Association of macular microhole and optic disc pit

Associação de microburaco macular e fosseta de papila

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ABSTRACT

Optic disc pit and macular microhole are two rare pathologies with an extremely low likelihood of coexistence, this paper will report an association of both pathologies in the same eye with the purpose of analyzing clinical manifestations, tests, angiography, OCT, retinography, biomicroscopy, treatment outcome and the connection between the optic disc pit and macular microhole.

Keywords: Retinal perforations/therapy; Fundus oculi; Optic disc/abnormalities; Tomography, optical coherence; Case reports

RESUMO

A fosseta de papila do nervo óptico e o microburaco macular são duas patologias raras, cuja probabilidade de coexistência se torna extremamente baixa, embora não haja relação fisiopatológica entre ambas, descreveremos um caso de associação das mesmas, acometendo comumente um olho, a fim de analisar as manifestações clínicas, os exames de OCT, angiografia, retinografia, biomicroscopia, o tratamento e a correlação entre ambas patologias.

Descritores: Perfurações retinianas/terapia; Fundo de olho; Disco óptico/anormalidades; Tomografia de coerência óptica; Relatos de casos

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INTRODUCTION

Optic disc pit maculopathy and macular microhole are two uncommon diseases whose chance of co-occurring appears to be extremely low.

Optic disc pit has an incidence of 1:11,000 and is a congenital malformation secondary to a developmental disorder of the primitive epithelium of the optic disc which occurs in the fifth week of embryogenesis\(^{(1,2)}\). Usually unilateral (in 95% of cases), this excavation of the optic nerve has an oval shape and is most often located in the temporal region of the optic disc; its colour ranges from shades of gray to yellow to black\(^{(3)}\). Symptoms occur only in the presence of serous macular detachment, which occurs in 25-75% of cases and affects visual acuity\(^{(3)}\).

The aetiology of serous macular detachment related to optic disc pit is undetermined. There are three possible sources of subretinal fluid: The movement of vitreous humour through the pit fissure to the subretinal space; the flow of cerebrospinal fluid from the subarachnoid space into the subretinal space; and the leakage of fluid through peripheral choriocapillaris into the subretinal space\(^{(4)}\). First described by Cairns and McCombe, macular microhole is a retinal defect consisting of a reddish lamellar fissure with well-defined edges. It is located in the inner layer of the retina, in the central region of the fovea, corresponding to the inner and outer segments of photoreceptors\(^{(5)}\). Its aetiology is undetermined, and since there is no causal factor directly related to its formation, it may be a primary condition. Symptoms usually include: central scotoma, mild to moderate impairment of visual acuity, and metamorphopsia. However, many patients remain asymptomatic. Diagnosis can be made by combining the findings of posterior pole biomicroscopy and optical coherence tomography (OCT). The prognosis is good, as the condition usually progresses with stability of symptoms and anatomic appearance\(^{(5,8)}\).

Even though optic disc pit and macular microhole have a low incidence, we describe a case where both conditions occurred simultaneously in the same eye. The aim of this paper is to describe the clinical manifestations, OCT scans, angiography, retinography, biomicroscopy, and treatment of the conditions.

Case report

VL, 30-year-old white female born in Nova Trento, Brazil, was diagnosed 3.5 years ago at another service with serous retinal detachment in the left eye (LE). Initial ophthalmic examination showed a visual acuity of 20/20 in the right eye (RE) and 20/40 in the LE. Biomicroscopy of the anterior segment showed no changes in both eyes. Fundoscopy showed the presence of a temporal optic disc pit in the RE. Evaluation of the fovea in the RE showed a reddish spot suggestive of macular microhole.

In the LE, detachment of the sensory retina was observed, affecting the macular region and reaching the temporal edge of the optic disc. The patient then underwent an OCT scan (Figures 1 and 2), which confirmed the temporal optic disc pit and the presence of a macular microhole in the RE. In the LE, the OCT clearly showed the optic disc pit with a communication between the subretinal space and the inferior temporal region of the optic disc, indicating a possible orifice communicating these structures. Furthermore, a loss of foveal contour was observed with serous detachment of the neurosensory retina, more pronounced in the region nasal to the fovea, and a content of average reflectivity was seen in the central macular region (Figure 1).

In the RE the approach toward the diagnosis of optic disc pit associated with macular microhole was expectant management, with frequent ophthalmic evaluations using Amsler grid every month and OCT every two months.

In the LE we performed laser photocoagulation in the temporal edge of the optic disc, with slow but the gradual improvement of the detachment; after five months, the macula was completely attached (Figure 1B).

The clinical picture remained stable for 1 year and 7 months. However, fundus examination showed a recurrence of the sensory detachment near the optic disc, affecting the inferior
macula in the LE. OCT confirmed the sensory detachment, with attachment of the posterior vitreous exactly in the region where the retina was elevated, suggesting the possibility of localised traction (Figure 2D). Serial OCTs showed the progression of the detachment; we then performed posterior vitrectomy with C3F8 gas infusion at a concentration of 12%. The patient recovered satisfactorily, with gradual decrease of the subretinal fluid and complete resolution of the condition in 6 months. After a follow-up of 9 months, the patient had stable vision (20/20) and an attached macula.

The RE had a 20/20 vision without visual impairment.

**DISCUSSION**

The optic disc pit is an uncommon congenital anomaly presenting as a yellow or gray oval lesion. It is usually unilateral and is found primarily in the temporal region of the optic disc. In more than 50% of cases, one or two cilioretinal arteries can be seen in the pit area(4). Changes in the colour of the pigment epithelium along the temporal edge of the disc and posterior vitreous detachment are findings associated with the condition(5).

In cases where an abnormal communication exists between the optic nerve and the retina, the passage of fluid between the two structures can occur. The origin of the fluid, whether from the vitreous humour, the cilioretinal arteries or the cerebrospinal fluid, remains unclear. The resulting maculopathy caused by fluid leakage can be observed on OCT as a separation of the retina in a two-layered structure. The fluid coming from the optic disc pit can accumulate in different layers of the retina, most commonly in the inner and outer nuclear layers. The appearance is reminiscent of retinoschisis, i.e. it overlies the sensory macular detachment(9). The latter occurs in 40-60% of patients and is considered the primary cause of visual impairment in patients with optic disc pit(23).

Macular microhole is a little known and infrequently diagnosed disease. It is identified by fundoscopy and confirmed by OCT and is described as a small red spot in the centre of the fovea measuring between 50 and 150 microns on average; it can lead to a mild loss of visual acuity, central scotoma and metamorphopsia(44).

In our case, the patient had optic disc pits in both eyes. In the RE, which had a 20/20 vision without symptoms or visual changes, fundus examination showed a reddish foveal spot suggestive of macular microhole, which was confirmed by OCT.

Given this diagnosis, we explained the patient that the condition generally does not progress and indicated a follow up with complete eye examinations and OCTs. In the literature there are no reports relating the two diseases. Despite their presentation in the same eye, the two conditions possibly have different physiological mechanisms.

The LE presented with an optic disc pit and serous macular detachment. Several types of treatment are proposed for this condition: laser photocoagulation alone, intravitreal gas injection, laser photocoagulation combined with intravitreal gas injection, and vitrectomy combined with gas injection, with or without laser photocoagulation(50).

In a joint decision with the patient, we opted for the simplest and less invasive alternative: laser photocoagulation in the temporal edge of the optic disc, in the region of fluid passage to the subretinal space. After laser therapy, the detachment improved slowly but completely. However, after a few months, the detachment recurred. The patient then underwent posterior vitrectomy with C3F8 gas infusion at a concentration of 12%.

The option to perform vitrectomy was based on the tomographic finding of vitreous attachment in the exact area of the retinal detachment, suggesting the possibility of localised traction. This, together with the other anatomical conditions present in cases of optic disc pit, is the aetiology of sensory retinal detachment. This traction effect had been previously mentioned by other authors(11,12); in our case, the OCT was very helpful in revealing the condition and was therefore essential for the decision to perform surgery.

Macular serous detachment associated with optic disc pit has a poor prognosis in untreated patients; chronic macular detachment can lead to visual acuity lower than 20/200 in 80% of cases(31).

In our case, the OCT was essential to confirm the diagnosis of macular microhole in the RE and to guide treatment in the LE, allowing detailed evaluation and monitoring of the improvement of sensory macular detachment after laser therapy. It also allowed us to observe the onset of recurrence and to diagnose the presence and the precise location of the traction, having been a key tool for choosing the appropriate type of treatment.

**REFERENCES**


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