–0.75 × 100, she achieved visual acuities of 20/32 O.D. and 20/25, O.S. Addition of +2.50 D enabled reading of 20/40 at near. Pelli–Robson contrast sensitivity was mildly reduced to 1.40 and 1.10 (log CS) in the O.D. and O.S., respectively, though contrast sensitivity under glare conditions—reduced by 0.30 and 0.15 log CS, respectively—was within age-matched normal limits.[2] Color vision, assessed individually in both eyes with the Farnsworth D–15 panel test, was normal. Pupils were equal, round, and reactive to light and accommodation, with no afferent pupillary defect noted.

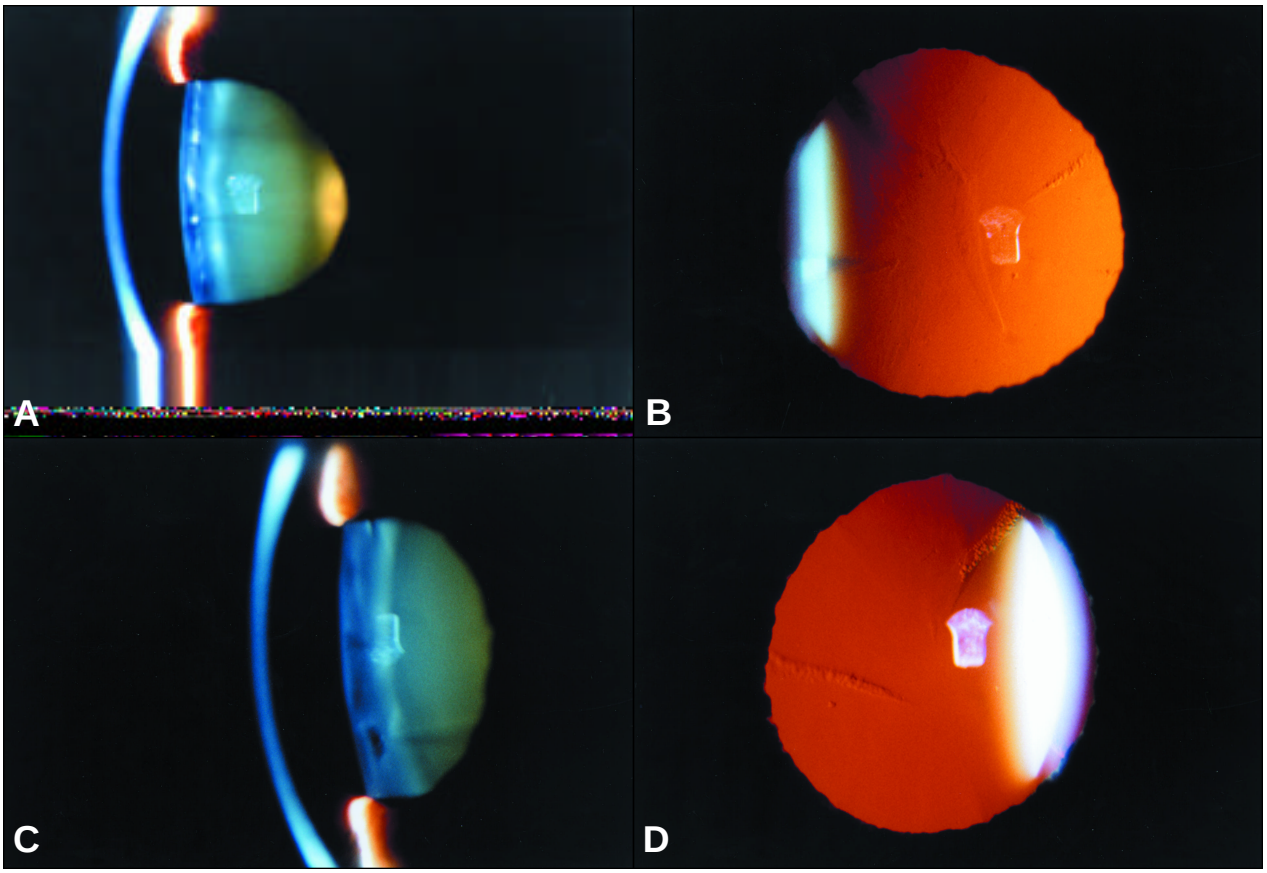

Figure 1 **A**, Slit-lamp biomicroscopy view of the right lens shows minimal cataractous change; **B**, retroillumination of the right lens; **C**, slit-lamp biomicroscopy view of the left lens shows minimal cataractous change; and **D**, retroillumination of the left lens.

Mild cataracts were present, as shown in Figure 1. This was graded with the LOCS III system, proposed by Chylack et al.,[3] in which nuclear opalescence and color are rated in a decimal scale from 0 to 7 in steps of 0.1, and cortical and posterior subcapsular cataracts are rated in a decimal scale from 0 to 6: nuclear opalescence (NO) 2.8; nuclear color (NC) 2.6; cortical (C) 3.2; posterior (P) 0.5, O.D. and NO 3.0; NC 2.9; C 3.0; P 0.5, O.S. Mild changes were also observed in the retinal pigment epithelium of both maculae. The right optic disk appeared normal, except for a temporally located optic nerve pit (*see* Figure 2). The left optic disk appeared to be duplicated inferiorly (*see* Figure 3). An optic nerve pit was also present in the true optic papilla. A cilioretinal artery was observed to exit from the optic nerve pit to subserve the inferior temporal retina. All the vessels subserving the superior vessel arcades exited the true optic disk. In addition, three vessels subserving the inferior vessel arcades were observed exiting from from the inferior lesion. Visual fields were normal in the right eye, but there was a dense inferior paracentral scotoma in the left, with a superior nasal step, superior arcuate defect, and enlarged blindspot (*see* Figure 4). Intraocular tensions were 15 mmHg OU. Since the optic nerve pits were located temporally instead of superiorly or inferiorly, they were probably congenital in origin rather than acquired, as from glaucoma. β–scan ultrasonography of the left eye and orbit was conducted, and this demonstrated the presence of only one optic nerve (*see* Figure 5), establishing the diagnosis of pseudo-doubling of the left optic disk.

Based on the symptoms of adaptational difficulties, improvement with refraction, normal glare loss, and contrast sensitivity, the patient's difficulties were attributed to refractive change, mild age-related macular degeneration, and early cataracts.

Discussion

Pseudo-doubling of the optic disk is an extremely rare condition. The authors are only aware of two previous reports in the literature.[4,5] In both cases,

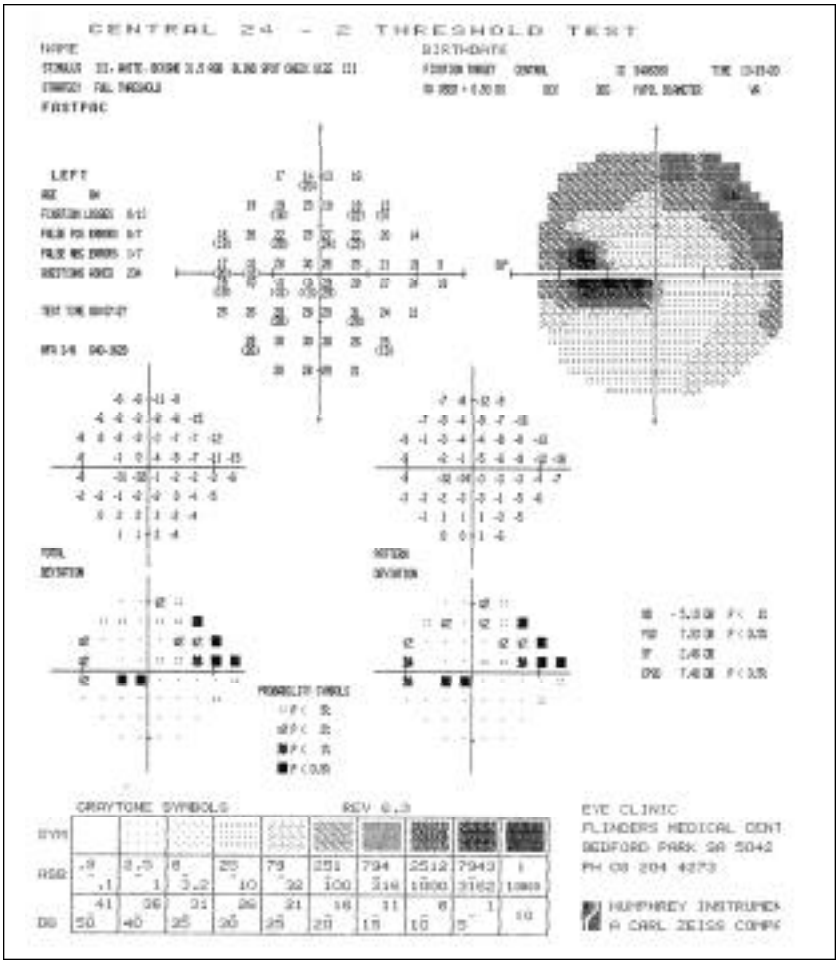

Figure 2 Humphrey 42–2 visual field of the left eye shows a paracentral arcuate scotoma.

the lesion resembling a second optic nerve head was located inferior to the disk within the region of the optic fissure, with apparent cupping and surrounding chorioretinal atrophy. The lesions reported in these three cases all resembled optic papillae in terms of size, shape, and vascularization. In two cases, the timing of vessel-filling during fluorescein angiography suggested the vessels were continuous with the central retinal vessels.[4,5]

The other eye may be normal,[4] although in this case the fellow eye was also affected by a congenital optic disk anomaly. Similarly, visual acuity may be normal or impaired.[4] Visual field defects have been reported and probably correspond to congenital nerve fiber bundle absence.[4] The paracentral visual field defect in this case is typical of that ween with an optic nerve pit,[6] and is likely to reflect this finding, rather than the presence of the disciform lesion, which corresponds to the superior nasal step with superior arcuate defect and enlarged blindspot.

Differential diagnosis

The circumscribed, disk-like appearance of this inferiorly located lesion—in combination with the presence of blood vessels and proximity to the true optic nerve head—makes it easily distinguishable from other retinal lesions, such as myopic (and other retinal), staphyloma, previous infection from toxoplasmosis, or those resulting from trauma. The primary differential diagnosis of pseudodoubling of the optic disk would be the presence of a true second optic disk, which can be detected by imaging the orbit with magnetic resonance imaging (MRI),[4] computed tomography (CT), or, more cost-effectively, with β-scan ultrasonography. In the literature, three cases of true optic disk doubling have been reported.[7-9] Brink and Larsen[5] suggested that a double-blind spot is also suggestive of true optic disk doubling. Fluorescein angiography may also be helpful as a true optic disk will show late hyperfluorescence, whereas a disciform lesion will not.[5]

Origin and classification of congenital anomalies of the optic nerve head

Although once considered to be part of a spectrum of disease, congenital anomalies of the optic disk such as optic disk coloboma, morning glory disk anomaly, peripapillary staphyloma, optic nerve pits, and optic nerve head dysplasia are now thought to be distinct clinical entities.[10-12]

Optic nerve head and chorioretinal colobomata represent a primary structural dysgenesis involving the proximal embryonic fissure, and may occur in conjunction.[13] The term *optic disk coloboma* should only be used for those cases with an inferiorly decentered white bowl-shaped excavation of the optic nerve head, without significant peripapillary pigmentary changes.[13]

Optic nerve pits, once considered to be part of a spectrum of disease with optic disk coloboma, are

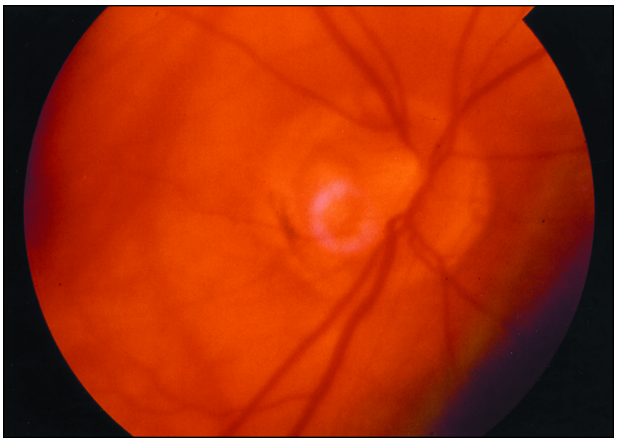

Figure 3 The right optic disk with a large optic nerve pit surrounded by nerve fibers in the temporal sector.

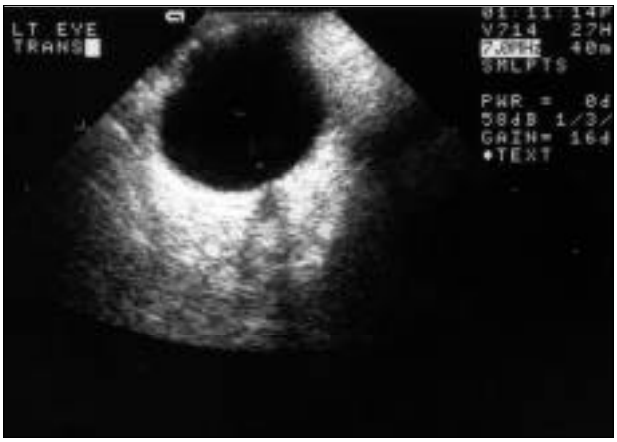

Figure 5 β-scan ultrasonograph shows the presence of only one optic nerve exiting from the left eye. There is no oblique entry, posterior globe protrusion, or evidence of any other abnormality.

not thought to be fundamentally distinct in their pathogenesis.[13] However, numerous cases of optic nerve pits occurring in the presence of true optic disk colobomata have been reported,[6,14,15] suggesting that the presence of an optic disk coloboma encourages optic nerve pit formation. Interestingly, colobomata manifest bilaterally as often as unilaterally (and may be asymmetrical),[15,16] whereas optic nerve pits are bilateral in only 15% of cases.[16] Cases of a coloboma in one eye and an optic nerve pit in the other have been reported.[15] Interestingly, optic nerve pits are associated with serous macular detachment, though the nature of the serous fluid—be it cerebrospinal, orbital, vascular, or vitreous in origin—is still debated.[13,17]

Morning glory optic disk anomaly, peripapillary staphyloma, megalopapilla, and congenital tilted

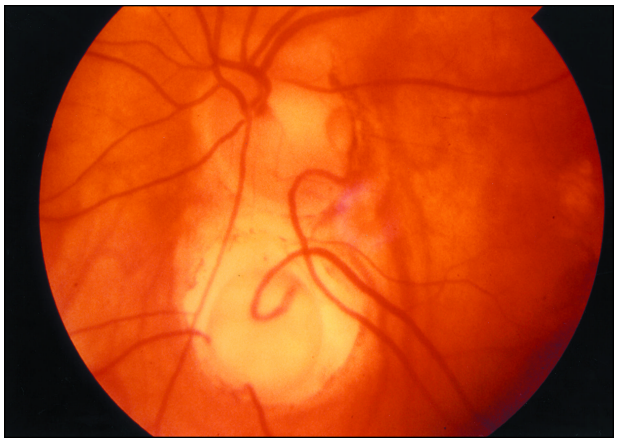

Figure 4 Apparent duplication of the left optic disk. The true optic disk is superior to the lesions resembling a second disk and contains an optic nerve pit abutting the temporal margin of the disk. The majority of vessels exit the true optic disk, but three major vessels subserving the inferior arcades exit the inferior lesion. A cilioretinal artery exits from the optic nerve pit in the true optic disk. Some retinal pigment epithelial changes with visible choroid vasculature are obscured in the macular area at the 3 o'clock position (on the right).

disk syndrome are all separate clinical entities.[10,13] Markedly deformed optic disks that cannot be otherwise classified are generally referred to as optic nerve head dysplasia. In addition, endocrinologic disorders such as growth hormone deficiency have been associated with other congenital optic nerve anomalies, such as optic nerve hypoplasia.[13] In this case, however, the patient experienced no systemic disease, nor are the authors aware of any direct association between pseudo-doubling of the optic disk and endocrinologic dysfunction.

The authors propose that pseudo-doubling of the optic disk is a chorioretinal coloboma, with optic disk involvement. This is supported by the observation that the disciform lesion is located inferior to the disk, in the region of the embryonic fissure. The chorioretinal coloboma, coincidentally, is of similar shape and size to the optic papilla, and there is sufficient optic nerve involvement to create vascular communication between the disciform lesion and the central retinal vessels. The existence of bilateral optic nerve pits in this case of pseudo-doubling of the optic nerve is consistent with the proposal that the disciform lesion is, indeed, a coloboma. As such, there may be an increase in the risk of rhegmatogenous retinal detachment associated with this type of (chorioretinal colobomatous) lesion.[17]

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