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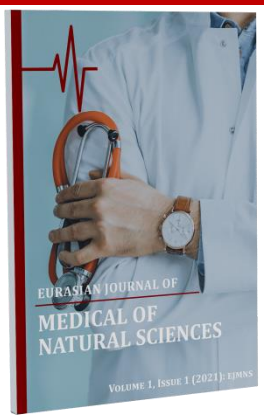

IMPROVEMENT OF SURGICAL TACTICS AND TREATMENT OF COMBINED INJURIES IN CHILDREN

Niyozbek Abdurakhmonov Khamdamjon ogli
doctoranx@gmail.com.
Gulomov Qahhorali
Assistants Ferghana Medical Institute of Public Health.
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ABSTRACT

Ocular coloboma defects are a frequent source of eye-related issues in children and can lead to notable vision problems. Recent progress in this field has mainly focused on pinpointing numerous genetic mutations associated with the condition and uncovering various molecular pathways. This has not only enhanced our knowledge of the origins of ocular coloboma but has also deepened our understanding of normal eye development. Despite these advancements, a significant portion of isolated ocular coloboma cases still lack a definitive genetic or environmental explanation.

Introduction. Ocular colobomas are a rare congenital eye abnormality characterized by a gap or hole in one or more structures of the eye, such as the iris, retina, or optic nerve. This condition can lead to various complications, including visual impairment, refractive errors, and increased risk of other eye conditions. Understanding the incidence of ocular colobomas and its associated complications is crucial for early detection, management, and prevention of potential vision-related issues. In this article, we will delve into the prevalence of ocular colobomas and explore the potential challenges and implications of this condition on ocular health. Eyelid coloboma is a rare congenital disease, which is described with the absence of a part of blepharon.

Methods. To delve into the topic at eyelid colobomas rate and its complications there were used several publications such as PubMed, ResearchGate, Scopus Elsevier, Cyberleninka and others, in order to this article be clear and more informational.

Results and discussion. In the past, the first official manuscripts discussing various types of colobomas in medical terms date back to the 17th century. For example, research on ocular colobomas was documented in a study by Aisha S. ALSomiry et al. It was noted that ocular coloboma can impact different parts of the eye such as the cornea, iris, lens, choroid, retina, and optic nerve. This condition can often occur independently without any systemic associations, and there may be noticeable differences in symmetry between the left and right eyes. The earliest mention of ocular coloboma in the iris was recorded in 1673 by Bartholin the Younger. Even today, the iris remains the most commonly affected site for ocular coloboma, typically found in the inferonasal quadrant of the eye. Iris colobomas are described

as wedge-shaped transillumination defects that can be complete (involving all layers of the iris) or incomplete (affecting only the stroma or pigment epithelium)[1]. It is evident that there are various types of congenital ocular colobomas, including eyelid colobomas that can be either congenital or acquired during a person's lifetime, often due to traumas. For example, Henry Smith and colleagues categorized congenital eyelid colobomas based on their location in their study, stating that "Eyelid colobomas are uncommon, full-thickness defects of the eyelid that are present from birth. They can be unilateral or bilateral, affecting one or both upper or lower lids, and can range from a small notch to a complete absence of the entire lid. Approximately one-third are isolated cases, while the rest are associated with other ocular and craniofacial abnormalities[2]. In addition, congenital eyelid colobomas can be categorized as isolated or associated, as detailed in the recent research by Sarah M. Jacobs et al. Eyelid colobomas are uncommon, congenital defects that involve full-thickness eyelid tissue. They can be unilateral or bilateral, impacting one or both upper or lower lids, and range from a small notch to a complete absence of the entire lid. Sample photos are provided below for better visualization.

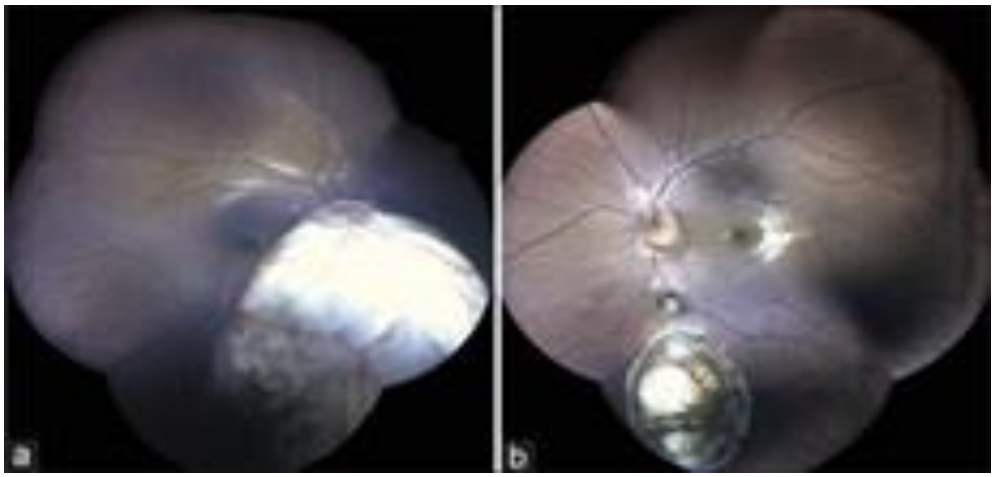

Figure 1. Here is a color fundus photograph displaying a retinochoroidal coloboma (RCC) that extends to the superior border of the optic disc. Additionally, there is another image showing multiple colobomas, including one in the inferior part of the optic disc, a small patch of RCC below the optic disc, and a larger RCC below all the colobomas with normal tissue in between[3].

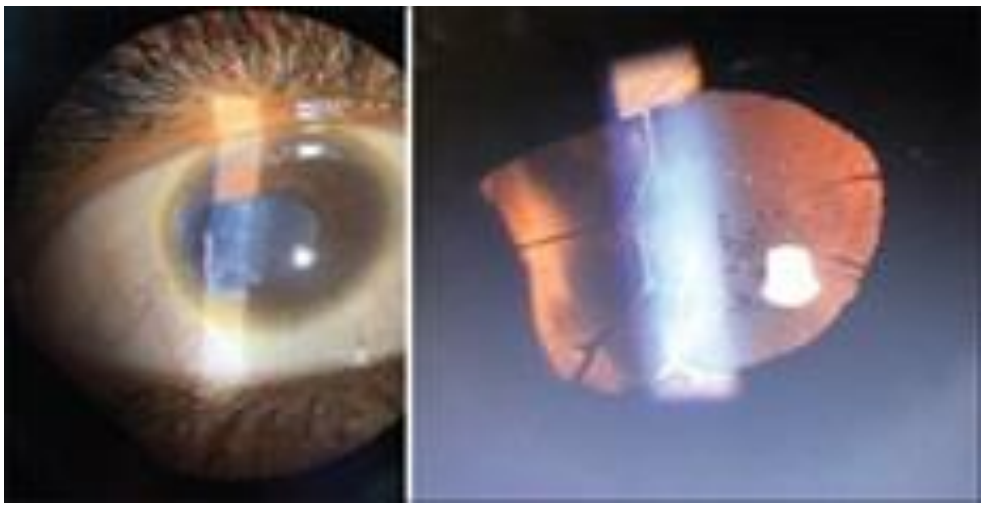

Figure 2. An image with magnification of the front part of the eye displaying microphthalmia, a coloboma in the iris, and persistent pupillary membranes [4].

Approximately one-third of cases present as a standalone occurrence, while the rest are linked to additional ocular and craniofacial abnormalities. [5]. A retrospective review of medical records of pediatric patients under 19 years old diagnosed with ocular coloboma in Olmsted County, Minnesota, from January 1, 1968, to December 31, 2007, led by Kelly M. Nakamura, BS, and colleagues revealed that thirty-three children were newly diagnosed with ocular coloboma. The annual incidence was 2.4 per 100,000 residents under 19 years old, with a prevalence of 1 in 2077 live births. The median age at diagnosis was 3.9 months (ranging from 2 days to 18.4 years), with 22 patients (67%) having unilateral involvement. Among them, 12 patients (36%) had anterior segment involvement only, 13 (39%) had posterior segment involvement only, and 8 (24%) had both. During a median ophthalmologic follow-up of 9.2 years (ranging from 13 days to 35.9 years), 19 patients (58%) had other ocular disorders, including amblyopia in 11 (33%) and strabismus in 10 (30%). Over a median medical follow-up of 16.8 years, 22 patients (67%) were diagnosed with nonocular disorders, including abnormal development in 12 (36%) and CHARGE syndrome in 4 (12%).6. According to findings from Danish researchers, they identified 415 patients with MO/AO/coloboma in the DNPR. The total number of live births between 1995 and 2012 was 1,174,299, with an average birth prevalence of MO/AO/coloboma at 3.6 per 10,000 live births and MO/AO at 1.2 per 10,000 live births. Extra-ocular abnormalities were noted in 32.1% of MO/AO cases and 21.7% of coloboma cases. Chromosome analysis was conducted in 36.1% of the cohort, revealing an abnormal karyotype in 14.7% of cases. Additionally, chromosome microarray analysis was performed in 8.7% of the cohort, with a possibly pathogenic copy number variation observed in 44.4% of cases.[7]. Moreover, ocular colobomas are not always isolated and can be linked to other syndromal diseases. Studies suggest that fifteen to thirty percent of colobomas may be associated with CHARGE syndrome. Shah et al., in a research conducted in the United Kingdom, demonstrated that the presence of systemic features is more prevalent in bilateral cases of AMC (Anophthalmia, microphthalmos, coloboma)[8]. Additionally, colobomas may be linked to other ocular structures, as evidenced by the research conducted by Gopal Lingam et al. The study indicates that the involvement of colobomas can be unilateral in 33-47.5% of cases or bilateral, with the bilateral cases being either symmetric or asymmetric. When macular involvement affects central fixation in both eyes, nystagmus can manifest as a presenting symptom. These eyes often exhibit significant microphthalmia as well. Furthermore, research by Nakamura has revealed that anterior segment involvement (iris and ciliary body) was isolated in 36% of cases, posterior segment involvement (chorio retinal and optic nerve) was isolated in 39% of cases, and both segments were involved in 24% of cases.[6]. It is crucial to note that the corneal diameter may not always accurately reflect the size of the eye. While eyes with microphthalmos typically have smaller corneas, it is possible for microcornea to coexist with normal-sized eyeballs. Precise evaluation of eye size can be achieved through ultrasonography, CT scan, or MRI. An eye is classified as microphthalmic when the axial diameter (adjusted for age) falls below the 95th percentile. [9]. In adults (and children over 13 years of age), an axial diameter less than 18.5 mm is classified as microphthalmic. It is important to note that in cases where there is ectasia

of the colobomatous area, ultrasonography measurements may appear falsely normal or even larger than normal if taken within the ectatic region. This is particularly significant in eyes with extensive colobomas affecting a significant portion of the posterior pole.

During development, the failure of embryonal fissure closure occurs when neuroectodermal hyperplasia causes the inner layer to evert at the edge of the embryonal fissure. If the neuroectoderm has not yet differentiated into the retina, the fissure may still close, resulting in an ectatic coloboma with intraconal cystic mass (ICM). However, if retinal differentiation has taken place, fusion does not occur, leading to the formation of a true orbital cyst. The size of the cyst can vary and may sometimes be mistaken for an orbital tumor due to its large proportions. In extreme cases, the eyeball may be severely microphthalmic or barely recognizable clinically, as it is displaced or pushed posteriorly by the cyst. The communication size between the cyst and the vitreous cavity can also vary. In most instances, the eye is non-functional with dysplastic retina, and management focuses on improving cosmesis.

While the iris is commonly affected in eyes with fundus coloboma, this association is not always present and does not necessarily correlate with the severity of the fundus coloboma. A complete iris coloboma presents as a defect inferonasally that merges with the pupil (resembling a keyhole iris), while a partial iris coloboma may manifest as a notch in the sphincter, a defect in the pigment epithelium, or heterochromia. Unlike traumatic iris defects, the margins of a coloboma are smooth.[10]. Discussing the complications of ocular colobomas can vary depending on their location and association with other conditions. For instance, according to findings from Rehan M. Hussain, MD, and colleagues, chorioretinal colobomas can present complications such as retinal detachment (RD). In their study, the incidence of RD (31%) falls within the reported range (2.4-42%) from previous research. The elevated rate of RD in our study may be attributed to a sequestration bias, as our center serves as a tertiary referral center for vitreoretinal diseases.[11]. Poor visual acuity can be another complication associated with colobomas. Additionally, colobomas may manifest independently or be linked to other systemic abnormalities, such as craniofacial anomalies like cleft lip, skeletal defects like thumb hypoplasia, and genitourinary anomalies such as horseshoe kidney.[13]. Colobomata associated with chromosomal abnormalities are more likely to have other systemic features.

Conclusion. To be in brief, Based on the research conducted on the incidence of ocular colobomas and its complications, it can be concluded that ocular colobomas are a relatively rare congenital eye abnormality. However, they can lead to various complications such as visual impairment, refractive errors, and increased risk of other eye conditions. Early detection and appropriate management are crucial in preventing or minimizing these complications. Further studies and awareness are needed to better understand the causes, risk factors, and optimal treatment strategies for individuals with ocular colobomas.

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