CASE REPORT

Optic Disc Drusen – Current Diagnostic Possibilities

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SUMMARY

Authors present the findings of two patients with optic nerve drusen, and an overview of current examination techniques in the diagnostics of optic nerve drusen including ultrasound examination, fundus photography, florescein angiography, computerized perimetry, auto-florescence fundus examination, examination of the nerve fibre layer using optical coherence tomography (OCT) or nerve fibres layer analyzer (GDx). In the first case, the patient was recommended to be supervised without any therapy. The second patient with regard to perimeter finding, loss of nerve fibre layer and increase of the intraocular pressure was prescribed local anti-glaucoma therapy.

Key words: acute posterior multifocal placoid pigment epitheliopathy (APMPPE), white dot syndromes, macular edema

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INTRODUCTION

The term optic disc drusen refers to the presence of hyaline deposits in the head of the optic nerve, in front of the lamina cribrosa (1). Pathogenically these are the consequence of a congenital or vascular disorder, forming by means of an accumulation of proteins and mucopolysaccharides in the deep parts of the disc in the case of a disorder of the axon metabolism (2, 3). Drusens also contain amino-acids, RNA, DNA and iron (4). They progressively calcify, increase in size and begin to protrude (3). Drusens represent one of the main causes of pseudoedema of the papilla of the optic nerve, and their presence must therefore be detected (10). Optic nerve drusens are generally linked to multiple vascular complications such as non-arteritic anterior ischemic optic neuropathy (NAION), central retinal artery occlusion (CRAO), central retinal vein occlusion (CRVO) (4), mainly in patients with risk factors such as arterial hypertension, use of contraceptives, migraine, high altitude and atrial septal defect (4). In young people a neovascular membrane may form adjacent to the disc of the optic nerve. Optic nerve drusens are generally associated with retinitis pigmentosa, pseudoxanthoma elasticum and angioid streaks (4). B-scan echography, fluorescence angiography, optical coherence tomography (OCT), autofluorescence and CT examination are used for the detection of drusens. A perimetric examination is essential for the assessment and monitoring of defects in the visual field, as well as OCT of the retinal nerve fibre layer (RNFL) and polarimetric analysis of the RNFL using a GDx instrument to monitor the thickness of the layer of nerve fibres in the peripapillary area.

CASE REPORT

Patient no. 1
In May 2012 a 59 year old man was examined in our clinic. The patient has been observed in the local ophthalmological outpatient department, where he was sent by a diabetologist for the purpose of evaluating a finding on the fundus in connection with diabetes and arterial hypertension. Upon examination, optic nerve drusens were diagnosed as a secondary finding, and the patient was sent to our workplace for an examination. From an ocular perspective the patient is asymptomatic. The patient has bilateral natural vision of 5/5, intraocular pressure (IOP) in the right eye 12 mmHg and in the left eye 17 mmHg. In the local finding the anterior segment is calm, the lenses slightly opaque. On the fundus the optic disc (or the optic nerve head; ONH) above the niveau, with visible drusens mainly in the upper section, capillaries with hypertonic-sclerotic changes, tortuosity of nasal venous branches, the other finding corresponds to the patient's age. We documented the finding on the fundus by mydriatic fundus camera (Fig. 1 and 2). In the B-scan echographic image, hyperechogenic deposits are visible on the surface of ONH – drusens (Fig. 3 and 4). In the visual field examined by computer perimetry (Oculus Centerfield) there is narrowing in the right eye in the lower, nasal and upper sections by 10-35 degrees, extended blind spot, in the left eye absolute scotomas in the peripheral area of the visual field. We examined the patient with the help of a blue-green light by means of a scanning system (Spectralis, Heidelberg Engineering, Germany), with the use of autofluorescence, in which the drusens appear as hyperfluorescent formations (Fig. 5). A bilateral diminution was determined in the RNFL in the upper and lower sectors by means of laser polarimetric analysis of the thickness of the nerve fibre layer (GDx VCC, Zeiss – meditec, USA), in which the curve on the TSNIT graph is pronouncedly flattened throughout its entire extent (Fig. 6). OCT RNFL examination using OCT Spectralis determined a diminution of the RNFL in the upper, lower and nasal sectors, with a more pronounced finding in left eye (Fig. 7 and 8). Examination by the Heidelberg Retinal Tomograph (HRT III, Heidelberg Engineering, Germany) was negative in all sectors according to the Moorfields Regression Analysis (MRA), the cup/disc (C/D) ratio equal

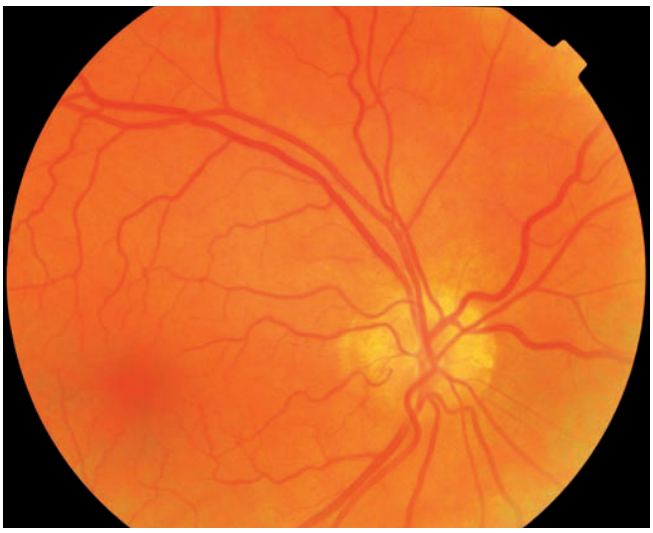

Fig. 1 Fundus photograph of the right eye (patient no. 1) Visible drusens on surface of the optic nerve head mainly in upper section and nasally, tortuosity of nasal venous branches

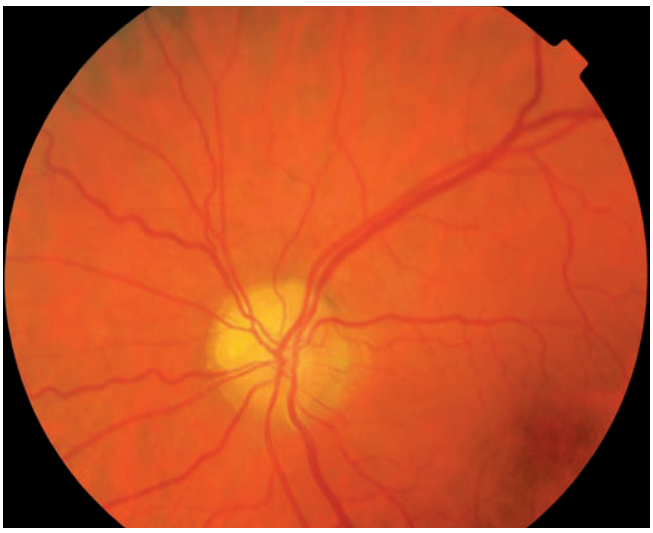

Fig. 2 Fundus photograph of the left eye (patient no. 1) Visible surface deposited drusens mainly in the upper nasal section, tortuosity of nasal venous branches.

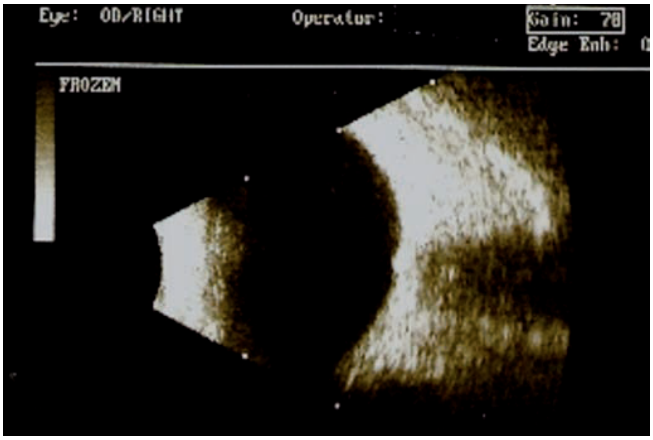

Fig. 3 B-scan ultrasound examination of the right eye (patient no. 1). Visible hyperechogenic formations – drusens on level of ONH.

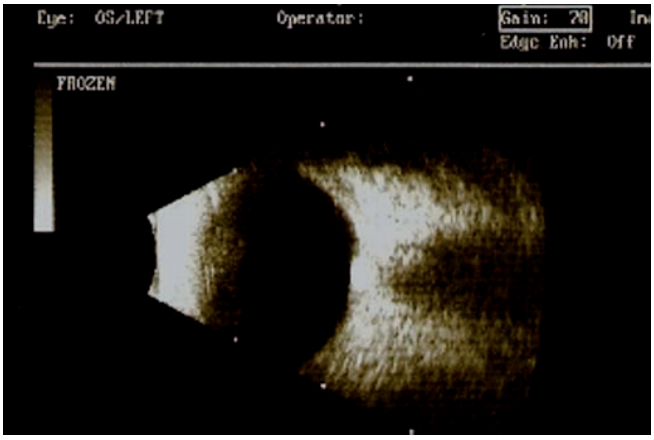

Fig. 4 B-scan ultrasound examination of the left eye (patient no. 1). Equivalent hyperechogenic formations – drusens at the level of ONH.

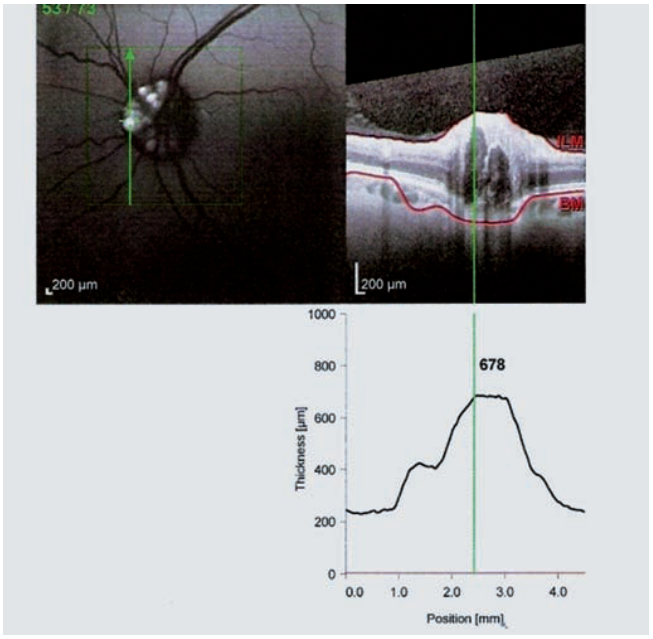

Fig. 5 Spectralis – fundus autofluorescence of the left eye (patient no. 1). Drusens are displayed as hyperfluorescent formations. In the right part of the image, a cross-section of anterior segment of optic nerve on OCT.

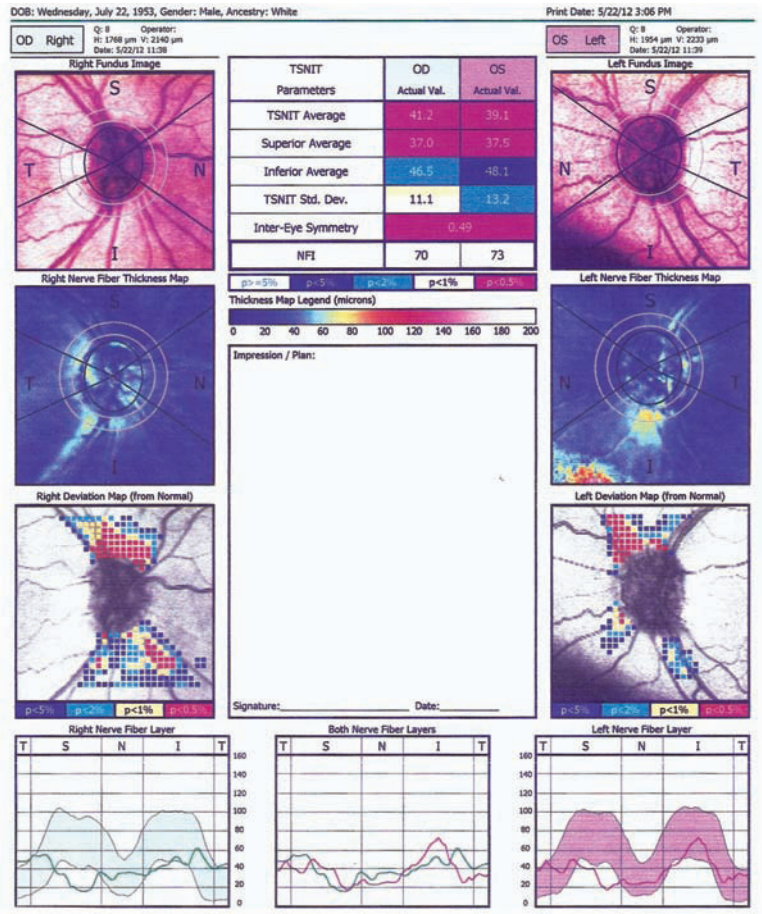

Fig. 6 GDx VCC examination (patient no. 1).
In the map of deviation, diminution of RNFL in upper and lower quadrants, flattened curves on TSNIT graph, NFI of right eye 70, left eye 73.

to zero, in both horizontal and vertical incision there are protruding drusens (Fig. 9). With regard to the presence of optic disc drusens together with the presence of risk factors with regard to potential vascular complications, we instructed the patient regarding the necessity of thorough adherence to the stipulated anti-hypertensive, hypolipidemic treatment and diabetic diet. We recommend regular checks, incorporating examination of vision, IOP, examination of fundus, perimetric examination and evaluation of RNFL by means of OCT RNFL and GDx.

Patient no. 2

In February 2012 a 26 year old female patient was treated in our clinic, who had been referred from the local ophthalmological outpatient department due to a positive perimetric finding and repeatedly measured high values of intraocular pressure (26 mmHg), with a concurrent finding of optic disc drusen. Natural vision was bilaterally 5/4. IOP bilaterally 24 mmHg. The finding in the anterior segment was physiological. Computer perimetry (Oculus Centerfield) determined bilateral narrowing of the visual field, on the right upper nasally by 10-30 degrees, lower nasally by 10-30 degrees and left eye narrowing of the visual field by 20-40 degrees. Bilaterally extended blind spot. On the fundus ONH with visible drusens, other finding

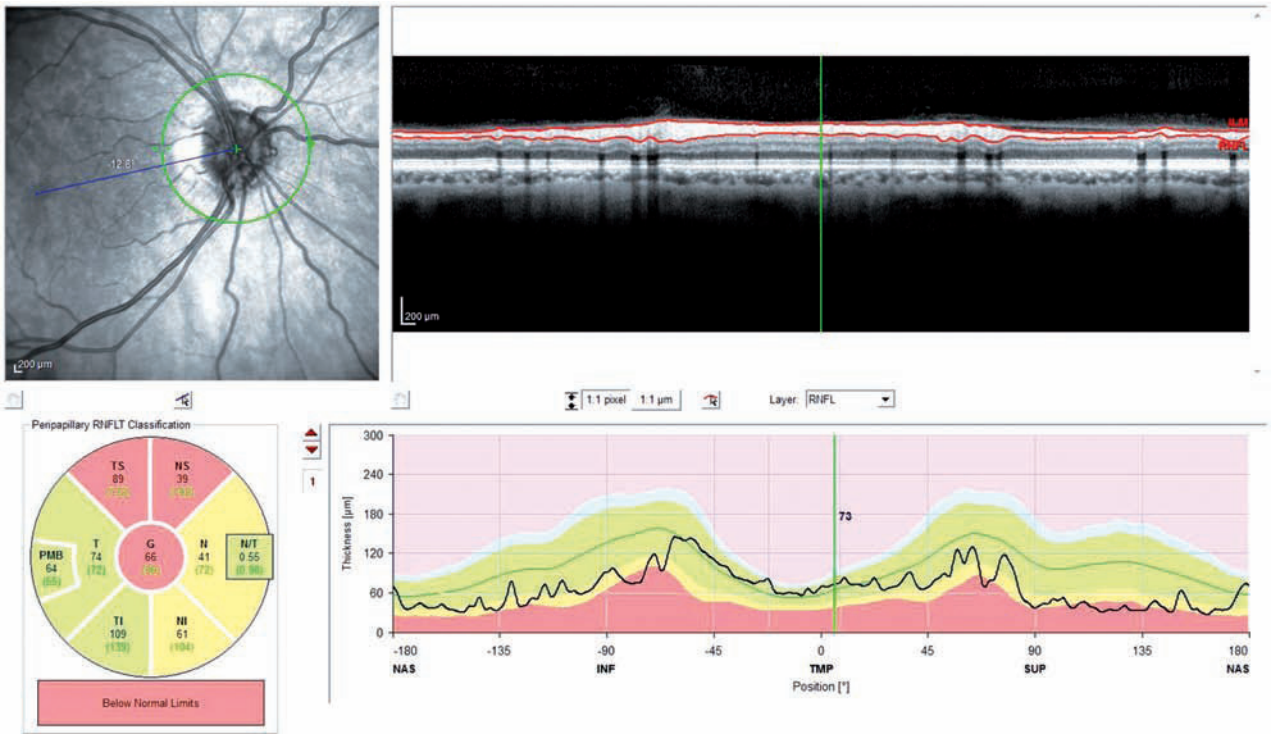

Fig. 7 OCT RNFL (OCT Spectralis), right eye (patient no. 1). Positive finding in upper section, borderline finding in nasal section.

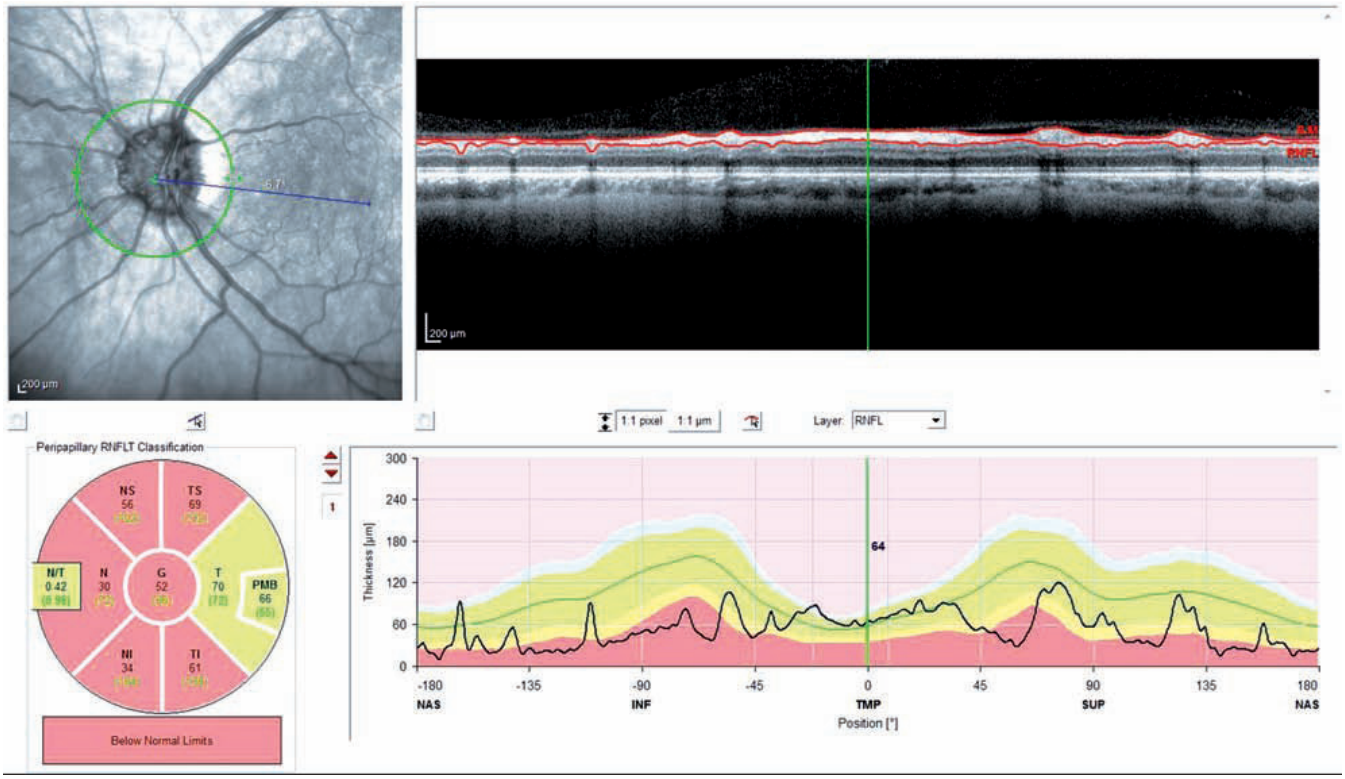

Fig. 8 OCT RNFL (OCT Spectralis), left eye (patient no. 1). Positive finding in upper, nasal and temporal sections.

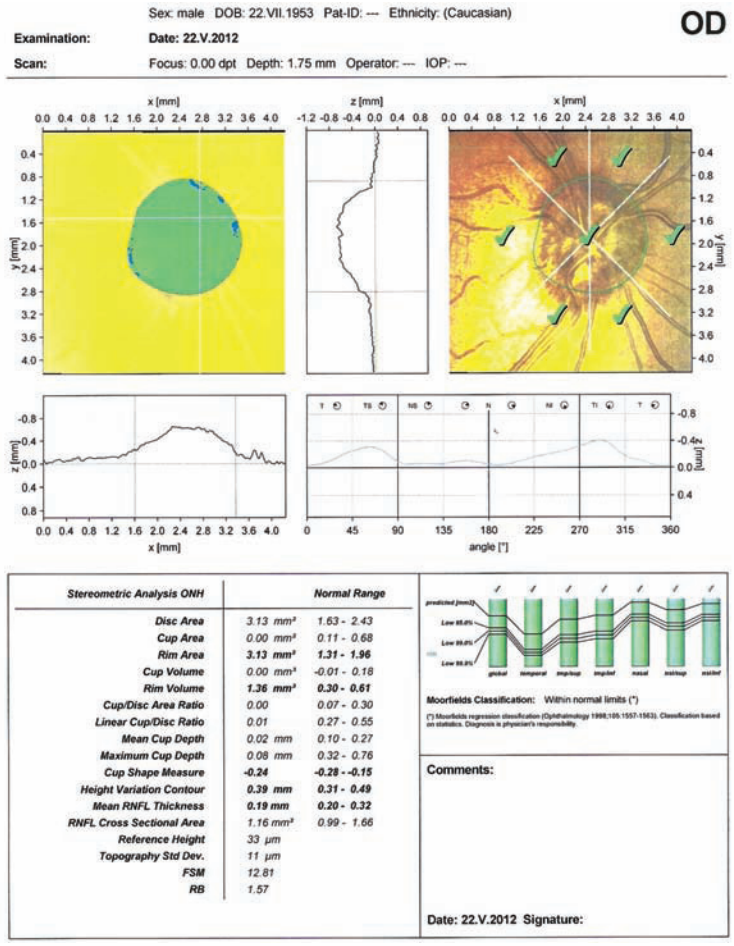

Fig. 9 HRT III examination, right eye (patient no. 1). According to Moorfields Regression Analysis negative finding in all quadrants, C/D ratio zero.

physiological. Upon examination by OCT Spectralis, numerous drusens are displayed, manifesting autofluorescence (Fig. 10 and 11). Finding documented by mydriatic fundus camera. Examination by OCT RNFL (Stratus, Zeiss) displayed a diminution of the RNFL in the upper sectors bilaterally. On GDx VCC diminution of RNFL in the right eye in the upper and lower temporal sector, in left eye diminution upper temporally. With regard to its low diagnostic significance, we do not conduct HRT III examination for ONH drusens. It is difficult to assess whether this represents changes caused by optic nerve drusens or whether the changes are a manifesta-

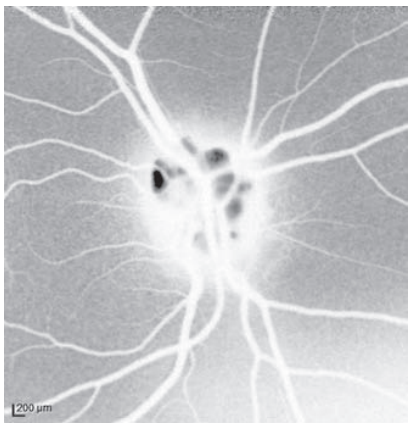

Fig. 10 Fundus autofluorescence (Spectralis), right eye (patient no. 2).

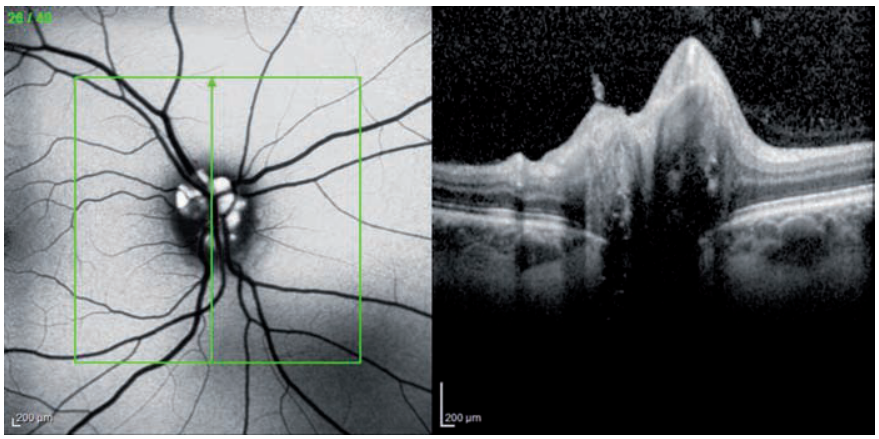

Fig. 11 OCT Spectralis – fundus autofluorescence and cross-section of DON, right eye (patient no. 2). Drusens are displayed by autofluorescence, in right section of image cross-section of DON.

tion of glaucoma damage to the ONH. We recommend the commencement of local antiglaucoma therapy and regular checks of IOP, perimeter, fundus and RNFL.

DISCUSSION

Optic disc drusens are determined in clinical practice as a secondary finding upon routine examination (7). The prognosis of optic disc drusens is considered good (8). The majority of patients with ONH drusens are asymptomatic. Severe disruption of vision is rare. Defects may occur in the visual field, which include defects in the lower nasal quadrant, extension of blind spot and peripheral narrowing of the visual field (4). Changes in the visual field in the case of optic nerve drusens are similar to glaucoma-related changes, which hampers interpretation of the damage to the optic nerve (6). Loss of central vision is rare upon haemorrhage from the peripapillary neovascular membrane. Diagnosis of drusens may be determined on the basis of the clinical picture in combination with ultrasound and CT examination. We did not perform a CT examination on our patients due to the burden of ionising radiation in the case of an unequivocal finding. B-scan echographic examination appears to be the most reliable method for detection of drusens (9). In addition to this, newer procedures of tomography of the optic nerve are also useful (12). Imaging by fundus autofluorescence (FAF) provides information beyond the framework provided by regular imaging methods. Imaging by fundus autofluorescence is an in vivo display technique for mapping of pathological accumulation of fluorophores on the fundus. The main source is fluorophores such as A2-E in lipofuscin granules. Lipofuscin is an autofluorescent and is suitable for noninvasive detection (5). The best quality images are obtained by using a scanning laser ophthalmoscope (5). The clinical value of this examination, based on simplicity, efficacy and non-invasiveness is useful (11). Fluorescence angiography may detect deep rooted drusens, and may help in differential diagnostics of pseudopapilledema or actual papilledema (12). In the case of uncertainty, evidence of drusens is applied with the help of CT examination (13), which at the same time excludes other more serious causes of calcification in ONH, such as retinoblastoma and glioma of the optic nerve (15). CT examination also displays small, deep-rooted drusens (10). OCT RNFL and GDx are used for quantitative monitoring of loss of RNFL. GDx reliably detects peripapillary constriction in connection with optic nerve drusens (16). HRT displays protruding ONH without excavation. An analysis of excavation, CD ratio and the neuroretinal rim is pronouncedly influenced by the presence of drusens (17). VEPs are abnormal in 41-97% of patients and correlate with the severity of damage to the peripapillary nerve fibres. However, the whole range of VEP abnormalities occurs in patients with drusens, and as a result VEP is not a reliable diagnostic modality for optic nerve drusens (4).

CONCLUSION

In specific cases we demonstrated possibilities in the diagnosis of drusens of the optic nerve. The most available method of detection of drusens is ultrasonography. At present modern OCT instruments are applied in diagnosis, which enable observation of the structure with high resolution and precision. These instruments are frequently combined with a HRA system, thus they are used for fluorescence angiography and also enable display with the help of autofluorescence.

The first presented patient, due to the presence of risk factors such as hypertension, hyperlipoproteinemia and diabetes mellitus, is at risk of possible vascular complications. Due to the increased risk of these complications it is necessary to observe patients with optic nerve drusens regularly.

Optic nerve drusens cause similar malfunctions in the visual field as we find in the case of glaucoma-related damage to the DOC. They may cause an elevation of IOP, they are accompanied by a diminution of the RNFL, they hamper interpretation of damage to ONH, and as a result patients with optic nerve drusens represent a diagnostic dilemma (6), just as in the case of the second presented patient. For patients with optic nerve drusens it is necessary to ensure regular examination of visual acuity, IOP, fundus, colour vision and the visual field. In patients with defects of the visual field it is recommended to monitor constriction of nerve fibres with the help of OCT RNFL (14).

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