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Macular Hole of the Left Eye in a 41-year-old Patient with Retinitis pigmentosa. A Case Report

Bożena Kmak[1], Tomasz Siewierski[1], Anna Szot[1], Sebastian Sirek[2, 3]

[1] Students' Scientific Society, Department of Ophthalmology, Faculty of Medical Sciences in Katowice, Medical University of Silesia in Katowice, Poland
[2] Department of Ophthalmology, Faculty of Medical Sciences in Katowice, Medical University of Silesia in Katowice, Poland
Head: Professor Ewa Mrukwa-Kominek, MD, PhD
[3] Kornel Gibiński University Clinical Centre in Katowice, Poland
Head: Professor Ewa Mrukwa-Kominek, MD, PhD

Summary: Retinitis pigmentosa is characterized by degeneration of the photoreceptors or retinal pigment epithelium and causes progressive vision loss. The disease can lead to night blindness, reduced field of vision and finally to complete loss of vision. The report describes a case of a patient diagnosed with retinitis pigmentosa who was admitted to hospital for further diagnosis and treatment. For several months, the patient reported a gradual decrease in visual acuity, especially in the left eye and visual impairments in poor lighting. Retinitis pigmentosa is a genetic disorder, therefore genetic counseling and screening of family members for retinitis pigmentosa is important. The specific pharmacological treatment is still unknown. In severe cases posterior vitrectomy is performed as a basic method of curing macular holes.

Key words: retinitis pigmentosa (RP), macular hole (MH), pars plana vitrectomy (PPV).

Introduction

Retinitis pigmentosa (RP) can be inherited in an autosomal dominant, autosomal recessive or X-linked manner. The incidence of the disease is 1 in 4000 people [1, 2]. Retinal dystrophy usually begins with damage to the peripheral photoreceptors and progresses towards the macula and fovea. Symptoms of the disease can appear in childhood or adulthood depending on the mode of inheritance. The natural course of RP causes 4–12% visual field loss per year [3]. Typical symptoms include abnormal pigmentation of the peripheral retina, pallor of the optic disc, and retinal vasoconstriction. Cataracts and cystic macular edema may also be noted [4]. In diagnostics, we use the following tests: visual field examination using a kinetic perimeter, macular optical coherence tomography (OCT) and fundus examination [5, 6].

The paper highlights the advances in experimental medicine and genetic counseling as important aspects in the treatment and early detection of the disease and inheritance by offspring.

Case report

A 41-year-old patient was referred to the Outpatient Clinic of Ophthalmology of the Medical University of Silesia Prof. K. Gibiński SUM in Katowice in October this year. The patient was diagnosed with retinitis pigmentosa at the age of 13. The purpose of the admission was diagnosis and treatment. For several months, the patient reported a gradual decrease in visual acuity, especially in the left eye and impairments of vision in poor lighting. His best corrected visual acuity (BCVA) was 5/5 in the right eye and 5/16 in left eye (Snellen charts, metric scale). The measured intraocular pressure was at 18 mmHg in the right eye and 17 mmHg in the left eye.

Fundus examination revealed a retinal arterial vasoconstriction, diffuse pigmented peripheral retinal lesions in the form of bone cells, waxy pallor of the optic disc, epiretinal membrane in the right eye, and macular hole (MH) in the left eye (Fig.1). Optical coherence tomography showed the epiretinal membrane of the right eye with a central retinal thickness of 288 μm and a lamellar MH of the left eye. One possible cause is a macular cyst (Fig. 2). Static visual field examination revealed severely narrowed, telescoped vision in both eyes (Fig. 3). The patient had no chronic diseases or addictions.

Due to the complexity of the clinical picture threatening the loss of the functional level of vision, close observation, regular ophthalmological check-ups together with an OCT examination of the macular area were recommended. The decision on pars plana vitrectomy (PPV) was postponed.

Discussion

In the clinical picture of RP, various types of macular holes can be observed, which may have a significant impact on the visual function of patients. Full and partial-thickness holes in the macula may be the result of retinal degeneration, loss of photoreceptor cells and structural changes in the retinal vascular layer. These changes may lead to the formation of full- and partial-wall holes, which may be observed during fundoscopy examination. Imaging

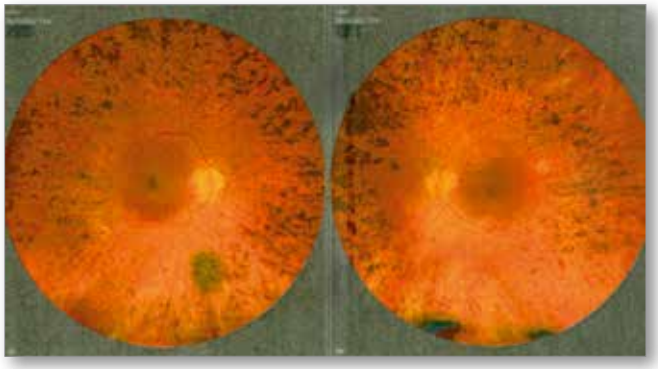

Fig. 1. Fundus examination – retinal arterial vasoconstriction, diffuse pigmented peripheral retinal lesions in the form of bone cells, waxy pallor of the optic disc, epiretinal membrane in the right eye, and macular hole in the left eye.

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studies such as OCT can provide detailed information on the characteristics of the holes and their impact on visual function [7, 8].

Full- and partial-thickness holes differ mainly in their structure and clinical characteristics. Full-wall holes are characterized by a complete lack of pigment tissue, which means that there are no layers of photoreceptor cells within the hole. Due to the lack of this tissue, they may cause greater visual disturbances, e.g. loss of central vision. It is often observed in advanced cases of RP, where the degeneration of retinal cells is more advanced and covers larger areas. Partial-wall holes may occur at different stages of the disease, both in its early and advanced stages they require monitoring, but may be easier to treat or may even not require surgical intervention at all, especially if they do not cause significant clinical symptoms [1, 9].

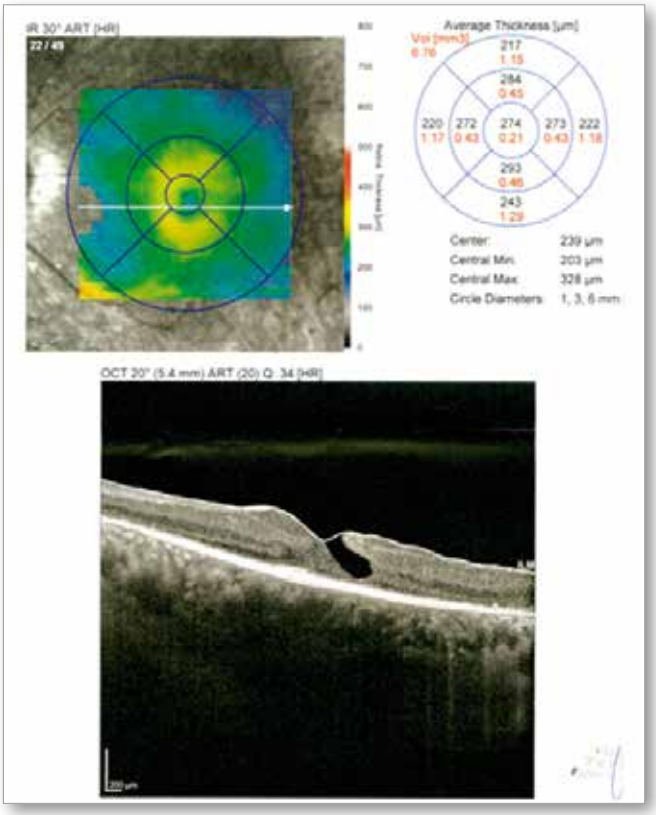

Fig. 2. Optical coherence tomography shows a macular hole of the left eye.

Retinitis pigmentosa is still a huge challenge when it comes to effective and applicable treatments with most patients being limited to supportive care only which is often inadequate [2]. However in recent years there have been a number of advancements in stem cell and gene therapy that could change the way this disease is approached by clinical specialists.

There are currently 131 drugs in clinical development for RP, with around 50% considered to be ATMPs (advanced therapy medicinal products). These therapies represent a highly important advance for the treatment of RP, with the potential for a curative effect and a significant reduction in the burden of disease [10]. The clinical application of these drugs is diverse, with several types in development which are suitable to treat RP patients across a different stage of the disease.

Gene therapies are likely to be applied in earlier stages of disease since in advanced disease, target photoreceptors will have largely degenerated and conventional gene therapy (to silence or mutate a gene) will no longer be effective [11]. Therapies utilizing stem cells in contrast are not reliant on the presence of remaining, viable photoreceptors, can work independently of gene mutation, and could be used throughout the course of the disease [12].

With most diseases prevention is equally, or even more important than the treatment itself. However with RP being an inherited disorder there are no known ways of its effective prevention [12].

Therefore, if one has a family history of retinitis pigmentosa it is important to contact a genetic counselor. That may present a challenge for a potential patient, as referrals to genetic specialists can take months if not years in some cases, leading to potential progression of the disease without proper diagnosis in the earlier stages [13].

A macular hole is a retinal defect located in the center of the fovea, causing significant vision impairment. The accumulation of vitelliform material induces chronic separation of photoreceptors from the retinal pigment epithelium layer, causing progressive

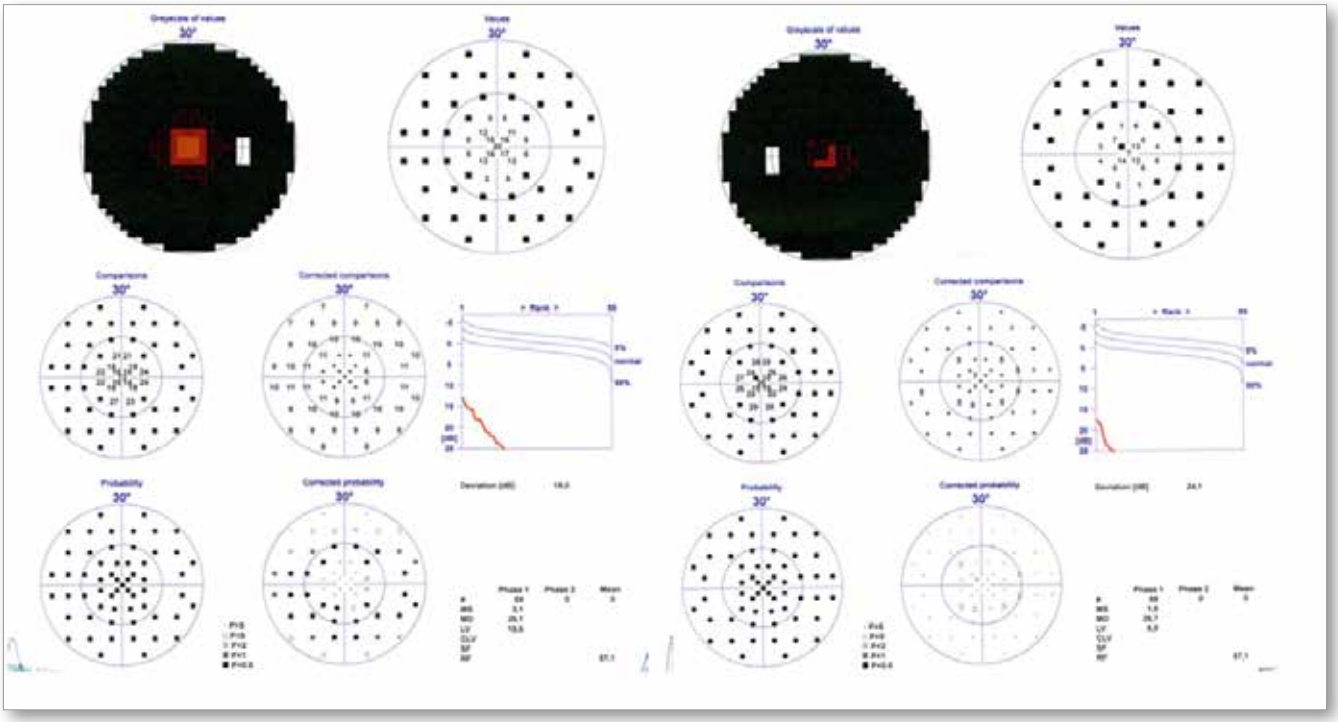

Fig. 3. Static visual field examination – severely narrowed, telescopic vision in both eyes. Static perimeter Octopus.

macular atrophy. The consequent retinal thinning is not only the cause of central vision loss that characterizes the natural history of the disease but also the presumed mechanism of MH development in these eyes [14]. The standard form of treatment with a high chance for anatomical closure (85–100%) is a vitrectomy. Pars plana vitrectomy is a commonly employed surgical technique that allows ab-interno access to the posterior segment of the eye. The procedure consists of partial or extensive vitreous removal, followed by the required retinal surgery [15]. However, it is questioned whether such a procedure is always necessary.

We know for example that in some cases stage I MH can resolve spontaneously [16].

In the case of our patient, taking into account his narrowed telescopic field of vision with preserved visual acuity along with the fact that the MH in his left eye is lamellar, the decision was made not to intervene surgically but to keep the patient under strict observation.

Conclusions

Retinitis pigmentosa is a diagnostic challenge for ophthalmologists. Counseling and testing of family members are important in prevention of RP. Due to the lack of causal treatment, the disorder is progressive. In the final stage of the disease, the level of vision is limited to the telescope field with a high level of visual acuity. The basic method of treating macular holes is vitrectomy. The macular hole in a patient with end-stage pigmentary degeneration poses a diagnostic and clinical challenge and is associated with a huge risk of irreversible loss of vision.

Disclosure
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Contact and reprint request:
Bożena Kmak, MD (e-mail: bozena.kmak@wp.pl)
Students' Scientific Society, Department of Ophthalmology, Faculty of Medical Sciences in Katowice, Medical University of Silesia in Katowice
35 Ceglana, 40-055 Katowice, Poland