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CASE REPORT

Kartagener syndrome

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Abstract: Kartagener syndrome is a rare, ciliopathic, autosomal recessive genetic disorder that causes a defect in the action of the cilia lining the respiratory tract and fallopian tube. Patients usually present with chronic recurrent rhinosinusitis, otitis media, pneumonia, and bronchiectasis caused by pseudomonal infection. Situs inversus can be seen in about 50% of cases. Diagnosis can be made by tests to prove impaired cilia function, biopsy, and genetic studies. Treatment is supportive. In severe cases, the prognosis can be fatal if bilateral lung transplantation is delayed. We present a case of a 66-year-old woman with chronic recurrent upper respiratory infections, pseudomonal pneumonia, and chronic bronchiectasis who presented with acute respiratory failure. She was diagnosed with Kartagener syndrome based on her clinical presentation and genetic studies. She expired on ventilator with refractory respiratory and multiorgan failure.

Keywords: chronic obstructive pulmonary disease, bronchiectasis, immotile cilia syndrome, situs inversus

Case report

A 66-year-old woman with a past medical history of chronic recurrent rhinosinusitis, otitis media, hearing loss, recurrent pseudomonal pneumonia, and bronchiectasis presented to our hospital with a few days' history of fever, productive cough with greenish phlegm, and significant shortness of breath despite being on home oxygen. She denied chest pain, nausea, vomiting, diarrhea, urinary symptoms, or headache. She was unemployed and denied any history of tobacco abuse. She had a sister who died of chronic progressive lung disease in her fiftieth decade.

Her vital signs were: respiratory rate, 26; oxygen saturation, 91% on 5 L of oxygen; blood pressure, 103/60; and temperature, 97.3 F. A physical exam revealed a frail woman in moderate respiratory distress with diffuse rhonchi and diminished air movement in both lungs. Heart sounds were more pronounced at the right sternal border. Other physical exam findings were unremarkable.

Her laboratory work-up was unremarkable except for mild leukocytosis and hypoxia. A chest X-ray revealed old bronchiectatic areas at the bases with air-fluid levels and new consolidation in the left mid and upper lung zones (Figure 1). The X-ray also revealed dextrocardia and right-sided stomach air confirming a case of situs inversus. She was treated with intravenous cefepime and piperacillin/tazobactam, oral steroids, and bronchodilators with chest physiotherapy. Since she did not improve with this management, bronchoscopy was performed. Alveolar lavage was negative for malignancy, but culture revealed positive results for *Pseudomonas aeruginosa*.

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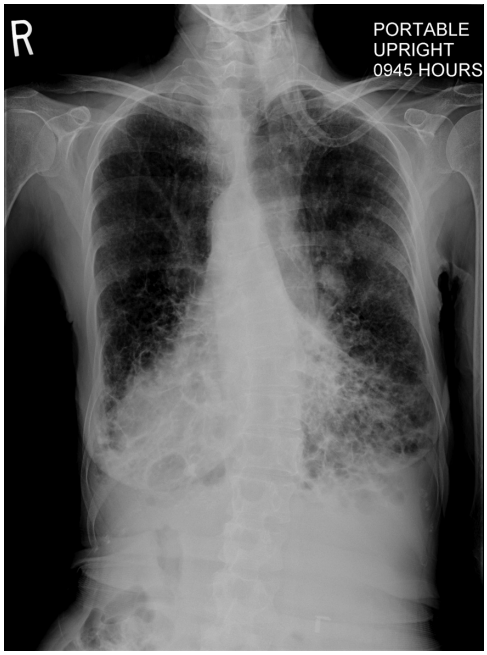

Figure 1 Bronchiectatic areas at the bases with air–fluid levels and new consolidation in left mid and upper lung zones. Dextrocardia and right-sided stomach air confirm a case of situs inversus.

A few days later, her respiratory failure deteriorated further and required endotracheal intubation. She remained on a ventilator for two weeks, then developed multiorgan failure which led to her death. She was diagnosed with Kartagener syndrome based on her clinical presentation and previous genetic studies that showed mutation in *DNAI1* and *DNAH5* genes.

Discussion

Immotile-cilia syndrome is a rare, ciliopathic, autosomal recessive genetic disorder that causes a defect in the action of the cilia lining the respiratory tract and fallopian tube.[1] As a result, patients usually present with chronic recurrent rhinosinusitis, otitis media, pneumonias, and bronchiectasis caused by pseudomonal infection.[2] When situs inversus, chronic sinusitis, and bronchiectasis occur together, it is called Kartagener syndrome.[3]

Diagnosis can be made by tests to prove impaired cilia function, biopsy, and genetic studies.[4] Measuring exhaled nasal nitric oxide which is mostly reduced or absent is a good screening test for immotile-cilia syndrome with a good negative predictive value.[5] Mucociliary transport, which is reduced in these patients, can be measured in situ by administering an inhalation aerosol of colloid albumin tagged with 99Tc.[6] Electron microscopy of a nasal or bronchial biopsy can reveal defected cilia structure.[7] Evaluation of sperm motility can help make the diagnosis in cases where other test results are equivocal. Genetic testing for mutations in the genes *DNAI1* and *DNAH5* is available through specialized laboratories. If biallelic mutations are present, the test is diagnostic. If only one allelic mutation is found, further testing may identify a transallelic mutation.[8]

Treatment of Kartagener syndrome includes daily chest physiotherapy, antibiotics with good pseudomonal coverage, and supportive pulmonary care. The effectiveness of DNase and other mucolytic agents such as hypertonic saline and acetylcysteine has not been fully assessed in these patients, but may be tried, particularly in patients with recurrent infections or ongoing respiratory symptoms.[9] Surgical intervention for bronchiectasis is rarely recommended, but can be beneficial when the disease is localized.[10] Prognosis can be fatal in severe cases if bilateral lung transplantation is delayed.[11]

Differential diagnoses for immotile-cilia syndrome include malignancy, interstitial lung diseases including idiopathic pulmonary fibrosis and idiopathic interstitial pneumonias, and other conditions associated with bronchiectasis which include acquired (foreign body aspiration, tumor, lymphadenopathy, chronic obstructive pulmonary disease, and mucoid impaction) and congenital bronchial obstruction (bronchomalacia, pulmonary sequestration, and yellow nail syndrome), recurrent infection (immunodeficiencies),

Table 1 Differential diagnoses of Kartagener syndrome

Conditions	Examples
Conditions associated with bronchiectasis	
– Acquired obstruction	Foreign body, tumor, lymphadenopathy, COPD, mucoid impaction, and connective tissue diseases
– Congenital obstruction	Bronchomalacia, tracheobronchomegaly, ectopic bronchus, pulmonary sequestration, pulmonary artery aneurysm, and yellow nail syndrome
– Immunodeficiency states with recurrent infections	IgG, IgA deficiencies, leukocyte dysfunction, other humoral immunodeficiencies
– Abnormal secretion clearance	ICS, cystic fibrosis, and Young's syndrome
– Miscellaneous disorders	Alpha-1 antitrypsin deficiency, recurrent aspiration pneumonia, inhalation of toxic fumes and dusts, and chronic rejection following organ transplantation
– **Malignancies**	Bronchoalveaolar carcinoma
– **Interstitial lung diseases**	Idiopathic pulmonary fibrosis Idiopathic interstitial pneumonias

Abbreviations: COPD, chronic obstructive pulmonary disease; ICS, immotile-cilia syndrome; IgA, immunoglobulin A; IgG, immunoglobulin G.

abnormal secretion disorder (cystic fibrosis), and other miscellaneous conditions (alpha-1 antitrypsin deficiency and connective tissue disease) (Table 1).[1,12–15]

Disclosure

The authors report no conflicts of interest or financial support in this work.

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