Living with Cystic Fibrosis into Adulthood

Ganesh Arun D.O., Patrick Passarelli M.D.

ArnotHealth

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Case Presentation

46 year old woman has a medical history of cystic fibrosis, chronic pancreatitis, type 1 diabetes presenting with shortness of breath for the last week. Associated dry cough. She was on tobramycin inhaler and oral cefepime for the last 2 weeks. Had accompanied severe abdominal pain, nausea, vomiting, diarrhea.

At age 12 presented to the hospital with coughing up a ½ cup of blood for 2 days. Her sputum culture grew Aspergillus and required aggressive treatment with Voriconazole.

At age 14 the sweat chloride test was initially negative. Decision was made to remove the tail of the pancreas that was necrotic and inflamed. Pseudocyst formed.

A Sweat Chloride test 3 years later at a different institution confirmed Cystic Fibrosis at the age of 17. A455E mutation, X-508, deletion, 5T- genetic mutation.

The diarrhea was concerning on hospital day 2 as it was increasing in frequency and had a foul odor. Clostridium Difficile was a possibility for her abdominal pain and came back positive for toxin B. Oral Vancomycin 125 mg for 10 days was started.

Her diet was advanced from full liquids to her regular diet and finished her 28 day course of inhaled tobramycin, azithromycin 500 mg every other day indefinitely for prophylaxis and a 10 day course of oral vancomycin.

Introduction

Cystic Fibrosis is an autosomal recessive disease that affects 1 in 2000 to 3000 births. The presentation includes chronic pancreatitis, recurrent lung infections, bronchiectasis, and elevated sweat chloride levels. The mutation is in the CFTR membrane transporter that is needed for proper regulation of chloride and ions into mucus.

Discussion

The most common genetic variant resulting in disease is ΔF508, but there are at least 1500 other genetic mutations.

The abnormal homozygous CFTR genotype results in abnormally thick secretions that are difficult to clear.

There is an increased concentration of chloride in sweat gland secretions, and sweat chloride testing is a primary diagnostic tool for CF.

Thickened secretions in the gastrointestinal tract impair flow of bile and pancreatic secretions, leading to pancreatic exocrine and endocrine deficiency, liver disease, and the development of malabsorption and maldigestion.

Often the greatest challenge to making the diagnosis is **failing to include CF in the differential diagnosis.**

The pillars of CF management are airway clearance, antibiotic therapy, nutritional support, and psychosocial support.

Conclusions

Cystic Fibrosis is normally a disease limited to children, however treatment has expanded to allow for living with this disease into adulthood. It is now the responsibility of clinicians to learn the mechanisms of lung and pancreas involvement to understand pain control and microbe eradication. Follow up with specialized clinics improve symptom management.

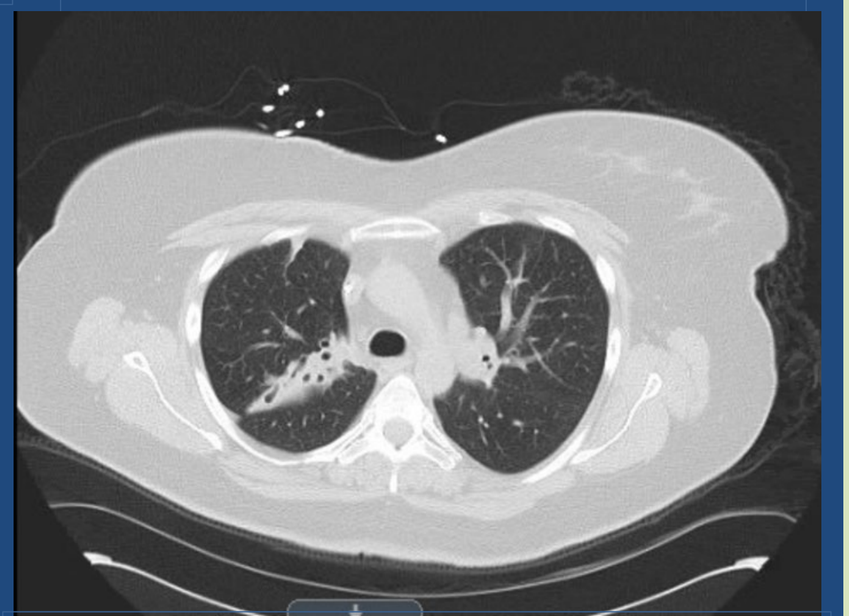

Figure 1: CT chest with IV contrast:
When compared to the most recent prior CT 1 month ago, the bilateral upper lobe opacities are without significant change in dimension however there is now bronchiectasis involving the lesions. The multiple pleural parenchymal nodules involving the left lung apex are not appreciated in the current study.

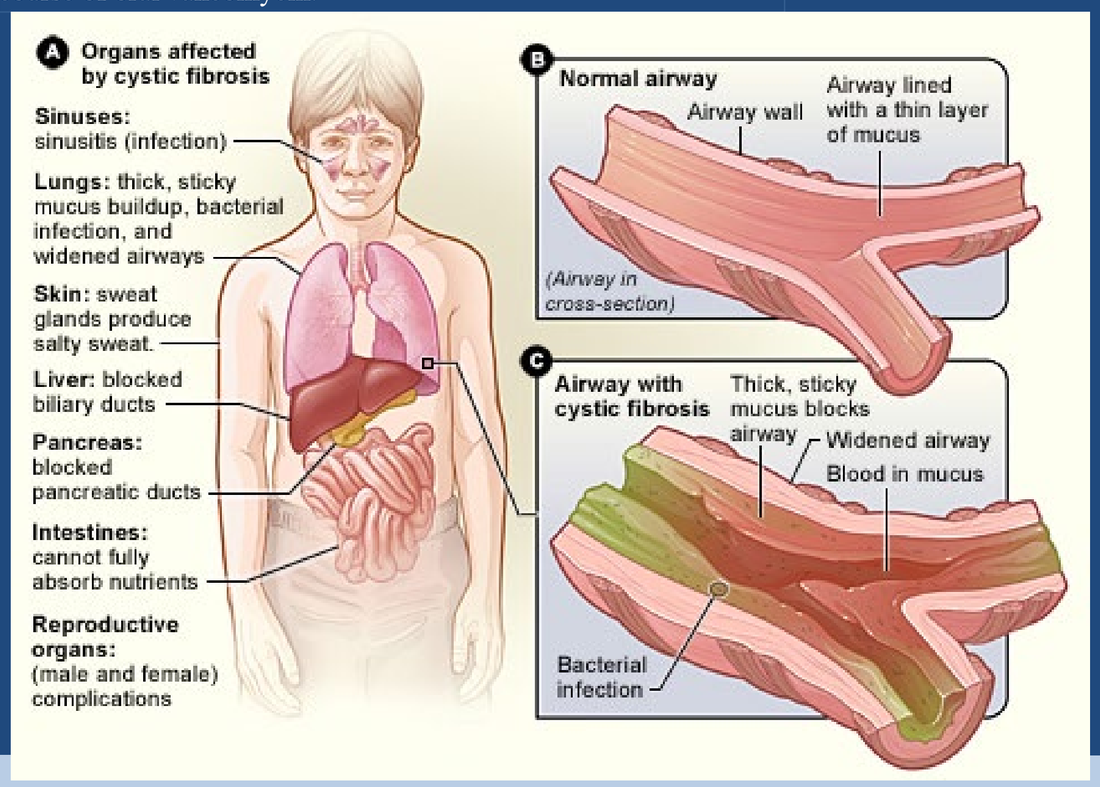

Figure 2: Cystic Fibrosis Organs Involved
https://upload.wikimedia.org/wikipedia/commons/9/9a/Cysticfibrosis01.jpg

Criteria for the diagnosis of cystic fibrosis

At least one of the following:
▪ One or more typical phenotypic features of CF:
• Chronic pulmonary disease
• Chronic sinusitis
• Characteristic gastrointestinal and nutritional abnormalities
• Salt loss syndromes
• Obstructive azoospermia
▪ History of CF in a sibling
▪ Positive newborn screening test
PLUS at least one of the following:
▪ Elevated sweat chloride concentration
▪ Two *CFTR* gene mutations known to cause CF on separate alleles*
▪ Abnormalities in NPD testing that are typical for CF¶

CF: cystic fibrosis; *CFTR*: cystic fibrosis transmembrane regulator gene; NPD: nasal potential difference.
* Using mutation classifications identified in the CFTR2 project.
¶ NPD testing measures abnormalities in ion transport across the nasal epithelium.

Table 1: Types of Cystic Fibrosis based on mutation location
https://www.cghjournal.org/article/S1542-3565(12)01308-0/fulltext

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