CLINICAL CASE

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CARTAGENE SYNDROME: PROBLEMS OF DIFFERENTIAL DIAGNOSIS

Introduction. Kartagener syndrome is a classic variant of primary ciliary dyskinesia, characterized by abnormal structure or function of cilia, leading to impaired mucociliary clearance, and manifested by chronic respiratory infections and incomplete or complete abnormal arrangement of internal organs. The rarity of this disease, the lack of a "gold standard" for diagnosis, the unavailability of high-tech diagnostic tests and low alertness of physicians, complicate the diagnosis, which leads to a late start of treatment and a decrease in the quality of life of such patients.

Description of a clinical case. The patient, 12 years old, was urgently admitted to the hospital with symptoms of iron deficiency anemia. During the physical examination, changes in the fingers of the hands of the "drumstick" type and nail plates of the "watch glass" type were recorded. From the anamnesis it was established that from the first days of life the child was diagnosed with dextrocardia, incomplete rotation of the intestine and annular pancreas. Since 1.5 years old the child has been regularly observed in medical institutions with repeated inflammatory diseases of the upper and lower respiratory tract, periodically undergoes treatment for diseases of the urinary system. Since 8 years old chronic bronchitis was diagnosed. During the examination a decrease in spirometry indicators, dextrocardia, chronic bronchitis changes in the lungs were revealed. The number of points on the PICADAR scale is 13. The set of clinical, laboratory and instrumental data, assessment on the PICADAR scale, allowed to establish the diagnosis of primary ciliary dyskinesia: Kartagener syndrome.

Conclusion. The presented clinical case clearly demonstrates the problems of differential diagnosis of Kartagener syndrome. Increasing the alertness and awareness of doctors about this disease can help in rapid diagnosis, timely treatment and improving the quality of life of such patients.

Keywords: Kartagener syndrome, primary ciliary dyskinesia, differential diagnosis, late diagnosis.

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Introduction. Ciliopathies are a group of rare hereditary diseases caused by mutations in proteins that determine the structure and functions of cilia. Due to the widespread prevalence of the ciliary epithelium in the body, cyliopathies are affected by various tissues and organs, including a number of states, including the Cartagener syndrome (KS) occupies a significant place.

Cartagener syndrome is a classic version of primary ciliary discinesia (PCD), characterized by pathology of the structure or function of the cilia leading to a violation of mucociliary clearance, and manifested by chronic infections of the respiratory tract and incomplete or complete abnormal arrangement of internal organs [11]. Currently, more than 50 genetic variants of the UK, characterized by clinical heterogeneity [3]. So, according to Kondratyeva E.I. Sub. (Moscow, 2024), patients with SK in 90% of cases are born of full-term, in 47% of cases are hospitalized in intensive care units in the neonatal period, suffer from recurrent bronchitis, pneumonia, sinusitis and otitis media in 86.6% -76% -51.6% of cases, respectively. In 28.3% of such patients, heart defects are recorded, in 5% - nasal polyposis, in 8.3% - kidney pathology [5].

Despite the presence of clinical recommendations, the diagnosis of the KS remains difficult due to the fact that the clinical picture is not specific only for the PCD, there is no “gold standard” diagnostics and all diagnostic recommendations include a combination of various anamnestic, functional, structural and molecular genetic methods [4]. Currently, the characteristic clinical- (quantitative assessment of the significance of clinical features is the PICADAR scale (from the English. Primary Ciliary Dyskinesa Rule) and instrumental data in conjunction with the results of special studies: measuring the nasal oxide (NO) in exhausted nasal air, high-speed digital. Video microscopy (to evaluate the mobility of ciliary viable cells), transmission electron microscopy (assessment of the state of ultrastructure of the axonema), immunofluorescent staining of various structural proteins and genetic testing [10]. Due to the inaccessibility for wide use of high -tech diagnostic tests in non -specialized hospitals, and the PCD are often diagnosed by the PDC is often diagnosed by late life dates or are not diagnosed at all, which leads to the late start of treatment or its absence.

The complexity of the diagnosis of PCD demonstrates the presented clinical case.

Clinical case. A child from fifth uncomplicated pregnancy, from young healthy parents who were not related. With a prenatal screening of 20 weeks, was revealed. A full-term baby with a

weight of 3600 g and a height of 53 cm was born from the second birth. Evaluation on the Apgar scale 7/7 points. Due to the manifestations of increasing respiratory disorders, she was transferred from the delivery room to the neonatal intensive care unit.

On the fourth day of life, the symptoms of intestinal obstruction developed, due to the incomplete turn of the intestines and the ring-shaped pancreas. During the examination, it was confirmed that the child has a right -handed heart, lumbar dystopia of the left kidney.

Throughout the first year of life, the condition of the child was stable. From the age of 1.5, she often (6-7 times a year) suffered from respiratory diseases, manifested in the form of laryngotracheobronchitis, bronchiolitis, and broncho-obstructive syndrome. During the convalescence period, a productive cough persisted for a long time. At the age of 5, an X-ray computed tomography examination revealed hypoplasia of the right lung. Upon repeated CT examination, at the age of 8 years, the changes characteristic of chronic bronchitis were determined: thickening of the bronchial walls, the presence of a parietal substrate (sputum). At the age of 9 years, a urolithiasis clinic appeared, and therefore, a lithotripsy was performed in the "NMIC of the health of children" (Moscow).

Preventive vaccinations were carried out according to the individual schedule. Allergic history without any special features. Neonatal screening was carried out, pathology was not detected.

At the age of 12, the child is admitted to the hospital of the regional children's clinical hospital named after N.V. Dmitrieva with signs f iron deficiency anemia.

During the physical examination, a change in the fingers of the hands according to the type of "drumsticks", a change in the nail plates according to the type of "watch glasses" was recorded (Fig. 1). Acrocyanosis of the fingers was observed, which increased in the standing position. The child's physique is asthenic. When calculating the body mass index, moderate malnutrition was detected (z-score -2.27). A number of dysembriogenesis stigmas have been identified: high palate, knee joint recurvation, and low nipple location. During auscultation of the lungs, uneven harsh breathing was detected, mainly dry diffuse wheezing is heard, more on inspiration. SaO2 96-99%. During auscultation of the heart, a short systolic murmur is heard to the right of the sternum in the fourth intercostal space. Physiological functions without special features.

Fig. 1. Changes in the fingers of the hands according to the "drumstick" type, changes in the nail plates according to the "watch glass" type

The child underwent a comprehensive examination. A general blood test revealed a decrease in the level to 66 g / l, MCV (from English Mean Corpuscular Volume) was 68.4 fl, single reticulocytes in the smear. In blood biochemistry, there is a decrease in serum iron levels to 6.0 mmol/L, serum ferritin to 2.5 mg/L, and other indicators (bilirubin, alkaline phosphatase, ALT, AST, creatinine, urea, protein, electrolytes, and CRP) are within reference values. Plasma coagulation levels are within the age range. When assessing the acid-base state of the blood, no changes were detected. Oxaluria is detected in urine: daily excretion was 209.8 mmol/day (the norm is up to 134.8 mmol/day). The study of humoral immunity (immunoglobulins A, E, M, G) is within the normal range. Screening for celiac disease is negative. The level of fecal calprotectin is <50 mcg/g. Coprology showed no signs of pathology, and no hidden blood was found in the stool.

Fibroesophagogastroduodenoscopy revealed fibrinous reflux esophagitis, cardia insufficiency, duodeno-gastric reflux, and a condition after gastroenteroanastomosis. A comprehensive ultrasound examination revealed: rotation and lumbar dystopia of the left kidney, concretions in the calyx-pelvic system of both kidneys up to 6 mm in size; uterine hypoplasia. Changes in the thyroid gland and abdominal organs have not been established. On an EchoCG, a congenital heart anomaly: a right-angled right-angled heart, pulmonary artery pressure less then 30 mmHg.

Computed tomography (CT) of the chest organs shows signs of congenital malformations: dextrocardia, hypoplasia of the right lung (due to the middle and lower lobes); transposition of the right upper lobe bronchus into the area of the middle lobe and lower lobe bronchus; signs of chronic bronchitic changes in the acute stage (thickening of the walls of the distal bronchi on the right; parietal substrate (sputum) in the lumen of large bronchi and trachea; multiple centrilobular nodules and Y–shaped structures with the formation of a "tree in the kidneys" pattern, lobular seals) (Fig.2 and 3). According to spirometry data, there was a marked decrease in forced vital capacity and forced exhalation volume after a salbutamol test without dynamics. An MRI scan of the brain revealed no pathology.

The child is rated on the PICADAR scale, the number of points is 13 ("diagnosis is valid") [2].

The combination of clinical, laboratory and instrumental data, assessed on the PICADAR scale, made it possible to establish the diagnosis of primary ciliary dyskinesia: Kartagenera syndrome.

Discussion. The presented clinical case demonstrates the complexity of the

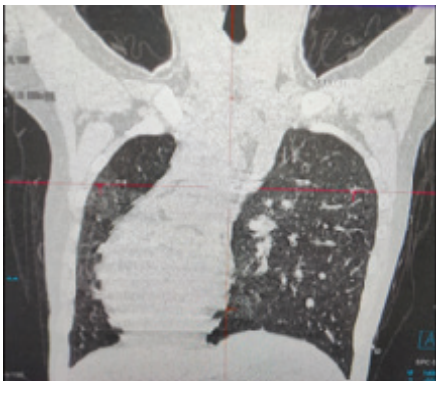

Fig. 2. Computed tomography of the chest. Dextrocardia (right-sided, right-formed heart)

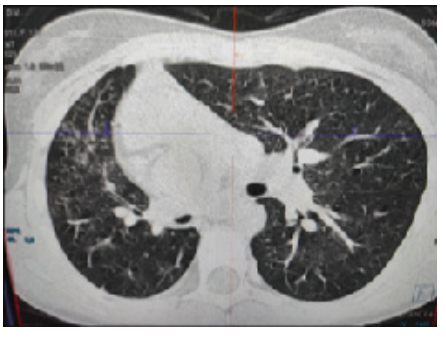

Fig. 3. Computed tomography of the chest. Thickening of the walls of the distal bronchi on the right; parietal substrate in the lumen of large bronchi and trachea; multiple centrilobular nodules and Y-shaped structures with the formation of a "tree-in-bud" pattern

diagnosis of PCD. The average age of verification of this disease is an important indicator of the effectiveness of the healthcare system in detecting a rare pathology in children [1,6]. Thus, according to A.A. Novak (Moscow, 2024), the average age of verification of the disease in the Russian Federation is 5.8 years, in the Australian cohort of patients, the diagnosis is established at the age of 6.4 years, in China-8.2 years, and in South Korea-11.8 years [3,13,7,8].

Due to the relatively low frequency of the KS, the variety of phenotypes, the lack of screening, low awareness and alertness of doctors, the diagnosis of PCD is often postponed for years, which significantly reduces the quality of life of such patients [12]. According to the clinical recommendations of the Russian Federation, as well as the diagnostic recommendations of the European respiratory and American thoracic societies, patients with constant productive coughing, the anomalies of the location of the internal organs and congenital heart defects are subject to an additional comprehensive examination using high -tech diagnostic tests for the verification of PCD [10,9].

In the case presented from the first days of life, the child was diagnosed with dectrocardia, incomplete intestinal rotation and ring-shaped pancreas. From the age of 1.5, the child is regularly observed in medical institutions with repeated inflammatory diseases of the upper and lower respiratory tract, and periodically undergoes for diseases of the urinary system. From the age of 8, chronic bronchitis was diagnosed. This child, despite the frequency of outpatient visits and inpatient hospitalizations, was not comprehensively examined, and did not take into account the dispensary accounting for chronic bronchitis.

Conclusion. The presented clinical case clearly demonstrates the problems of the differential diagnosis of the KS. An increase in the awareness of doctors about this disease can help in quick diagnosis, timely treatment and improve the quality of life of such patients.

The authors declare no conflict of interest.

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A FAMILY CLINICAL CASE OF COMBINATION OF TWO MENDELIAL DISEASES: SPINOCEREBELLAR ATAXIA TYPE 1 AND HYPOPHOSPHATEMIC RICKETIS

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The combination of two genetic syndromes in a single patient is a rare occurrence. This article describes a clinical case of a rare combination of two Mendelian diseases: spinocerebellar ataxia type 1 and hypophosphatemic rickets in a single Yakut family. Given the low incidence of both diseases, this finding is of scientific and practical interest. The paper discusses a clinical observation of family members examined in 2012 and 2025. This clinical example is also relevant for practicing physicians. It is necessary to develop algorithms for monitoring complications of spinocerebellar ataxia and phosphate diabetes and to identify pathogenetic therapy.

Keywords: spinocerebellar ataxia type 1, hypophosphatemic rickets, bone deformity, phosphate diabetes, family case.