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SHORT COMMUNICATION

Optic disc drusen in tilted disc

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PURPOSE. To investigate if a congenital anomaly of the head of the optic nerve like such as tilted disc can be a risk factor for the development of optic disc drusen.
METHODS. The study was performed retrospectively on the files of 47 patients with optic disc drusen. The diagnosis was confirmed by fluorescein angiography and B-scan ultrasonography. The authors examined the fundus photographs and the fluorescein angiographies of these patients looking for the presence of tilted discs.
RESULTS. Two of the 47 patients with optic nerve drusen had tilted discs as well, about twice the expected rate. Both cases presented a parapapillary hemorrhage.
CONCLUSIONS. The concomitant presence of tilted disc and optic disc drusen can have a cause-effect relationship. The axonal crowding in a scleral canal of reduced size, as seen in tilted disc, can compress the nerve fibers against the stiff lamina cribrosa, producing a chronic optic neuropathy leading to drusen. (Eur J Ophthalmol 2005; 15: 647-51)

KEY WORDS. Optic disc drusen, Tilted disc, Risk factor, Small optic canal, Axonal transport

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INTRODUCTION

Anomalous elevations of optic disc are challenging for the ophthalmologist and other diagnoses besides papilloedema must be considered. Optic disc drusen and tilted disc are two causes of pseudopapilloedema that can mimic papilloedema from incremented intracranial pressure. These two diseases have distinct ophthalmoscopic as well as clinical features, so that a differential diagnosis is generally easy (1, 2).

Optic disc drusen arise from accumulation of hyaline material within the optic nerve head. It is believed that a chronic anomalous axonal transport in the ganglion fibers of the optic nerve causes damage to the axons and deposition of extracellular material in the optic disc that calcifies, giving origin to the ophthalmoscopically visible drusen (3, 4).

Tilted optic disc is an anomalous conformation of the distal part of the optic nerve: the nerve enters in the eye in an extremely angulated fashion, so that the supero-temporal portion of the optic disc appears elevated while the infero-nasal part is depressed (5, 6). The shape of the head of the optic nerve is lenticular and in some way hypoplastic; the fibers of the optic nerve have to bend to pass the stiff lamina cribrosa and the optic canal has a reduced, D-shaped aspect (7).

Due to the anomalous course of the optic nerve in its distal tract, we can suppose that the axonal transport in the ganglion are fibers can be impaired at least in some cases of tilted disc. We hypothesized that this anomalous conformation of the optic disc could be a risk factor for the development of optic disc drusen.

MATERIALS AND METHODS

We performed a retrospective analysis of the data of the patients in whom a diagnosis of optic disc drusen was made from January 1997 to September 2003 in our service of fluorescein angiography.

Out of 5,965 patients, 53 had a diagnosis of drusen of the optic disc. In order to not include doubtful cases in the study, stringent inclusion criteria were established.

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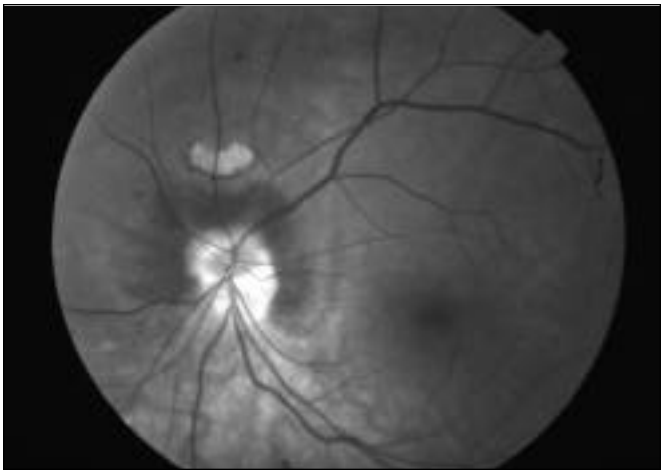

Fig. 1 - Case 1. The left eye of this patient shows tilted disc, inferior crescent, depigmentation of the inferior fundus, a subretinal hemorrhage encircling the superior two thirds of the optic disc, and a semilunar white lesion superior to the disc (resorbing hemorrhage).

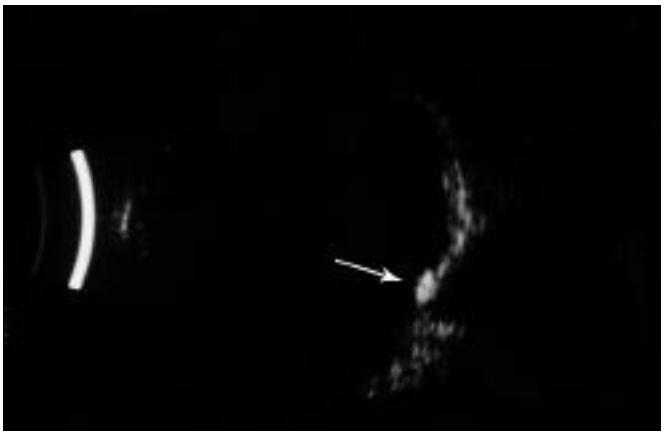

Fig. 2 - Case 1. B-scan ultrasonography reveals high reflectivity echoes on an elevated optic disc head, corresponding to calcified drusen (arrow).

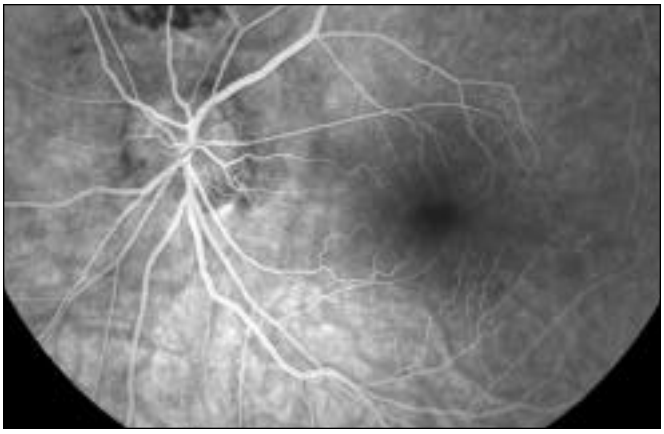

Fig. 3 - Case 1. Fluorescein angiography, performed 1 month later, shows enlarged and polilobated aspect of the optic disc bearing drusen.

The inclusion criteria were as follows: 1) ophthalmoscopic evidence of colloid bodies in the optic disc; 2) autofluorescence of the supposed drusen (based on preinjection photographs); 3) ultrasonographic finding of high reflectivity lesion in the optic disc typical of calcified drusen.

The presence of at least two of these three features was necessary for inclusion in the study. Based on these criteria, 6 of the 53 patients with a previously diagnosis of drusen of the optic disc were excluded.

A diagnosis of tilted disc was made when at least three of the following four ophthalmoscopic signs were present: 1) lenticular and oblique optic disc; 2) situs inversus of retinal vessels; 3) parapapillary conus; 4) depigmentation of the infero-nasal fundus. In addition, to make the diagnosis of tilted disc reliable, the presence of myopic astigmatism was necessary. Fundus photographs and fluorescein angiography were available for all patients. B-scan ultrasonography was performed in 40 of the 47 patients.

Two patients among the 47 with optic disc drusen had tilted disc as well. These patients are described in the following.

Case reports

Case 1

A 53-year-old woman complained of distorted vision of her left eye that began 1 month before the examination. Her ophthalmologist diagnosed a subretinal parapapillary hemorrhage and requested fluorescein angiography. Visual acuity was 20/20 (-5.50 cyl -1.50 20°) in the right eye and 20/25 (-8 cyl -1.00 155°) in the left eye. The fundus of the right eye showed a typical tilted disc, but no evidence of optic disc drusen. In the left eye the disc was elevated in its superior half. An inferior crescent was seen together with depigmentation of inferior fundus.

A subretinal peripapillary hemorrhage encircled the superior two thirds of the optic disc (Fig. 1). No sign of subretinal neovascular membrane was found. Superior to the hemorrhage an oblongated white area corresponded to resorbed blood. The macula was normal. B-scan ultrasonography showed high reflectivity lesion on the head of the optic disc, typical of calcified drusen (Fig. 2). One month later, when the hemorrhage was almost completely resorbed, the polycyclic contour of the left optic disc was apparent, especially in fluorescein angiography (Fig. 3).

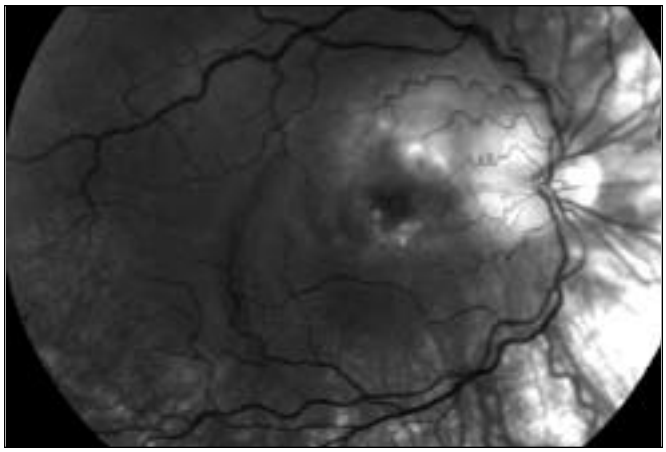

Fig. 4 - Case 2. The right optic disc has an horizontal axis and is elevated in its temporal half. The nasal fundus was depigmented. A grayish subretinal membrane was evident supero-temporally to the disc.

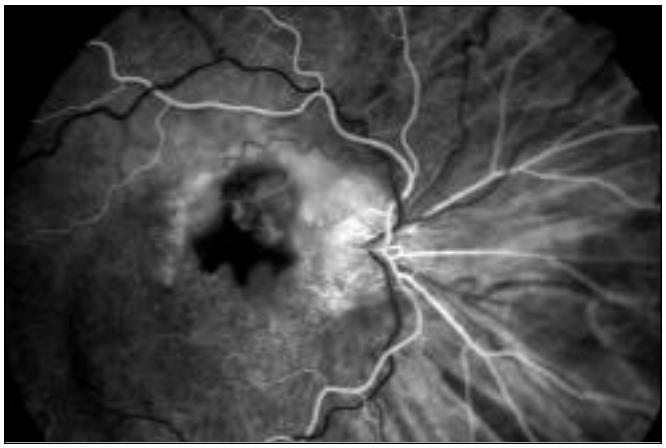

Fig. 5 - Case 2. Fluorescein angiography in arterial phase shows the contour of a parapapillary subretinal neovascular membrane as well as the presence of subretinal blood.

Case 2

A 19-year-old woman presented with abrupt reduction of visual acuity in the right eye. Corrected visual acuity was 20/400 (-8 cyl -2 80°) in the right eye and 20/30 (-7.50 cyl -1.50 70°) in the left eye.

The right optic disc presented anomalous elevation in the temporal portion; the disc showed irregular contour and oval shape (Fig. 4). There was a small temporal conus, situs inversus of the retinal vessels, and depigmentation of the fundus, especially in the infero-nasal quadrant.

Supero-temporal to the disc a slightly elevated grayish subretinal membrane was seen encircled by a subretinal hemorrhage. The supero-temporal portion of the disc showed autofluorescence. In fluorescein angiography the parapapillar subretinal membrane was easily seen as well as the hyperfluorescent, polycyclic optic disc (Fig. 5). B-scan ultrasonography confirmed the presence of optic disc drusen (Fig. 6). The left optic disc was tilted, but no evidence of drusen was found. The neovascular subretinal membrane was successfully treated with argon photocoagulation, but the visual acuity did not improve.

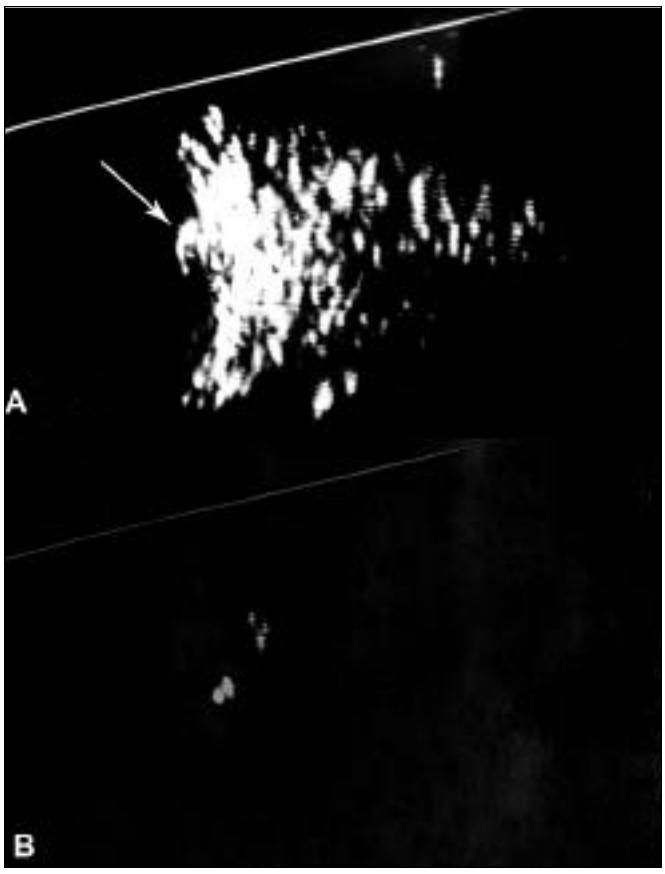

Fig. 6 - Case 2. B-scan ultrasonography discloses a high reflectivity lesion on the optic nerve head (A, arrow). The lesion is evident even at highly reduced system sensitivity when any other structure of the eyeball disappears on the display, confirming the presence of calcium in the drusen (B).

DISCUSSION

Tilted disc is a congenital anomaly characterized by an exceedingly inclined entrance of the optic nerve in the optic canal (5, 6). Consequently the optic disc appears lenticular in shape with the inferior part depressed and the

superior pole elevated. Other features of tilted disc are parapapillary conus, situs inversus of retinal vessels, depigmentation of the infero-nasal part of the fundus, and oblique myopic astigmatism. All these findings are probably derived from an abnormal closure of the embryonic fissure (8).

Optic disc drusen have been considered a congenital disease as well (9, 10).

However, the appearance of drusen in optic nerve heads that previously seemed unaffected or the surfacing of drusen once buried have given evidence that optic disc drusen often lack their heredofamilial character and are progressive.

Today is believed that drusen of the optic nerve head represent a degenerative and progressive optic neuropathy depending on a particular conformation of the optic canal (11). An abnormality in the axoplasmic transport can produce disintegration of axons and accumulation of axoplasmic material outside the nerve fibers producing the ophthalmoscopically visible drusen (3, 4).

The concomitant presence of tilted disc and optic disc drusen that we found in two patients can have a cause-effect relation. In the tilted disc there is a disproportion between the retinal opening that has normal size and the scleral opening that is reduced and has a D-shape; in addition, tilted disc has been considered a sectorial hypoplasia of the optic disc (7).

However, also in eyes with optic nerve drusen the scleral canal and the optic disc are smaller than in normal eyes (12-14) and the primary pathology of optic disc drusen is probably a dysplasia of the optic disc (15) so that their formation can be related to the reduced size of the optic nerve in its distal part (2).

Thus, the axonal crowding in a scleral canal of reduced size present in the tilted optic discs could produce a relative compression and the consequent chronic optic neuropathy that leads to the development of drusen.

A certain resemblance occurs with anterior ischemic optic neuropathy that has a risk factor in a small optic disc (or an optic disc with a small cup) (16, 17): in this case the small disc favours the occurrence of an acute neuropathy that is vascular in nature, while the small tilted disc can be a risk factor for the occurrence of a chronic optic neuropathy secondary to anomalous axonal flow.

We found tilted disc and optic disc drusen to be associated in a slightly greater number of cases than can be expected from the relative prevalences of the two diseases. Prevalence of optic disc drusen in general population is about 0.34% (18), but in cadaver eyes the finding of drusen is higher, being between 0.41 and 2% (19, 20), probably because disc drusen are often buried and not visible at ophthalmoscopy. Tilted disc is a rather frequent anomaly and it is found in 1.6 to 1.7% in population-based surveys (21, 22). Thus, if the two diseases are not correlated between them we would find about 1 tilted disc in every 50 subjects bearing optic disc drusen. We found 2 cases of tilted disc in 47 subjects with optic disc drusen: about twice the expected rate. However, our sample is small and no epidemiologic conclusion can be drawn.

The cases described were complicated by subretinal hemorrhages, developing near the superior pole of the disc. One of these two patients also showed subretinal neovascularization which is a complication of both optic disc drusen and tilted disc while in presence of drusen neovascularization develops in the perioptic area (2, 23); in tilted disc it is found in the posterior pole, on the border of the ectasia (24-26). Thus, the pathogenetic mechanism of subretinal neovascularization in these cases seems to be related to the disc drusen.

In conclusion, it is conceivable that crowding and bending of nerve fibers in a reduced optic canal as seen in tilted discs cause elevation of the superior part of the optic nerve head as well as impairment of the axoplasmic flow. In some cases, the consequence can be the accumulation of hyaline material in this portion of the optic disc and production of drusen.

However, more extensive studies are needed to confirm that tilted disc could be a risk factor for optic disc drusen.

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