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Case Report

A Retinoschisis and Serous Detachment of the Macula Revealing an Optic Disc Pit: A Case Report

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Abstract

Introduction: Optic disc pits (ODPs) are a rare congenital anomaly of the optic disc. They are thought to be a result of incomplete closure of the superior edge of the embryonic fissure. The main complication is the serous maculopathy: optic disc pit maculopathy (OPDM). We present an unusual case of patient with ODPM.

Aim: Underline a rare cause of serous maculopathy and discuss its treatment options.

Observation: 25-year-old patient, male, previously healthy who consults for rapidly progressive vision loss on his right eye quantified at 1/10.

Results: The fundus examination objectified a deeply pigmented circular hole in the inferotemporal neuroretinal rim of the optic nerve.

Macular OCT showed splitting within the neurosensory retina, intraretinal cysts suggesting a retinoschisis with a serous macular detachment.

A pars plana vitrectomy was done with intravitreal injection of gas and peeling of the internal limiting membrane (ILM).

On follow up examinations, a visual improvement was noted, restoration of foveal outer retinal layer structure and diminution of intraretinal fluid.

Discussion: One of the initial treatments was oral corticosteroids and acetazolamide. This treatment proved ineffective in a majority of cases.

Others reports described treating ODPM with argon laser photocoagulation at the temporal disc margin. The time for improvement was often long. Intravitreal gas injection has been proposed. The pneumatic displacement will cause reattachment of the macula.

The majority of the published literature on ODPM favors pars plana vitrectomy (PPV) as the treatment of choice. Several reports described successful anatomical and visual restoration in patients with ODPM who underwent PPV with or without ILM peel, endolaser and gas tamponade.

In our case, a PPV was done with intravitreal injection of gas and peeling of the ILM. A visual improvement was noted, restoration of foveal outer retinal layer structure was documented by OCT and diminution of intraretinal fluid.

Conclusion: The mechanism of pathogenesis of the subretinal fluid in OPDM been reached. This makes it difficult to determine the optimal surgical technique. Typically, ODPM is treated with PPV with or without juxtapapillary retinal laser photocoagulation, gas tamponade and ILM peeling.

Keywords: *Optic Disc Pit Maculopathy; Treatment; Vitrectomy*

Introduction

Optic disc pits (ODPs) are a rare congenital anomaly of the optic disc. Wiethe was the first author to describe them in a 62 year old female, in 1882 [1].

They are considered as part of a spectrum of congenital cavitary anomalies of the optic disc, which also includes optic disc coloboma, morning glory and extrapapillary cavitation [2].

They are often unilateral but may be bilateral in up to 15% of patients [3,4]. They are thought to occur sporadically with no known risk factors in particular but possible autosomal inheritance has been suggested in some pedigrees with multiple affected members [5,6]. No specific gene has been associated with ODP formation. Men and women are affected equally with an estimated incidence of 1 in 11,000 people [3,7].

Even though pathophysiology of optic disc pits is not completely clear, optic pits are thought to be a result of incomplete closure of the superior edge of the embryonic fissure.

Some hypothesize suggests that optic disc pits and colobomas can share the same pathophysiology, while others authors considered optic disc pit as a variant of optic disc coloboma [8,9].

Contrary to optic disc coloboma, Optic disc pits in general are less commonly associated with systemic associations and they are rarely found simultaneously with retinochoroidal or iris colobomas [3,9].

It is well known that both optic disc coloboma and pit can develop macular complications [10].

We present an unusual case of patient with optic disc pit associated with a retinoschisis and serous detachment of the macula.

Case Report

It is about a 25-year-old patient, male, previously healthy, he denied any previous ocular pathology and surgeries. There was no family history of ocular diseases. He consults for rapidly progressive vision loss on his right eye quantified at 1/10.

Anterior segment biomicroscopy, extrinsic ocular motility and pupillary light reflex were normal. Goldmann applanation tonometry was 12 mmHg in both eyes.

The dilated fundus examination and retinography (Figure 1) objectified, at the right eye, a deeply pigmented circular hole in the inferotemporal neuroretinal rim of the optic nerve. It is a small, round and grayish excavated defects and typically unilateral. It occupies approximately 1/8 of the size of the disc.

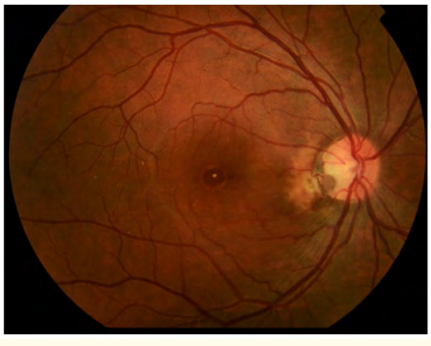

Figure 1: *Fundus photograph of an inferotemporal optic nerve pit.*

The macula and the temporal juxtapapillary retina appeared elevated.

A radiological exploration was done: Macular spectral-domain optical coherence tomography (OCT) (Figure 2) showed splitting within the neurosensory retina, intraretinal cysts suggesting a retinoschisis with a serous macular detachment.

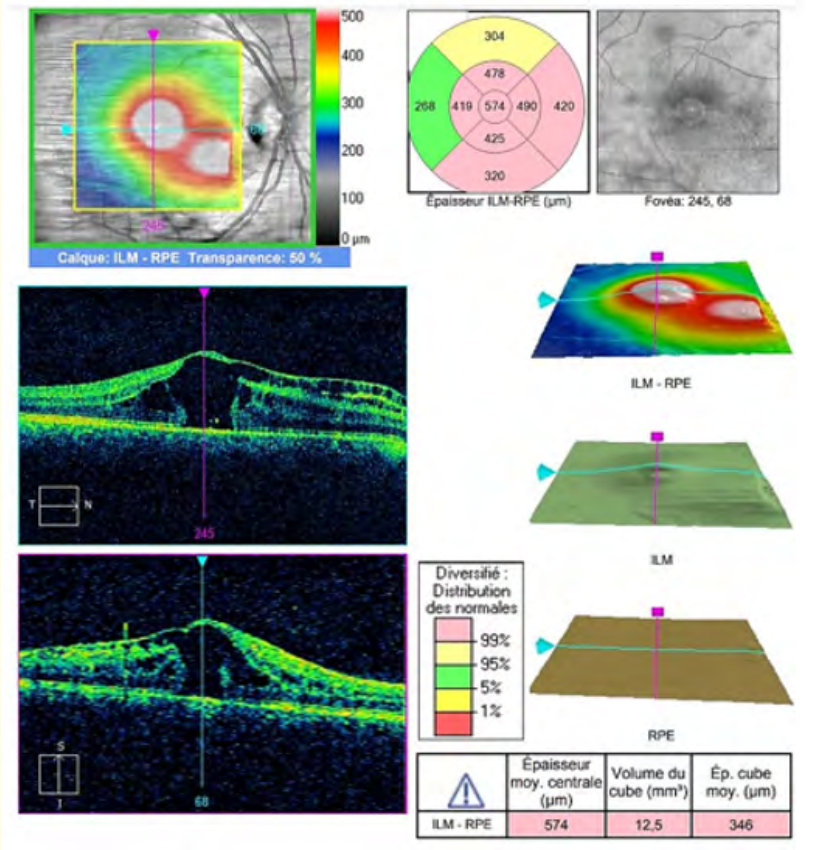

Figure 2: *Macular OCT showing splitting within the neurosensory retina, sub and intraretinal fluid.*

OCT of optic nerve (Figure 3) showed retinochoroidal-scleral excavation of the nerve. A defect in inferotemporal was detected with an intraretinal and subretinal fluid, extending towards the ODP.

AGF with fluorescein (Figure 4) objectified focal leakage in the fovea.

Therapeutically, a pars plana vitrectomy was done with intravitreal injection of gas and peeling of the internal limiting membrane.

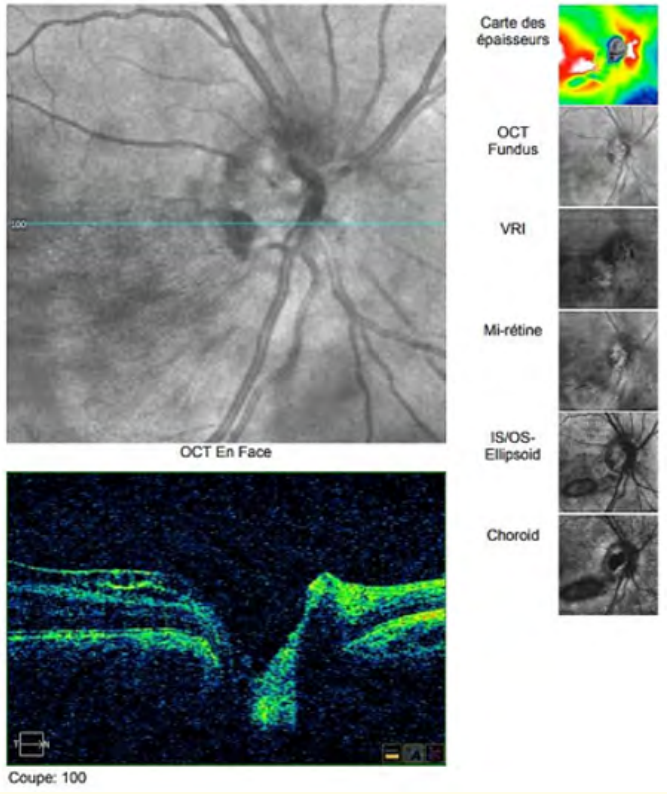

Figure 3: *A horizontal OCT scan through the optic disc showing retinochoroidal-scleral excavation of the nerve with a defect in inferotemporal and an intraretinal and subretinal fluid, extending towards the ODP.*

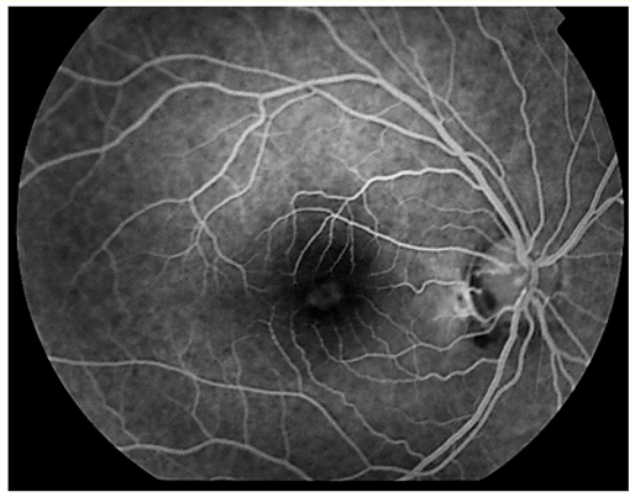

Figure 4: *Fundus fluorescence angiography showing focal leakage in the fovea.*

On follow up examinations, after surgery, a visual improvement was noted, visual acuity increased to 3/10, restoration of foveal outer retinal layer structure was documented by OCT and diminution of intraretinal fluid (Figure 5).

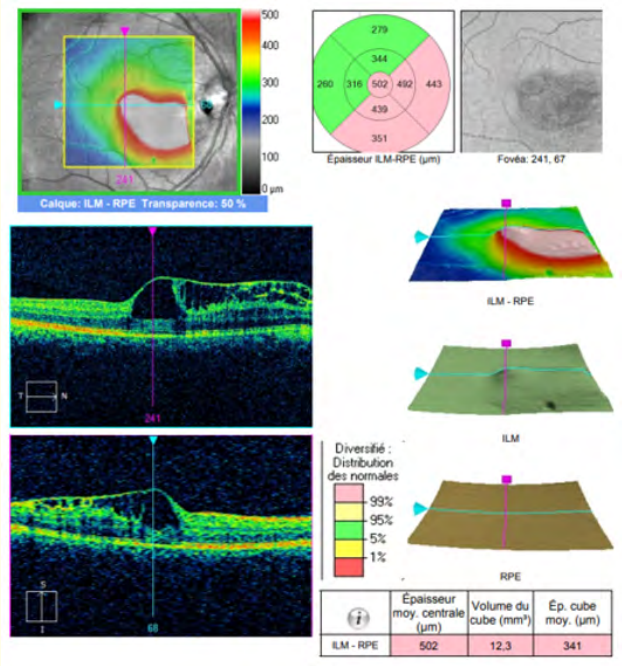

Figure 5: *Macular OCT showing the evolution of the optic disc pit maculopathy after surgery: restoration of foveal outer retinal layer structure with diminution of intraretinal fluid.*

Discussion

Optic nerve pits are a rare anomaly of the optic disc. they have typically been an incidental finding on routine dilated fundus exam. Rarely, they may cause visual field defects or maculopathy.

Histologically, an ODP is a herniation of the surrounding dysplastic retina and fibrous tissue into a collagen-rich excavation that extends into the subarachnoid space through a defect in the lamina cribrosa [3]. That creates an anomalous communication between the intraocular and extraocular spaces, a feature shared by all congenital cavitary anomalies of the optic disc [3,4].

A patient who presents with congenital optic disc pit (CODP) is at risk for developing optic disc pit maculopathy (ODPM). It is characterized by intraretinal and subretinal fluid at the macula.

Patients with temporally pits are at the greatest risk for developing serous maculopathy [11]. It occurs in patients between 30 and 40 years old [12,13].

The origin of this fluid remains unclear, and four several different mechanisms of pathogenesis have been proposed for ODPM.

The first possible source is the vitreous through the formation of a macular hole [14,15]. Several studies reported on the passage of gas or silicone oil from the vitreous cavity to the subretinal space in eyes with ODP [16,17]. However, glycosaminoglycans, which are a

component of the vitreous, were not found in the subretinal fluid in Collie dogs [18] and a direct connection between the vitreous and subretinal space through the ODP was not demonstrated by imaging or histology.

The second possible source of fluid is the cerebrospinal fluid, from the subarachnoid space through the ODP defect. Several OCT studies have shown direct communication exists between the subarachnoid space and the subretinal space [19,20].

The third possible source of fluid is leakage from blood vessels at the ODP [21]. This hypothesis was based on the finding of late hyperfluorescence at the ODP in fluorescein angiography, as well as in the area of macular elevation in eyes with ODPM [22,23]. However, some patients with ODP-M do not demonstrate this late hyperfluorescence [24].

The fourth possible source of fluid is from the choroid, through the Bruch's membrane and peripapillary atrophy [25].

These theories have led to a multitude of interventions intended to treat it. However, no consensus regarding the optimal treatment for ODPM exists.

One of the initial treatments was oral corticosteroids and acetazolamide. This treatment proved ineffective in a majority of cases and resorbed fluid tended to reappear following discontinuation of treatment [26,27].

Others reports described treating ODPM with argon laser photocoagulation at the temporal disc margin. Though laser photocoagulation provides a strong chorioretinal barrier that would prevent the passage of fluid from the optic pit to the underlying subretinal space [28]. The time for improvement was often long [29,30] and the location of the laser treatment could also cause significant visual field defects.

Intravitreal gas injection has been proposed as a treatment option for ODPM. The pneumatic displacement will cause reattachment of the macula [31].

This technique was used in small series, other series used the combination of intravitreal gas injection and laser photocoagulation temporal to the disc. The results are summarized in table 1.

Studies	Technique	Results
Lincoff., *et al*. [31]	Intravetreal gas injection C2F6	Initial improvement but later recurrence.
Akiyama., *et al*. [32]	Intravetreal gas injection SF6	Retinal reattachment was only achieved in about half of the cases.
Lei., *et al*. [33]	Intravetreal gas injection with laser photocoagulation C3F8	Improvement and reduction in fluid in all eyes, and complete resolution of intraretinal and subretinal fluids in 75% of eyes.

Table 1: *A summary of the results of studies treating the ODPM with intravitreal gas injection.*

Another alternative approach for these patients is macular buckling surgery. It includes a scleral sponge at the posterior aspect of the globe behind the macula along the 6 o'clock to 12 o'clock meridian creating a buckling effect under the macula [34]. Its complexity has prevented it from gaining much popularity [35].

The majority of the published literature on ODPM favors pars plana vitrectomy (PPV) as the treatment of choice. PPV is performed, with or without internal limiting membrane peel, with or without endolaser and gas tamponade [36]. One study found the use of combination PPV, laser photocoagulation and intravitreal gas injection improved VA in 90% of patients with a complete resolution of maculopathy in 70% [37].

Several reports described successful anatomical and visual restoration in patients with ODPM who underwent PPV with or without endolaser to the temporal disc margin and gas tamponade [38,39].

In our case, a pars plana vitrectomy was done with intravitreal injection of gas and peeling of the internal limiting membrane.

A visual improvement was noted, visual acuity increased to 3/10, restoration of foveal outer retinal layer structure was documented by OCT and diminution of intraretinal fluid.

the internal limiting membrane (ILM) peeling is another controversial surgical method. Although many reports have advocated its use, the benefits of this surgical technique remain debatable. Few cases of PPV, ILM peeling and tamponade with gas or air have been reported to achieve successful resolution of ODPM [40,41].

Some authors have proposed that in addition to inducing PVD, the surgeon should look for any glial tissue overlying the ODP and carefully peel it off during surgery. In one report on nine eyes undergoing PPV, laser photocoagulation and gas tamponade. ODPM resolved in six out of six patients in which the glial tissue surrounding the optic pit was removed. It only resolved in two out three patients in which the glial tissue was not removed [42].

Additionally, there are several research that suggested sealing of the ODP during surgery to prevent passage of fluid into the intraretinal and subretinal spaces can prove beneficial [43].

Recently, researchers have proposed a new surgical method in ODPM treatment: the creation of inner retinal fenestrations temporal to the optic disc. This technique is based on the theory that there is a pressure gradient pushing fluid from the inner retina into the submacular space and this creates retinal fenestrations that allow for the diversion of fluid back into the vitreous [44,45].

Conclusion

The ODP is a rare congenital anomaly. The main complication is the serous maculopathy.

The mechanism of pathogenesis or the origin of the subretinal fluid been reached. This makes it difficult to determine the optimal surgical technique. There are no established guidelines for the treatment of ODPM.

Typically, PPV is the treatment of choice for ODPM with or without juxtapapillary retinal laser photocoagulation, gas tamponade and ILM peeling.

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