

Case 15

Caso 15

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Figure 1 - Chest x-ray in PA.



Figure 2 - Picture of the patient's left hand.

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CASE

Female patient, eight years old, attends the PA of the HC-UFMG with fever, productive cough, vomiting, and dyspnea, with seven days of evolution. On physical examination presented digital clubbing, respiratory effort, SatO₂ of 67% in room air, and bilateral and diffuse crepitations. Patient in follow-up in the Pediatric Pneumology outpatient clinic, with previous cultures of oropharynx secretion positive for *Staphylococcus aureus* sensitive to oxacillin (OSSA), *Burkholderia cepacea*, and *Stenotrophomonas maltophilia*.

Based on clinical history and images, the most likely diagnosis is:

- primary ciliary dyskinesia;
- acute viral bronchiolitis;
- cystic fibrosis with pulmonary exacerbation;
- obliterating bronchiolitis.

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IMAGE ANALYSIS



Figure 3 - Picture of the patient's left hand showing digital clubbing (red circles), alterations also known as "hippocratic fingers" or "drum sticks," common in patients with cystic fibrosis due to hypoxemia.

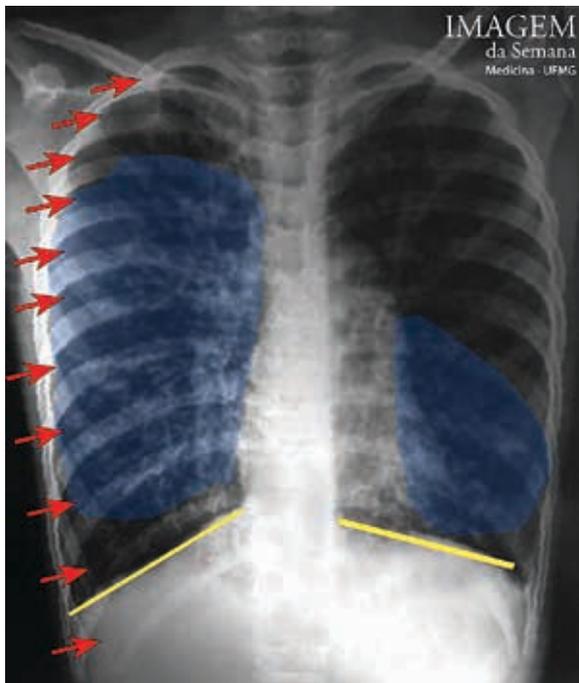


Figure 4 - Chest x-ray where pulmonary hyperinflation is observed – visualization of 11 intercostal spaces (red arrows), extended beyond the rectification of diaphragmatic domes (yellow lines). Reticular opacities are also present (blue shadows) around cystic images in the upper lobes, compatible with peri-bronchial thickenings and bilateral infiltrates, with evidence of bronchiectasis. Notable increased cardiac area due to hyperinsufflation. In later stages, increased cardiothoracic index might occur due to the emergence of cor pulmonale.

DIAGNOSIS

In patients with cystic fibrosis (CF), the pulmonary exacerbation can manifest with cough, increased sputum, changing in color of secretions, weight reduction, hypoxia, and fever. Hisses, crepitations, and snoring fuzzy can be auscultated.

On primary ciliary dyskinesia, the patient presents history of repeating respiratory tract infections, medium otitis, bronchitis, and rhinosinusitis due to genetic alteration in mucociliary transport. The Kartagener syndrome is the classical genetics manifestation of the disease.

The acute viral bronchiolitis (BVA) is an infection of viral etiology, prevalent until two years of life, manifesting itself predominantly in infants below the age of six months. There is initial involvement of the upper airways, progressing to lower airways in a few days. The diagnosis is clinical and the infection is self-limiting. Because it is a viral infection, BVA may reoccur.

The obliterating bronchiolitis is a syndrome of chronic obstruction of the airflow associated with inflammatory lesion of the small airways. In children, most often, it is preceded by infection of the lower airways, mainly by adenovirus, with fever, cough, wheezing, and tachypnea.

DISCUSSION OF THE CASE

CF is a chronic and progressive autosomal recessive disease that affects the exocrine glands of multiple organs. The triad of clinical characteristics consists of repeating pulmonary infections, pancreatic failure, and growing deficit. There is no cure for the disease; however, support measures are needed to slow the evolution of lung damage.

The gold standard test for diagnosing patients with CF is the sweat test. Sodium levels exceeding 60 millimoles per liter, in two dosings, associated with characteristic clinical picture indicates that the person is a carrier of the disease. After diagnosis, ambulatory multidisciplinary outpatient follow-up starts with a pulmonologist, gastroenterologist, nurse, dietitian, and physiotherapist.

Pulmonary involvement is largely responsible for morbidity. Thus, the analysis of culture of airway with tracheal aspirate, oropharynx swab, or sputum

is performed in patients aiming at identification of potentially pathogenic bacteria such as *Pseudomonas aeruginosa* (PA), *Staphylococcus aureus* sensitive or resistant to oxacillin (OSSA and ORSA, respectively), *Haemophilus influenzae* (HI), and *Burkholderia cepacia* complex (BCC). The identification and early treatment of lung infections are essential to slow down disease evolution to bronchiectasis and lung function loss.

The initial lung injury is characterized by dilation and hypertrophy of mucous glands, followed by the emergence of metaplasia, plugs of mucus in peripheral airways, secondary ciliary alterations, and lymphocytic infiltrate in the submucosa. There is evolution to bronchiectasis, with repeated obstruction and infection. Exocrine pancreatic insufficiency is characterized by chronic diarrhea, bulky stools, and bright, greasy, and fetid evacuations. Sterility and digital clubbing are other clinical manifestations of the disease.

The survival rate has been increasing over the years. It is estimated that life expectancy, which was in average of 35 years, could reach 50 years with early diagnosis by neonatal screening.

RELEVANT ASPECTS

- CF is a severe autosomal recessive disease that affects exocrine glands in multiple organs (especially lungs and pancreas);
- the most common clinical manifestations are persistent chronic cough, chronic diarrhea, and malnutrition;
- the diagnosis is made by clinical history and positive sweat test;
- there is no cure for cystic fibrosis yet;
- it is important, during the outpatient follow-up, to take supporting measures to slow the progression of the disease.

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