THE STORY OF A PAPILLA – A CASE REPORT

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This study was presented at the 22nd annual congress of the Czech Ophthalmological Society in Prague in 2014.

The authors of the study declare that no conflict of interest exists in the compilation and theme of this professional communication, and that it is not supported by any pharmaceuticals company.

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SUMMARY

Purpose: To present a case report with „unclear" and sudden decrease of left eye visual acuity and bilateral visual fields defects.
Methods: A case report.
Case presentation: A 66-year-old woman was referred to our Center of Neuroophthalmology and Orbitology by a neurologist for a history of sudden decrease of visual acuity of her left eye 3 years ago. From September 2009, she was examined at various and not only ophthalmology departments. One by one the optic nerve neuritis, traumatic, compressive or toxic neuropathy and also nutritive neuropathy because of vitamin B_{12} deficiency were excluded. The patient underwent also a genetic examination for Leber's hereditary optic nerve neuropathy, but this diagnosis was not confirmed. On magnetic resonance imaging, an atrophy of both optic nerves was described, with no further progression found during the follow-up examination after one year. In available patient's medical records we found out that on optical coherence tomography scans optic disc drusen of the both eyes are visible, but this wasn't described in the records. Also, an examination of Visual Evoked Potential was performed - this confirmed the diagnosis of optic disc drusen. However, our patient was further examined for visual lost of the left eye. At the time of presentation (January, 2014), her best-corrected visual acuity of the right eye was 0.5, and counting fingers at 50 cm distance with correct light projection in the left eye. Static perimetric examination demonstrated bilateral and concentric narrowing of visual fields. The eyes were parallel, with no limitation of their movements in any direction. The patient was without diplopia, the direct pupil reactions to the light were sluggish bilaterally, and anterior segments of both eyes were with no pathologies. Examination of the fundus revealed bilateral findings of pale optic disc with absent optic cup and indistinct "lumpy" margins. Waxy pearl-like irregularities of the papila of both eyes were visible even without pupil dilatation. Bilateral optic disc drusen were confirmed by ultrasonography, fundus autofluorescence and spectral-domain optical coherence tomography.
Conclusion: Optic disc drusen are often asymptomatic, frequently it is an accidental finding during the biomicroscopy of fundus due to ordinary eye examination. Rarely, optic disc drusen can cause blood circulation failure on the optic disc with typical defects of the visual field. That's why we shouldn't forget the optic disc drusen in the differential diagnosis considerations.
Key words: optic disc drusen, decrease of the visual acuity, visual field defect, autofluorescence, neuritis, neuropathy, perimetry, ultrasonography

Čes. a slov. Oftal., 71, 2015, No. 2, p. 116–121

INTRODUCTION

The issue of optic disc drusen is well known in professional circles. Nevertheless, on the basis of our experiences, its diagnosis in regular practice continues to cause problems and is frequently overlooked. We wish to draw attention to the issue in question by means of this case report.

OWN CASE REPORT

A 66 year old female patient was referred to our workplace by a neurologist for a consultation examination due to an anamnesis of sudden decrease of visual acuity in the left eye, persisting for longer than 3 years. Since September 2009 she had been examined at various, not only ophthalmological centres, where various possibilities of the cause of decrease of visual acuity in the left eye were progressively excluded: optic nerve neuritis, traumatic, compressive or toxic neuropathy and also nutritive neuropathy due to vitamin B12 deficiency. The patient also underwent a genetic examination for Leber's hereditary optic nerve neuropathy, but this diagnosis was not confirmed. On repeated magnetic resonance imaging (MRI), an atrophy of both optic nerves was described within their full scope, with no further progression found during the follow-up examination one year later. In the available documentation which the patient brought, it ensued that the patient had been examined at a private ophthalmology centre, where examination of the papillas had been conducted by optical coherence tomography (OCT). Optic disc drusens of both eyes were visible on the OCT scans, but that this was not described in the documentation and the patient had not been informed of the finding. In 2011 an examination of visual evoked potentials (VEP) was also performed, at which a diagnosis of optic disc drusen was determined, nevertheless the our patient was examined further for loss of vision in the left eye.

At the examination at our workplace, the patient had best corrected visual acuity of 0.5 with -2.0 D in the right eye, and counting fingers at a distance of 50 cm with clear light projection in the left eye. A perimetric examination demonstrated bilateral, concentric to tubular constriction of the visual fields, with a more pronounced finding in the left

eye (fig. 1). The eyeballs were parallel in the primary position, with no limitation of their movements in any direction. The patient was without diplopia. Direct photoreactions of the pupils were sluggish bilaterally. The anterior segments of both eyes were without pathologies, on the fundus we determined bilaterally diffuse pale optic discs, which were above the niveau and had indistinct "lumpy" magins. Biomicroscopically, even without artificial mydriasis, numerous superficial drusens were visible on the discs of the optic nerves, forming waxy, pearl-like irregularities (fig. 2). Bilateral optic disc drusens were confirmed by also by ultrasonography (USG), fundus autofluorescence and spectral-domain optical coherence tomography (SD-OCT) (fig. 4).

DISCUSSION

Optic disc drusens are small, acellular deposits of calcium, amino-acids and mucopolysaccharides, situated on or in the papilla of the optic nerve near the lamina cribrosa, which they may sometimes infiltrate. According to this, superficial or deep drusens are described (5, 6, 8, 9, 11, 20, 23). Their pathogenesis is not entirely clear (9, 16). A number of authors state an abnormal axonal metabolism, leading to the formation of calcareous crystals in mitochondria as the cause of the origin of drusens, as well as damage to the axons themselves with the extrusion of mitochondria into the extracellular area and subsequent accumulation of calcareous deposits (16, 20, 21, 22). Seitz et al. (19) state that drusens may occur also due to a reduction of the axoplasmic flow secondarily in congenital anomalies of the disc of the optic nerve. Optic disc drusens may occur in both adults and children (9). According to Friedman et al. (4), the prevalence of clinically distinguishable optic disc drusens within the population is only 0.3%, with an equal ratio of men and women. However, autopsy findings demonstrate a higher prevalence, up to 2.4%. The study by Auw-Haedrich et al. (1) states that this discrepancy is due to the high prevalence of undiagnosed optic disc drusens.

Optic disc drusen was first described by Müller in 1858 (9). It may by unilateral or bilateral, as a separate disease with an incidence of sporadic or autosomal dominant heredity with incomplete penetration, or may be associated with other ocular pathologies. These are most commonly retinitis pigmentosa (in approximately 10%), angioid streaks without pseudoxanthoma elasticum (in 25%) and glaucoma, and of general illnesses Alagille syndrome in children (hereditary intrahepatic cholestasis, in up to 95%), Usher syndrome type I (in 35%) and type II (in 8%), angioid streaks with pseudoxanthoma elasticum (in 21%) and syphilis (9, 16, 20).

In the clinical picture, the finding on the fundus dominates (9), as was the case with our patient. In adults the originally smooth elevation on the disc is altered over the course of years. Initially one or two bright yellowish-white spots are visible at the edge of the disc, which are later circularly added to by other drusens. The appearance of the disc is then characteristic, according to Otradovec (16) it has a typical "lumpy" edge and a waxy, pearl-like surface. Drusens appear as oval, slightly irregular growths on the edge of the disc, sometimes overlapping onto the surrounding retina. Superficial drusens are yellowish-white, are sharply defined and their size ranges from small dots to larger granules. Their diagnosis as a whole is problem-free, in unclear cases ultrasound examination of the eye may help, especially upon reducing the signal gain (gain <60 dB), in which drusens are pronouncedly hyperechogenic (9, 16). Deeply embedded drusens are of a dull appearance, and their definition is not sharp. They are frequently embedded so deep that they are not perceptible whatsoever ophthalmoscopically. Sometimes the edges of the disc, especially nasal, are furrowed, not sharply defined, and their surface is undulating (6, 9, 16, 23). Although the drusens do not merge, they have a tendency to cluster over the course of time, and their conglomerations may fill the entire disc of the optic nerve (9).

In the majority of cases this represents an asymptomatic affliction, frequently concerning a chance finding at a regular eye examination (6, 9, 23). The finding on the anterior segment is physiological and good central visual acuity is generally preserved (9). However, the compression on the veins exerted by the drusens may cause their more pronounced filling, as well as the occurrence of vascular links or haemorrhages on the disc. The presence of mainly deep drusens on the papilla may be the cause of circulatory malfunctions in the channel of the posterior ciliary arteries, with subsequent sudden and permanent blind spots in the visual field, of the character of a bundle of nerve fibres (ty-

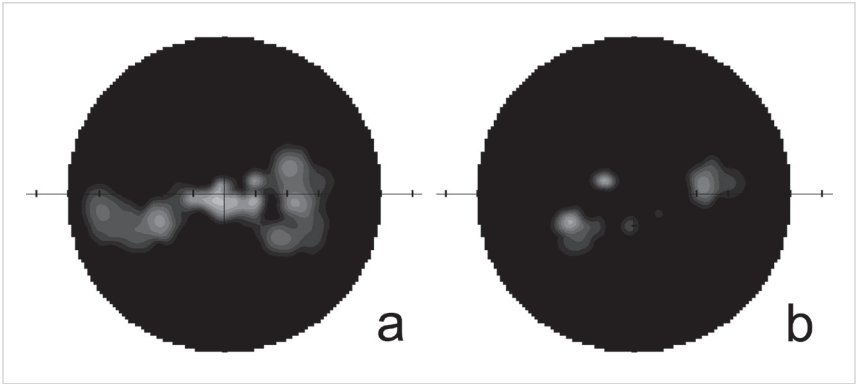

Fig. 1 Perimetric finding. Concentric constriction of the visual field of the right eye a), paracentral residues of visual field of left eye b)

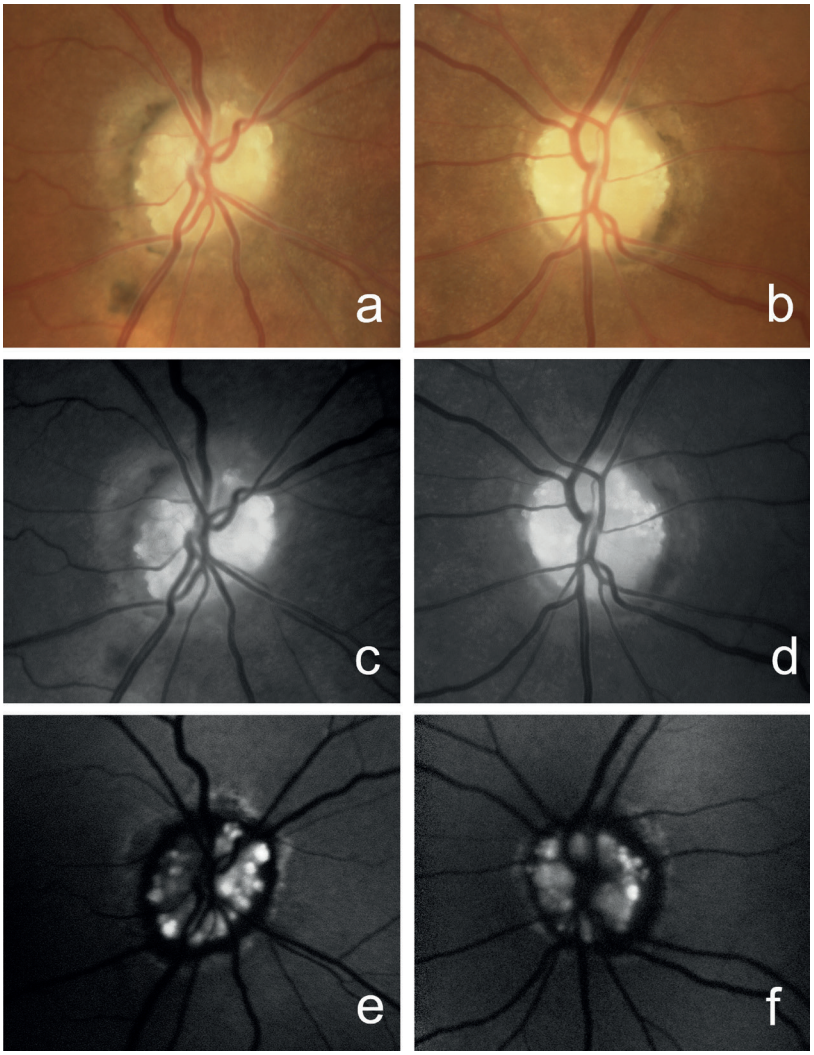

Fig. 2 Optic disc drusen. Colour images of papilla of optic nerve in right a) and left eye b), red-free images of papilla of right c) and left eye d), autofluorescence of optic disc drusens of right e) and left eye f)

pically arcuate and sectoral scotomas), followed by partial atrophy of the disc (2, 3, 9, 14, 15, 16, 23, 24). The scope of blind spots in the visual field may range from an enlarged Marriott's point to extensive defects of the visual field, as was the case in our patient, who we otherwise treat for glaucoma optic neuropathy (17, 20). In the case of a patient with an anamnesis of sudden, painless decrease of visual acuity and with a finding on the perimeter, after the exclusion of other acute afflictions such as retrobulbar or intraocular neuritis, anterior ischemic optic neuropathy or retinal vein occlusion, it is appropriate to consider optic disc drusens. Silverman et al. (20) in their study state that in addition to capillaries, drusens may also compromise the axons of the surrounding ganglion cells of the retina, and thereby impair the axoplasmic flow, with their subsequent cell death. In exceptional cases optic disc drusen may be accompanied also by momentary obnubilations, which are otherwise typical of congestive papilla (9, 16, 23).

In addition to biomicroscopic examination of the ocular fundus and the aforementioned USG examination, in the diagnosis of optic disc papilla it is possible to use the method of autofluorescence of the fundus, which is useful mainly in differentiating superficial drusens and edema of the papilla. In addition we can evaluate the retinal nerve fibre layer (RNFL) and document cross-sections of the papilla performed using SD-OCT or newly enhanced depth imaging OCT and swept source OCT. Using these methods it is possible to display deep layers of the retina, choroid and sclera, and to supplement the information obtained by USG or examination by autofluorescence of the fundus (10, 12, 13, 20). In the case of uncertainties it is possible to supplement fluorescence angiography, which is useful mainly in differentiating deep drusens and edema of the papilla (18, 20, 25). In unclear cases it is also possible to use examination by computer tomography or MRI, which, as stated in a study by Žiak et al. (25), at the same time excludes other more serious causes of calcification of the disc, such as retinoblastoma, optic glioma and others. Žiak et al. (25) also state that the results of VEP examination correspond to the seriousness of damage to the peripapillary nerve fibres, and demonstrate

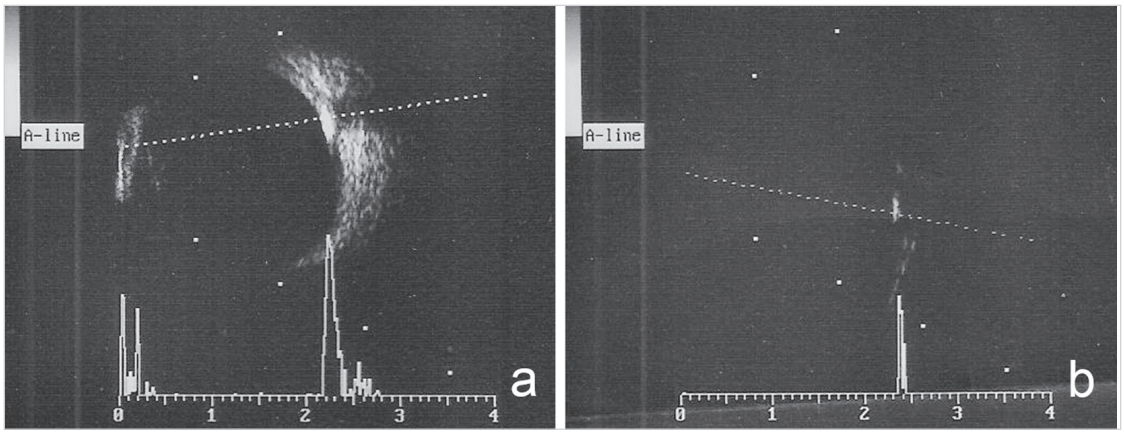

Fig. 3 Ultrasonography of right eye. Display of optic disc drusen upon normal signal gain (gain 68 dB) a), hyperechogenicity of optic disc drusen upon reduced signal gain (gain 40 dB) b)

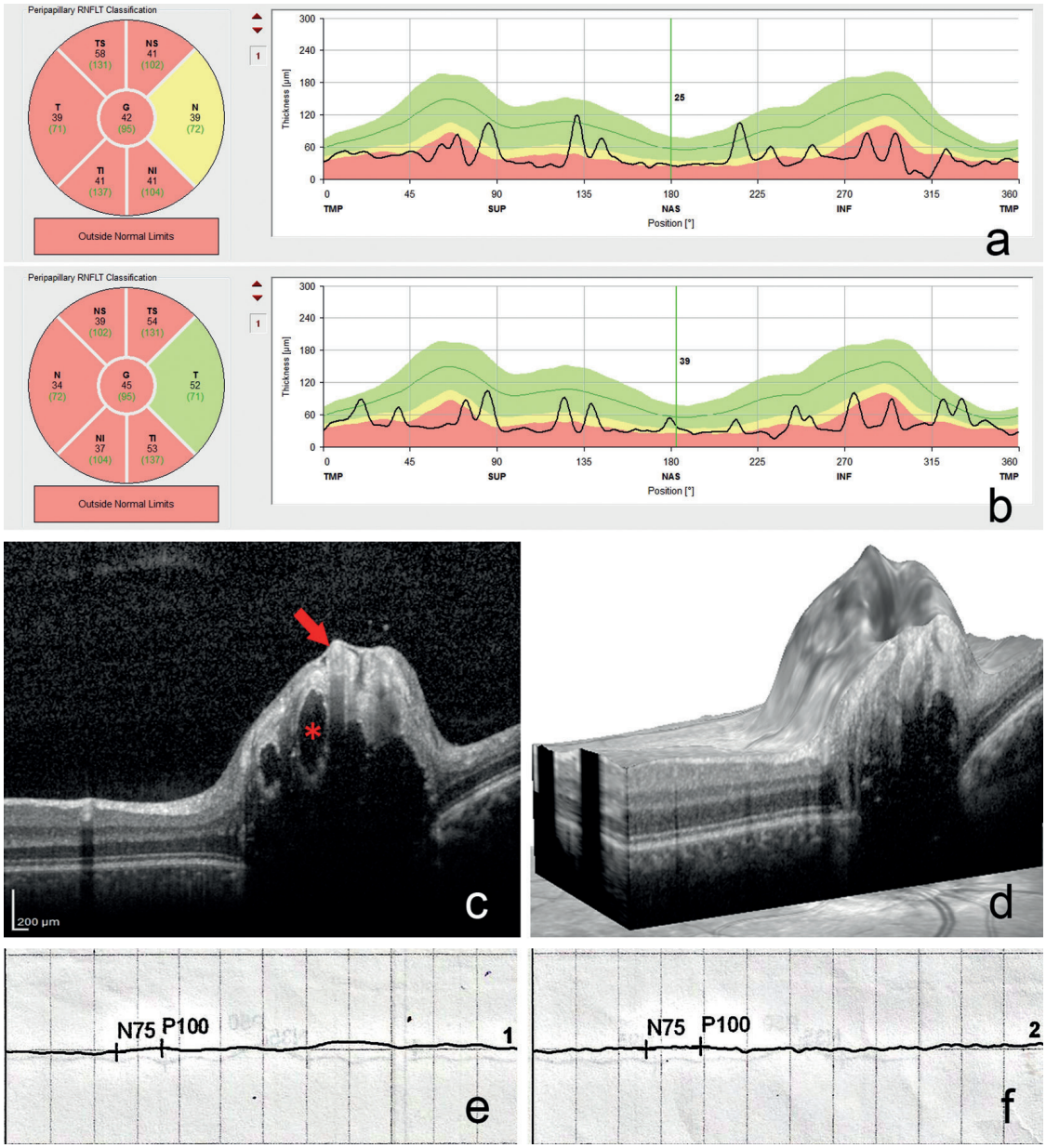

Fig. 4 Decrease of thickness of retinal nerve fibre layer on optical coherence tomography of right a) and left eye b). Cross section of papilla of optic nerve of right eye (arrow points to drusen, star in lumen of capillary) c), papilla of right eye in three-dimensional display d), flattened curve of visually evoked potentials of right e) and left eye f)

a wide range of abnormalities, as a result of which they are not reliable for the diagnosis of optic disc drusen.

The sovereign diagnostic method in regular clinical practice, especially in the case of deep drusens, remains ultrasonography, which is gradually being supplemented by constantly developing optical coherence tomography (7, 9, 12, 13, 18, 20).

In the differential diagnostics of optic disc drusen we must consider especially incipient or chronic congestion (6, 9).

To date no treatment or prevention of drusens is known. The most serious complications of optic disc drusens include anterior ischemic optic neuropathy, retinal vein occlusion and venous stasis retinopathy complicated by cystoid macular edema (9).

CONCLUSION

Optic disc drusen is frequently asymptomatic, in rarer cases however it may cause circulatory malfunction on the disc of the optic nerve, with distinctive and often extensive blind spots in the visual field. Following the exclusion of acute afflictions on the papilla, in a patient with an anamnesis of sudden, painless decrease of visual acuity and with a pathological finding on the perimeter, we should consider optic disc drusens, which in their position on the surface or in the papilla may be the cause of compression of capillaries, with subsequent circulatory malfunction. We can thus save the patient further lengthy and unnecessary examination.

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