

COMBINATION OF PATHOLOGY OF THE BRAIN AND THE OPTIC NERVE

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ABSTRACT

Background: The optic disc drusen is one of the most common anomalies of the optic nerve. The optic disc drusen is often found in combination with systemic and congenital diseases. The recognition of the combination congenital pathology of the brain and the optic disc drusen is considerably difficult and affects the patient's life prediction and quality.

Objective: The relevance of assessing the lesion of the visual analyzer with optic disc drusen and the possibility of identifying concomitant pathologies, increases with the introduction of the latest diagnostic techniques.

Methods: The patient, 50 years old, complaining of episodic blurred vision. Standard ophthalmic examination, optical coherence tomography, computer perimetry, ultrasound B-scanning of the orbit and optic nerve, MRI of the head on with contrast were carried out.

Results: The diagnosis revealed the drusen of the optic nerve head and concomitant congenital neurological pathology. The patient was consulted by a neurosurgeon with the aim of delivering a diagnosis and determining treatment tactics. The diagnosis was made: Arteriovenous malformation of the deep arteries of the sections of the left parietal lobe. After assessing the size of the arteriovenous malformation, its location in the functionally important area of the brain, it was decided to follow up the patient in dynamics.

Conclusion: Thus, when verifying the optic disc drusen, it is possible to identify concomitant congenital pathologies of the brain. These facts indicate the need for neurological examination of patients with anomalies of the optic nerve and confirm the importance of further study of this problem.

Keywords: Drusses of the optic nerve disk, arteriovenous malformation

Introduction

Optic disc drusen (optic disc) is an optic nerve congenital pathology characterized by progressive degeneration of neurons into intercellular hyaline, calcified formations located in the prelaminar part of the optic nerve. Optic disc drusen are found in 0,3% - 2,4% of the population.^{1,2} Clinically, in most cases, asymptomatic for patients, optic disc drusen are the cause of functional disorders in the optic nerve and optic neuropathy.^{3,4,5,6} With progression of optic neuropathy in 71-92,5% of cases, optic disc drusen lead to the development of optic nerve atrophy.⁷ Changes in visual fields are an important diagnostic criterion for optic neuropathy. With optic disc drusen, they are diagnosed in 24-87% of cases, in the form of slowly progressive defects in the peripheral and paracentral visual fields.⁸ With the advent of optical coherence tomography, foreign researchers have found that the loss of the visual field with optic disc drusen correlates with the thickness of the retinal nerve fiber layer.^{9,10,11,12} However, only a few publications describe neurological manifestations in this pathology. In 2016-2017, Russian scientists conducted a study where, in a group of 108 people with verified optic disc drusen, 51

patients had neurological symptoms, and 6 patients had congenital brain pathology.^{1,13} According to the opinion of Auw-Haedrich et al. hereditary dysplasia of the optic nerve head and its blood supply is primary in the pathogenesis of optic disc drusen.¹⁴ The Finnish researcher H. Erkkila, based on a survey of 50 children with optic disc drusen, established an unusual state of the retinal vessels. There was a large number of vessels, their earlier branching and sharp tortuosity, as well as an increased frequency of the presence of a cilioretinal artery. The author associates these anomalies with the manifestation of mesodermal dysplasia.¹⁵ Researchers also paid attention to the abnormal angioarchitectonics of the optic disc drusen. The leading role of vascular dysplasia in the pathogenesis of optic disc drusen was suggested in 2008 by Lam B. The researcher, describing large blood vessels connecting superficial and deep vessels in the disc, increased capillarity of the disc, suggested increased extravasation of plasma proteins and deposition of extracellular material.¹⁶ The optic disc drusen is often found in combination with systemic and ocular diseases such as retinitis pigmentosa, elastic pseudoxanthoma and Alagill's syndrome.

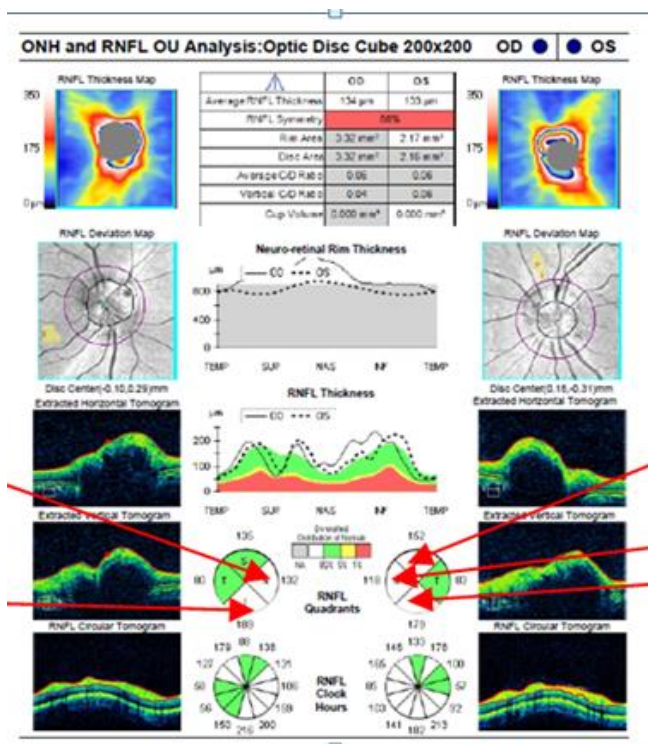


Figure 1. The protocol of the optic nerve disc of patient K. Thickening of the retinal nerve fiber layer is indicated by red arrows.

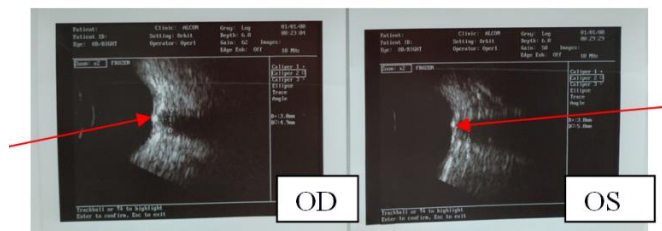


Figure 2. B-scan of the orbits of patient K. The increase in acoustic density in the optic nerve disc of both eyes is indicated by red arrows.

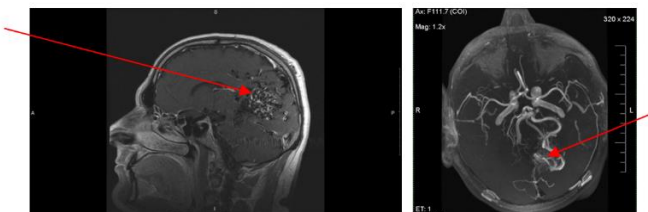


Figure 3. MRI of the brain of patient K. Arteriovenous malformation is indicated by red arrows.

This article presents an interesting clinical case of a combination of optic disc drusen with arteriovenous malformation. According to histological data, congenital vascular malformation occurs in about 0.1% of the population. In most cases, it is asymptomatic and detected as an accidental finding during magnetic resonance imaging (MRI) or computed tomography of the orbits (CT scan).^{17,18,19} The diagnosis of optic disc drusen becomes more informative with the advent of ophthalmic methods of structural and topographic assessment of the visual analyzer, such as spectral optical coherence tomography, computed laser retinotomography, video ophthalmography, as well as ultrasound B-scanning of the orbit and optic nerve, fluorescent angiography of the retina, CT scan of the

orbits, MRI the brain and orbits. However, when verifying the optic disc drusen, it is possible to identify concomitant pathologies.^{20,21,22}

Methods

In the S. Fyodorov Eye Microsurgery Federal State Institution, Moscow, a patient, 50 years old, presented complaints of episodic blurred vision and headache attacks.

The standard ophthalmological examination researches were carried. The computer perimeter was performed under the visual field testing program within 120 points (Humphrey Field Analyzer II ("Carl Zeiss Meditec Inc.")). The optic disc and layer of the retina nerve fibers were measured by the protocol Optic Disc Cube 200x200 and RNFL Thickness Analysis, the ganglion cells were scanned by the protocol Ganglion Cell Analysis: Macular Cube 512x128 by using OCT (CIRRUS HD-OCT 5000 ("Carl Zeiss Meditec Inc.")). The patient was directed to ultrasound B-scanning of the orbit and optic nerve (Ultra Scan, Alcon) and MRI scanning of the head revealing the contrast (SIEMENS SYMPHONY 1,5 Tesla).

Case Report

Initial examination revealed no decrease in visual functions: visual acuity OD = 1.0; visual acuity OS = 0.8 cyl -1.0 ax 170° = 1.0. Biomicroscopy: the eyes are calm, the conjunctiva is pale pink, the cornea is transparent, the anterior chamber is of medium depth, the iris is structural, and the lenses are transparent. When examining the fundus of the optic nerve disc in both eyes, pale pink, with irregular, scalloped contours, penetrates into the vitreous body, on the optic disc surface rounded white-yellow inclusions. According to the data of computer perimetry, expansion of the blind spot zone in both eyes. The patient was referred for optical coherence tomography. According to the data of optical coherence tomography, the morphometric parameters of the optic disc were changed. Cup volume reduced to zero for both eyes. The ratio of the Rim area to the Disc area is 1:1. There was a thickening average thickness of the retinal nerve fiber layer (RNFL) is OD=70 μm, OS=72 μm and RNFL in all quadrants except for the upper one on the both eyes (Figure 1).

The thickness of the retina is normal, the thickness of the retinal ganglion cells and the inner plexiform layer is normal. A preliminary diagnosis was made: optic disc drusen in both eyes. In order to confirm the optic disc drusen, an ultrasonic B-scan of the orbits was carried out. Ultrasonic B-scanning of the orbits in the optic nerve disc area revealed bodies of increased acoustic density. However, the 30 ° test was positive in both eyes (Figure 2). In order to exclude stagnant optic disc edema, the patient was prescribed MRI of the brain and orbits (Figure 3).

Conclusion MRI: a picture of large size arteriovenous malformation of the left parieto-occipital region against the background of incomplete posterior trifurcation of the left internal carotid artery, which feeds from the enlarged left posterior cerebral artery and drains into the system of the great cerebral vein. Conclusion of MRI-angiography: asymmetry of the vertebral arteries (D> S) without signs of narrowing, asymmetry of the transverse and sigmoid

sinuses (D<S) with signs of unsharp narrowing on the left (susp. Hypoplasia of the right transverse sinus). Signs of obstructed venous outflow. MRI signs of structural changes in the optic nerve region. The patient was referred for a consultation with a neurosurgeon. The diagnosis was made: Arteriovenous malformation of the deep arteries of the left parietal lobe (Spetzler-Martin - 4-5 degrees). Considering the large size of the arteriovenous malformation, the location in a functionally important area of the brain and the absence of serious clinical manifestations, the patient is not indicated for surgical treatment. Radiosurgical treatment is impossible due to its large size.

Conclusion

This paper presents a rare clinical case. When the diagnosis of the optic disc drusen was made, concomitant congenital neurological pathology was revealed, which became an accidental finding on MRI. The patient is currently under the dynamic supervision of a neurosurgeon. These facts indicate the need for neurological examination of patients with anomalies of the optic nerve and confirm the importance of further study of this problem.

Acknowledgement

None.

Conflict of Interest

The authors declare that this study has no conflict interest.

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