



Airway Clearance Indications: Genetic Disorders of Mucociliary Clearance

Cystic Fibrosis (CF)

Cystic fibrosis (CF) is the most common of three recognized genetic disorders of mucociliary clearance: 1.) cystic fibrosis, 2.) primary ciliary dyskinesia (PCD), and 3.) pseudohypoaldosteronism (PHA). As the classification implies, all three are characterized by profound disruption of the mucociliary clearance system, although the underlying pathophysiology and long term prognosis vary between them.

Cystic fibrosis is a genetic disorder of the exocrine system that affects the sweat glands and mucus secreting glands, including those in the lungs and pancreas. Mutations in a gene called CFTR cause metabolic abnormalities that result in the production of large quantities of extremely thick, sticky, tenacious mucus. Cystic fibrosis is a multi-system disorder, but the most serious illness is primarily related to lung and pancreatic complications.

Cystic fibrosis is a recessive disorder, meaning both parents must carry a CFTR mutation for a child to have the disease. Mutations of the CFTR gene are very common in the Caucasian population, with as many as 1 in 30 individuals being symptomless and unwitting carriers of cystic fibrosis. When two carriers have a child, there is a 1 in 4 chance that the baby will be born with cystic fibrosis. This happens in 1 out of every 2,500 to 3,000 Caucasian births, meaning that there are approximately 30,000 individuals currently living with cystic fibrosis in the United States. Cystic fibrosis is less common in other ethnic groups, affecting about 1 in 17,000 African Americans and 1 in 31,000 Asian Americans.

What Happens in Cystic Fibrosis?

Effective mucociliary clearance requires mucus of the right amount and consistency working in a coordinated fashion with ciliary activity to keep airways clean and free of infection. The thick, sticky mucus that is the hallmark of cystic fibrosis gets stuck in the airways, completely blocking them and overwhelming the mucociliary clearance system. Additionally, cough clearance is not effective in cystic fibrosis because the mucus is tenacious and adheres doggedly to the airway wall rather than being expelled during cough. Compounding the respiratory problems, most people with cystic fibrosis also have digestive problems because thick, sticky mucus interferes with the function of the pancreas, which is responsible for producing insulin and other enzymes required to digest food. Problems with digestion can lead to compromised nutritional status, which is known to contribute to respiratory disease.

Lack of a functioning mucociliary clearance system coupled with ineffective cough causes frequent, serious respiratory infections in individuals with cystic fibrosis. By early adulthood, most will have significant bronchiectasis, a condition characterized by enlarged, damaged, weakened airways that serve as reservoirs for bacteria-laden mucus and further infection. Advanced lung disease in cystic fibrosis is the primary cause of disability and death and many people with cystic fibrosis will require lung transplant in early to mid adulthood.



How Airway Clearance Therapy Can Help in Cystic Fibrosis

Because people with cystic fibrosis lack a functioning mucociliary clearance system, daily, lifelong airway clearance therapy is required. Keeping the airways clear of excess secretions and thereby reduce the incidence of inflammation and/or infection and is crucial to maintaining respiratory health. Airway clearance therapy using High Frequency Chest Wall Oscillation (HFCWO) has been demonstrated by clinical study to promote excess mucus clearance and improve bronchial drainage. Shear forces are created by HFCWO treatment that mechanically releases adhered secretions from the walls of the pulmonary tract. HFCWO has also been shown to reduce the viscosity of secretions which significantly improves mobilization of excess mucus. By replicating cough, HFCWO can effectively mobilize pulmonary secretions from smaller airways to larger airways where they can be coughed out, swallowed or suctioned.

Symptoms of Cystic Fibrosis

- Persistent wet cough
- Excessive production of sputum
- Recurrent lung infections, sometimes with unusual pathogens (germs)
- Wheezing or shortness of breath
- Very salty-tasting skin;
- Failure to thrive
- Frequent greasy or bulky stools or difficulty with bowel movements

For More Information on Cystic Fibrosis:

1. National Heart, Lung and Blood Institute cystic fibrosis fact sheet:
http://www.nhlbi.nih.gov/health/dci/Diseases/cystic_fibrosis/cystic_fibrosis_what.html
2. Cystic Fibrosis Foundation: http://www.cystic_fibrosisf.org/
3. GeneReviews overview of cystic fibrosis:
<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=cf>
4. Medline Plus cystic fibrosis fact sheet: <http://www.nlm.nih.gov/medlineplus/cysticfibrosis.html>
5. March of Dimes cystic fibrosis fact sheet: http://www.marchofdimes.com/pnhec/4439_1213.asp