Helping patients cope

A life-threatening genetic disease that affects the respiratory and gastrointestinal systems, cystic fibrosis has no cure, but patients with the disease are living longer than ever before. Find out what you need to know about the latest advances in diagnosis and treatment so you can help your patients get the care they need.

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CYSTIC FIBROSIS (CF) is an inherited autosomal recessive disorder of the exocrine glands that affects the lungs and digestive system of children and adults. Approximately 30,000 people in the United States have been diagnosed with CF, and it affects all races (primarily Caucasians). Girls and boys are affected equally. Additionally, it’s estimated that about 1 in every 31 Americans are carriers of the defective CF gene, without actually having the disease.

This disease was once considered strictly one of childhood because many children who had it didn’t survive past their early years. Currently, the estimated survival age for people with CF extends to the mid to late 30s. In fact, more than 40% of patients living with CF are age 18 or older. The great increase in life expectancy of patients with CF over the past few decades is due to many advances in diagnosis and treatment.

In this article, I’ll discuss the genetics of CF and its effects on various organ systems. I’ll also take a look at how the disease is screened for and diagnosed. Finally, I’ll review current treatments and therapies for CF, as well as how to best assist your patients in coping with this chronic disorder.

It’s in the genes
To understand how CF develops, you need to understand inherited autosomal recessive genetic disorders. All genes come in pairs. A recessive gene is one that’s expressed only when both copies of the gene are present. Carriers of the CF gene have one normal gene and one abnormal gene in the pair, but don’t have the actual disease. In order for a child to inherit CF, he must inherit both copies of the defective gene, one from each parent (see Inheritance of CF).

If both parents are CF carriers, each pregnancy has a 25% chance of producing a child with CF. If the child doesn’t inherit the disease, there’s a 50% chance that he’ll be a CF carrier like his parents.

So what exactly is CF? Patients with CF have a mutation in a gene that encodes a protein called cystic fibrosis transmembrane conductance regulator (CFTR). This protein is responsible for transporting sodium and chloride ions across cell membranes in the epithelial lining of the ducts of exocrine glands, which include sweat glands, sinuses, bronchial mucosa, the pancreas, salivary glands, the intestines, the biliary tract, and the reproductive tract. Abnormal function of
with cystic fibrosis
CFTR in these cells leads to abnormal transport of water and salt across the epithelial surfaces. This causes abnormally thick secretions in many different organs and disrupts their normal functions.

**Early warning**
Signs of CF generally appear at birth or during early infancy. The first sign of CF may occur right after birth, when the passage of meconium (the dark greenish feces that have collected in the intestines of a full-term fetus) is delayed. Infants with the disorder typically develop recurrent respiratory infections or failure to thrive, prompting an exploration into a CF diagnosis. Children may also exhibit signs of pancreatic insufficiency early in life, resulting in the passing of frequent, fatty, bulky, foul-smelling stools (steatorrhea) and poor growth despite a good appetite.

**Systems affected by CF**
The effects of CF can involve multiple organs. Over 1,000 different mutations of the CF gene have been identified, each one causing different signs and symptoms, as well as variable progression of the disease. The following organ systems are typically affected in people with CF.

**Respiratory system**
Patients with CF have thick and viscous airway secretions that result in a continuous cycle of obstruction, infection, and inflammation. Thick mucus is very difficult to expel by coughing, and it becomes stagnant and clogs the patient’s airways (see *What the lungs look like in CF*). This excessively thick mucus is the perfect breeding ground for bacteria. The resulting chronic infection leads to irreversible lung damage.

Additional respiratory symptoms include nasal polyps and chronic sinusitis. Air trapping in the lungs can lead to development of a barrel-shaped chest. Clubbing of the fingers and toes may also occur.

**Gastrointestinal (GI) system**
Abnormally thickened pancreatic fluids can block the pancreatic ducts, preventing the secretion of essential digestive enzymes into the intestine. Pancreatic insufficiency affects 85% to 90% of patients with CF, preventing the body from being able to digest nutrients. Fats, protein, and fat-soluble vitamins (A, D, E, and K) are malabsorbed, leading to nutritional deficiencies, delayed growth, weight gain, and steatorrhea. Plugged pancreatic ducts lead to pancreatic damage.

Gastroesophageal reflux disease is common in patients with CF due to hypersecretion of gastric acid and hyposecretion of bicarbonate. Chronic coughing and postural drainage techniques used to mobilize respiratory secretions and aid expectoration can further aggravate this condition. Additional GI complications related to CF include distal intestinal obstructive syndrome, constipation, and liver disease.

**Reproductive system**
In both men and women, CF affects the reproductive system. Puberty may be delayed
in both boys and girls. Nearly all men with CF are sterile due to absent or malformed vas deferens. Women may have decreased fertility caused by thick cervical secretions blocking sperm movement. A variety of fertility treatments are available to help these patients achieve pregnancy. Genetic counseling is required to help couples understand the risk of passing on the disease.

Endocrine system
An additional CF complication is cystic fibrosis-related diabetes (CFRD), which can occur in 20% to 30% of patients over age 20. The main cause of CFRD is insulin deficiency due to obstruction of the pancreatic duct. However, impaired glucose metabolism is also a factor. Most patients with CFRD require insulin therapy to manage their diabetes.

Sweating out the diagnosis
For the past 40 years, the gold standard for diagnosing CF has been the quantitative pilocarpine iontophoresis sweat test, which is used to measure the amount of chloride in the patient’s sweat; in someone with CF, the concentration of chloride in sweat is very high.

During this simple procedure, pilocarpine is applied to a small area of the arm or leg; an electrode is placed over the area to deliver a weak electric current and stimulate sweat. Sweat is applied to a gauze pad or filter paper and sent to the lab for analysis. Diagnosis is made as follows:
- sweat chloride concentration less than 40 mmol/L is normal
- concentration between 40 and 60 mmol/L is borderline
- concentration greater than 60 mmol/L is diagnostic of CF.

Genetic testing may be performed if the patient’s sweat test is positive or borderline to confirm the diagnosis. The presence of CF mutations in both CFTR genes also confirms the diagnosis.

Newborn screening tests can be performed on blood to evaluate for CF. The screening test evaluates levels of immunoreactive trypsinogen, which if elevated can indicate CF and prompt additional testing. The CDC recommends that all states consider adding CF testing to their newborn screening blood tests. Detecting CF early helps improve the child’s growth and cognitive development, decrease incidences of hospitalization, and improve survival.
What treatments are available?

Patients with CF typically receive care from a team of physicians, nurses, dietitians, respiratory therapists, and social workers at a CF care center. The goals of treatment include prevention and treatment of lung and digestive problems, ensuring proper nutrition, promoting physical activity, and ensuring appropriate psychosocial support. The following are specific ways in which you can help CF patients achieve these goals.

• **Antibiotic therapy.** Progressive lung damage due to chronic infection and inflammation is responsible for the majority of morbidity and mortality in patients with CF. Prevention and appropriate treatment of lung infections is the main goal of CF treatment. Because of excessive thickened airway secretions, pulmonary infections are common and difficult to treat. Your patient’s sputum culture and sensitivity results will determine the appropriate antibiotic therapy (see Medications to treat CF).

  The most common organisms that cause lung infections in patients with CF are *Staphylococcus aureus*, *Haemophilus influenzae*, and *Pseudomonas aeruginosa*. Given orally, intravenously, and by inhalation via nebulizer, antibiotics are indicated when the patient has acute pulmonary exacerbations and to suppress bacterial flora during remissions. A patient with CF may require higher than normal doses of antibiotics, a combination of two different antibiotics, and a longer duration of therapy to clear his lung infections.

  Patients chronically infected with *Pseudomonas* experience increased lung inflammation and decreased lung function. The Cystic Fibrosis Foundation published new guidelines for the treatment of this pathogen in November 2007. Chronic use of inhaled tobramycin in patients with CF ages 6 and up with moderate-to-severe lung disease is recommended to decrease exacerbations and improve lung function. This treatment is also recommended for patients with asymptomatic or mild lung disease to decrease exacerbations. Chronic use of azithromycin has also been recommended for CF patients ages 6 and up with *P. aeruginosa* that’s continually present in airway cultures to improve lung function and reduce the number of exacerbations.

• **Airway clearance techniques.** A vital component of CF pulmonary treatment is effective airway clearance maneuvers to help the patient expectorate thick airway secretions. Not clearing these secretions can lead to obstructed airflow, impaired gas exchange, and infection.

  Your patient can choose from various airway clearance maneuvers, including postural drainage techniques, positive end-expiratory pressure therapy, a high-frequency chest compression vest, and a flutter device.

  Sessions of airway clearance maneuvers typically take 1 hour to allow for coughing and expectoration of mucus. Teach your patient to perform these sessions before meals because frequent coughing can trigger vomiting. He’ll typically require airway clearance once or twice a day for maintenance. During times of acute pulmonary exacerbation, airway clearance maneuvers are done more frequently.

  Also encourage your patient to exercise regularly, as this helps to clear the airways, maintain overall health, and minimize CF complications. He may participate in any type of exercise program or sport he enjoys.

• **Additional pulmonary therapies.** A variety of medications are used to promote airway clearance, improve lung function, reduce inflammation, and increase oxygenation. Dornase alfa nebulizer solution can be used as mucolytic therapy to thin secretions so they’re easier to expectorate. Hypertonic saline given via nebulizer helps to draw more water into the airways, thinning mucus and making it easier to expectorate. Bronchodilators administered by nebulizer,
such as albuterol, are given to open the airways and improve lung function. Administering mucolytic therapy and bronchodilators before airway clearance techniques may improve the outcome. Oral nonsteroidal anti-inflammatory drugs are also used in the treatment of CF. The Cystic Fibrosis Foundation recommends chronic use of oral ibuprofen to slow the loss of lung function in patients ages 6 and up with forced expiratory volume in the first second greater than 60% predicted.

- **Nutritional management.** Maintaining proper nutrition is essential in the treatment of CF. As most patients with CF have increased nutritional needs, encourage a high-calorie, high-protein diet to ensure proper growth and development.

  Patients with pancreatic insufficiency need to take pancreatic enzyme supplements with food to help aid nutrient absorption. Instruct your patient to take these supplements before or during all meals and snacks. Patients with pancreatic insufficiency require daily vitamin supplementation with a multivitamin and supplements of vitamins A, D, E, and K because they have difficulty absorbing these fat-soluble vitamins.

  During acute pulmonary exacerbations, shortness of breath and increased work of breathing can make meeting nutritional needs difficult for a patient with CF. Short-term or long-term supplemental parenteral or enteral feedings may be necessary to meet his caloric needs. Because your patient with CF is at a risk for dehydration, instruct him to increase fluid and salt intake during hot weather and while exercising.

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### Medications to treat CF

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<th>GI system</th>
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<td><strong>Lactulose</strong></td>
<td>Given to prevent constipation.</td>
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<tr>
<td><strong>Multivitamins, iron; vitamins A, E, D, and K when deficient</strong></td>
<td>Given because the production and absorption of vitamins are decreased by the thick secretions. Give water-soluble vitamins for better absorption.</td>
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<tr>
<td><strong>Pancreatic enzyme concentrates such as pancrelipase (Viokase)</strong></td>
<td>Given to decrease the fat and bulk of stools. Give immediately before or with meals and snacks to increase the absorption of nutrients.</td>
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<tr>
<td><strong>Ursodeoxycholic acid or ursodiol</strong></td>
<td>Used to treat and possibly prevent progressive liver disease in CF. It improves the viscosity of biliary secretions.</td>
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<th>Respiratory system</th>
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<td><strong>Bronchodilators (albuterol, terbutaline)</strong></td>
<td>Used to open airways. They’re used before chest physiotherapy (CPT) and to relieve symptoms; administered via nebulizer.</td>
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<tr>
<td><strong>Dornase alpha (Pulmozyme)</strong></td>
<td>Used with standard CF therapies to improve pulmonary function and to decrease risk of respiratory tract infections requiring parenteral antibiotics; administered via nebulizer.</td>
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<tr>
<td><strong>Aerosol antibiotics (gentamicin, ticarcillin, tobramycin)</strong></td>
<td>Administered via nebulizer after CPT. Aerolized antibiotics decrease systemic absorption.</td>
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<tr>
<td><strong>Oral and I.V. antibiotics</strong></td>
<td>Prolonged administration with high doses given to treat infection; specific drug based on culture sensitivity.</td>
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Is a transplant necessary?
Lung transplantation is becoming more common in CF patients with severe lung disease. The patient may receive healthy transplanted lungs from a cadaver or lung lobes from two different living donors. Success rates of lung transplants in patients with CF equal or exceed those with other lung disorders; 90% of CF patients are alive 1 year after transplant, and 50% survive longer than 5 years. Specific eligibility requirements for lung transplant are determined by the transplant center, and the wait for donor lungs can be very long.

Complications of lung transplantation in patients with CF can include organ rejection and an increased risk of infection and malignancies due to having to take immunosuppressant drugs. However, successful transplants can greatly improve the patient’s quality of life.

Helping patients and families cope
Patients with CF and their families require extensive psychosocial support. Parents must learn to cope with having a child with a chronic life-shortening illness, manage daily treatments and medications, and endure the financial hardships that accompany the high cost of managing this disease. The Cystic Fibrosis Foundation (http://www.cff.org) is an excellent source of support with access to support groups, up-to-date treatment options and research materials, and financial assistance options. The patient’s CF care center with its team of expert professionals is another great support system for the family.

As a patient with CF ages, he must learn the importance of his daily treatment plan to slow disease progression and begin to take an active role in his own care to foster a sense of independence. As he gets older, acute illness will become more frequent as the disease process worsens. As the patient’s condition deteriorates, sensitive discussions regarding end-of-life care must take place.

Patients and their families can make informed choices regarding end-of-life care with the assistance of the healthcare team.

Slowing the progression
As a chronic, life-shortening disease that affects multiple organ systems, CF causes frequent illnesses and hospitalizations for the patient who has it. He must learn how to effectively incorporate his individualized range of therapies into his everyday life to help slow disease progression. Encourage your patient that advances in treatment options, including lung transplantation, are improving quality of life and extending life expectancy for people with this disease.

Learn more about it