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CASE REPORT

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Retinal detachment in chorioretinal coloboma – a challenge for the vitreoretinal surgeon

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ABSTRACT
Chorioretinal coloboma is a rare, congenital condition of the eyeball. This anomaly of the eye is a result of improper differentiation of eyeball structures during the embryonic and fetal period. This malformation gives a high risk of other ophthalmic diseases such as choroidal neovascularization, glaucoma, cataract or – the most serious and the most common from all of these – retinal detachment. Proper and quick diagnostics, urgent treatment and cooperation with experienced vitreoretinal surgeons are necessary and can improve the prognosis. We present a clinical case report of retinal detachment in 23-year old patient with chorioretinal coloboma, hospitalized in the Department of Ophthalmology in University Clinical Centre in Gdansk, treated by *pars plana* vitrectomy with endolaser therapy and silicone oil endotamponade.
KEY WORDS: retinal detachment, vitrectomy, coloboma, chorioretinal coloboma

INTRODUCTION
Chorioretinal coloboma is a rare congenital developmental anomaly of the eye (Figure 1). It affects 0.14% of the population [1]. It results from abnormalities in the process of optic cup morphogenesis during the closure of the embryonic fissure [2]. Chorioretinal coloboma is characterized by the absence of part of the retinal pigment epithelium and choroid, with the presence of abnormally thin neural retina (Figure 2). The etiology of the condition involves genetic, infectious, teratogenic and environmental factors [3].

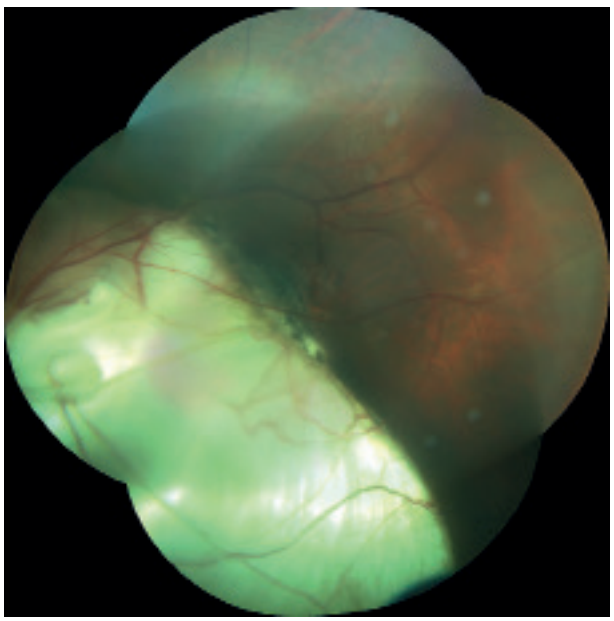

Figure 1. Chorioretinal coloboma of the left eye

This developmental anomaly of the eye may coexist with other fetal anomalies including microphthalmia and anophthalmia. It can also occur in association with genetic syndromes such as COACH (**c**erebellar vermis aplasia, **o**ligophrenia, congenital **a**taxia, **c**oloboma, and **h**epatic fibrosis), CHARGE syndrome (**c**oloboma, **h**eart defects, choanal **a**tresia, **r**etardation of growth and psychomotor development, **g**enital and/or urinary abnormalities, and **e**ar abnormalities or deafness), Joubert syndrome (cerebello-oculo-renal syndrome), cat eye syndrome, or tuberous sclerosis [4, 5]. Chorioretinal coloboma has also been reported in lethal anomalies including branchio-oculo-facial syndrome (BOFS), Fraser syndrome (cryptophthalmos, organ malformations mainly in the respiratory and urinary tracts), and others [6, 7]. Coloboma on the fundus most typically involves the inferior nasal quadrant of the retina and choroid, as this area of the embryonic fissure is the last to close during fetal development [8]. The changes also affect other areas of the eyeball.

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The most commonly used classifications of coloboma are those proposed by Ida Mann (Table I) and Lingam Gopal (Table II), based on the anatomical location of fundus changes secondary to coloboma in relation to the location and morphology of the optic disc [9].

Chorioretinal coloboma (Figure 3) is associated with the risk of ocular complications including cataract, secondary glaucoma, amblyopia, nystagmus, strabismus, and subretinal neovascularization [8]. Patients with coloboma are also at an increased risk of developing rhegmatogenous retinal detachment. The reported prevalence of this complication ranges from 8% to 50% [10]. The condition requires urgent surgical treatment. The management of retinal detachment in chorioretinal coloboma, which is devoid of pigment epithelium, is a major challenge for ophthalmologists, and requires experience in vitreoretinal surgery.

CASE REPORT

A 23-year-old male patient with iris and chorioretinal coloboma in both eyes, additionally complicated by nystagmus, divergent strabismus and amblyopia in both eyes, presented to the Department of Ophthalmology at the University Clinical Center (UCK) in Gdansk with a sudden deterioration of vision in the right eye.

Table I. Ida Mann's classification

Type	Ida Mann's classification
1	Coloboma extending above the optic disc
2	Coloboma extending up to superior border of disc
3	Coloboma extending below the lower border of disc / separated from the disc by a narrow retinal area
4	Coloboma involving the disc only / isolated optic disc coloboma
5	Coloboma present below the disc with normal retina above and below the coloboma
6	Pigmentation present in the periphery
7	Coloboma involving only the periphery

Table II. Lingam Gopal's classification

Type	Lingam Gopal's classification
1	Normal disc outside fundus coloboma
2	Disc outside the fundus coloboma and abnormal
3	Disc outside the fundus coloboma and independently colobomatous (peripheral coloboma + independent disc coloboma)
4	Disc within the fundus coloboma and normal
5	Disc within the fundus coloboma and colobomatous
6	Disc shape not identified but blood vessels seen emanating from the superior border of the large fundus coloboma

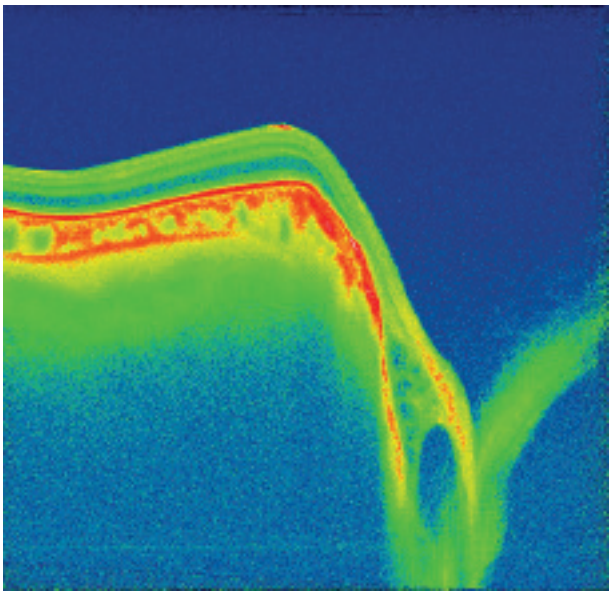

Figure 2. OCT

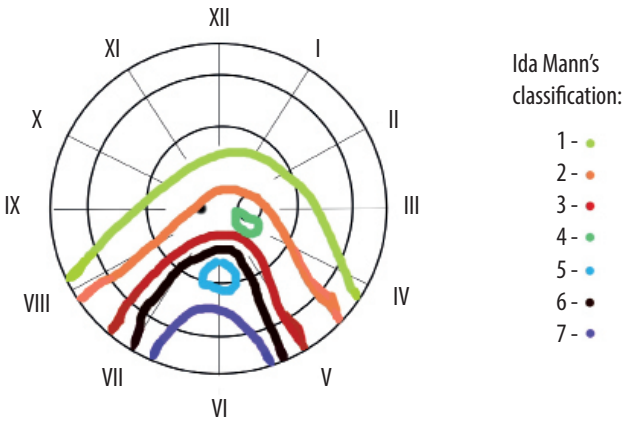

Figure 3. KIda Mann's classification of coloboma

Ophthalmologic examination revealed rhegmatogenous retinal detachment in the right eye. On admission, the best corrected visual acuity (BCVA) in Snellen charts was 0.08 in the right eye, and 0.125 in the left eye. The man reported that his vision in both eyes was similar during childhood. His medical history revealed craniofacial trauma suffered a few months before, and surgical intervention to remove congenital cataract in both eyes along with IOL implantation in the posterior chamber performed over six months previously in a different medical center. Ophthalmoscopic examination was difficult because of poor mydriasis and nystagmus. The visible area of the fundus in the right eye was dominated by a pale cavity of the choroidal coloboma, involving the posterior pole together with the optic nerve, and a bulla of detached retina located in the superior temporal quadrant. The patient was referred for urgent vitrectomy.

23-gauge *pars plana* vitrectomy of the right eye was performed under general anesthesia. During the procedure, extensive chorioretinal coloboma was visualized, involving the optic disc, the macula, and the inferior nasal retinal sector. The bulla of detached retina involved primarily the superior temporal sector and did not communicate with the posterior pole of the retina. A macular hole was identified in the choroidal coloboma cavity within the thin parchment-like retina (Figure 4). The routine procedure of perfluorodecalin administration into the vitreous chamber was not performed because of severe deformation of the posterior wall of the eyeball and the macular hole. To drain subretinal fluid, a controlled opening was made in the temporal retinal sector outside the coloboma (Figure 5). During fluid-air exchange, complete retinal reattachment was achieved. Also, an attempt was undertaken to drain residual fluid from the area superior to the macular hole. The peripheral retinal tear was secured with laser impacts (Figure 6). Because of nystagmus, endotamponade with silicone oil with a viscosity of 5000 centiStokes (cSt) was performed. The intra- and postoperative course was uncomplicated. The patient was discharged and instructed to take prescribed medications (diclofenac, 1% tropicamide and tobramycin/dexamethasone) postoperatively 4 times a day, and to position the head face down with a tilt to the left side for a period of 14 days.

The first postoperative follow-up assessment was scheduled two weeks after vitrectomy. Visual acuity in the right eye was found to have improved to 0.125 BCVA. An elevated intraocular pressure of 43 mmHg was observed in the right eye. Consequently, hypotensive treatment was started, with topical dorzolamide/timolol and brimonidine administered twice daily to the right eye, and oral 250 mg acetazolamide taken once daily for 5 days. Ophthalmoscopic evaluation revealed retinal reattachment. At the next follow-up examination (3 weeks after surgery), the intraocular pressure was found to be normalized at 12 mmHg. Topical hypotensive treatment of the right eye was maintained. Visual acuity remained stable and, as the patient reported, returned to the level before retinal detachment and was close to the visual acuity of the left eye (0.125 BCVA).

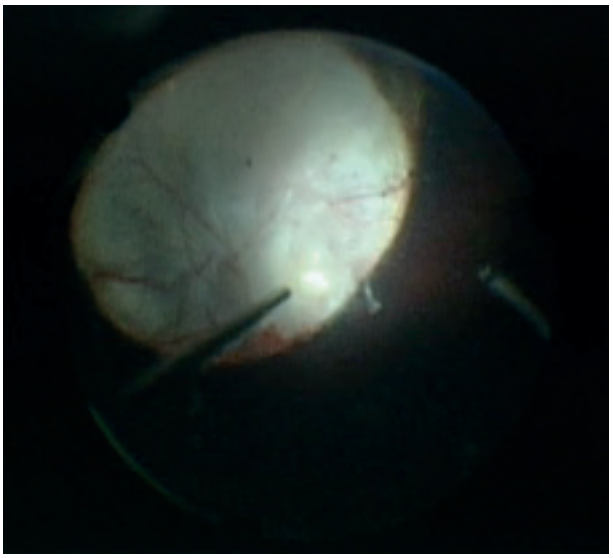

Figure 4. *Pars plana* vitrectomy

Currently, the patient remains under the care of the Retinal Outpatient Clinic at the University Clinical Center (UCK). At the last follow-up visit, which took place 12 months after the surgical procedure, slight retinal elevation was noted in the inferior retinal quadrants within the chorioretinal coloboma. Since the eye was stable, the intraocular pressure was normalized, the level of visual acuity was relatively good and constant, and the patient reported no symptoms, a decision was taken not to attempt another ophthalmic surgical intervention, and to leave the silicone oil in the vitreous chamber for as long as possible.

DISCUSSION

Chorioretinal coloboma is a congenital defect of the eye associated with a high risk of multiple complications. One of them is rhegmatogenous retinal detachment, which can lead to the loss of vision. The prevalence of rhegmatogenous retinal detachment among patients with coloboma is higher than in the general population, and estimated at 8-50% [10]. The first cases of this complication were reported by Mannhardt in 1897 and by Gilbert in 1902 [11, 12]. In 1925-1926, Wagener, Gipner and Komoto put forth the hypothesis that chorioretinal coloboma was a risk factor for retinal detachment [13].

Severe chorioretinal coloboma leading to extensive retinal detachment is most commonly associated with lethal malformations, as noted in Schubert's 2005 study [11]. In adult patients with moderately severe and isolated coloboma, retinal detachment occurs most typically in the 2nd and 3rd decades of life [11]. The case reported here is of a 23-year-old man with chorioretinal coloboma of moderate severity.

Retinal tears in the eyes with chorioretinal coloboma may be located within or outside the coloboma (i.e. in healthy tissue), or in both these areas [14]. The reported case can be classified as retinal detachment with a tear located within the area of chorioretinal coloboma.

We diagnosed the lesions as type 1 in Ida Mann's classification and type 5 according to Gopal's classification [9]. The treatment of choice in cases of retinal detachment accompanying lesions of this type is *pars plana* vitrectomy. Other available methods include the use of an encircling band, scleral buckling, laser therapy or cryotherapy.

According to Vuković *et al.* the most common and effective method for treating retinal detachment in coloboma patients is *pars plana* vitrectomy with gas or silicone oil endotamponade, which makes it easier to locate the tear and stabilize the eye [1].

Performing endolaser therapy during vitrectomy may pose a challenge. Achieving effective laser impacts in the coloboma site devoid of pigment epithelium often proves impossible. Vuković highlights that laser therapy is more effective in

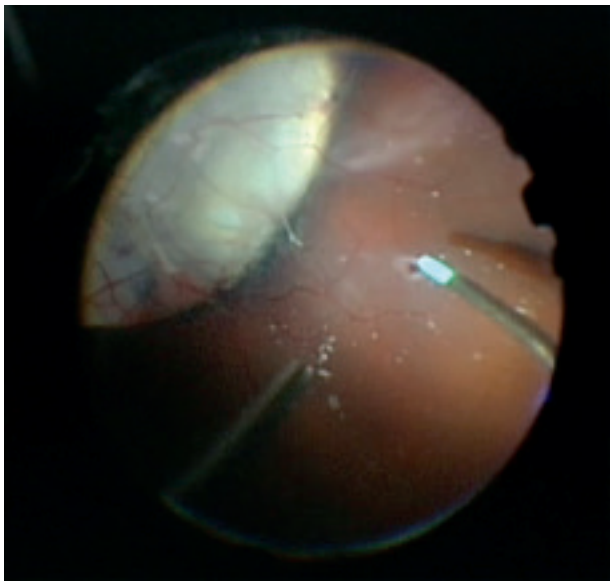

Figure 5. *Pars plana* vitrectomy

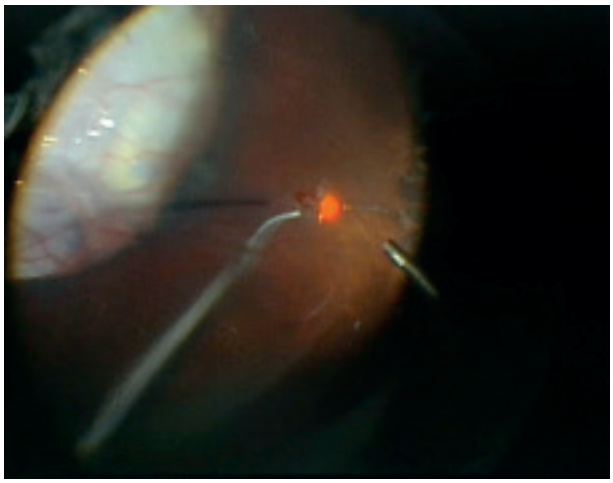

Figure 6. *Pars plana* vitrectomy

cases of tears located out of the coloboma site [1]. In our case, the opening in the retina outside the coloboma was successfully managed by laser impacts around it. Consequently, it is recommended that endolaser therapy is performed around tears located outside the anomalous areas [1, 15]. Some authors recommend 360° laser therapy of the peripheral retina and around the coloboma (at the border between healthy retina and the coloboma edge) [1, 15].

Vitrectomy as the treatment of choice for retinal detachment secondary to chorioretinal coloboma is also recommended by Hocaoglu *et al.* [16]. According to the researchers, vitrectomy with silicone oil endotamponade allows complete reattachment of the retina and prevents the recurrences of retinal detachment. Similar conclusions are reported by Ramezani *et al.* [10].

Another method of surgical treatment for retinal detachment is the use of an encircling band with scleral buckling. According to Vuković *et al*., an advantage of this method is faster patient recovery [1]. Unlike vitrectomy with silicone oil endotamponade, it is a single-stage procedure. However, given the difficulty in accurately locating retinal tears in coloboma eyes, it may prove impossible to place the scleral buckle correctly, rendering the procedure ineffective [1, 8, 17]. Furthermore, patients treated with scleral buckling surgery are exposed to a greater risk of developing vitreoretinal proliferations leading to retinal redetachment, which is also highlighted by Ramezani *et al.* [10]. Vitrectomy reduces the risk of distant complications and helps avoid repeated surgical interventions (except for silicone oil removal).

Small retinal detachment can be treated with laser therapy or cryotherapy performed on an outpatient basis. Applying these methods in patients with chorioretinal coloboma requires very good patient cooperation and precise location of tears after achieving effective mydriasis. They are most commonly indicated for localized peripheral detachment not involving the coloboma site [1, 14]. In our patient, these criteria were not met.

In the reported case of retinal detachment with chorioretinal coloboma, the treatment of choice was 23-gauge vitrectomy with 5000 cSt silicone oil endotamponade. During vitrectomy, it was possible to accurately locate the retinal hole, which could not be done by slit lamp examination (nystagmus, poor mydriasis). Gas tamponade was not considered to be an appropriate technique because nystagmus would have made it impossible for the patient to maintain proper eye position. Consequently, an effective tamponade would not have been achieved.

Endolaser treatment was performed at the border between healthy retina and coloboma, but could not involve the retina devoid of pigment epithelium at the site of coloboma. Prophylactic peripheral 360-degree laser treatment was not done in view of the potential risk of complications including retinal fibrosis and shortening within the area of intense laser operation. In the coloboma cavity, against the white background of the sclera, macular peeling in the thin transparent retina was impossible to perform. Despite these difficulties and limitations, the postoperative outcome turned out to be satisfactory. Complete reattachment of healthy retina was confirmed. Follow-up evaluation of the operated eye showed no evidence of vitreoretinal proliferation, either epiretinally or subretinally. Also, we achieved satisfactory visual acuity in the treated eye.

After approximately a year of patient follow-up, we decided not to perform another vitrectomy to remove silicone oil from the vitreous chamber and manage the small area of retinal detachment observed in the inferior quadrants. Subjecting the patient to another surgical procedure would have carried the risk of retinal deterioration including proliferative vitreoretinopathy processes. Effective laser therapy in an area of the retina where the pigment epithelium is absent would have been difficult, with reattachment of detached retina unlikely to achieve. The risk of retinal redetachment after sili-

cone oil tamponade removal was reported by Pal and Gopal [18, 19]. Removing the silicone oil tamponade from the eye with chorioretinal coloboma – with coexisting macular hole in our case – would have been associated with a high risk of repeated retinal detachment and the development of advanced proliferative vitreoretinopathy.

CONCLUSIONS

In patients with chorioretinal coloboma, retinal detachment is an urgent ophthalmic condition associated with a high risk of vision loss. The clinical case reported above shows that *pars plana* vitrectomy with silicone oil endotamponade is the treatment of choice in patients with chorioretinal coloboma involving an extensive area of the posterior retinal pole. Furthermore, endolaser therapy and silicone oil endotamponade are recognized as the best methods for managing patients who are difficult to cooperate, also in cases of coexisting nystagmus. The choice of surgical technique and successive stages of vitreoretinal surgery in patients with chorioretinal coloboma require ophthalmic surgeons to have extensive experience and, as a consequence, have an impact on the outcome of the procedure.

DISCLOSURE

The authors declare no conflict of interest.

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