KARTAGENER SYNDROME: AN UNUSUAL CONDITION WITH NUMEROUS DIFFICULT OPTIONS : CASE REPORT AND REVIEW OF LITERATURE

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Authors' contributions

This work was carried out in collaboration between all authors. Author GYA designed the study, wrote the protocol and wrote the first draft of the manuscript. Author SLA managed the literature searches and manuscript editing and manuscript review. All authors read and approved the final manuscript.

ANNOTATION. Approximately 50 % of patients with primary ciliary dyskinesia have Kartagener syndrome, which classically described by a triad including dextrocardia, chronic sinusitis and bronchiectasis. Resynchronous dyskinesia, mucociliary clearance disturbances cause disturbances in the drainage function of the airways, which is the main cause of recurrent infections of the upper and lower respiratory tract - chronic rhinosinusitis, otitis media, chronic bronchitis with the formation of bronchiectasis, recurrent pneumonia. The given example of clinical observation showed the whole complexity of diagnosis of the Kartagener syndrome caused by the rarity of this disease.

KEYWORDS: Kartagener syndrome; Genetic disease; Pneumonia; Ciliary dyskinesia; Dextracardia

INTRODUCTION

Kartagener syndrome is a variety of genetically determined autosomal recessive disorder, primary ciliary dyskinesia, characterized by ciliary dysfunction and impaired mucociliary clearance [2, 6]. Approximately 50% of patients with primary ciliary dyskinesia have Kartagener syndrome, which is classically described by a triad, including dextracardia, chronic sinusitis and bronchiectasis [3, 7, 8]. M. Kartagener first recognized this clinical triad as a separate congenital syndrome and in 1933 presented its detailed description [5]. The frequency of the syndrome estimated as 1:

15000–30000 cases [1, 9]. Dextracardia can be combined with the normal disposition of other organs (situs inversus solitus), in some patients there is a mirror arrangement of all internal organs (situs inversus totalis) [1]. Ciliary dyskinesia, impaired mucociliary clearance can cause impaired drainage function of the airways, which is the main cause of recurrent infections of the upper and lower respiratory tract - chronic rhinosinusitis, otitis media, chronic bronchitis with the formation of bronchiectasis, recurrent pneumonia [4, 10, 11]. There is no evidence-based treatment for patients with Kartagener syndrome. Long-term prophylactic and therapeutic courses of antibiotic therapy are usually used, mucolytic are used, and in case of bronchoobstructive disorders inhalation of bronchodilators, glucocorticosteroids are used. If necessary, use surgical methods of treatment [10]. Next, we present a clinical case from our practice.

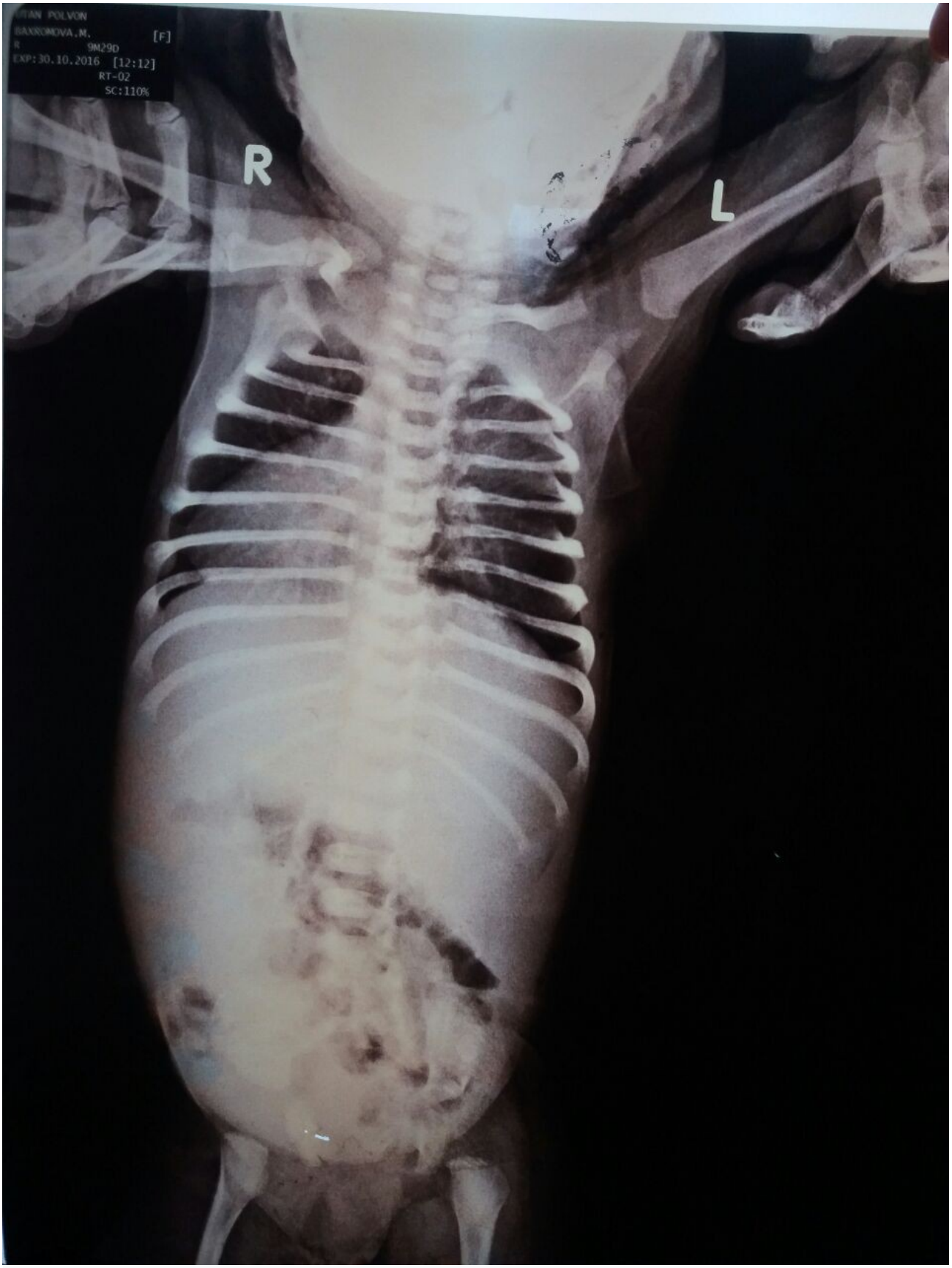
CASE REPORT

Patient M., born 12.09.2016, was admitted to the emergency department of the 1st clinic of the Tashkent Medical Academy on November 2, 2016, complaining of coughing, shortness of breath, fever, difficulty breathing. The patient immediately hospitalized to the neonatal pathology department. The child, together with the head of the department, examined by a pediatrician and diagnosed “Out of hospital pneumonia, acute phase, respiratory failure 2 degrees, perinatal injury of the central nervous system (PICNS), congenital heart failure (Patent foramen ovale, false chordae tendinae)”.

From the anamnesis of the disease: according to the mother has been ill since birth. After discharge from the hospital on day 4, the child became restless, coughing and shortness of breath appeared. Immediately returned to the pediatrician. Recommended inpatient treatment at the place of residence. The child hospitalized. In the hospital, a chest x-ray, echocardiography, ultrasonography of internal organs and other laboratory tests performed.

Diagnosis: Community acquired pneumonia, acute phase, cardiovascular insufficiency 1 degree. Dextracardia. Congenital heart failure. Atrial septal defect. Appointed antibiotic therapy, hormone therapy, mucolytic. Despite the therapeutic measures, the child’s condition remained severe and, according to the indication, transferred to our clinic for in-depth examination and inpatient treatment. In the emergency department experts, examine the child. Studies conducted echocardiography, neurosonography, chest X-ray, ultrasound of internal organs.

Diagnosis: Community acquired pneumonia, lobar, acute phase. Cardiovascular insufficiency. False chordae tendinae. Dextracardia.



Pic.1. Plan x-ray film of chest and abdomen

From the anamnesis of life: it known that the child is from the third pregnancy, 2 urgent deliveries. First child died on day 3 of life. Weight at birth 3200 gr. The navel dropped on day 15; day 3 discharged from the hospital. Heredity not burdened. Status praesens: General condition of severe. The right physique. Musculoskeletal system without pathology. The skin is clean, pale, with a marble shade. Subcutaneous fatty tissue is poorly developed. Large spring 3.0 x 2.5 cm, does not swell, pulsates. Nose breathing is moderately difficult. Tonsils are friable. The arms are moderately hyperemic. Cough wet, painful, and obsessive.

With auscultation of the lungs - breathing hard. In the lower sections of the left lung moist fine wheezing. The respiratory rate - 60-62/minute. In the act of breathing, the auxiliary muscles are involved. When percussion in the lungs is determined by the dulling of sound. With percussion, the heart is determined on the right. The boundaries of relative cardiac dullness within the age norm. Heart rate is 144. The liver is percussion determined on the left, increased by + 2.5 +, 2.0 + 1.5 sm, the spleen on the right increased by + 5.0 + 4.5 sm. Additional examination methods conducted: Radiograph of the chest organs in a direct projection. Determined by the right-sided position of the heart and gas bubble of the stomach. Pulmonary drawing is strengthened, infiltrative foci on the left. The left dome of the diaphragm is higher than the right. The sinuses are free. Conclusion: Dextracardia. Signs of pneumonia.

Ultrasound of the abdominal organs: Situs inversus and totalis. Reverse location of the abdominal organs.

Complete blood count: Red blood cells - 2.8 mcL. Hemoglobin – 10 g/dL. Color indicator - 0.76. Leukocytes - 11.0 × 10^9/L, eosinophils 9%, neutrophils 2,000/mm, monocytes 3%. ESR 14 mm/hour.

Clotting time from 4 min 30 sec to 5 min 00 sec.

Urinalysis: Density - 1010. Protein - traces. Sugar is negative. Leukocytes - 8-10 in sight. Erythrocytes - 1-2 in sight. Biochemical analysis of blood: Total bilirubin - 17.5 μmol / l. Urea - 5.9 mmol / l.

Based on all the above results of the examination, a final diagnosis was made: “Community acquired pneumonia, lobar form, acute phase. Kartagener syndrome. Respiratory failure II degree. Perinatal injury of the central nervous system (PICNS), with excitation syndrome. Congenital heart failure. Atrial septal defect. False chordae tendinae. Hepatosplenomegaly. Moderate anemia.

For several days, the patient underwent a complex, symptomatic treatment: antibacterial, mucolytic, hormonal therapy, bronchodilator, detoxification therapy.

As a result, the patient's condition improved - shortness of breath decreased, the amount of sputum separated, and the body temperature returned to normal. The patient discharged in satisfactory condition under the supervision of a pediatrician at the place of residence. Thus, our observations have shown the complexity of diagnosing Kartagener syndrome due to the rarity of this disease. It should also be noted the severity of the treatment of this disease.

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