

## **SYNDROME KARTAGENER IN CHILDREN**

**(clinical case of observation)**

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**Annotation.** Primary ciliary dyskinesia (Kartagener syndrome) refers to inherited rare diseases, characterized by the reverse location of internal organs, the development of secondary chronic diseases of the bronchi, hypoplasia of the sinuses and the development of chronic rhino-sinusitis.

In the article the peculiarities of clinic, diagnostics and treatment of children with the syndrome of Kartagener. A clinical case of diagnosis of this pathology in a child in the first year of life is presented. Early diagnosis and regular adequate conservative therapy contribute to the slow progression of the disease without signs of bronchiectasis.

**Keywords:** Kartagener syndrome, primary ciliary dyskinesia.

The development of chronic non-specific lung diseases in children is an urgent problem, as they begin in childhood, are transformed into different age periods of life, leading to a decrease in working capacity and quality of life.

In some cases, chronic lung disease develops against the background of congenital and hereditary lung diseases, when the first symptoms of lung damage are observed from early childhood. Among the hereditary diseases include syndrome Kartagener. The first description of the syndrome belongs to the Kiev doctor A.K. Zivert, and then this syndrome was described by the Swiss therapist M. Kartagener. They observed patients with a combination of reverse arrangement of internal organs, the presence of purulent sinusitis and bronchiectasis.

Kartagener syndrome refers to genetically determined conditions with a predominantly autosomal recessive type of inheritance and is considered as the main form of primary ciliary dyskinesia (PCD) [1, 2, 3, 4]. When studying genealogical features in persons with signs of dysmorphogenesis, an increase

in the severity and number of signs of dysplasia or their transmission by inheritance in an unchanged form is noted. Patients inheriting a variety of genetic defects of their parents, already in the prenatal and early postnatal period, morphofunctional changes in various organs and systems, aggravated with the development of disorders and insufficiency of compensatory mechanisms [20].

Primary ciliary dyskinesia is based on congenital defects in the structure of cilia of the ciliated epithelium of the respiratory tract and similar structures that lead to a violation of their motor activity [5, 6].

In the human body, cilia and flagella are present in many organs and systems. These are the ciliated epithelium of the respiratory tract, and the cells of the cortical organ of the ear, flagella of sperm, ciliated cells of the ependyms of the ventricles of the brain, photoreceptors of the retina of the eyes; cells lining the biliary tract, cells of the renal tubules, cells lining the fallopian tubes.

In addition, there are cilia located on the embryonic node, which provide rotation of internal organs in the period of intrauterine development, resulting in the reverse arrangement of internal organs in half of patients.

Violation of the activity and mobility of the cilia of the ciliated epithelium of the respiratory tract leads to disruption of the normal cleansing function of the respiratory system, the secret stagnates and gets infected, which contributes to the formation of recurrent and chronic infections: chronic bronchitis, chronic sinusitis; may develop chronic otitis media with hearing loss [7,8, 9, 11, 12].

In adulthood, immobility of spermatozoa causes infertility in men in patients with PCD, and ectopic pregnancy is often observed in women [9, 10]. Abnormal functioning of cilia located in other organs and systems leads to the formation of such extremely rare manifestations as retinopathy pigmentosa,

biliary cirrhosis of the liver, internal hydrocephalus, and polycystic kidney disease [8].

The frequency of occurrence is 1 in 2265 to 1 in 40000 of the population [11]. Among patients with bronchiectasis -13%, among patients with inverted internal organs -25% [11].

The most common and classic form of PCD is Zivert-Kartagener syndrome (situs inversus - with the reverse location of internal organs, chronic bronchiectasis, hypoplasia of the sinuses or sinusitis). It accounts for 50-60% of this pathology [6]. But it is possible to have an incomplete abnormal arrangement of internal organs (situs ambiguus - heterotaxy, when the location of the main internal organs differs from their normal position (situs solitus), and from their complete mirror image (situs inversus).

According to research data, in the presence of situs inversus, the diagnosis is established earlier than without the reverse arrangement of internal organs [13, 14].

Available literature data on the outcome of primary ciliary dyskinesia is ambiguous. In some cases, the progression of chronic bronchopulmonary process with the development of respiratory failure is noted [15, 16]. There are data from the results of a prospective study, when no progression of the process was detected and the stability of functional indicators was noted [1, 7]. There are publications on patients with the syndrome of Kartagener who have reached old age in the literature [17].

In the classic development of the disease, difficulty in nasal breathing is noted from the first days. In the family history, there may be the presence of chronic bronchitis or sinusitis in relatives, cases of male infertility, abnormal location of internal organs.

In the neonatal period, full-term infants may develop respiratory distress syndrome with oxygen demand from 1 day to 1 week [18, 19]. Children have frequent rhinitis частые риниты with damage to the

maxillary sinuses and recurrent exudative otitis media with hearing loss, repeated bronchitis and pneumonia. Exacerbations of the bronchopulmonary process are noted several times a year, the clinical symptoms are pronounced and indicate the totality of the defeat of the respiratory tract. Later, Mucopurulent or purulent endobronchitis is diagnosed, then limited pneumosclerosis develops with a deformity of the bronchi, which can lead to the formation of bronchiectases. Character of morphological changes in the lung, the syndrome of Kartagener ambiguous. Bronchological studies have shown that bronchiectases are not an obligate sign of this syndrome. Some children are diagnosed with deforming bronchitis or chronic bronchitis without structural deformation of the bronchi [21].

The main goals of therapy for Kartagener syndrome are to prevent the progression and/or development of bronchiectasis and restore/preserve normal pulmonary function, as well as nasal breathing and hearing. It is recommended to use various methods that help to clean the airways and nasal passages. In case of exacerbation of chronic bronchopulmonary process, it is recommended to prescribe antibacterial drugs in accordance with the sensitivity of the isolated microflora [22]. Antibacterial therapy is also used for sinusitis exacerbations, if there is no effect from irrigation treatment [22]. Antibacterial therapy is prescribed in accordance with the sensitivity of the microflora to drugs, including, if necessary, reserve antibiotics. If necessary, designate bronchospasmolytic inhalation and mucolytics orally. Patients with primary ciliary dyskinesia are observed by a district doctor and a pulmonologist with regular examination of the function of external respiration (FVD), saturation (saturation of body tissues with gas), EchoCG with Doppler analysis and the periodic monitoring MSK of the chest cavity. In connection with the defeat of the nasopharyngeal mucosa, the patient is regularly examined by an otolaryngologist, according to the indications of a surdologist and a cardiologist.

Observational data [21] showed that timely and systematic conservative therapy allows not only to avoid dramatic outcomes, prevent the progression of the process, but also to maintain a sufficiently high level of social adaptation of patients.

As an example, we present our own observation of a child with primary ciliary dyskinesia (Zivert-Kartagener syndrome), diagnosed in the first year of life.

A child of 7 months was admitted to the hospital in the early age with complaints of fever rising to subfebrile numbers, difficulty breathing, coughing, shortness of breath. From the mother's medical history, there is a burdened obstetric and gynecological history (long-term treatment due to infertility), an unfavorable course of pregnancy. The baby was born on time with a weight of 3500gr, the newborn period was relatively smooth. Subsequently, the mother began to notice that the child's nasal breathing was somewhat difficult, and there was a mucous discharge from the nose. Repeatedly appealed to the local pediatrician, who prescribed symptomatic treatment intranasally, fenistil, which had the effect. At 7 months, the child after contact with a patient with an acute respiratory infection, against the background of existing symptoms, the temperature rose to 38<sup>0</sup> C, there was a cough, pronounced breathing difficulties, and therefore was hospitalized.

Upon admission, the child's condition is moderate. Musculoskeletal system is without deformation. The chest is cylindrical shape, and both halves participate symmetrically in the act of breathing. In the lungs, there is a percutaneous-pulmonary sound, auscultation revealed changes with the presence of scattered dry and wet wheezes. The heart area is not changed, but the heart borders are shifted to the right. Auscultation heart tones are clear, rhythmic. On palpation, the abdomen is soft, without pain.

The patient was diagnosed with acute bronchitis, which was confirmed by X-ray examination.



Pic. 1. X-ray examination of the chest: increased pulmonary pattern in the basal corneal zone, pulmonary fields without focal-infiltrative shadows. The sinuses are free. The heart is located on the right. Conclusion:dextrocardia, acutebronchitis.

The child was consulted by an otolaryngologist, a diagnosis of acute rhinosinusitis was made, and a genographic examination of the paranasal sinuses was carried out, which revealed an underdevelopment of the frontal sinuses, which is one of the manifestations of Ziwert—Kartagener syndrome [2,23, 24].

In subsequent conducted and histological examination of the chest: basal peribronchial infiltration, fibrous strands in the basal parts of both lungs, single foci of weakly expressed infiltration in the parenchyma of the "left" lung. Enlargement of axillary lymph nodes are on the both sides. The arc and ascending aorta, the heart chambers are turned to the right, the liver is turned to the left.

A biopsy of the ciliated epithelium of the nasal mucosa was performed. Conclusion: severe violations of the ciliary function of the epithelium.

The child was diagnosed with: Kartagener syndrome (primary ciliary dyskinesia).

At discharge, recommendations are given, including regular monitoring by a pulmonologist, district doctor and otolaryngologist, if necessary, a surdologist and cardiologist. It is recommended to carry out methods that help to clean the respiratory tract (washing the nasal passages with a hypertonic solution of sodium chloride), regular anti-relapse therapy, which includes methods that improve the function of the bronchi (drainage massage, postural drainage), carrying out inhalations with mucolytics and bronchospasmolytics. Carrying out anti-bacterial therapy and according to indications, but with prescribing in accordance with the sensitivity of the microflora to drugs after bacteriological examination of the mucus of the throat and nose, including, if necessary, reserve antibiotics. Treatment in the hospital for exacerbations and deterioration of the condition.

In the dynamics of observation, the child develops according to his age, procedures are constantly performed to improve nasal breathing and bronchial function, periodically bronchodilators and mucolytics. Signs of respiratory infection are observed 2-4 times a year, when the child receives antibacterial treatment. MSCT of the chest showed no changes in the dynamics of bronchiectasis formation.

Thanks to timely and adequate conservative therapy, the child has now been able to achieve relief of symptoms of the disease, promote normal development.

**Conclusion.** Primary ciliary dyskinesia (Kartagener syndrome) refers to inherited rare diseases, characterized by the reverse location of internal organs, the development of secondary chronic diseases of the bronchi, hypoplasia of the sinuses and the development of chronic rhinosinusitis. Early diagnosis and regular adequate conservative therapy

contribute to the slow progression of the disease without signs of bronchiectasis.

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