CLINICAL VIGNETTE

Congenital Tracheobronchomegaly

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Case Presentation

A 31-year-old male with no significant past medical history was seen in clinic for evaluation of cough and shortness of breath. The cough had been present for ten years and the shortness of breath had been present for two years. He also noted a hoarse voice intermittently for over ten years. He had been treated with multiple courses of antibiotics and prednisone with some temporary relief. The cough was productive throughout the day, worse in the morning when he woke up and also worse when supine than sitting up. He had tried albuterol, inhaled corticosteroids, and inhaled long-acting beta-agonists with no improvement. He was a prior smoker and had smoked half a pack daily for about 11 years and quit smoking 1.5 years ago. His physical exam was notable for inspiratory rhonchi in the right middle lung field. A CT of the chest was notable for severe tracheomegaly and diffuse airway dilation. Saccular diverticula of the airway were also noted. These findings were consistent with a diagnosis of congenital tracheobronchomegaly.

Given this diagnosis, he was started on flutter valve therapy and nebulized saline. He was also diagnosed with allergic rhinitis and started on saline sinus rinses and budesonide nasal irrigation. Although these interventions improved his symptoms, he continued to have recurrent infections in the winter months and was started on prophylactic azithromycin in those months with good outcome. On two year follow up, his symptoms remain under good control with resolution of his shortness of breath, decrease in his cough and sputum production, and normal pulmonary function tests.

Figures 1 and 2. CT scan of the chest demonstrating diffuse dilation and saccular diverticula of the airways.

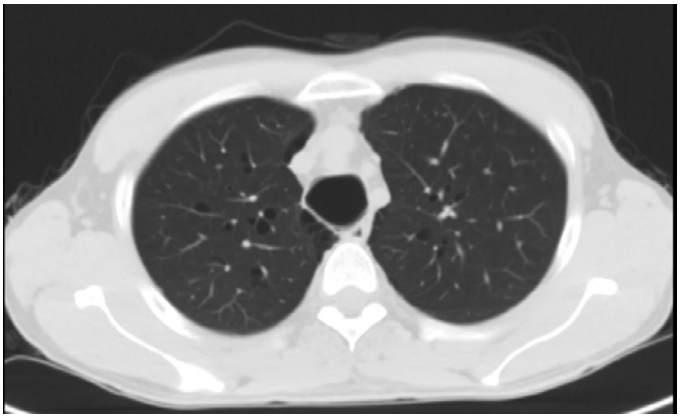

Figure 1

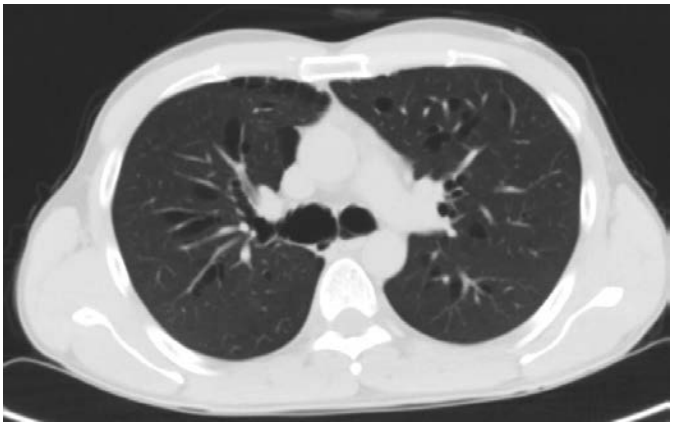

Figure 2

Discussion

Congenital tracheobronchomegaly, also known as Mounier-Kuhn syndrome, is a disorder characterized by an abnormally enlarged trachea due to the absence of elastic fibers in the trachea and main bronchi with thinning of the smooth muscle layer.[1] Estimated prevalence is 0.4% to 1.6% in patients with pulmonary symptoms with male predominance of 8:1.[1,2] Patients diagnosed with Mounier-Kuhn syndrome have been reported as young as 18months to 86-years.[1] Congenital tracheobronchomegaly has been associated with Ehlers-Danlos syndrome, Marfan syndrome, Kenny-Caffey syndrome, cutis laxa, ankylosing spondylitis, ataxia-telangiectasia, and light chain deposition disease.[2] Bronchiectasis is present at rates of 60-88% in reviews.[3] GERD is also common (29% of cases).[3] Tracheomalacia is present in two-thirds of patients.[3] Progressive hoarseness, caused by vocal cord paralysis, possibly due to changed crycoarytenoid joint, has also been described.[1] It is unclear whether congenital tracheobronchomegaly is progressive, as long term follow up data is limited.[4]

Airway diverticula and increased airway diameters reduce cough efficacy, resulting in retention of secretions and recurrent infections.[2] Typical symptoms include dry or productive cough, purulent sputum, dyspnea and hemoptysis.[1] The recurrent respiratory complaints are frequently misdiagnosed as COPD or TB.[4] In one systematic review of 128 cases of congenital tracheobronchomegaly, 25.8% of the patients had been previously diagnosed with COPD with only 10 out of 33 of them having history of smoking. In the same review 10.9%

had been treated for TB, though in half of those cases, the presence there was no documentation of mycobactermia.[4]

Congenital tracheobronchomegaly can be diagnosed by chest x-ray or chest CT. Diameters of the trachea, right main bronchus and left main bronchus on chest x-ray that exceed 3.0, 2.4, and 2.3 cm respectively are consistent with a diagnosis of tracheobronchomegaly.[5] On axial CT, mean tracheal diameters exceeding 21.8mm in men and 19.4mm in women are diagnostic of tracheobronchomegaly.[2]

Treatment options include mucolytic therapy and chest physiotherapy, including percussion and postural drainage, to help facilitate airway clearance and prevent recurrent infection.[1] Influenza and pneumococcal vaccinations should be encouraged.[3] Long term azithromycin can be considered for recurrent infections.[3] Smokers should be advised to quit, is a higher prevalence of smokers than non-smokers in patients diagnosed with tracheobronchomegaly, although whether smoking worsens the disease is unclear.[3] Tracheal stenting and tracheobronchoplasty have been explored with promising results. One study of 10 patients who underwent tracheal stenting, 8 had clinical improvement and were offered tracheobronchoplasty. Tracheobronchoplasty consisted of plication and fixation of the redundant posterior membranous wall to a tailored mesh, resulting in a stiffened posterior membranous wall. Of the 7 that underwent tracheobronchoplasty and 1 patient that opted for chronic stent placement, there was an improvement in their FEV1 (median FEV1 improved from 1.31L to 1.71 L (P=0.54) as well as their subjective dyspnea scores. The most common complication of tracheal stenting was increased secretions, occurring in 50% of the patients that underwent tracheal stenting.[6]

Outcomes after lung transplant for congenital tracheobronchomegaly is limited, as there have been only three cases reported. A 59-year-old female underwent bilateral lung transplant which was complicated by prolonged respiratory failure and tracheostomy placement requiring surgical closure due to the absence of longitudinal fibers failing to constrict the tracheal stoma.[7] She had a prolonged hospital course with recurrent pneumonias, malnutrition, and skin breakdown ultimately ending in sepsis with withdrawal of care.[8] A 49-year-old male underwent double lung transplant with reasonable quality of life for 5 years before dying of infection in year 6.[9] A 51-year-old male remains asymptomatic at 26 months with excellent exercise capacity.[10]

Conclusion

Tracheobronchomegaly is a disease in which airway enlargement results in ineffective airway clearance. Diagnosis is based on increased airway diameter on chest imaging or direct bronchoscopy. Mainstays of therapy include airway clearance technique and smoking cessation. Tracheal stenting, tracheobronchoplasty, and lung transplantation are also reasonable considerations in severe cases.

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