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DIAGNOSTIC AND THERAPEUTIC MODALITIES IN THE MANAGEMENT OF OPTIC DISC PIT MACULOPATHY- CASE REPORT AND LITERATURE REVIEW

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Abstract

Optic disc pit is an oval depression that is usually located in the temporal sector of the papilla nervi optici. It can be asymptomatic, but can also be the cause of maculopathy, optic disc pit-associated maculopathy (OPM). Its etiology is still not fully known, but modern imaging techniques in ophthalmology (OCT and OCT-A) have helped to better understand the pathophysiology of this entity.

The purpose of this paper is to present a case with optic disc pit maculopathy, but also to give an overview of the pathogenesis, clinical aspects, imaging methods and therapeutic modalities in the management of patients with this pathology.

***Keywords*:** optic disc pit, pars plana vitrectomy, carbonic anhydrase inhibitors, optic disc pit maculopathy.

Introduction

Optic disc pit (OP) is an oval depression of the optic nerve papilla that is most often found in its temporal part. The size of the pit can be from 1/10 to 7/10 of the size of the disc [1].

It was first described in 1882 by Weithe in a 62-year-old woman, bilaterally [2].

This entity was later correlated with the appearance of maculopathy in a large number of patients. Usually this papillary change is asymptomatic, but it can lead to visual field disturbances such as enlargement of the blind spot or appearance of paracentral scotoma as well as central vision affection. Decreased eye function is most often due to maculopathy, which is a complication associated with OP. Maculopathy is characterized by the presence of subretinal fluid, retinoschisis and/or the appearance of a lamellar or complete macular hole, changes that damage the retinal pigment epithelium (RPE) and cause atrophy of the neurosensory retina [3].

OP affects both sexes equally and is a rare entity with an incidence of 1:11,000 people [4]. It is mostly unilateral (85% of cases), sporadic, single, but sometimes multiple pits can be present on the same disc. It occurs in all age groups from 3-82 years, with mean age being 35 years [5].

Certain authors suggest a possible autosomal dominant inheritance in some patients with unilateral OP [6].

Due to the common morpho-functional characteristics with other congenital anomalies of the optic disc, OP belongs to the group of so-called congenital optic disc cavitary anomalies, together with: coloboma of the optic disc, extra-papillary cavitations and the morning glory anomaly [7].

The appearance of maculopathy associated with OP is a frequent manifestation, it is found in 84% of patients, especially those where OP is located temporally. As many as 20% of patients with OPM are asymptomatic, and the remaining symptoms usually present between the third and fourth decade of life, however, cases with a much earlier onset have also been described [4,8].

Visual acuity is in a wide interval from 6/6 to counting fingers, with a mean value of 0.3 [4].

The etiology and pathogenesis of OP and OPM are still not fully understood. Congenital OP is assumed to occur as a result of inadequate closure of the upper edges of the embryonic fissure. In this way, it remains as a potential communication between the vitreal compartment and the retina, through which fluid penetrates and causes stratification of the neurosensory retina. According to these hypotheses, depending on the site of the communication, an accumulation of subretinal or intraretinal fluid will occur

[8,9]. Another assumption is that OP is a herniation of the dysplastic retina through the defective excavation of the lamina cribrosa and is the communication of the intraocular with the subarachnoid space [10,11].

The origin of the fluid in OPM is also a controversial topic. Some authors think that the fluid originates from the vitreal compartment due to the process of liquefaction and posterior detachment of the vitreous (PVD), which previously caused pit tamponade. Brown et al confirmed this hypothesis by injecting dye into the vitreous cavity and the same was found in the subretinal space, in dogs with this pathology [12].

Another data that supports this is the presence of silicone oil or gas in the subretinal space in OPM patients after vitrectomy. However, the presence of OPM in individuals without PVD also points to the fact that this is not the only source of the fluid. Another hypothesis for a possible source of fluid in OPM is the subarachnoid space, through the OP, which is thought to be in communication with this compartment. A patient with OP after pars plana vitrectomy (PPV) and injected silicone oil was described, part of which migrated into the subarachnoid space [13].

However, other studies have not confirmed the association between these two compartments. A third, less likely theory is the association of OPM with the presence of a cilioterinal artery. It is interesting that the appearance of OP is also associated with other diseases, such as: incontinentia pigmenti, encephalocele, Algille and Aicardi syndrome, ocular trauma, etc. [8].

Materials and methods

During the preparation of this paper, a search of the PubMed library, a database of medically relevant data, was performed by entering keywords: optic disc pit, pars plana vitrectomy, carbonic anhydrase inhibitors, optic disc pit maculopathy. From the obtained results, those that show the modern attitudes in the management of patients with this condition were chosen. They were analyzed and arranged didactically in this paper in order to present the new knowledge from this field of ophthalmology.

Case report

A 73-year-old male, patient came to the PHI UC for Eye Diseases in Skopje, due to a gradual and progressive decrease in vision in the left eye over the past few months. From the anamnestic data, we saw that he has hypertension and was previously operated on for cataracts in both eyes. Biomicroscopic examination, motility and tonometry were without pathological findings. The best-corrected visual acuity of the right eye was 0.9; and on the left 0.1. During the fundoscopic examination, the right eye was normal, while on the left, a small-oval excavation was observed in the lower-temporal sector of the papilla, as well as an elevation of the neurosensory retina in the macular region. The RPE beneath the edematous retina was discolored and the entire zone had a bronze appearance. To document the changes, a color fundus photograph and an optical coherence tomography of the macular region were made (Figures 1 and 2). A diagnosis of optic disc pit maculopathy was made.

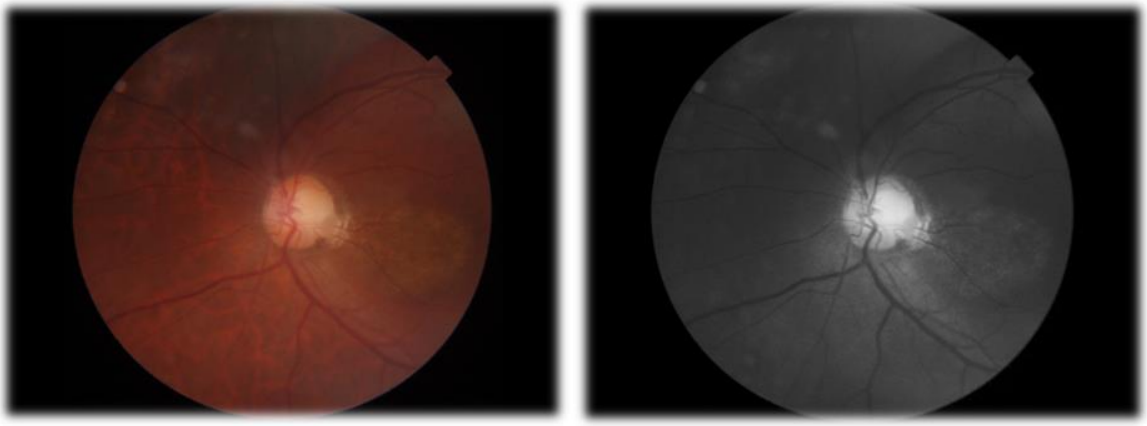
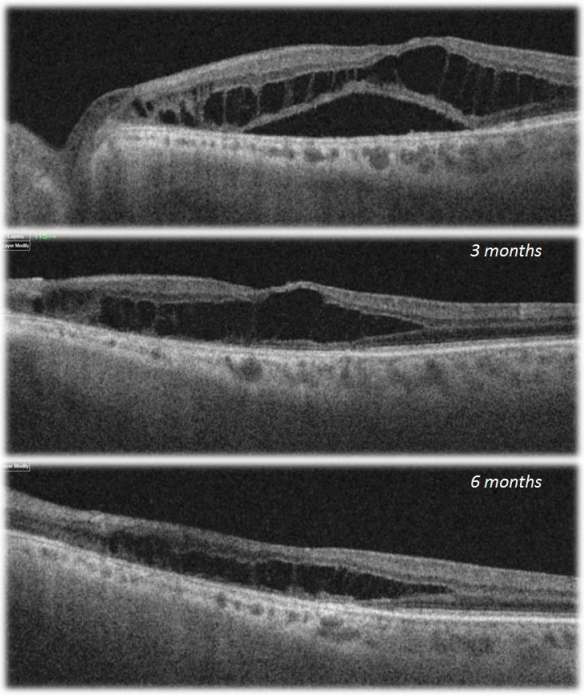


Figure 1. Color fundus photograph, left, optic disc pit in the lower temporal sector of a papilla, oval in shape, through which the cilio-retinal artery passes. A zone with a bronze color in the macular region - maculopathy, a consequence of the long-term presence of intraretinal and subretinal fluid. Red-free photograph, right, delineation of optic disc pit and granular hypodense aspect of papillo-macular zone.

The condition and the possibilities for its treatment were explained to the patient. Due to the general state of health, age and moderately preserved visual acuity, a decision was made for conservative treatment with low doses of systemic and local application of carbonic anhydrase enzyme inhibitors: Tbl.Acetozolamide and Sol.Dorsolmaide for several months. At subsequent follow-ups at 3 and 6 months, a significant reduction of intraretinal and subretinal fluid was observed (Figure 2) with an improvement in visual acuity of two lines according to the Snellen optotype, and it was 0.3 before publication of this paper. Due to the possible side effects of long-term use of Acetozolamide, the patient is regularly monitored.



Macular schisis and disorganization of the outer retinal layers, a large amount of subretinal fluid.

Reduced retinal thickness and complete resorption of subretinal fluid and partial reduction of intraretinal cystic spaces.

Significantly reduced retinal thickness, regression of intraretinal fluid, without the presence of subretinal fluid.

Figure 2. Follow-up of regression of cystoid spaces and subretinal fluid with OCT in the patient, underthe influence of local and systemic carbonic anhydrase enzyme inhibitors.

Discussion

The discovery of the morpho-functional characteristics of OP and OPM in the past decades is especially due to the development of a large number of imaging methods in ophthalmology. Perimetry, fundus autofluorescence (FAF), fundus photography; as well as fluorescein angiography (FFA), optical coherence tomography (OCT) and optical coherence tomography with angiography (OCT-A) helped to understand the mechanisms of OP occurrence, as well as the characteristics that are an integral part of OPM. The visual field shows the presence of arcuate scotomas as a consequence of displacement of nerve fibers by OP, and the appearance of central or paracentral scotomas in those patients with OPM [14].

FAF reveals RPE changes due to the long-term presence of subretinal and intraretinal fluid [15].

FFA shows OP as a hypofluorescent zone, which in the late stages of the angiogram turns into hyperfluorescent as a result of staining. Hyperfluorescence in the late stages is also found in the retinal schisis and subretinal space in patients with OPM, which is in favor of fluid circulation in these spaces [16].

However, the greatest insights into the morphology of OP and OPM have been obtained with OCT. With this tool, the vitreoretinal interference in patients with OP, the appearance of vitreomacular traction, vitreous detachment as well as the structural analysis of OPM have been observed.

Gowdar et al. with OCT showed a connection between the macular schisis and the lamina cribrosa which supports the theory that the fluid in OPM originates from the cerebrospinal fluid. OCT today also plays a major role during vitreoretinal surgery in these patients, for better visualization of the structures during the procedure [10,17].

OCT-A indicated microvascular involvement in the maculo-papillary area in patients with OPM. Adams et al. observed the presence of irregular blood vessels around OP in patients with OPM using SS-OCTA [18].

There is still no unified opinion among ophthalmologists about the treatment of this condition. The observation of such patients, without any additional treatment, indicated that spontaneous regression of the maculopathy occurs over time in as many as 25% of cases [8,19].

Several authors have shown cases of different ages where gradual and complete resorption of the intraretinal and subretinal fluid occurred over a period of several months to several years, with an improvement in visual function.

They emphasize that preserving the integrity between the IS/OS layers of the retina is of exceptional importance in such cases for achieving satisfactory visual results. Whether the foveal center is affected or not, is of great importance in the prognosis.

Treatment with laser photocoagulation along the temporal edges of the pit has been attempted in the past. The following were used: xenon photocoagulation, argon photocoagulation and krypton laser therapy. The obtained results were with a certain anatomical success, however, without a significant improvement of the visual function, or even a deterioration in the visual field as a consequence of the destructive action of the laser [8].

Acetozolamd has been used for the first time in pediatric patients with OPM. The idea is that with the help of a carbonic anhydrase enzyme inhibitor, the production of cerebrospinal fluid is reduced, so in patients in whom there is thought to be communication between these two compartments, this reduction would contribute to a reduced flow of fluid in the intraretinal and subretinal space. Today, this method is still used either as a primary treatment or as an adjuvant in pars plana vitrectomy. However, in patients with long-term treatment with this medication, potential systemic side effects are possible [20].

Gas tamponade with or without laser photocoagulation has recently been shown to be a possible therapeutic option. Lei et al. showed 9 cases how the combination of intravitreal application of C3F8 gas and laser photocoagulation of the temporal part of the disc is a good therapeutic modality both in terms of anatomical and functional benefits. The application of gas tamponade without laser photocoagulation by different authors has led to more or less satisfactory results [8,21].

Macular buckling is also a potentially good therapeutic procedure, with a success rate of over 85%. The principle is to press and push the macular region with a sponge placed behind the posterior pole, which would lead to a reduction in vitreomacular traction. Due to the specificities in performing this procedure, this method of treatment is not widely accepted nowadays [22].

In contrast to buckling, today, pars plana vitrectomy (PPV) occupies a major place in the operative treatment of these cases with an anatomic success rate of over 75%. There are different vitrectomy approaches in OPM such as: PPV with removal of posterior hyaloid, PPV with peeling of internal limiting membrane (ILM) and tamponade with SF6, PPV with inverted ILM flap, PPV with peeling of glial tissue, closure of OP using autologous blood or blood clot, PPV with endolaser and gas/oil tamponade, and PPV with retinal fenestration [5,8,23].

The existence of different approaches to vitrectomy in patients with OP indicates that there is still no consensus and uniform opinion on the optimal treatment of this pathology. Future studies of a larger number of patients should answer which technique is superior over others and when it is indicated.

Knowing the fact that the condition in a certain assessment of patients is self-limiting, in contrast to the risky operative techniques leads to the consideration of an individualized approach in each patient, realizing the possible benefits and risks when choosing one therapeutic option over another. So in our case, due to the general health, the age of the patient and the relatively preserved visual acuity, we decided on a conservative treatment of the condition for a period of one year with a systemic and local inhibitor of the carbonic anhydrase enzyme, which resulted in a moderate morpho - functional improvement of the condition.

Conclusion

The existence of multiple therapeutic options indicates that there is no perfect solution in the treatment of patients with optic disc pit maculopathy. Each therapeutic modality brings with it its own advantages, but also risks and disadvantages.

Therefore, an individualized approach to the patient, for now, may be the most appropriate course of action.

That is, when making the final decision, the doctor should consider the general health of the patient, the potential risks and benefits of such treatment, the visual needs and the degree of visual disability of the patient; before deciding whether to just follow it or apply a more invasive treatment method.

The patient, on the other hand, should be adequately informed and familiar with the overall situation, in order to actively participate in making the final decision. Finally, research in the future should help in taking a uniform attitude among ophthalmologists for the treatment of patients with this pathology.

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