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CONGENITAL AUTOSOMAL RECESSIVE CATARACT IN A SAKHA CHILD

The article presents a clinical case of congenital autosomal recessive cataract, first identified in a 3-year-old Sakha child. Congenital cataract is a relatively rare pathology found in children, but it is often the cause of visual impairment and blindness. The restoration of a child's eyesight depends on the early detection and treatment of the disease. Studies show that 30 to 50% of congenital cataracts are caused by genetic mutations.

Keywords: congenital cataracts, child, malformation, genetics.

Introduction. A cataract is a disease that causes clouding of the lens (any light-scattering clouding of the lens). Congenital cataracts, also known as neonatal cataracts, are intrauterine clouding of the lens. According to statistics, congenital cataracts cause blindness in children from 5 to 20% [1,3,4,9]. There are unilateral and bilateral cataracts. They can be classified according to morphology, suspected or definite genetic cause, the presence of specific metabolic disorders or associated ocular abnormalities or systemic features [8,13]. Congenital cataracts are phenotypically and genotypically heterogeneous and can occur alone or in combination with other systemic diseases. Significant progress has been made in identifying the molecular genetic basis of cataracts [6].

The eye begins to develop on day 22 of pregnancy. The lens develops from the superficial ectoderm. Most of fibroblasts growth factors produced in the vitreous are required for differentiation of secondary lens fibers, since lens polarity is determined by fibroblast-regulating growth factor [1,2,6]. PAX6, PITX3, c-Maf and FOXE3 are genes that encode proteins that play the role of a transcription factor in lens development. Mutation of either protein results in defective lens production. The anterior epithelial cells of the lens retain their morphology and proliferative capacity, while the posterior epithelial cells form the primary fiber of the lens [2,9].

In many children with congenital cataracts, the etiology has not been identified, but many authors are inclined to an autosomal dominant type of inheritance. The most common cause of most bilateral congenital cataracts is a genetic mutation. According to epidemiologists, a quarter of all congenital cataracts are hereditary [8]. More than fifteen genes involved in the formation of cataracts have been identified, and inheritance is most often autosomal dominant. Variation in cataract phenotype results from mutations in the *CRYAA, CRYAB, CRYBB1, CRYBB2, CRBB3, CRYGC* and *CRYGD* genes [2,7,8,11]. Congenital autosomal recessive cataract is one of the most common hereditary diseases among the Turkic-speaking population of Yakutia (Eastern Siberia, Russia). Our geneticists under the leadership of Ph.D. Barashkova N.A. have identified the molecular genetic basis of this disease: a mutation in the *FYCO1* gene and carriage of the c.1621C>T mutation [5].

The mutation affects the structure of the eye lens. Studies by some authors indicate that half of genetic mutations are affected by so-called proteins - crystallins; in 20 percent of cases they affect connexins, growth factors and lipid metabolism [11]. The variety of clinical manifestations of congenital cataracts may be due to the fact that a mutation of one gene leads to different phenotypic changes in different families. At the same time, different genetic mutations can manifest themselves in the same way, and this fact suggests that there are other factors involved in morphological changes [1,2,7].

Surgical intervention at an early age and subsequent vision correction can contribute to the timely social adaptation of the child. Late surgery can cause sensory deprivation and cause complications such as strabismus, nystagmus, and the formation of incorrect fixation. To restore a child's vision, the sooner the operation is performed, the better the prognosis [1,4]. Despite early surgical treatment, complications may subsequently develop in children [1,2,14].

Congenital cataracts can be caused by infections that a woman comes into contact with during pregnancy. The main infections that have an increased risk of developing cataracts include rubella virus, cytomegalovirus, herpes simplex virus, and toxoplasmosis.

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For preventive purposes, it is necessary to exclude contact of a pregnant woman with infectious patients, and also to minimize the effects of alcohol, smoking, teratogenic drugs, and radiation. Early detection of chromosomal abnormalities allows a decision to be made to terminate the pregnancy. There is no specific prevention of congenital cataracts [2,10,12].

Thus, congenital cataracts require early recognition and surgical intervention to ensure good clinical outcomes.

Purpose of the study: to describe a clinical case of congenital cataract in a 3-year-old child.

Clinical example. Patient Z., 3 years old, Sakha, a child from the 8th pregnancy, 4 births, the pregnancy proceeded in the first half - toxicosis, in the second half - edema, anemia, gestational diabetes mellitus, suffered from acute respiratory viral infection at 26 weeks. Delivery at term, birth weight – 3350g, body length – 53cm. Apgar score – 8/9 points. He was discharged home on the 6th day in satisfactory condition under the supervision of a local pediatrician at his place of residence. The child grew and developed according to his age. Got preventive vaccinations according to the calendar. There is no hereditary history of eye diseases. Past diseases: frequent acute respiratory viral infections, chalazion.

At the age of 9 months, parents noticed that the child brings small objects and toys closer to his face and examines them, tilting his head to the side. Due to the absence of an ophthalmologist at the district hospital, the child was not examined. At the age of 3 years, he was sent to the Republican Hospital No. 1 - National Center of Medicine (RH No. 1-NCM) with a diagnosis of: Frequently ill child. Visual impairment.

After an examination by an ophthalmologist, the diagnosis was made for the first time: Congenital zonular bilateral cataract. Sent to the ophthalmology department of RB No. 1-NTsM for examination and decision on the issue of surgical treatment. A consultation with a geneticist and an allergist-immunologist is recommended.

Ophthalmological examination: The eyeball is in the correct position, full mobility. Eyelids: on the right side of the upper eyelid there is a trace of a small infiltrate, protruding above the level - 0.1 mm, painless, lacrimal openings are normal. The conjunctiva is calm. The cornea is transparent. The anterior chamber is of medium depth. The moisture in the anterior chamber is transparent. The pupil is round, 3 mm in diameter, reaction to light is friendly. The iris is not changed. The lens is cloudy, more on the left, inhomogeneous, irregularly shaped, in the center. Vitreous body: without features. Fundus: optic disc is pale pink in color, the boundaries are clear. The arteries are slightly narrowed, the course is not changed. The veins are of normal caliber, their course is not changed. The retina is not changed.

Results of instrumental research:

Skiascopy: P30, OD=18.55mm, OS =18.5mm, IOP Icare OD=13 mmHg; OS =13 mmHg

Ultrasound OU: compaction of the lens capsules, the vitreous body is acoustically transparent, the retina is adjacent, the contour is smooth.

Genetic research data:

1. Conclusion of a molecular genetic study in the proband: as a result of a molecular genetic study for a major mutation in the *FYCO1* gene, a homozygous carriage of the c1621C>T mutation was revealed, causing the hereditary disease cataract 18 (autosomal recessive congenital cataract 2).

2. Conclusion of a molecular genetic study of the mother. As a result of a molecular genetic study for a major mutation in the *FYCO1* gene, carriage of the c.1621C>T mutation in a heterozygous state was revealed, causing the hereditary disease cataract 18 (autosomal recessive congenital cataract 2).

3. Conclusion of a molecular genetic study in the father: as a result of a molecular genetic study for a major mutation in the *FYCO1* gene, carriage of the c.1621C>T mutation in a heterozygous state was revealed, causing the hereditary disease cataract 18 (autosomal recessive congenital cataract 2).

Based on complaints, examination data, instrumental and laboratory data, a clinical diagnosis was made: Congenital autosomal recessive incomplete cataract (Q12.0).

The child was sent to the Helmholtz National Medical Research Center for Eye Diseases for surgical treatment.

Conclusion: Based on the presented clinical case, we can conclude that the diagnosis of the pathology and surgical treatment were carried out late (the child is 3 years old). Regardless of the type of cataract, early detection and treatment is necessary, since the restoration of vision in the child depends on this.

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