

BILATERAL CONGENITAL CATARACT IN A NEWBORN- LITERATURE REVIEW AND CASE REPORT

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Abstract

Congenital cataract is the leading cause of childhood blindness. The etiology for its occurrence can be different: metabolic diseases, genetic factors, infectious agents, but mostly idiopathic.

The purpose of this paper is to present a case of congenital cataract in a 10-day-old infant who presented with leukocoria in the first days of life, as well as to provide an overview of modern diagnostic and therapeutic modalities in the management of this ophthalmic condition.

In the preparation of this topic, medically relevant data obtained from three large databases were used: PubMed, Embase and Scopus. Keywords such as: congenital cataract, newborn, leukocoria, red reflex were used during the search. The obtained data were selected and processed, while the latest and most relevant knowledge from this field was included in the preparation of this paper.

Rapid recognition of this condition is important in order to preserve good visual function and avoid the potential complications of its presence: deprivation amblyopia, strabismus, nystagmus and glaucoma.

Keywords: congenital cataract, leukocoria, red reflex, newborn.

Introduction

Congenital cataract, although quite rare compared to that in the old population, takes a leading place in childhood blindness. It is estimated that around 200,000 children worldwide are blind as a result of this condition [1].

By definition, any lens opacification observed within the first two months of birth is considered a congenital cataract. According to other authors, a congenital or neonatal cataract is considered to be one that occurred intrauterinely or in the first year of life. Cataracts that occur later in childhood are known as developmental or juvenile [2].

In the congenital type, the clouding is present in the embryonic or fetal nucleus. Depending on the localization and extent of opacification, it can be asymptomatic and have no repercussions on visual acuity, up to a significant impairment of visual function and the appearance of deprivation amblyopia. Cataracts that are centrally located and dense have the greatest impact on vision. They cause deprivation of the visual function of the organ of vision during the crucial period of its development. Several studies have shown that the mother's health, education and hygienic-social conditions have a great influence on the occurrence of this condition in the newborn [3,4].

It can be unilateral or bilateral, be an independent entity or in conjunction with other dysmorphisms and syndromes. The prevalence is much higher in low-standard and developing countries. The genetic environment also plays a major role in the occurrence of congenital cataracts. In other words, the development of the lens cristalina begins on the 22nd day of gestation, it has an ectodermal origin, so a large number of genes are involved in the synthesis of the various proteins that participate in the construction of the lens. PAX6, PITX3, c-Maf, FOXE3 are just some of the genes that play a role in this complex genetic-biochemical process [5].

A mutation of any of the genes involved will result in the synthesis of a mutated protein that would not perform the function for which it was intended and would undergo processes of denaturation and degeneration, which are the basis for the occurrence of lens opacification-cataract and loss of lens transparency. Some of the congenital cataracts have a family history, an autosomal dominant mode of inheritance, a smaller part of them X-linked inheritance, and most of them are sporadic [6].

Case report

A ten-day-old newborn was brought for ophthalmological evaluation at the PJU UC for eye diseases in Skopje due to observed leukocoria, bilaterally, in the maternity hospital. From the medical reports, data were obtained that it is a male child born on time, with a birth weight of 3000g and a body length of 50cm, without any dysmorphisms of the limbs and body, Apgar score 8/10.

During the neonatal examination by the pediatrician, leukocoria was observed, which is why the parents were advised to call for an ophthalmological examination. During the initial macroscopic inspection, bilaterally, milky white discoloration in the pupillary area and absence of red reflex during direct ophthalmoscopy of miotic pupils was observed in the newborn.

Due to the age of the patient, any evaluation of the visual function was impossible. Next, pupil dilation was approached with Sol.Cyclopentolate hydrochloride 0.5%, a blepharostat was placed and the presence of nuclear and posterior polar cataract bilaterally with suppression of the red fundus reflex in the central parts was noted with a manual biomicroscope. Due to the density of the cataract, a more detailed evaluation of the fundus was not possible. In order to obtain more information about the ophthalmological condition, an ultrasonography was performed, where the morphology of the structures from the posterior segment of the eye, on both sides, were with normal echographic characteristics.

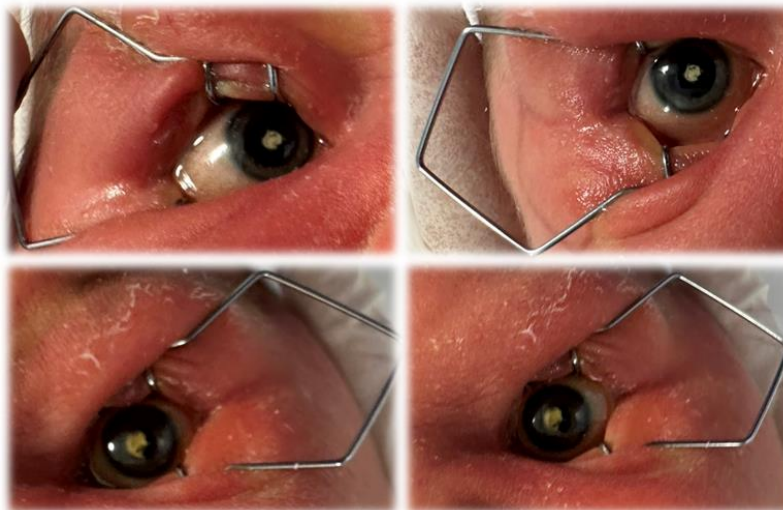


Figure 1. Macroscopic view of right and left eye respectively, centrally located congenital cataract with leukocoria.

From the mother's anamnestic data, we received information that the entire course of the pregnancy was orderly, with regular gynecological controls. Regarding her comorbidities, she is being treated for hypothyroidism, under therapy with Levothyroxine tablets, and TSH and FT4 were constantly within the normal range during pregnancy.

The parents denied the occurrence of congenital cataract in another family member, during their ophthalmological evaluation no opacifications of the lens cristalina were observed. In order to rule out an infectious genesis for the occurrence of congenital cataracts, the most common causes such as: Toxoplasmosis, rubella, cytomegalovirus, herpes simplex, syphilis (TORCH), as well as Hepatitis B and HIV were examined in the serum of the mother and the child. Laboratory analyzes were negative for all the listed causes.

In addition, the blood count, serum calcium and phosphorus values, erythrocyte galactokinase as well as the analysis of metabolites in urine, in the newborn, were without deviations from reference values. Taking into account the entire pediatric, laboratory and ophthalmological evaluation, and at the same time consulting medically relevant literature on this issue, a diagnosis of congenital bilateral cataract in a

newborn, with unknown-idiopathic etiology, was made. Due to the specificity of the case, the age and type of the cataract, but also the bilateral eye affection, advice was given for surgical treatment in a specialized center for this ophthalmic condition, all in order to reduce the risk of possible complications, both operative and related to the disease itself. : deprivation amblyopia, strabismus, nystagmus, glaucoma.

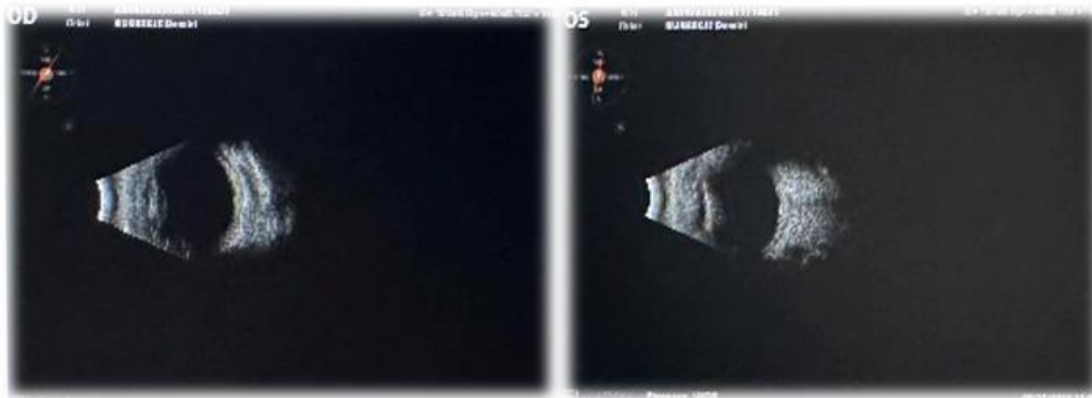


Figure 2. Normal posterior segment echographic features of the right and left eye, respectively.

Discussion

The etiological factors for congenital cataract are numerous. Most of the unilateral cataracts and more than half of the bilateral ones, to date, still remain with an unexplained etiology - idiopathic. Genetic factors are one of the most discussed in recent decades. Namely, a large part of congenital cataracts have an autosomal-dominant mode of inheritance and are usually not associated with other syndromes or systemic diseases. However, neonatal cataracts may be associated with trisomies (Down, Edwards, Patau), chromosomal deletions, X-linked diseases, and a number of multisystem syndromes. Examples of this are: Lowe-syndrome, Alport syndrome, myotonic dystrophy, ichthyosis, Stickler syndrome and others [7,8,9].

Various metabolic diseases that present themselves in early childhood due to osmotic damage to the lens can be the cause of newborn and developing cataracts: galactosemia, galactokinase deficiency, hypoglycemia or diabetes [10,11].

Infections that disrupt the development process of the lens during the intrauterine period have a great impact on the occurrence of congenital cataracts, especially in underdeveloped countries. Rubella virus infection is a particularly significant etiopathogenetic factor. Apart from rubella, toxoplasma, syphilis, herpes and cytomegalovirus (TORCH) are indicated as causes not only of congenital cataract but also of a number of other ophthalmic and neurological disorders such as: microphthalmus, glaucoma, iris atrophy, chorioretinitis, pigmentary retinopathy, etc. [12,13]. Persistent hyperplastic primary vitreous and persistent hyaloid artery with invasion of mesoblastic tissue in the lens cristalina are ophthalmic conditions that can lead to a special type of congenital cataract, posterior-polar. This type of cataract has its own specifics in the clinical picture (rings of a cut onion) and in the method of its surgical treatment [14].

Leukocoria is the most important clinical sign that can be first recognized in the first days of life in children with congenital cataract. Another very significant sign is the absence of eye contact with an object that appears to be at a close distance from the newborn. As an accompanying finding in such patients, a change in the size of the eyeballs (microphthalmos or buphthalmus) as well as the appearance of nystagmus can be found [15].

In older children, strabismus, bringing objects quite close when they want to see them, is of essential importance to raise the suspicion of a possible congenital cataract.

During the ophthalmological examination, attention should also be paid to possible dysmorphisms of the ocular and periocular region as well as other accompanying diseases and conditions in order to rule out a possible syndromic disease. Relevant anamnestic data such as regarding possible infections of the

mother during pregnancy or the occurrence of congenital cataract in another family member can be of great importance regarding the possible etiology.

Unlike preschool and school children, in whom the determination of visual acuity is performed using signs and symbols, in newborns it is more difficult and can only be evaluated using visual evoked potentials and optokinetic nystagmus [16].

A crucial part of the ophthalmological examination is direct ophthalmoscopy and the absence of a red reflex even with a miotic pupil in congenital cataracts that have a central location. In addition, if visualization of the posterior segment of the eye is impossible, ultrasonography of the eye can help in excluding other possible ophthalmic conditions presenting with leukocoria [17].

Timely diagnosis of congenital cataract is very important for prevention of deprivation amblyopia, as a result of inadequate development of the optical system in children with cataract, as it is dependent on photoinduction. According to Birch et al. the period up to 6 weeks in a newborn with unilateral congenital cataract is the so-called latent period, until the removal of the cataract will not lead to deprivation amblyopia [18]. Lambert and Drack state that this period in bilateral congenital cataracts is up to 10 weeks [19].

After this latent period, deprivation of visual development occurs, so it is believed that after 14 weeks of life, this suppression will lead to a pronounced degree of amblyopia in the newborn.

Operative treatment, for now, is the only option that can prevent blindness or a high degree of amblyopia in these patients. However, preoperative evaluation and appropriate assessment of surgical technique and decision-making when and how to perform surgery are of great importance for outcome. Depending on the age of the patient, an appropriate assessment of the diopter of the IOL should be made, taking into account the length of the eye bulb, the size of the cornea, its refractive power, the type of cataract, and whether it is present in both eyes or is unilateral. The most commonly used formula for determining the appropriate IOL power in young children are Holladay 1 and SRK/T. The IOL in children can be placed in the sulcus, although there are studies that indicate that placing the IOL in the capsular bag reduces the risk of posterior capsular opacities (PCO) [20,21].

In children under 4 or 6 years of age, posterior capsulorrhexis and anterior vitrectomy are of great importance for the prevention of possible retinal ablation and the appearance of dense PCO. Today, polymethyl-methacrylate IOLs with a heparin surface that are biocompatible are used, as well as flexible acrylate hydrophobic lenses that can be implanted through small incisions. Monofocal lenses are more suitable compared to multifocal lenses in childhood. It should be emphasized that part of the studies today are of the opinion to leave the eye aphakic, and the refractive error is corrected with a contact lens. However, in unilateral cataract, contact lens application in operative aphakia leads to high anisometropia and anisocoria, two major amblyogenic factors. In contrast, in eyes with anterior segment dysgenesis, microphthalmia, persistent hyperplastic primary vitreous, or corneal diameter less than 10 mm, operative aphakia is considered a safer procedure to avoid postoperative complications, especially glaucoma [22,23].

Because the intraocular environment in children's eyes is highly reactive to a foreign body, during IOL implantation, adequate heparin, corticosteroid, and nonsteroidal anti-inflammatory support are required to prevent possible postoperative fibrinous-type uveitis.

Although surgical treatment is the only way to deal with this condition, it brings with it certain complications. Like any intraocular surgery, postoperative endophthalmitis is a possible acute complication. Because of the tight adhesion with a "ligamentous system" between the anterior hyaloid and the posterior lens capsule, posterior capsulorrhexis and anterior vitrectomy are recommended in young children in order to prevent retinal detachment, a possible postoperative complication.

This risk is particularly pronounced in patients with persistent hyperplastic primary vitreous, which itself has firm adhesions with the retina and is a major risk factor for postoperative vitreo-retinal traction. The immunogenicity of the intraocular environment in young children is the cause of possible postoperative uveitis, with the appearance of posterior synechiae, fibrin membranes and the possibility of seclusion and occlusion of the pupil, iris bombae and the appearance of secondary glaucoma. Growth and migration of the residual lenticular epithelium can lead to PCO, which will require an additional YAG-laser capsulotomy, and such migration has the possibility of spreading to the anterior surface of the vitreous after performing the same. Therefore, children with intraoperative capsulotomy and anterior vitrectomy have a

significantly lower risk of developing postoperative PCO. However, one of the most common and serious postoperative complications in this condition is glaucoma [24].

It occurs immediately after surgery or several years later. Risk factors for its occurrence are: small cornea, ocular abnormalities, type of IOL, age at surgery, nuclear type of cataract and aphakia. According to different sources, the representation is between 20-60% of operated cataracts in childhood. It can be quite refractory to topical therapy, and trabeculectomy is also often without the desired result. Most often, long-term success in postoperative glaucoma in childhood is achieved by placing an Ahmed valve or a Baerveldt implant [25].

Conclusion

In this paper, we describe a case of early diagnosis of congenital bilateral cataract in a 10-day-old infant. The quick and timely diagnosis of this condition is of essential importance for the prevention of deprivation amblyopia and low vision. Screening for newborn cataracts must be carried out in maternity hospitals, through the recognition of leukocoria and the absence of a red reflex. Operative treatment is currently the only possibility to solve this condition, while having an individualized approach to each patient depending on a large number of factors that may affect the operative and post-operative period. Patients with this condition need to be followed up throughout their life post-operatively, with the aim of timely recognition and appropriate treatment for possible post-operative complications, especially secondary glaucoma.

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