ARCHIVES OF HELLENIC MEDICINE: ISSN 11-05-3992

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ARCHIVES OF HELLENIC MEDICINE 2019, 36(3):430–431
ΑΡΧΕΙΑ ΕΛΛΗΝΙΚΗΣ ΙΑΤΡΙΚΗΣ 2019, 36(3):430–431

Medical Imaging Quiz – Case 55

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An 18-year-old man presented to the emergency department due to persistent cough and hemoptysis. He lived abroad in a rural area and he had never visited a physician before. Relatives referred multiple respiratory infections.

Physical examination revealed temperature of 37.3 °C, pulse rate: 85/min, respiratory rate: 18/min, blood pressure: 128/85 mmHg, and pathologic auscultatory sounds. Finger clubbing was remarkable. A chest computed tomography (CT) was performed and revealed multiple bronchiectasis (fig. 1). Laboratory investigation showed elevated ALT, AST, and sputum culture revealed *Pseudomonas aeruginosa*.

Comment

Cystic fibrosis (CF) is an autosomal recessive genetic disease that affects the exocrine function of the lungs, liver, pancreas, and small bowel resulting in progressive disability and multi-system failure. CF is the most common genetic disease affecting European population with an incidence of approximately 1:2,000–3,500 live births.

Pulmonary manifestations of CF includes bronchiectasis, pneumothorax, recurrent bacterial infection and pulmonary arterial hypertension. CF causes dysregulation of pancreas (exocrine and endocrine insufficiency, fatty replacement of pancreas, pancreatitis, pancreatic cysts), liver (hepatic steatosis, focal biliary and multilobular cirrhosis, portal hypertension, cholelithiasis, sclerosing cholangitis and gastrointestinal tract (distal intestinal obstruction syndrome, meconium ileus).

The diagnosis may be suspected antenatally due to the presence of echogenic bowel on antenatal ultrasound, or due to genetic testing of the parents. Diagnosis of CF after birth is tested with a sweat test (positive sweat chloride test Cl >60 mEq/L) or immediate genetic testing. The diagnosis usually becomes evident in infancy, with presentations including meconium ileus, rectal prolapse and recurrent pulmonary infection. Delayed diagnosis of CF is rare nowadays.

CF is due to a defect of the cystic fibrosis transmembrane regulator (CFTR) gene located on chromosome 7q31.2. This gene encodes for a transmembrane protein known as CFTR which is responsible for regulating chloride passage across cell membranes. There are at least six classes of mutations, the commonest being ∂F508 (66–70%).

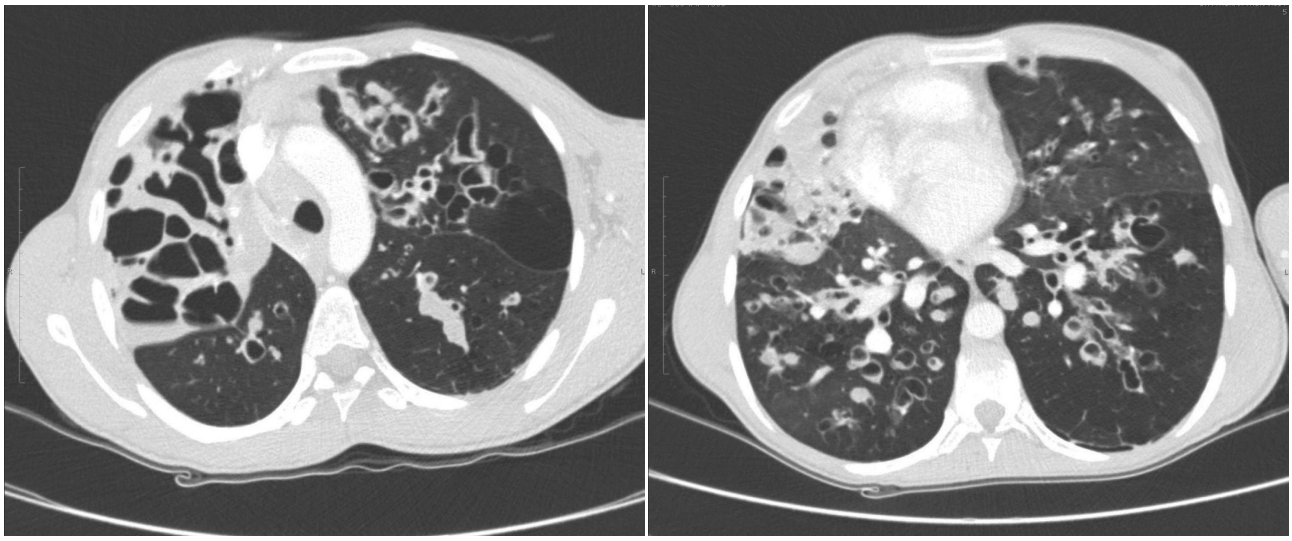

Figure 1. Chest computed tomography (CT) image revealing multiple bronchiectasis.

In skin, unlike elsewhere, CFTR is responsible for the influx of chloride and increases the sodium channel activity, thus controlling the influx of sodium. The net effect of a normally functioning CFTR is to resorb sodium and chloride. In CF patients, this is lost and therefore the characteristic increase in salt content of sweat (thus the sweat test).

In tissues other than skin, CFTR is responsible for efflux of chloride and inhibition of the sodium channel's activity which controls the influx of sodium. Therefore, under normal circumstances, salt and chloride remain in the lumen and keep water there osmotically. In CF patients, too little chloride is pumped out, too much sodium is reabsorbed with osmotic re-absorption of water from the lumen. The result is iso-osmotic, but low volume, secretions, which tend to dry out, or be thick as they still contain all the other constituents.

Early initiation of multi-disciplinary treatment is essential and responsible for the dramatic increase in life expectancy. Treatment options include prolonged courses of antibiotics, oral and inhaled corticosteroids, pancreatic enzyme supplementation, vitamin supplementation, insulin, physiotherapy, lung transplantation while gene therapies are under research.

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Diagnosis: *Delayed diagnosis of cystic fibrosis*