Optical Coherence Tomography (OCT) Diagnostic of Retinitis Pigmentosa - Case Study

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ABSTRACT
Background: Retinitis pigmentosa (RP) is a set of inherited rod-cone degenerative diseases that clinically presents with similar signs and symptoms. Mutations in one of more than 70 genes are involved. Patients will commonly present with bone-spicule pigment formation, waxy optic nerve pallor, and attenuated blood vessels in the posterior pole. Symptoms often begin with progressive night blindness, mid-peripheral visual field defects, and eventual tunnel vision. Central vision loss will ultimately occur following loss of rod function. Complete blindness is uncommon. Objective: The aim of this article is to present two cases of retinitis pigmentosa (mother and daughter) through ophthalmologic exams in our clinic. The next aim is to show how to manage a low vision service and to treat cystoid macular oedema as a complication of retinitis pigmentosa. Methods: All medical reports are shown in this article. Every diagnostic tool as well as report is a part from our archived history of the patients and has been thoroughly analysed. We also reviewed available literature using the key words retinitis pigmentosa, cystoid macular oedema, gene therapy. Case presentation: A 38 year old female patient for a low vision consultation. The patient was legally blind secondary to retinitis pigmentosa, which was diagnosed in her late 20s. She reported gradually progressive hazy central vision and decreasing peripheral vision in both eyes as well as severe night blindness. Other than the diagnosis of retinitis pigmentosa in both eyes, the patient had no other remarkable ocular conditions. Findings at that visit included unaided distance visual acuities VOD: 0,04 VOS: 0,06. Pupils were round with brisk responses. Extraocular muscle motility was full in both eyes. Confrontation visual fields were noted as temporal loss in the right eye and superior and temporal loss in the left eye. The perimetry test could not be performed due to the lack of correspondence of the patient even after a couple repetitions of the perimetry. She had normal ocular adnexa and quiet lids, conjunctiva, and sclera in both eyes. Corneas in both eyes were noted as clear epithelium, clear stroma, and clear endothelium. Anterior chambers had normal depth, irises with no pathological findings in both eyes; lens incipient sclerotic. Intraocular pressures were noted as 22 mmHg in both eyes with Icare, 21mmHg and 19 mmHg with applanation tonometry; pachymetry corretional factor was +1 on both eyes. The vitreous was clear in both eyes. Both optic nerves were measured as 0.4 cup-to-disc ratios with no disc edema, disc hemorrhages, notching, or thinning noted. Waxy disc pallor and attenuated blood vessels were observed in both eyes. The macula in both eyes had retinal pigment epithelium (RPE) changes with no edema or hemorrhages. Bone spicule changes were noted 360 in the periphery of both eyes with no holes or tears(Figure 1a+1b+1c+1d), Conclusion: We presented two cases of retinitis pigmentosa – the mother with diagnosed RP more than 15 years ago in need for low vision rehabilitation service and the daughter that got diagnosed after our initial examination and with complications in visual impairment through cystoid macular oedema. Keywords: Optical Coherence Tomography (OCT) Diagnostic of Retinitis Pigmentosa.

1. BACKGROUND
Retinitis pigmentosa (RP) is a set of inherited rod-cone degenerative diseases that clinically presents with similar signs and symptoms. Mutations in one of more than 70 genes are involved. Patients will commonly present with bone-spicule pigment formation, waxy optic nerve pallor, and attenuated blood vessels in the posterior pole. Symptoms often begin with progressive night blindness, mid-peripheral visual field defects, and eventual tunnel vision. Central vision loss will ultimately
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occur following loss of rod function. Complete blindness is uncommon. There is currently no cure for RP. Treatment is primarily aimed at slowing progression of the disease that include interventions for secondary complications, retinal prosthesis implants, and gene therapy. RP is one of the most common causes of severe vision impairments and can significantly affect daily life, impacting activities such as driving, reading, and mobility. Low vision rehabilitation services are available to improve visual function through devices, to assess safe mobility, and to connect patients with resources to maintain levels of independence (1).

2. OBJECTIVE
The aim of this article is to present two cases of retinitis pigmentosa (mother and daughter) through ophthalmologic exams in our clinic. The next aim it to show how to manage a low vision service and to treat cystoid macular oedema as a complication of retinitis pigmentosa.

3. MATERIAL AND METHODS
All medical reports are shown in this article. Every diagnostic tool as well as report is a part from our archived history of the patients and has been thoroughly analysed. We also reviewed available literature using the key words retinitis pigmentosa, cystoid macular oedema, gene therapy.

4. CASE PRESENTATION
An optical coherence tomography (OCT) scan showed an ischaemic macula and no evidence of macular edema or subretinal fluid in both eyes (Figure 2). The OCT of the optic disc showed thinning of the GCL+IPL (Figure no 3). The patient reported a history of testing in her late 20s in Russia when she was diagnosed with retinitis pigmentosa (amnestic informations with no documentations). She did not recall the name of the test or doctor who performed the test. From her description, an electroretinogram (ERG) was most likely performed, which she reported resulted in positive findings for retinitis pigmentosa. The patient was also mentioned receiving injections in Russia "in the eye" that were as explained for treatment of the condition. The patient’s younger daughter accompanied her to the low vision consultation. Knowing that this is a hereditary disease, she is also interested in an exam. The mother regarded herself as a very independent woman and reported living alone with no husband and taking care of the daughter. She was fully aware of the condition and her main goal for low vision rehabilitation was reading and a desire to be fit with a stronger reading prescription. She also wants to show her daughter that it is in fact possible to maintain everyday life with handing all the daily tasks with no big help. She was currently using several different pairs of reading glasses, all of which she found unhelpful. At the low vision examination, entering unaided distance visual acuities were 0.04 in the right eye and 0.06 in the left eye. Near visual acuities with her preferred +2.50 diopter(D) readers in both measured with single letters at 15 centimeters(cm). The patient was allowed to hold the near chart at any distance to evaluate her preferred working distance. With both eyes open...
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using the +2.50D readers, the patient read with difficulty. The patient initially reported that she preferred using the +2.50D readers combined with an addition +2.00 D for reading. However, when tested with this, the patient was unable to hold the material at the correct distance and struggled to read. A trial frame refraction did not result in an improvement in visual acuity in the left eye but did result in an objective and subjective improvement in the right eye to 0.1 with +2.0 -1.50 x 075. Full-time wear of single vision distance glasses with polycarbonate lenses were recommended for improved clarity of vision and protection. A prescription for glasses was released to the patient. The daughter mentioned seeing her falling from the stairs and hitting object while walking, therefore orientation and mobility training was discussed and strongly advised to the patient for safe navigation, and a possibility of using a cone while walking. The patient was receptive to the recommendation as she was aware of her limitations due to reduced peripheral and central vision. Private orientation and mobility instructors were also discussed, but financially difficult for the patient to pursue. We also decided to test the mother for the RPE65-mediated retinal dystrophy so she could be treated with Luxturna, but after the genetic testing performed in „Sveti dub“ clinic in Zagreb, the mother was a heterozygous for PRPF3 c.1481>T.p. (Thr495Met), which is patogenic and a heterozygous for PROM1 c.2401G>T.p.(Ala801Ser) which is a variant of uncertain significance (VUS), and not a candidate for the above mentioned treatment. Findings for the patients daughter at the examination: VOD:1.0 cc -1,0 d sph -0,75 dcyl axis 75, VOS: 0,7 cc -1,0 d sph -1,0 dcyl axis 87; Pupils were round with brisk responses. Extraocular muscle

Figure 3. OCT of the patient no 1 (mother)–Optopol Revo nx 130

Figure 4a, 4b, 4c and 4d. Foto Fundus + autofluorescence of the patient No 2 (daughter)–Centrevue Eidon FA
motility was full in both eyes, optic disc round with no visible
oedema but seemingly waxy C/D 0.4, macular field with vis-
ible foveal reflex, thinning of the retinal periphery with low pig-
mented atrophic zones especially visible in the mid periphery
(Figure 4a+4b+4c+4d). We decided to send the patient for
an ERG where a retinal cell demage was detected, which is
reflected in the reduction of amplitude and duration of the b
wave especially pronounced in the late adapted 3.0 flicker
response. After reviewing the OCT images of the patient’s
macula and finding that there was cystoid macular edema
in the outer plexiform layer, we decided to apply Triamcino-
lone acetate subtenonialy with a positive response subjectivly
and clinicaly (Figure 5, Figure 6). After reviewing the OCT
of the optic dics (Figure 7) and aplanation tonometry (TOD
23mmHg TOS 20 mmHg with no pachymetry correcting
factor) we prescribed brinzolamide solution and educated the
young patient about the proper application. We put an
emphasis on the proper education of the daughter and mental
health, as well as the importance of regular follow ups.

5. DISCUSSION
Retinitis pigmentosa (RP) is a disease that affects to rod
photoreceptors and subsequent loss of cone function in both
eyes. There are more than 3,000 genetic mutations in ap-
proximately 70 known genes that are associated with R.P. (2)
This condition can present as an autosomal dominant, auto-
somal recessive, X-linked, or unknown pattern inheritance.
Approximately 2.5 million people worldwide are affected by
RP. There was no significant gender predilection found in lit-
erature. The pathophysiology of RP begins with rod photo-
ceptor degenerationand leads to cone photoreceptor de-
generation as well. Mutations can occur in many processes
involving the rod photoreceptors, ranging from rod visual
transduction to metabolism and RNA processing. Patients
with RP initially present with nytalopia as rod photorecep-
tors primarily involve vision in dim illumination. Symptoms
are initially noticed in dim light settings and commonly begin
during adolescence. In advanced disease, patient will have
central vision loss in daylight as the cones degenerate further.
Eventually, all photoreceptors will be lost, leading to complete
blindness. Fortunately, complete blindness was found to be
rare in past studies. However, most patients with RP are clas-
sified as legally blind (20/200 or worse in the better seeing eye
or 20 degrees field or less in the better seeing eye) by the age
of 40.(3) One common cause of vision loss is cystoid macular
edema(CME). The mechanism of CME in RP is incompletely
understood with several theories proposed. One hypothesis
suggests that because RP is associated with an increased prev-
ance of anti-retinal antibodies, CME is the result of an au-
toimmune reaction. This autoimmune phenomenon may ex-
plain the efficacy of steroids in the treatment of CME (4) An
alternative hypothesis proposes that dysfunction of the outer
blood-retinal barrier at the RPE, perifoveal capillary plexus,
or both increases vascular permeability causing fluid leakage
in the macula. Carbonic anhydrase inhibitors may treat CME
by enhancing the pumping mechanism at the level of the
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RPE.(5) In addition to its anti-inflammatory properties, steroids may also work at the level of the blood-retinal barrier in RP as it has been shown experimentally to reduce breakdown of the blood-retinal barrier in diabetic retinopathy.

Diagnosis of RP is often based upon characteristic appearances, but ancillary testing can also be performed. A full-field or multifocal electroretinogram (ERG) is often performed to confirm diagnosis. The scotopic a- and b-wave amplitudes measuring mainly rod function are reduced in all types of RP and photopic (cone) b-wave amplitudes are gradually reduced as well. Gene therapy for retinal degenerations has been an area of great research. Luxturna, voretigene neparvovec (AAV2-hRPE65v2), is the first U.S. FDA-approved gene therapy for RPE65-mediated retinal dystrophy. Positive outcomes have included improved light sensitivity, visual fields, and mobility in dim lighting. It carries an adeno-associated virus vector containing the human RPE65 complementary DNA that is injected subretinally. Inclusion criteria consist of patients with visual acuity 20/60 or worse or visual field less than 20 degrees.(6) Patients also need a confirmed genetic diagnosis of biallelic RPE65 gene mutations as the cause of RP. Unfortunately, RP associated with RPE65 mutation has been found to be rare, approximately 2% of autosomal recessive RP.(6)

Low vision rehabilitation services are crucial for patients in all stages of RP. In early stages, proper education about the disease symptoms and prognosis is critical. Keeping patients independent with low vision rehabilitation helps maintain confidence levels, emotional health, and satisfaction with life.

6. CONCLUSION

We presented two cases of retinitis pigmentosa – the mother with diagnosed RP more than 15 years ago in need for low vision rehabilitation service and the daughter that got diagnosed after our initial examination and with complications in visual impairment through cystoid macular oedema. The aim of the review was to emphasize the obligatory education of the patient for the now and in the future possible treatments as well as helping the patient to maintain a full and independent lifestyle.

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REFERENCES


Figure 7. OCT of the optic disc of the patient No 2 (daughter)–Optopol Revo nx 130