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Case report

BILATERAL UVEAL COLOBOMA IN COMBINATION WITH A SYSTEMIC SYNDROME – A CASE REPORT

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Abstract

Coloboma of the uvea is a relatively rare ophthalmic entity, which occurs as a result of a disturbance in the process of closing the embryonic optic fissure. It can be hereditary, sporadic or part of systemic syndromes. Each of the structures of the uveal tract, alone or together, unilaterally or bilaterally, can have the presence of a coloboma. In this paper, we describe the case of a 9-year-old girl with bilateral coloboma and accompanying ectrodactyly of the lower limbs, probably as part of a syndrome - Ectrodactyly-ectodermal dysplasia-cleft syndrome. The patient is regularly monitored with biomicroscopic and funduscopic examination supplemented with posterior optical coherence tomography and perimetry, with the aim of timely recognition of possible ophthalmic complications.

Keywords: coloboma, uvea, ectrodactyly, congenital malformation

Introduction

Uveal coloboma is a relatively rare congenital ophthalmic malformation, with an incidence of 0.5-0.7 per 10,000 births[1]. Iris coloboma was described for the first time in 1673 by Thomas Bartholin[2]. It is thought to occur as a consequence of dysfunction in the closure of the embryonic optic fissure during the period of 5-7 weeks of embryogenesis[3]. The word coloboma is of Greek origin meaning crippled, shortened, with a defect. It can be isolated to one uveal structure (e.g. iris) or extend through the entire uveal tract and affect the retina. It is most often localized in the lower nasal quadrant, unilaterally or bilaterally. In some cases, there is a hereditary component, mostly sporadic, but it can also be part of systemic syndromes[4]. Colobomas that affect only the iris, depending on their size, can be asymptomatic to the appearance of photophobia. It can also affect the lens and the zonular system. Posterior colobomas, depending on the size, can be asymptomatic, up to the appearance of defects in the visual field and affect vision when presented in the area of the macula or papilla. A possible complication in the presence of large posterior colobomas is retinal ablation as a result of degeneration and presence of trophic rhegmas in the area of the coloboma. Large colobomas that affect all structures of the uveal tract might disrupt the overall ocular development leading to the appearance of microphthalmia or anophthalmia[5].

Case report

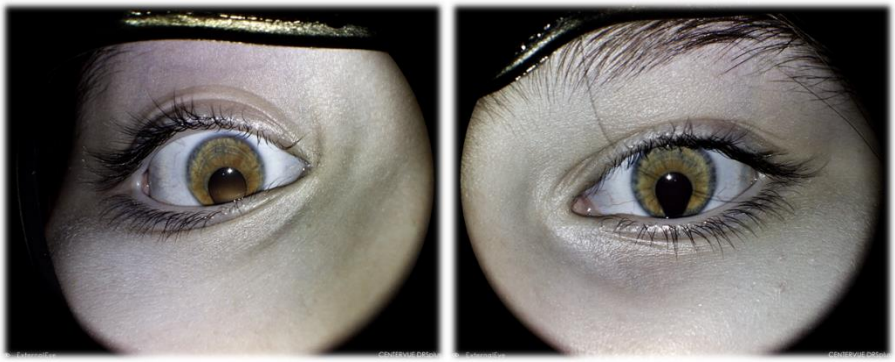

Fig. 1. Anterior segment photograph of right and left eye respectively, bilateral iris coloboma, keyhole phenomenon

A 9-year-old girl has been followed for several years at the PHI University Clinic for Eye Diseases in Skopje, due to bilateral coloboma of the uvea. For the first time, she came for an ophthalmological examination 5 years ago with the appearance of photophobia and reduced vision during a routine preschool examination. Refractometric values bilaterally showed a moderate degree of myopia, and the best-corrected visual acuity of both eyes was 6/6. In both eyes, a split of the iris was observed at 6h with a keyhole-shaped, through which ciliary zonules were observed (Figure 1). Other structures of the anterior segment were without changes.

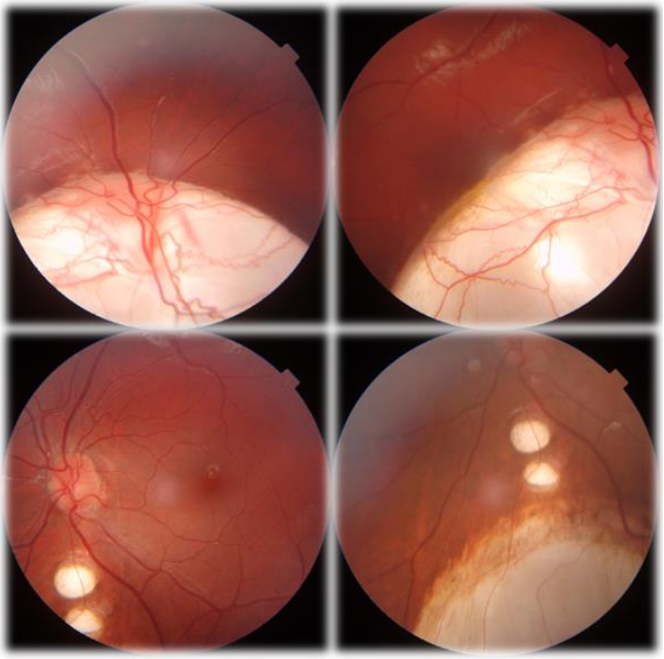

Fig. 2. Native fundus photograph of the right (top) and left eye (bottom). On the right eye, a wide zone of choroidal defect can be observed, which extends to the level of the macula, tilted disc, blood vessels clearly visible above the sclera. Clear demarcation line between healthy chorioretinal tissue and coloboma. Image below, left eye, small oval tissue defects under which a broad zone of chorioretinal coloboma with a clearly demarcated border can be seen. Papilla and macula without pathological changes

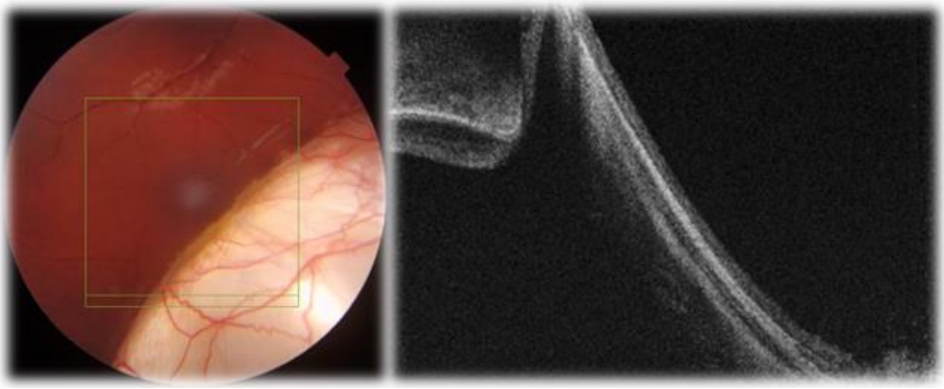

Fig. 3. Fundus photograph and OCT of the right eye. Transition zone between retina and coloboma. Staphylomatous posterior excavation, with absence of chorioretinal structures.

On indirect ophthalmoscopy of the right eye, a chorioretinal defect extending from the papilla to the periphery with visible sclera and a tilted disc was present. The border between the coloboma and the unaffected retina was clearly demarcated by a pigmented border. The defect extended temporally to the macular region, without affecting the fovea. In the left eye, two smaller choroidal colobomas of almost 1PD in size were observed next to the papilla, below which there was a wider zone of chorioretinal defect, with a rudimentary retina, increasing in diameter towards the periphery (Figure 2). An optical coherence tomography (OCT) was performed on the macula and the lesions, where a lack of chorioretinal tissues in the area of the coloboma, with staphylomatous excavation of the underlying sclera, was noted on the right eye (Figure 3). At the level of the small colobomas, in the left eye, the retinal tissue was present, rudimentary, with cystic changes in the outer layers and absence of choroid (Figure 4). The native fundus photograph showed morphological characteristics described above, and absolute scotomas were observed in the visual field in the area of the tissue defects (Figure 5).

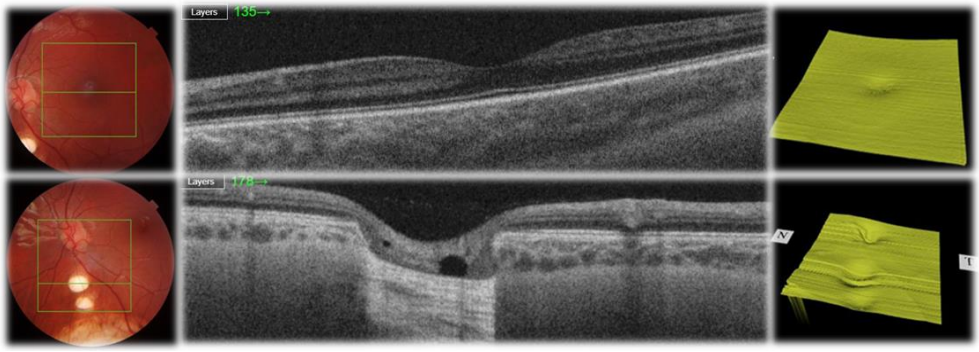

Fig. 4. Fundus photograph, OCT scan and posterior segment topography. Image above, macula with preserved foveal depression and normal topo/tomogram features. Image below, OCT through one of the colobomas. Choroidal defect, collapsed retinal layers, degenerative cystic changes in the outer retinal layers

The patient was examined by a pediatrician-dysmorphologist, during which the presence of ectrodactyly on both feet was noted. For esthetic and functional reasons, reconstruction of the fingers was performed by a plastic surgeon. In consultation with the dysmorphologist, a possible syndrome was suspected: Ectrodactyly-ectodermal dysplasia-cleft syndrome (EEC). The patient has been regularly monitored for 5 years in our Clinic, without any complications of the ophthalmological condition until the moment of publication of this paper.

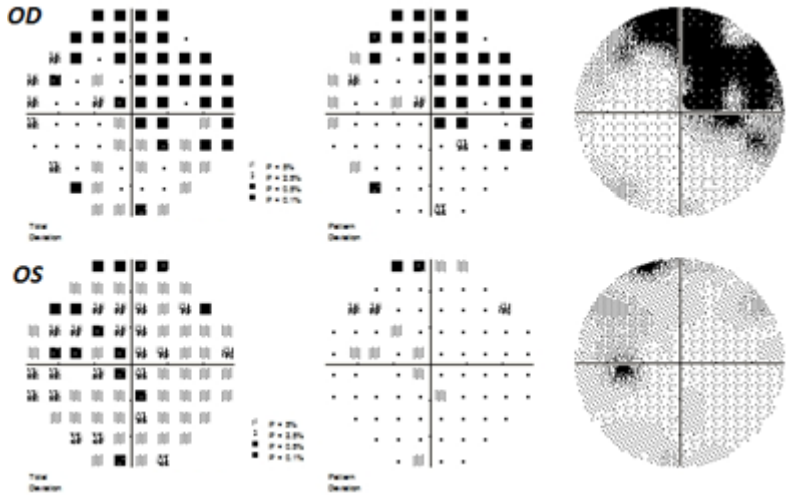

Fig. 5. Visual field (30-2) of right and left eye, respectively

Right eye-scotoma in the upper temporal quadrant that extends through the superior parts of the upper nasal quadrant (corresponds to the tissue defect in the lower sector of the right eye). Left eye - with relative scotoma in the upper temporal part of the visual field immediately next to the papilla, and absolute scotoma in the same quadrant at 30° (corresponding to tissue defects in the lower sector of the left eye).

Discussion

Uveal coloboma occurs as a result of a disruption of the complex genetic-biochemical processes that participate in the closure of the embryonic optic fissure. Epigenetic factors are also considered to be involved in the formation of this ophthalmic abnormality[3]. According to Uhumwando *et al*., the condition is somewhat more common in males[6]. It may exist alone or in association with other ocular and/or systemic abnormalities. The most commonly associated ophthalmic conditions with this tissue defect are: nystagmus, strabismus, cataract, microphthalmia, microcornea, myopia and posterior staphyloma[2]. In our case, in addition to bilateral uveal coloboma, moderate myopia and posterior staphyloma of the right eye were also present. In addition to ocular abnormalities, colobomas are also associated with various genetic syndromes such as Ascher, CHARGE or Goltz[7-9]. According to Maumenee *et al.*, almost one-third of patients with coloboma have some systemic abnormality[10]. It was also present in our case. The girl had a lack of fingers on the lower limbs - bilateral ectrodactyly. The remaining examinations by a pediatrician-dysmorphologist indicated normal psycho-physical development. Due to the presence of bilateral ectrodactyly of the lower limbs and uveal coloboma, it was considered that this case was a syndrome, of the group of ectodermal dysplasia (a large group of syndromes that are followed by embryonic dysgenesis of structures of ectodermal origin)[11].

Although coloboma is a stationary condition without the possibility of progression, depending on its localization and size, it can lead to certain ophthalmic complications. Already in embryonic development, large colobomas can cause instability of the ocular structures and affect the overall ocular development, resulting in microphthalmia or complete anophthalmia[5]. Widespread posterior colobomas affecting the papilla lead to its twisting-tilting. If they are present at the level of the macula, they significantly reduce vision and are responsible for the appearance of anisometropia, amblyopia with subsequent strabismus and nystagmus. Cases of choroidal neovascularization at the edge between healthy retinal tissue and coloboma have been described. The reason for this is considered to be a pre-existing vascular anomaly and regrouping of the retinal pigment epithelium with its cleft at the level

of the retino-colobomal junction[12]. However, the most common complication that can seriously impair vision is retinal detachment. It occurs in up to 40% of eyes with a large choroidal coloboma[6]. The cause of its appearance is supposed to be the abnormal and atrophic retinal tissue. Under the action of vitreo-retinal forces, the formation of a rhegma occurs or it is trophically created, which will result in the ablation of a sector or the entire retina. Postvitrectomized eyes with coloboma have an increased risk of reablation probably due to the absence of retinal pigment epithelium and rudimentary retinal tissue[13,14]. Most often, rhegma is formed intracolobomally in young patients and extracolobomally in adults. Therefore, patients with large posterior colobomas need annual ophthalmological controls with monitoring of the retinal periphery with indirect ophthalmoscopy, all with the aim of timely recognition of these lesions. In patients with a higher risk, prophylactic laser photocoagulation at the edge of the coloboma is recommended[15]. Our case is regularly ophthalmologically monitored twice a year with the intention of timely recognition and prevention of possible ophthalmological complications. As of the time of publication of this paper, no high-risk intra- or extracolobomal lesions are visible that would require the performance of prophylactic laser photocoagulation.

Conclusion

In this paper we have described a patient with bilateral coloboma of the uvea, a relatively rare ophthalmic condition. Regular ophthalmological examinations are important for monitoring patients with this entity in order to promptly recognize and treat possible complications, especially the occurrence of rhegma and retinal detachment. Every patient with a coloboma of the uvea should be evaluated in detail by a pediatrician-dysmorphologist to determine whether the coloboma is consistent with a systemic syndrome or not.

Conflict of interest statement. The authors declare no conflict of interest.

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