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Family Case of The Clinical Course of Cartagener's Syndrome in Children

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Abstract: Kartagener's syndrome (KS) is a rare hereditary disease characterized by a triad of symptoms: primary ciliary dyskinesia, situs inversus, and chronic respiratory infections. This article presents a family case of KS in children, emphasizing the clinical features, diagnostic challenges, and management strategies. The study analyzes the genetic aspects, pathophysiology, and progression of the disease in affected siblings. Special attention is given to respiratory complications, recurrent infections, and the impact on the quality of life. Early diagnosis and comprehensive therapeutic approaches, including airway clearance techniques and antibiotic prophylaxis, are crucial for improving long-term outcomes in children with KS.

Keywords: Kartagener syndrome, situs inversus, primary ciliary insufficiency.

Introduction: Kartagener syndrome (variants: Siewert-Kartagender syndrome, Kartagener syndrome) is a rare hereditary disease in humans, related to the group of ciliopathies. This syndrome is also known as Ciliary dyskinesia, primary (CILD). This syndrome was first described in 1904 by the Kyiv doctor A.K. Siewert, and later a more detailed description of this pathology and its familial forms was made by the Swedish doctor M. Kartagener in 1933. In Kartagener syndrome, there is a congenital combined malformation with a triad of symptoms (reverse arrangement of internal organs; chronic bronchopulmonary process (chronic bronchitis, pneumonia with the development of bronchiectasis); rhinosinusopathy (rhinosinusitis, nasal polyposis, recurrent otitis media) [2,3,5].

Kartagener syndrome is a common form of primary ciliary dyskinesia (PCD), which is based on structural defects of the cilia of the ciliated epithelium of the respiratory tract mucosa with the development of their immobility, which is confirmed by the saccharin test,

phase contrast, light and electron microscopy, and the radionuclide method.

The result of PCD is the formation of a chronic inflammatory process in the respiratory tract [1,2,3,4,5]. Another important manifestation of PCD is a violation of the motor activity of spermatozoa (in men) or villi of the oviduct funnel (in women), which leads to infertility [3,5].

The disease is inherited autosomal recessively, with an incidence rate of 1: 50,000 in the population as a whole [2,5].

PURPOSE OF THE RESEARCH

To study is the clinical course of Kartagener syndrome in children of the Uzbek population.

Materials and Methods

In our case, there was a clinical observation of a family case of Kartagener syndrome. At the Tashkent Medical Academy 1 - clinic, in the intensive care unit, in 2021, a brother and sister with this pathology were treated.

RESULTS AND DISCUSSIONS

Boy Umarov Sh., aged 1 month, had a wet cough and shortness of breath upon admission. It is known from the anamnesis that the child is from the 8th pregnancy, which proceeded against the background of the threat of termination at 12 weeks, anemia, colpitis, edema, uterine fibromatosis. From 3 term deliveries. Birth weight - 2580 g, body length - 49 cm. The condition at birth was severe due to respiratory failure, due to which on the first day of life the boy was transferred to the intensive care unit of the first clinic at TMA, where artificial ventilation of the lungs was performed for 80 hours. During the examination, pneumonia was detected on the chest X-ray. During dynamic observation, atelectasis of the upper lobe of the left lung was detected on the X-ray. ECHO-CS revealed VSD (ventricular septal defect) in the muscular part 5 mm, PFO (open oval window) - 3 mm. At the age of 1 month, the boy was transferred to the TMA with the diagnosis: Congenital pneumonia, severe, complicated by atelectasis of the upper lobe of the left lung, protracted course. Congenital heart disease: VSD in the muscular part, primary adaptation phase. NC0 degree. At the time of admission, an objective examination revealed a condition of moderate severity due to respiratory failure. Nasal breathing was difficult due to mucous discharge. Dyspnea of a mixed nature with a respiratory rate (RR) of 54 per minute was noted. Percussion over the lungs noted a box shade of pulmonary sound. On auscultation, weakened breathing, various wet and dry wheezing rales on both sides were heard over the lungs. The heart was determined by percussion to be on the right. The liver was determined by percussion to be on the left, of normal size. During the examination, an X-ray of the chest organs on the left revealed inflammatory infiltration of the lung tissue, mirror rotation of the internal organs, dextrocardia, the liver was located on the left, and the gas bubble of the stomach was on the right.

ECHO - CS revealed a left-formed right-located heart. VSD in the muscular part is 5 mm, OOO - 3 mm, with left-to-right blood shunt. Complete transposition of the internal organs was revealed by ultrasound. Despite the complex therapy, including inhaled glucocorticosteroids (puli-micort) for 1.5 months after the disappearance of lung tissue infiltration, the patient continued to have broncho-obstructive syndrome (BOS). A differential diagnosis was made between infectious causes of BOS, gastroesophageal reflux, and cystic fibrosis. Cystic fibrosis was excluded (sweat chlorides - the Macrodact system - 47 mmol / l (No. up to 80), feces for trypsin - positive in a dilution of 1: 160). Geneticist's conclusion: Kartagener syndrome.

The girl Umarova B., aged 11, upon admission complained of a wet cough with the separation of yellow sputum, shortness of breath, nasal congestion, and rapid fatigue. From the anamnesis it is known that the girl was born from the 2nd pregnancy, 2 term deliveries. According to the mother, until the age of 7, the girl often suffered from acute respiratory viral infections, bronchitis, nasal congestion remained almost constantly. Since the eighth year of life, recurrent bronchitis with an obstructive component has been bothering her. In 20014, dextrocardia was first detected on the ECG, and in 2021, on the chest X-ray, the inverse arrangement of the internal organs. Since the age of 10, the girl has been registered with an ENT doctor with a diagnosis of Chronic rhinosinusitis. During an objective examination, the child's condition was assessed as moderate. Nasal breathing was difficult due to nasal congestion. Mixed dyspnea was noted with a respiratory rate of -22 per minute. Percussion revealed a boxed shade of the pulmonary sound over the lungs. Auscultation revealed weakened breathing, dry wheezing on both sides. The heart was percussion determined on the right. The liver was percussion determined on the left, of normal size. During examination, signs of bronchitis and dextrocardia were revealed on the chest X-ray. The gas bubble of the stomach was located on the right, and the liver on the left. The paranasal sinus radiograph revealed signs of bilateral sinusitis. The echocardiogram showed a left-formed right-sided heart. The ultrasound showed complete transposition of the internal organs. Spirometry described signs of minimal obstruction (prolonged expiratory time). The girl was consulted by

an ENT doctor with the following conclusion: Chronic rhinosinusitis, exacerbation. Based on all of the above examination results, the final diagnosis was: Kartagener syndrome: Situs viscerum inversus. Primary ciliary insufficiency. Secondary chronic obstructive bronchitis, exacerbation. Chronic rhinosinusitis, remission. Both children received complex, symptomatic treatment: antibacterial, mucolytic, bronchodilator, including inhaled corticosteroids (pulmicort, beclazone). After discharge from the hospital, it was recommended to continue the use of inhaled corticosteroids for a long time. Physiotherapy, massage and kinesitherapy were also carried out.

CONCLUSION

Thus, our observation showed the complexity of diagnosing Kartagener's syndrome, due to the rarity of this disease, as well as a number of errors in conducting instrumental examination. For example, the sticker on the radiograph is placed in such a way that the heart is located on the left, which was observed in this case during the examination of the girl. It is also necessary to note the severity of the therapy of this disease. In our case, both children had persistent BOS, despite the long-term use of glucocorticosteroids, against which repeated episodes of broncho-obstruction were noted.

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