Bronchiectasis in cystic fibrosis and pregnancy

Bronșiectazii în fibroza chistică și sarcină

Abstract

Cystic fibrosis is an autosomal recessive disease with a prognosis determined by the extent of pulmonary lesions. Here we present a rare case of a patient diagnosed with cystic fibrosis in the pediatric service, but who came for the first time to the Pneumology Hospital of Iasi. Respiratory failure, recurrent pulmonary infections and associated metabolic modifications generate a high vital risk during pregnancy.

Keywords: *cystic fibrosis, pregnancy, respiratory failure*

Rezumat

Fibroza chistică este o afecțiune autozomal recesivă al carei prognostic este determinat de gradul de extindere al leziunilor. Prezentăm un caz rar al unei paciente cunoscută cu diagnosticul de fibroză chistică în serviciul de pediatrie dar la prima prezentare la Spitalul de Pneumologie din Iași. Insuficiența respiratorie, infecțiile pulmonare recidivante, precum și modificările metabolice asociate imprimă un risc vital crescut în sarcină.

Cuvinte-cheie: *fibroză chistică, sarcină, insuficiență respiratorie*

Adina Turcanu, Traian Mihăescu, Cristian Cojocaru

"Grigore T. Popa" University of Medicine and Pharmacy of Iasi, Faculty of General Medicine

Clinic of Pulmonary Diseasesrgery, Coltea Clinical Hospital, Bucharest, Romania

Corresponding author:
Adina Turcanu,
E-mail: turcanu.adina@umfiasi.ro

Introduction

Cystic fibrosis is a genetic autosomal recessive disease, which occurs most frequently because of the DF508 mutation, which subsequently determines its defective gene to produce an abnormal protein called CFTR. Its consequences can be at pulmonary and digestive level. This disease is currently being diagnosed on 70 000 adults and children all over the world, and new 1 000 cases are identified every year. 70% of the patients receive this diagnostic by the age of two[1-3].

Case presentation

Herein below we present the case of a patient aged 21 years old, living in an urban area, who has been admitted at the Pneumology Hospital of Iasi for: dyspnea at small efforts, 3rd degree mMRC, productive cough accompanied by expelling mucus and pus (150 ml/day).

Hereditary and collateral antecedents showed that the patient had a sister who died due to a chronic pulmonary disorder.

The personal physiological antecedents confirm the existence of an ongoing pregnancy (10 weeks), and the medical history revealed that in 2011, at the age of 12, she was diagnosed with cystic fibrosis with pulmonary events via CT scan (cystic bronchiectasis) and was tested positive for the sweat test – Na = mEq/l, Cl = 160 mEq/l. By the age of 18, the patient was repeatedly admitted in the paediatrics departments for recurrent bronchial and pulmonary infections.

The patient works in the sewing industry, in a dusty and puffy environment, benefitting, since she came of age, from medical care, but only from her family doctor, who recommended aerosol-therapy, thoracic tapotement and medical gymnastics.

The clinical exam reveals an influenced, underweight overall condition (BMI = 17.3 kg/m^2), pale facies, teguments and mucous, nail clubbing, connective and fat tissue poorly represented, hypotonic, hypotrophic muscular system.

The exam of the respiratory system highlights a hyposthenic thorax, physiological vesicular murmur bilaterally roughened, bilateral basal crackles.

Following the case history and the clinical examination, the possible diagnostics considered are: cystic fibrosis with pulmonary events, idiopathic bronchiectasis over infected with bacteria or virus.

The conducted paraclinical investigations revealed the following pathological values: a peripheral oxygen saturation of 88%, neutrophilic leukocytosis, mild thrombocytopenia, low serum Fe and Hb, and normal erythrocytes indices (possibly also due to the pregnancy), presence of the systemic inflammatory syndrome, normal values of the urea, creatinine, and LDH.

The arterial blood gas performed under oxygen administration, 2 litres per minute, highlights the existence of hypoxia and alkalosis (PO_2=63 mmHg, Ph = 7. 498, Na^+=134.1 mmol/L, K^+=3.45 mmol/L, Ca^{2+}= 0.77 mmol/L, PCO_2 – could not be processed).

At the same time, new samples of sputum have been collected, on which the bacteriological exam for BAAR has been conducted, with negative result every time. The bacteriological exam of the sputum identified under the microscope the existence of gram-negative cocci, gathered together, significantly associated to inflammatory cells, and the culture confirmed the existence of *Staphylococcus aureus*, sensitive only to teratogenic antibiotics during the pregnancy (lyncomycin, vancomycin, linesolid, chloramphenicol, teicoplanin).

The examination of the pulmonary function detected a severe mixed ventilatory dysfunction (CV=52.4%, FVC= 54.1%, FEV1= 32.0% of the predictions).

No chest X-ray has been conducted because the patient was pregnant and it was not deemed necessary.

The genetic test necessary to confirm the cystic fibrosis was recommended, but it was not performed due to financial reasons.

The clinical, anamnestic and paraclinical data led to the diagnostic of cystic fibrosis with pulmonary events, bilateral bronchiectasies over infected with methicillin-resistant *Staphylococcus aureus*, chronic respiratory deficiency and ongoing pregnancy, 10 weeks gestation.

Regarding the treatment, the patient has been explained the potential teratogenic effects of the efficient antibiotics necessary to be administered, as well as the existence of the option of therapeutic abortion, since the pregnancy was unlikely to reach term and the complications could be severe.

The patient refused being administered any type of antibiotic that could influence the condition of the foetus and declared that she was aware of the existing risks that would result from changing the treatment, including of the risk of death, of both the foetus and herself. In addition, the patient declares that she knows the illness and the outcome, since her sister had died of the same disorder, but she wanted to have the child, regardless of the consequences.

Considering the patient's option, it was decided to administer aerosols with acetylcysteine, amoxicillin and clavulanic acid, 1 gram every 8 hours, aerosols with Colistin, respiratory gymnastics, thoracic tapotement, oxygenotherapy 2-3 litres/minute.

Following the hydroelectrolytic rebalance, the patient was discharged with the advice to continue the therapy with aerosols, acetylcysteine and phosphomicine, 1 sachet 3 times a day, for 10 days.

During the pregnancy, after 2-3 weeks the patient has returned for admission with phenomena of respiratory deficiency, accompanied by expelling mucus and pus, fever. Each time, apart from the previously discussed treatment, the patient was advised to undergo home therapy with oxygen. Upon every admission, the patient declared that she was not able to obtain an oxygen concentrator at home. Each gynaecologic and pulmonary check-up raised doubts about the neuro-psychiatric development of the foetus, given the chronic hypoxia of the mother.

In the 30th week of pregnancy, the patient is brought by an ambulance from home, with mild phenomena of respiratory deficiency, fever, and change in the overall condition. She is admitted in the Intensive Care Unit of the Pneumology Hospital. During the night, she develops a febrile episode of 39° C. The gynaecological hospital on duty was contacted and it was performed an emergency C-section. Following the C-section, the antibiotic treatment was administered, focusing on Staphylococcus aureus with double association.

Two months after the birth, the patient is oxygen-dependant (3 litres/minute), she has 3rd-4th degree dyspnea mMRC, asthenia, cough accompanied by expelling of mucus and pus. The specific treatment with dornase alpha cannot be administered due to financial reasons and of the fact that she falls under the category of patients that cannot benefit from free treatment. It is stated that it was attempted to rank the patient in the class of pregnant or post-partum in order to achieve the approval from the Health Insurance House for free treatment within the cystic fibrosis programme, but because of the bureaucracy, the patient exceeded the period of pregnancy and post-partum by the time a part of the documents were approved, so the advised treatment could not be administered. The child (a baby girl) is healthy, but she is always exposed to her mother's recurrent infections.

Dicussions

Although fertility in female patients with cystic fibrosis is present, and they can reach term under proper medical surveillance, in this case, it was recommended therapeutic abortion right from the beginning, since the pulmonary function was severely impaired and the drug-resistant *Staphylococcus aureus* can lead to the death of both the mother and the child. The refusal of the patient to take any medication that could have prevented the occurrence of malformations and the contraindications of the gynaecologists, made the pulmonologists temporise the proper treatment[2,4,5].

The outcome of the patients with cystic fibrosis in the absence of the advised treatment is severe. In the case presented herein, the impossibility to administer the proper medication led to the change of patient's pulmonary function, who became dependant to oxygen therapy. She was eventually included in the adult programme for cystic fibrosis. ■

References

1. Pop L, Popa Z, Cilt C. Ghid de management in mucoviscidoza. *Ro Med J.* 2009; 2:98-111.
2. http://www.cff.org/AboutCF/ (accessed 7.01.2013).
3. Flume PA, O'Sullivan BP, Robinson KA et al. Cystic Fibrosis Pulmonary Guidelines Chronic Medications for Maintenance of Lung Health. *Am J Respir Crit Care Med.* 2007; 176: 957-969.
4. Sanders DB, Bittner RC, Rosenfeld M et al. Failure to Recover to Baseline Pulmonary Function after Cystic Fibrosis Pulmonary Exacerbation. *Am J Respir Crit Care Med.* 2010; 182: 627-632.
5. Rosenfeld M, Emerson J, Williams-Warren J et al. Defining a pulmonary exacerbation in cystic fibrosis. *J Pediatr.* 2001; 139: 359-365.