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SHORT COMMUNICATION

Unusual coexistence of bilateral keratoconus and optic disc pit: A case report

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PURPOSE. *To report the unknown coexistence of bilateral optic disc pit and keratoconus.*
METHODS. *A 23-year-old man with bilateral keratoconus underwent complete ophthalmology screening, with an unexpected detection of undiagnosed optic disc pit in both eyes. Computerized corneal topography (CT), Orbscan, corneal pachometry, endothelial microscopy, and optical coherence tomography (OCT) examination were performed.*
RESULTS. *The corneal CT showed a keratoconus pattern in both eyes, evolved in the right eye with a minimum corneal pachometry of 336 µm in the right eye and 405 µm in the left eye. Mean endothelial cell density was 1937 cells/mm^2 in the right eye and 1912 cells/mm^2 in the left eye. The OCT scans showed the presence of the disc pit in both eyes with a normal macular thickness and profile in the right eye, and in the left eye an augmented retinal thickness in the nasal macular zone due to retinal œdema and schisis, with an initial detachment of the neuroepithelium in the parapapillary area starting from the optic pit.*
CONCLUSIONS. *This is the first clinical report of bilateral optic disc pit and keratoconus. Further investigations will be necessary to assess if there is a possible pathogenetic correlation between these two ocular pathologies or if this is an unusual coexistence of separate entities. (Eur J Ophthalmol 2008; 18: 134-7)*

KEY WORDS. *Computerized corneal topography, Keratoconus, Optical coherence tomography, Optic disc pit*

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INTRODUCTION

Optic disc pit is a rare congenital and circumscribed colobomatous anomaly of the optic disc (1). Its incidence is estimated to be about 1 case in 11,000 ophthalmic patients (1). Literature data suggest different ocular pathologies associated with keratoconus as corneal dystrophies (granular dystrophy, Terrien and pellucid marginal amyloidosis, posterior polymorphous dystrophy, Avellino dystrophy, Fuchs dystrophy, latex dystrophy), congenital pathologies (Leber congenital amaurosis, floppy eyelid syndrome, bilateral progressive essential iris atrophy, keratoglobus, retrolental fibroplasia, ankyloblepharon, aniridia, blue sclera disease, Axenfeld anomaly, pigmentosus retinitis), and allergic diseases (atopic reaction, spring like conjunctivitis). Moreover, several systemic diseases are described to be associated with keratoconus, such as 21-hydroxylase deficiency, Down syndrome, thalassemia, Albers-Schönberg disease, Crouzon syndrome, Ehlers-Danlos and Marfan syndrome, neurofibromatosis, osteogenesis imperfecta, elastic pseudoxanthoma, pigmentosus xeroderma, articular hypermobility, and mitral valve prolapse.
The following report describes the coexistence of two entities that could be related: keratoconus and optic disc pit.

Case report

A 23-year-old man with bilateral keratoconus underwent complete ophthalmic examination with digital slit lamp examination, computerized corneal topography (CSO, Eye Top 2005, Florence, Italy), Orbscan (Orbscan IIz, Bausch & Lomb, US), corneal pachometry (Pachmate, DGH Technolo-

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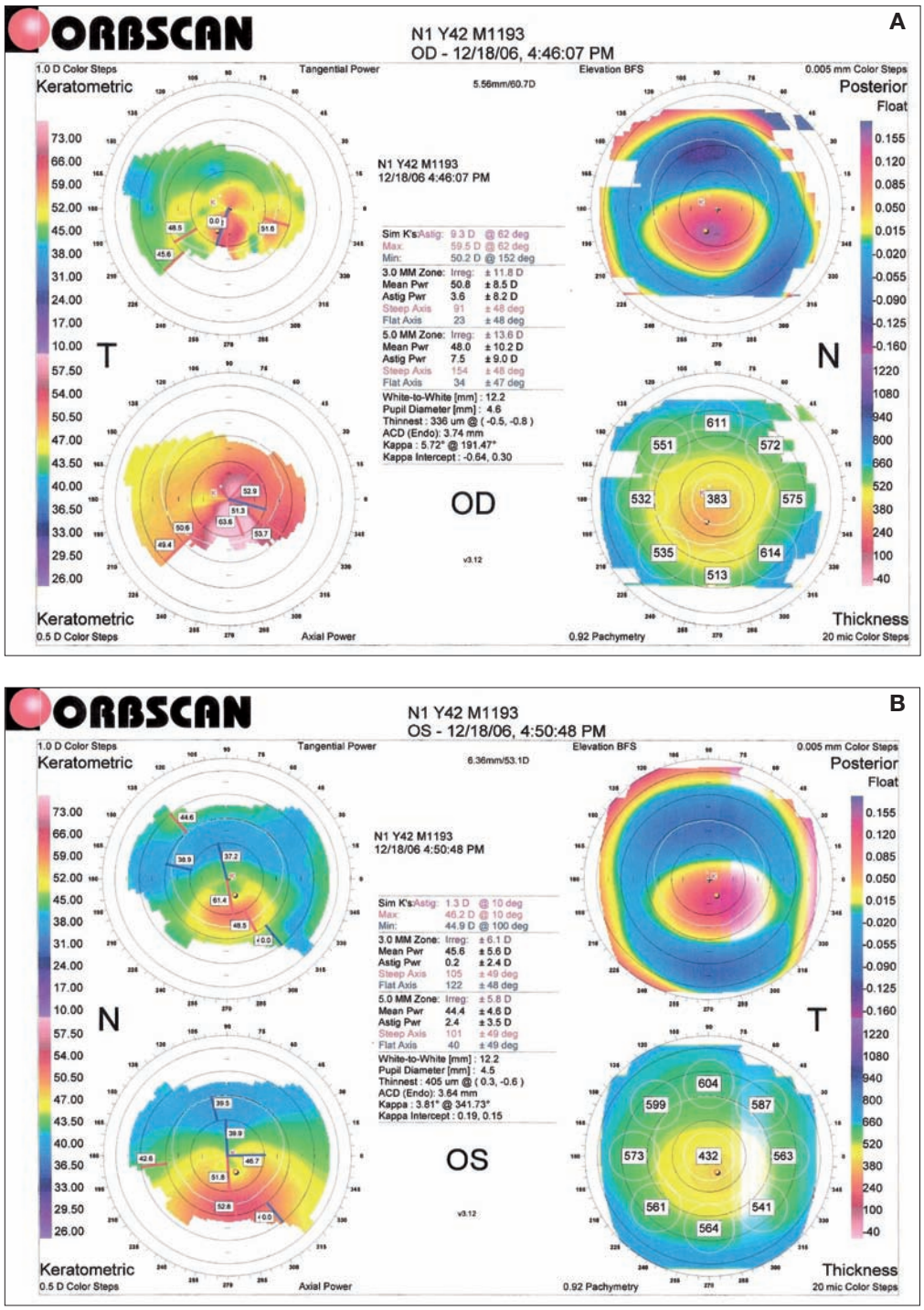

Fig. 1 - *Orbscan examination of right eye* **(A)** *and left eye* **(B)** *showing keratoconus topographic patterns, low corneal pachometry values, and typical posterior float in both eyes.*

gy Inc., Exton, PA, USA), endothelial microscopy (Konan Specular Microscope, Japan), uncorrected (UCVA) and best spectacle-corrected visual acuity (BSCVA) measurement, and fundus evaluation. Digital slit lamp examination showed bilateral corneal ectasia with light corneal opacity and Vogt striae in the right eye. The computerized corneal topography map and Orbscan evaluation showed a keratoconus pattern in both eyes, more advanced in the right eye (RE) (Fig. 1). BSCVA was 20/25 with –0.50 sph –2.50 cyl 90° in the RE and in the left eye (LE) 20/20 with –0.75 sph –2 cyl 85°.

Corneal pachometry showed a minimum thickness of 336 µm in the RE and 405 µm in the LE. Mean endothelial cell

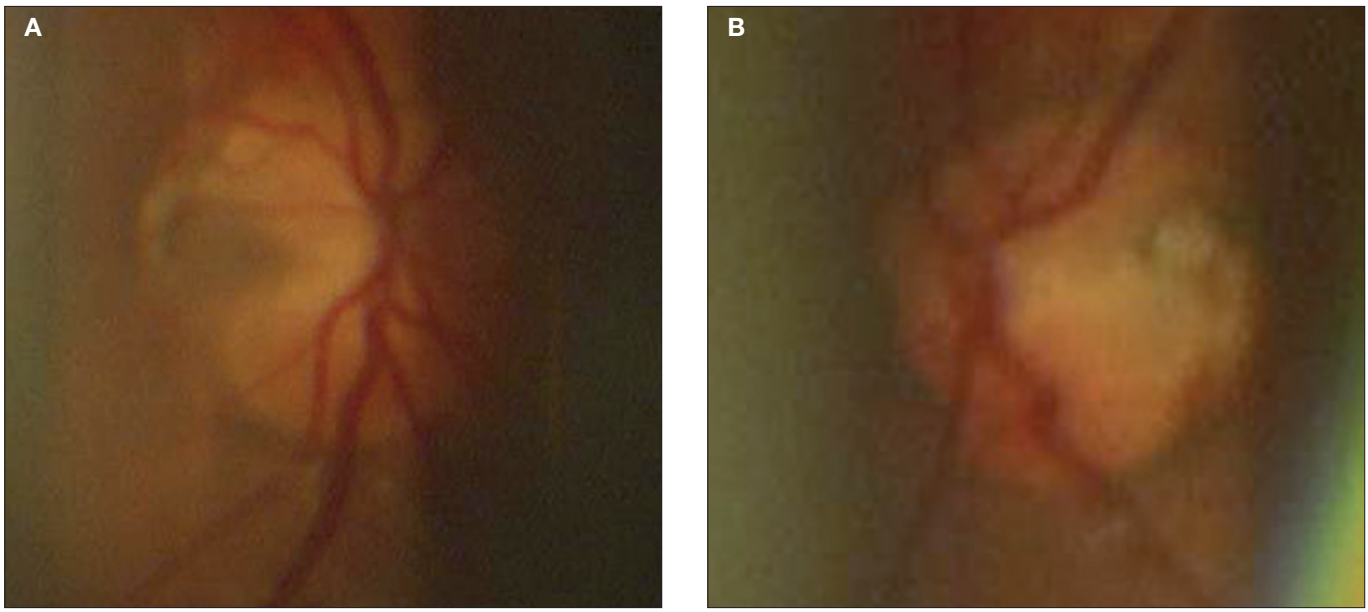

Fig. 2 - *Ophthalmoscopic digitalized image of disc of the right eye* **(A)** *and left eye* **(B)***: optic disc pit in both eyes is clear.*

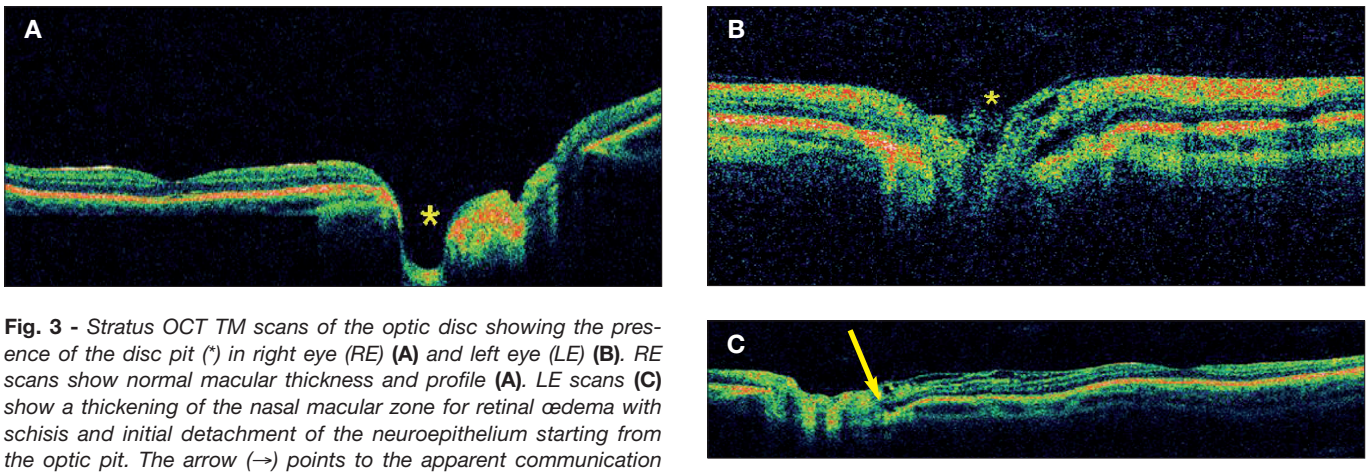

Fig. 3 - *Stratus OCT TM scans of the optic disc showing the presence of the disc pit (*) in right eye (RE)* **(A)** *and left eye (LE)* **(B)***. RE scans show normal macular thickness and profile* **(A)***. LE scans* **(C)** *show a thickening of the nasal macular zone for retinal œdema with schisis and initial detachment of the neuroepithelium starting from the optic pit. The arrow (→) points to the apparent communication between optic pit and subretinal space.*

density was 1937 cells/mm^2 in the RE and 1912 cells/mm^2 in the LE.

Ocular fundus examination showed an optic disc pit in both eyes with light retinal œdema (Fig. 2). Therefore, optical coherence tomography (OCT) (Stratus OCT^{TM}, Carl Zeiss Meditec, Dublin, CA, USA) retinal examination was performed. The OCT scans showed normal macular thickness and profile in the RE, and an augmented retinal thickness in nasal macular zone due to retinal oedema and retinoschisis with an initial detachment of the neuroepithelium in the para-papillary area starting from the optic pit in the LE (Fig. 3).

The patient underwent periodic eye examination showing no progression of the pathologies.

DISCUSSION

Optic disc pits can be localized in any part of the disc surface, but are more frequent in the temporal portion of the optic disc (1, 2). Optic pits have a round, oval, or fissure-like shape and the anomalous vessel course that often emerges

from the depth of the slit is a useful marker of their presence (1). Some authors have proposed a defective closure of the fetal fissure (3, 4) or an incomplete development of the primitive optic disc. Mann (5) suggested that optic pits are a consequence of an anomalous differentiation of the neuroectodermic folds of the primitive papilla at the stage of 15 mm when the retinal differentiation begins at the posterior pole (4). This hypothesis explains the more frequent temporal localization of optic pits (6) and justifies the presence of poor differentiated retinal tissue in the depth of the pits, observed in the histologic findings (7). Optic disc pits are often associated with other abnormalities of the ocular fundus, such as disc (8) or retinochoroidal (2) colobomas, full thickness macular hole (9), enlargement of optic disc (10), the presence of cilioretinal vessels (2), and a situs inversus (11). Temporal disc pits are clinically symptomatic because they often cause serous detachment of the macula, while non-temporal pits often remain unrecognized for the lack of clinical symptoms (1). The visual acuity of eyes with optic pits may be influenced by the presence of associated macular lesions. The vitreous presence and adhesions between vitreous and retina in some places of the vitreoretinal interface in the papillomacular area, as OCT scans showed (Fig. 3, b and c), are probably the cause of macular schisis (9). In our case, the visual acuity and the refractive changes must be related to the keratoconic cornea too. The pathogenesis of keratoconus remains unclear but different hypothesis have been proposed: mechanical, inflammatory, allergic, genetic, and dysmetabolic theories have been argued (12). Keratoconus may be associated with a variety of connective tissue diseases and inheritance. The contemporaneous presence of keratoconus and optic disc pit is difficult to explain. Keratoconus is a slowly progressive disease while optic disk pit is present in the first years of life. On the other hand, some observations must be considered. The mesenchymal structures contributing to the development of the corneal stroma are neural crest derivatives (13) and neuroectodermic folds of the primitive papilla are developed directly from neural crest (6). Is this embryogenic relationship the key of the pathogenetic mechanism of these two different ocular entities? To our knowledge, this is the first case report of coexistence of optic disc pit and keratoconus and probably this is an unusual clinical presentation of separate entities. Nevertheless, the bilateral presence of corneal and retinal diseases should be suspected for a pathogenetic link between these two ocular disorders.

Further investigations will be necessary to assess the potential pathogenetic correlation between these two entities.

The authors have no proprietary interest in any element of this case.

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