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Reporte de caso: Paciente Peruano de 52 años con Fibrosis Quística

A Case Report: 52 Year Old Peruvian Patient with Cystic Fibrosis

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Abstract

The Cystic fibrosis (CF) is an autosomal and recessive genetic disease. It affects much more frequently the population of Caucasian origin, where their incidence ranges from 1 in 3,000 in Caucasians to 1 in 8,000 in newly Hispanic live births. By 2018, 30,775 patients with cystic fibrosis, of them less than 10% of patients worldwide had more 40 years and none had been diagnosed after 40 years of age. We present below the case of a 52-year-old Peruvian adult with CF. This would be one of the patients with the latest diagnosis worldwide and the longest patient in Peru and one of the longest in Latin America.

Keywords: cystic fibrosis, cystic fibrosis in adults.

Resumen

La fibrosis quística (FQ) es una enfermedad genética autosómico y recesiva. Afecta con mucha más frecuencia a la población de origen caucásico, donde su incidencia varía de 1 entre 3 000 en caucásicos a 1 entre 8 000 en hispanos recién nacidos vivos. Para el 2018 se encontraban registrados 30 775 pacientes con fibrosis quística, de ellos menos del 10% de pacientes a nivel mundial tenían más de 40 años y ninguno había sido diagnosticado luego de los 40 años de edad. Presentamos a continuación el caso de un adulto peruano de 52 años con FQ. Este sería uno de los pacientes con el diagnóstico más tardío a nivel mundial y el paciente más longevo del Perú y uno de los más longevos de América Latina.

Palabras clave: fibrosis quística, fibrosis quística en el adulto.

Introduction

The Cystic fibrosis (CF) is an autosomal and recessive genetic disease characterized by the abnormal ion transport, caused by a defect in the gene encoding a transmembrane conductance regulatory protein: cystic fibrosis transmembrane conductance regulator (CFTR) (1). It affects the population of Caucasian origin much more frequently, where its incidence varies from 1 in 3,000 in Caucasians to 1 in 8,000 in Hispanic live newborns (2).

A research indicates that the patients diagnosed in adulthood have a relatively better lung function, a lower frequency of digestive involvement and the sweat test can be negative or positive reaching a mean age at death of 31 years for those diagnosed in childhood and 40 years old in the group diagnosed in adulthood (3) (4). By 2018, 30 775 patients with cystic fibrosis were registered, of them less than 10% of patients worldwide were over 40 years old and none had been diagnosed after 40 years of age. We present below the case of a 52-year-old Peruvian adult with CF who gave his informed consent for this report. This would be one of the patients with the latest diagnosis worldwide and the longest-lived patient in Peru and one of the longest-lived in Latin America (4) (5) (6) (7).

Description

He went to the hospital with a history of "pulmonary tuberculosis" at the age of 12 years diagnosed with chronic cough and mucopurulent sputum, receiving treatment for 4 months. Since then, he has persistent mucous expectoration. When he was 17 years old, he was diagnosed with "chronic sinusitis." He was also diagnosed with bronchiectasis at the 40 years old and reports monthly episodes of hemoptoic expectoration of approximately 5-10 mL. The patient enters to the hospital with the diagnosis of bronchiectasis and probable pulmonary tuberculosis. He was awake, overweight, and had thick crackles scattered in both lung fields. The vital functions are normal and the rest of physical examinations without any alterations. He does not present acropaquias.

Analytical study: Hemoglobin = 16.0g / dl, Hematocrit 47.6%, Leukocytes = 10.8 x109 / L, neutrophils 64.1%, eosinophils 1.3%, platelets 255x109 / L. Total proteins, glycemia, urea,

creatinine, sodium and potassium within the normal ranges. C Reactive Protein 2.9 mg / dl. HbA1C: 8%.

The multiple sputum examinations to investigate the presence of Mycobacterium tuberculosis were negative. As part of the study of bronchiectasis, rheumatology markers were performed: The Negative antinuclear antibody (Ac) (ANA), Negative Ac Anti Cytoplasm (ANCA) negative, Ac Anti DNA negative, Anticardiolipin negative, mRNP / sm, sm, SS-A , SS-B, ScL-70, Jo-1, ds-DNA. The liver function study was within the normal ranges and the hepatitis B and C study was negative. The Tumor markers (AFP, Cyfra 21.1, Neurospecific Enolase) were within normal values.

The study of microbiological agents for the cytomegalovirus, toxoplasma, herpes virus type 1 and 2 all negative. The serology study for HIV Non-Reactive, HTLV 1 and 2 are Negative. Due to the history of not having children, a spermiogram was requested with the following result: Azoospermia and hypospermia. After this, the electrolytes in sweat were requested: First test: 81 mmol / L of NaCl and second test: 83 mmol / L of NaCl (both positive) performed by the Iontophoresis technique (Pilocarpine) with the Nanodut AC-081 kit. The stool analysis negative for inflammatory reaction and no steatorrhea found. The Immunolgobulin assay A, M, G, and E: Within Normal Values. In the computed tomography of the paranasal sinuses, the brain, the thorax, the abdomen and the pelvis, we found: turbinate hypertrophy and thickening of the mucosa of the left maxillary sinus and hypoplasia of the same sinus, as well as a hydro-aerial level in the right maxillary sinus. At the pulmonary level, multiple cystic bronchiectasis is observed, some of them with content, some bilateral central and peripheral cylindrical bronchiectasis (Figure 1). The rest of the organism did not show alterations. The Sputum results reported that the bacterial growth corresponded to the E. Coli. The sensitivity to antibiotics over 9 years has not varied, remaining sensitive to amikacin, aztreonam, ceftazidime and cefepime as fundamental medications. Mushrooms were not isolated. The echocardiogram showed no alterations in myocardial function or cardiac chambers, no increase in pulmonary artery systolic pressure was observed.

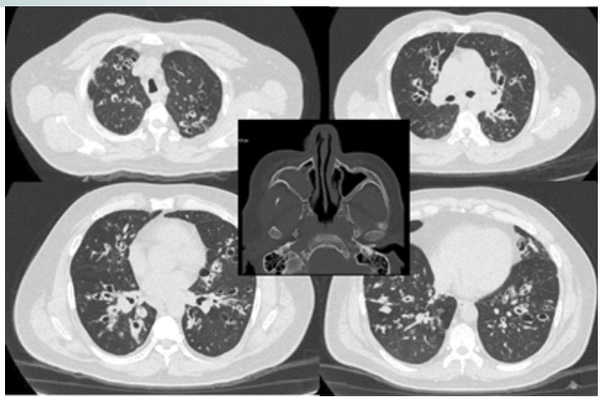

Figure 1. Image obtained from the Multi-slice Spiral Tomography of the chest corresponding to December 2019: The cystic bronchiectasis is observed, some with content and bilateral diffuse bronchioloectasias. The Axial tomography that corresponds to the maxillary sinuses showing the occupied right maxillary sinus

For chronic pulmonary infection associated with E. coli, he received inhaled suppressive antibiotic treatment with amikacin at a dose of 1 gram every 12 hours in a first cycle of 28 days with treatment and 28 days without treatment. The sputum bacteria have not been eradicated. No variation was found in the resistance and sensitivity profiles in the antibiogram. There have been no exacerbations requiring hospitalization. He received daily inhaled treatment with alpha dornase 2.5 mg / 2.5 mL once daily. The inhaled medication is received through a PARI LC® Plus nebulizer.

In the pulmonary function tests we found a progressive deterioration of the pulmonary function that currently corresponds to a very severe obstructive alteration (FEV 34%) with a decrease in the forced vital capacity (56%), with deterioration of approximately 20% per year. The total lung capacity by plethysmography is increased (156%) at the expense of residual volume and functional residual capacity (210%) and with a moderately decreased Carbon Monoxide Diffusion (hemoglobin-corrected DLCO of 45%). The patient uses high doses of ipratropium bromide (MDI 20 ug / puff: dose 4 puff every 4 hours) with the intention to reduce the functional residual capacity. Other studies such as the requested amylase and lipase were in normal ranges and in the ultrasound renal study microlithiasis was found without clinical significance. Currently, the patient is stable, with an occasional cough, little

bronchorrhea, no hemoptysis, he carries out his daily activities, but he shows greater limitation. He is evaluated every three months in the external pneumology office, at the date he has not presented exacerbations, aggravation or required hospitalization. He tolerates the medication very well and he has not reported or shown adverse reactions.

Discussion

The CF patients diagnosed in adulthood generally have milder symptoms, with fewer complications, making the diagnosis more difficult. In this patient, the suspicion of cystic fibrosis was based on the presence of: bronchiectasis and infertility, which is why the sweat chlorine test was performed, which confirmed the clinical diagnosis, although the diagnosis of pulmonary tuberculosis was initially strongly considered due to that the prevalence of tuberculosis is high in Peru. The susceptibility to chronic infections present in CF conditions the development of bronchiectasis and the progressive destruction of the lung parenchyma, while alterations in the vas deferens are associated with the presence of infertility in men. Although the main cause of bronchiectasis in Peruvian adults is that caused by sequential lesions after pulmonary tuberculosis, other differential diagnoses such as CF, primary ciliary dyskinesia, and primary immunoglobulin deficiencies should not be completely ruled out and should always be considered as diagnoses. The study of other bacteria in sputum is important as a cause of chronic infections and functional and structural deterioration of the lung parenchyma.

Although the diagnosis of CF in this case is clear, a limitation is not have a complementary genetic tests for this disease that help us to know which variants in the mutations are associated with greater survival and less systemic compromise (5). This patient could be the oldest adult with CF in Peru and be among the oldest in Latin America, unfortunately, there is no Peruvian or Latin American registry of CF patients (7).

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