Test for Helico bacterium from 18.10.2023: Hp +

Colonoscopy from 12.10.2023: The lumen of the cecum was narrowed by 1/3 due to edema of the bauginia flap, the mucosa of the bauginia flap was sharply edematous and hyperemic, with multiple ulcers of 2.0 cm with fresh undermined edges, the crater was covered with fibrin. The aperture of the flap was spasmodic, impassable for the apparatus. The rest of the examined areas of the colonic mucosa were without peculiarities. For biopsy 2 fragments of the ileum mucosa, 3 fragments of the mucosa of the Bauginia flap were taken. Conclusion: Terminal ileitis. Crohn's disease. Active stage with ulcers and stricturing.

Biopsy of small intestine mucosa fragments from 20.10.2023: biopsy material contains fragments of small intestine mucosa with the picture of chronic active ileitis, with focal cryptitis, with focal hyperplasia of peyer's plaques.

Clinical diagnosis: Inflammatory bowel disease. Crohn's disease with extraintestinal manifestations. Juvenile arthritis (seronegative), polyarthricular variant, grade 3 activity, X-ray grade 2. FC 3. Autoimmune hepatitis. Osgood-Schlatter disease.

Treatment was prescribed: ward regime, table #5, adalimumab (Humira) 40mg p/k, pancreatin 1 capsule 3 times a day with meals, omeprazole 20 mg 2 times a day (8 h-20 h) for 14 days.

On 20.11.2023 the child was discharged with improvement.

Recommendations at discharge: observation of the district pediatrician, table #4, exemption from physical training (exclude physical activity), contraindicated insolation, contraindicated vaccination and administration of gamma globulin, contraindicated hypothermia and bathing in open water, immunomodulators, contact with animals, contraindicated physical and mental trauma. Examination of gastroenterologist and rheumatologist once a month, ophthalmologist once every 3 months, planned hospitalization in Pediatric Endocrinology and Gastroenterology Department every 3 months, cardiac ultrasound once every 3 months, chest Computed Tomography once every 6 months.

Adalimumab (Humira) 40 mg once every 2 weeks. (every 2 weeks) constantly, calcium preparation, vitamin D constantly for 3 months, 1 month break, mesalazine (salofalc) 1g 3 times a day constant intake, Nexium 20 mg once a day for 3 weeks, pancreatin (Creon, Micrazyme) 10 TE 3 times a day with meals for 3 weeks, urodesoxycholic acid 250 mg at lunch, 500 mg in the evening.

**Conclusion.** This article presents a case of the course of two severe autoimmune diseases (Crohn's disease and juvenile rheumatoid arthritis) in a male Sakha adolescent. Such clinical examples are rare in the publicly available literature. The management of this child requires a multidisciplinary approach and joint therapy by gastroenterologists and rheumatologists to achieve a durable remission.

## References

1. Belkin A.S., Dyakonova E.Y., Surkov A.N. [et al.]. Bolezn' Krona u detej: sovremennoe sostoyanie problem [Crohn's disease in children: the current state of the problem]. Pediatrics. G.N. Speransky Journal. 2022:100(6):78-85 (In Russ.).]

2. Kagramanova A.V., KnyazevO,V,, Parfenov A.I. Bolezn' Krona: do i posle 1932 goda [Crohn's disease: before and after 1932]. Terapevtich. arhiv. [Therapeutic archive. 2023; 95(2): 193-197 (In Russ.).]

3. Kalatina YU.E., Sorokin D.V., Volosnikov D.K. Klinicheskie i laboratornye osobennosti bolezni Krona u detej [Clinical and laboratory features of Crohn's disease in children]. Pediatrich. vestnik YUzhnogo Urala [Pediatric Bulletin of the Southern Urals. 2020:1:66-71 (In Russ.).]

4. Tarasova D.D., Shilova L.N., Koroleva. Klinicheskij sluchaj sochetaniya ankiloziruyushchego spondilita i bolezni Krona [A clinical case of a combination of ankylosing spondylitis and Crohn's disease]. Medicin. Alfavit [Medical alphabet. 2021:33:53-57 (In Russ.).]

5. Shkurko T.V., Kagramanova A.V., Knyazev O.V. Bolezn' Krona. K 90-letiyu opisaniya [Crohn's disease. To the 90th anniversary of the description]. Problemy social'noj gigieny, zdravoohraneniya i istorii mediciny [Problems of social hygiene, health care and the history of medicine. 2022:30(5):1162-1166 (In Russ.).]

6. Crohn B.B., Ginzburg L., Oppenheimer G.D. Regional ileitis. JAMA. 1932:99:1323-1328.

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# COMBINATION OF MILLER FISHER SYNDROME AND UNSPECIFIED PERIPHERAL T-CELL LYMPHOMA

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The article presents a clinical case of combination of Miller Fisher syndrome and unspecified peripheral T-cell lymphoma, which is an aggressive disease with an extremely poor prognosis. The pathogenetic treatment of the identified syndrome did not cause an improvement in the patient's condition and did not affect the expected unfavorable prognosis of the primary malignant disease, for which chemotherapy was not carried out due to the extremely serious condition of the patient. This case emphasizes the importance of an oncological search in patients with Miller Fisher syndrome and the mandatory determination of onconeural antibodies used in the diagnosis of paraneoplastic neurological syndrome, which occurs in malignant tumors of various histogenetic types.

**Keywords:** unspecified peripheral T-cell lymphoma, paraneoplastic neurological syndrome, Guillain-Barré syndrome, Miller Fisher syndrome.

Paraneoplastic syndrome (PNS) is a clinical and laboratory manifestation of a malignant tumor, caused by nonspecific reactions from various organs and systems or ectopic production of biologically active substances by the tumor. It is

registered in 10-18.5% of cases in colon cancer, small cell lung cancer, breast and ovarian cancer and malignant lymphomas [6]. It is noteworthy that paraneoplastic syndrome can manifest before the tumor is detected or sometimes simultaneously with its clinical manifestations [2]. After radical treatment of a malignant tumor, this syndrome may disappear and reappear with tumor relapses [1].

Domestic and foreign literature describes various manifestations of PNS (hematological, endocrine, neurological, gastroenterological, nephrological, dermatological, etc.).

Recently, clinicians have been particularly interested in Guillain-Barré syndrome (GBS), which has many clinical forms that differ in the characteristics of the pathological process, the primary point of application of autoimmune aggression (nerve sheath or axon core), prognosis of recovery and clinical manifestations. Most often (70-80%) all over the world, including in Russia, acute inflammatory demyelinating polyneuropathy (AIDP) is diagnosed as part of GBS, in which autoantibodies attack the myelin sheath of the nerve. In the second most common place (5-10%) are axonal forms – acute motor and motor-sensory axonal neuropathies (AMAN and AMSAN), characterized by primary damage to the axons of peripheral nerves and differing from each other in involvement (AMSAN) or intactness (AMAN) of sensitive fibers. Other forms of GBS (Miller Fisher syndrome (MFS), pharyngo-cervico-brachial, acute pandysautonomia, paraparetic, sensory) are diagnosed extremely rarely (1-3%). In the Russian Federation, among all forms of GBS, Miller-Fisher syndrome accounts for 2-5% [4,13].

MFS has three defining features: external ophthalmoplegia (ocular weakness resulting in impaired eye movements and consequent diplopia), ataxia (incoordination of the limbs movement), hypo- or areflexia (absence of tendon reflexes). In severe cases, tetraparesis and paralysis of the respiratory muscles may occur [15]. It has been established that the disease can develop after viral and bacterial infections [8]. The occurrence of areflexia in MFS is associated with a decrease in the acetylcholine content in the terminals and peripheral nerves, which is clinically manifested by polyneuropathy [5, 12]. The study of ataxia in this disease using modern neuroimaging methods made it possible to detect changes in the signal from cerebellar structures [9] and suggest a combination of sensitive and cerebellar ataxia due to both central and peripheral demyelination [12]. Damage to the oculomotor nerves is explained by the formation of anti-GQ1b antibodies, which are detected in the blood of the vast majority of patients with MFS [11, 12, 16]. Gangliosides are the components of the myelin sheaths that envelop nerve fibers and ensure the speed of transmission of nerve impulses. According to their chemical structure, gangliosides are lipopolysaccharides containing a sialic acid residue. Many bacteria (the intestinal bacterium *Campylobacter jejuni*, the causative agent of pneumonia – *mycoplasma pneumoniae* and viruses (Epstein-Barr virus, cytomegalovirus)) and others are very similar in composition to gangliosides. When these microorganisms enter the body, the immune system may mistakenly react not only to viruses, but also on gangliosides of nerve cells, which leads to the production of autoantibodies to its own neurons and the development of various clinical forms of Guillain-Barré syndrome.

Many researchers have found that with the development of malignant tumors, specific antitumor antibodies, called onconeural antibodies, begin to be produced. Due to the antigenic identity of some tumor antigens and components of nervous tissue, these antibodies, associated with onconeural antigen-specific T lymphocytes, attack components of normal nervous tissue [7,14].

We present our own clinical observation of Miller Fisher syndrome in a patient with nodal unspecified T-cell lymphoma.

Patient E., 63 years old, was hospitalized to department of medicine of Republic Hospital No.1 – National Center of Medicine (RH No.1- NCM) with complaints of numbness in the lower and upper extremities, inability to move without assistance, severe weakness, fever up to $38^0$C, loss of body weight by 10 kg, cough, shortness of breath.

From the anamnesis: ill for about 2 months, when the cervical and supraclavicular inguinal lymph nodes enlarged, there was a daily increase in body temperature to $38^0$C. After 1 month from the moment of illness, the patient developed numbness in the extremities, diplopia, unsteadiness when walking, weakness of the right half of the face, cough and shortness of breath during physical activity. In 2 months of illness due to increasing weakness in the legs, the patient stopped walking. The patient was hospitalized in the central district hospital, from where he was sent to RH No. 1-NCM with a diagnosis of lymphadenopathy of unknown origin.

Objectively: at admission the patient's general condition is considered as serious. The skin is clean, of normal color, acrocyanosis is noted. On palpation, the cervical, axillary and inguinal lymph nodes on both sides are enlarged, dense in consistency, painless. Lung auscultation – clear breath sounds on both sides, weakened in the lower sections. Heart sounds are rhythmic and muffled. Blood pressure – 100/60 mmHg. Pulse 96 bpm.

Neurological status: alert and oriented in his own personality, place and time. Emotionally labile. The pupils are equal, and reactive to light. Bilateral converging strabismus is noted, limited upward and outward movements of the eyeballs, ptosis on the right, binocular diplopia. Sensitivity on the face is preserved, the trigeminal points are painless. The face is asymmetrical: the right corner of the mouth is drooping, lagophthalmos on the right, smoothness of the frontal and nasolabial folds on the right. Dysphagia when taking liquid food. Dysarthria. There are no oral automaticity reflexes. The pharyngeal reflex is absent. Tongue in the midline. Flaccid deep tetraparesis (muscle strength reduced to 2 points). Diffuse muscle hypotonia. Deep and periosteal reflexes of the arms and legs are reduced. Hypotrophy of the leg muscles. Hypoesthesia of the "sock" type. It is impossible to check in the Romberg pose. The knee-to-heel, finger-to-nose tests cannot be checked due to paresis. There are no meningeal or pathological signs. Tension symptoms are negative. Controls the functions of the pelvic organs.

***Examination data:***

**Complete blood test**

*Component Value*

Red Blood Cell Count 3.76 x $10^{12}$/l
Hemoglobin $10^9$ g/l
White Blood Cell Count 11 x $10^{9}$/ l
Eosinophilia 3%
Platelet Count 307x$10^{9}$/l.

**Biochemical blood test**

ALT 10 units/l
AST 11 units/l
LDH 163 units/l
Total protein 79 g/l
Albumin 30 g/l
Creatinine 68 μmol/l
Urea 2.5 mmol/l
Total bilirubin 10.2 μmol/l
Alkaline phosphatase 92.3 units/l
Ferritin 216 μg/l
Glucose 8.35 mmol/l
Potassium 2.8 mmol/l
Sodium 135 mmol/l
Triglycerides 2.05 mmol/l
Total cholesterol 4.17 mmol/l

**Coagulogram**
Fibrinogen 3.88
Prothrombin index 03%
Prothrombin time 12
APTT 27.30 sec.
INR 1.10.

Thyroid stimulating hormone (TSH) level is 1.36 mIU/l, the level of free thyroxine (free T4) is 12.80 pmol/l (both hormones are within normal limits).

Serological tests (enzyme immunoassay) for syphilis, human immunodeficiency virus, hepatitis B and C are negative.

***Conclusion of nerve conduction study (NCS)***: signs of damage to the motor and sensory fibers of the median, radial, tibial and peroneal nerves, such as severe axonal demyelinating polyneuropathy, more pronounced in the nerves of the legs (M-waves absent).

***Ultrasound of peripheral lymph nodes:*** submandibular, mental lymph nodes are not enlarged, cervical lymph nodes are enlarged (largest on the right 18x6 mm, on the left 16x7 mm), posterior cervical (right largest 26x15 mm, left 28x16 mm), supraclavicular (right largest 10 mm, left largest 19x11 mm), subclavian (right 10 mm, left 10.2 mm), axillary (right largest 41x16 mm, left 34x19 mm), inguinal (largest on the right 28x9.4 mm, on the left - 28x10 mm) lymph nodes.

***Result of ultrasound of the abdominal organs:*** hepatosplenomegaly, hardening of the kidney sinuses, enlarged lymph nodes in the hilum of the liver and spleen.

***Conclusion of computed tomography of thorax***: hilar and peripheral lymphadenopathy, right-sided paracostal pleuritis.

***Conclusion of CT scan of the brain***: signs of dyscirculatory encephalopathy.

For diagnostic purposes, an open biopsy of the enlarged cervical lymph node was performed. Microscopic description of the material: fragments of lymphoid tissue with a disturbed structure, diffuse focal proliferation of atypical medium-sized lymphocytes is noted. Tumor cells with expression of CD4, CD5, bcl-2, CD2, CD43, EBV, in the absence of pax5, CD79a, CD10, CD15, CyclinD1, CD138, CD30, EMA, MUM1, bcl-6, CD1a, CD246, CD56, CD57, TdT. Expression of CD34 in vessels and CD23 in preserved areas of the network of follicular dendritic cells was revealed. ***Conclusion:*** Unspecified T-cell lymphoma.

The patient underwent 5 sessions of discrete plasmapheresis, intravenous immunoglobulin therapy, antibiotic therapy and antiviral treatment without the desired result.

The patient's condition sharply deteriorated due to signs of respiratory failure, and therefore was transferred to mechanical ventilation. Despite the treatment, due to worsening respiratory failure, patient expired on the 22nd day after hospitalization.

**Conclusion.** Thus, taking into account the data of clinical, laboratory and instrumental research methods, the results of an immunohistochemical study of a cervical lymph node - unspecified peripheral T-cell lymphoma, an indication of a recent history of viral infection, acute development of external ophthalmoplegia, hyporeflexia and ataxia, as well as NCS data, confirming the presence of polyneuropathy, allowed us to establish the diagnosis: Unspecified peripheral T-cell lymphoma combined with Miller Fisher syndrome.

The temporary coincidence of the development of neurological disorders with T-cell lymphoma most likely suggests their paraneoplastic nature. Although MFS is classically associated with infectious diseases, primarily *Campylobacter jejuni* and *Haemophilus influenza* [17], clinical observations of the development of MFS in patients with lung cancer [10] and Burkitt's lymphoma [8] have been described in the literature.

In the case we presented, attention is drawn to the severe course of neurological disorders, namely the development of severe flaccid paresis with the development of respiratory failure, damage to facial muscles. According to the opinion of Professor O.S. Levin, such cases may be the result of the “superposition” of MFS on Guillain-Barré syndrome [3].

A limitation of our observation is undoubtedly the lack of serum testing for antibodies to gangliosides, which are detected in 95% of patients with MFS; examination of cerebrospinal fluid to determine protein-cell dissociation. In this regard, we cannot reliably state the development of neurological disorders within T-cell lymphoma, but only pay attention to their combination. Consequently, the answer to the question whether the Miller Fisher syndrome that developed in the patient can be considered a paraneoplastic syndrome associated with unspecified peripheral T-lymphoma remains controversial and doubtful. It should be noted that many studies have established the trigger role of Epstein-Barr viruses and cytomegalovirus infection in the occurrence of both lymphomas and Miller-Fisher syndrome. In this patient, both viruses were detected in the replication phase, which could serve as a trigger for the simultaneous development of two completely different diseases. In addition, the patient did not have onconeural antibodies, which are used as one of the criteria for identifying definite or probable paraneoplastic neurological syndrome. However, this clinical example demonstrates the possibility of co-occurrence Miller Fisher syndrome with unspecified peripheral T-cell lymphoma, which is an aggressive disease with an extremely poor prognosis. The pathogenetic treatment of the identified syndrome did not cause an improvement in the patient's condition and did not affect the expected unfavorable prognosis of the underlying malignant disease, for which chemotherapy was not carried out due to the extremely serious condition of the patient.

This case emphasizes the importance of an oncological search in patients with Miller Fisher syndrome and the mandatory determination of onconeural antibodies used in the diagnosis of paraneoplastic neurological syndrome, which occurs in malignant tumors of various histogenesis.

## References

1. Bolotnaya L.A., Serbina I.M. Paraneoplasticheskie dermatozy [Paraneoplastic dermatoses] Mezhdunarodnyj medicinskij zhurnal. [International Medical Journal. 2008; 3: 86–90. (In Russ.).]

2. Evtushenko S.K. Paraneoplasticheskie nevrologicheskie sindromy (klinika, diagnostika i vozmozhnosti lecheniya) [Paraneoplastic neurological syndromes (clinic, diagnosis and treatment options)]. Mezhdunarodnyj medicinskij zhurnal. [International Medical Journal. 2011; 8 (46). (In Russ.).]

3. Levin O.S. Polinevropatii: klinicheskoe rukovodstvo. [Polyneuropathies: clinical guidelines]. Moscow: MIA, 2016: 480 p. (In Russ.).]

4. Piradov M.A., Suponeva N.A. Sindrom Gijena – Barre: Diagnostika i lechenie. Monografiya. [Guillain–Barre syndrome: Diagnosis and treatment]. Monograph. Moscow: MEDpress-inform. 2011: 72-73. (In Russ.).]

5. Ponomarev V.V. Autoimmunnye zabolevaniya v nevrologii. [Autoimmune diseases in neurology]. Minsk: Belorusskaya nauka, 2010: 259. (In Russ.).]

6. Fursova L.A., Lihacheva E.M. Paraneoplasticheskie spinal'nye sindromy (nauchnyj obzor i lichnoe nablyudenie). [Paraneoplastic spinal syndromes (scientific review and personal observation)]. Mezhdunarodnyj nevrologicheskij zhurnal [International Neurological Journal. 2017; 4 (90): 69-76. (In Russ.).]

7. Schneider N.A., Dykhno Yu.A., Ezhikova V.V. et al. Paraneoplasticheskij nevrologicheskij sindrom. [Paraneoplastic neurological syndrome]. Novosibirsk. 2016:172. (In Russ.).]

8. De Silva NL, Weeratunga P, Umapathi T, et al. Miller Fisher Syndrome Developing as a Parainfectious Manifestation of Dengue Fever: A Case Report and Review of The Literature. Journal of Medical Case Reports 2019; 13(1):120.

9. Gentile S., Messina M., Rainero I. et al. Miller Fisher syndrome associated with Burkitt's lymphoma // Eur J Neurol. 2006 Apr;13(4):430. doi: 10.1111/j.1468-1331.2006.01254. x.

10. Hela J., Mariem K., Saloua F. et al. Mill-

er Fisher Syndrome revealing a lung carcinoma: Paraneoplastic origin of Miller Fisher Syndrome? Rev Neurol (Paris). 2019;175(5):332-334. doi: 10.1016/j.neurol.2018.07.01211. Lee SY, Lee YH, Chun BY, Lee SY, Cha SI, Kim CH, Park JY, Lee J. An adult case of Fisher syndrome subsequent to Mycoplasma pneumoniae infection. Journal of Korean Medical Science. 2013;28(1):152-155. https://doi.org/10.3346/jkms.2013.28.1.152

12. Maddison P. Anti-GQ1b IgG antibody syndrome without ophthalmoplegia: clinical and immunological features. Journal of Neurology, Neurosurgery, and Psychiatry. 2001; 71:282-283.

13. Muñiz AE. Multiple cranial nerve neuropathies, ataxia and areflexia: Miller Fisher syndrome in a child and review. The American Journal of Emergency Medicine. 2017;35(4):661. doi: 10.1016/j.ajem.2016.07.042.

14. Pelosof LC, Gerber DE. Paraneoplastic syndromes: an approach to diagnosis and treatment. Mayo Clinic Proceedings. 2010 Sep;85(9):838-54.

15. Ryu WY, Kim YH, Yoon BA, et al. Pattern of Extraocular Muscle Involvements in Miller Fisher Syndrome. Journal of Clinical Neurology. 2019; 15(3):308-12.

16. Snyder LA, Rismondo V, Miller NR. The Fisher variant of Guillain-Barre syndrome (Fisher syndrome). Journal of Neuroophthalmology. 2009; 29:312- 324.

17. Spyromitrou-Xioufi P., Ntoulios G., Ladomenou F. et al. Miller Fisher Syndrome Triggered by Infections: A Review of the Literature and a Case Report. J Child Neurol. 2021 Aug;36(9):785-794. doi: 10.1177/0883073820988428.

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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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