

## Cystic Fibrosis: Review Questions

*Subramanyam Chittivelu, MD, FCCP*

### QUESTIONS

Choose the single best answer for each question.

- Each of the following statements regarding the genetics of cystic fibrosis (CF) is correct EXCEPT:**
  - The CF gene codes for a protein called cystic fibrosis transmembrane regulator (CFTR)
  - CFTR is largely expressed in epithelial cells of the airways, the pancreas, the biliary system, the genitourinary system, and the sweat glands
  - Although there are a few hundred mutations that exist for CF, the most prevalent mutation among northern Europeans is W1282X
  - Severity of lung disease and liver disease cannot be predicted by genotype
- Which of the following exocrine glandular ducts are NOT obstructed in cystic fibrosis?**
  - Pancreas
  - Lungs
  - Sweat glands
  - Glands of the uterine cervix
- Which of the following is NOT a pulmonary complication of CF?**
  - Massive hemoptysis and pneumothorax
  - Atelectasis and acute respiratory failure
  - Allergic bronchopulmonary aspergillosis and hypertrophic pulmonary osteoarthropathy
  - Lymphangiomyomatosis and chylothorax
- Which of following statements regarding the mechanism of action of recombinant human deoxyribonuclease (rhDNase) in CF is correct?**
  - rhDNase cleaves the extracellular DNA from the neutrophils in sputum to make it less viscous
  - rhDNase's potent antibacterial action prevents the growth of pseudomonads and staphylococci
  - rhDNase has antibacterial, bronchodilator, and mucolytic actions
  - rhDNase corrects the defect in the CF gene on chromosome 7
- Each of the following statements regarding pregnancy in women with CF is correct EXCEPT:**
  - Impairment of pulmonary function is the major predictor of both maternal and fetal outcome
  - CF patients have an increased risk for premature delivery
  - The maternal and fetal outcome is good for most cystic fibrosis patients
  - The pregnancy need not be a planned one

*(turn page for answers)*

---

*Dr. Chittivelu is Assistant Professor, Department of Internal Medicine, Division of Pulmonary and Critical Care Medicine, and Clerkship Director in Internal Medicine, Texas Tech University Health Sciences Center, Amarillo, TX.*

#### EXPLANATION OF ANSWERS

- (C) Although there are a few hundred mutations that exist for CF, the most prevalent mutation among northern Europeans is W1282X.** CF is an autosomal recessive disorder of adults and children. More than 700 genetic mutations contribute to the development of CF. The most prevalent mutation of CFTR is deletion of a single phenylalanine residue at amino acid 508 ( $\Delta F508$ ). This mutation is responsible for the high incidence of CF in northern Europeans. The W1282X mutation is most common among the Ashkenazi Jewish population.
- (C) Sweat glands.** Most CF patients have 3 distinct abnormal characteristics. First, the ducts of the mucus-secreting glands are obstructed due to an increase in viscosity of these secretions leading to glandular dilatation and destruction. Second, CF patients are prone to chronic bacterial colonization and infections. Third, the sweat glands are not obstructed in CF patients because in serous glands such as sweat glands there are abnormal concentrations of inorganic ions, rather than glandular obstruction with thick mucus. The quantitative pilocarpine iontophoresis sweat test is a uniformly accepted method for diagnosing CF. The sweat gland ducts must be patent for this test. Obstruction of airways leads to bronchiectasis and atelectasis; pancreatic duct obstruction leads to pancreatitis and malabsorption; and plugging of bile ducts leads to obstructive jaundice.
- (D) Lymphangiomyomatosis and chylothorax.** There is no link between lymphangiomyomatosis and CF. Lymphangiomyomatosis may cause chylothorax. Hemoptysis (either scanty or profuse) and spontaneous pneumothorax are well-known complications of CF. Atelectasis can result from the mucus plugging of airways. Acute or chronic respiratory failures are also seen commonly in CF patients, as are pulmonary infections due to pseudomonads or staphylococci. CF patients are prone to develop allergic bronchopulmonary aspergillosis, which requires treatment with corticosteroids. Hypertrophic pulmonary osteoarthropathy, which is a triad of digital clubbing, arthritis, and periostitis, can occur in CF and is clearly associated with severity of pulmonary disease. In hypertrophic pulmonary osteoarthropathy, the long and tubular bones are most often affected. Hypertrophic pulmonary osteoarthropathy is treated with anti-inflammatory agents for symptomatic relief.
- (A) rhDNase cleaves the extracellular DNA from the neutrophils in sputum to make it less viscous.** Recombinant human DNase is given in aerosol form in a dose of 2.5 mg once or twice daily. The rhDNase decreases the viscosity of sputum by digesting the neutrophil DNA. Although rhDNase can reduce the frequency of infections, the need for parenteral antibiotics, and the length of hospital stay, it does not have any antibacterial or bronchodilatory actions. Various mucolytics have been used in CF patients to clear the thick secretion. Mucolytics can be used in both oral and aerosol forms to liquefy the hyperviscous airway secretions. N-acetylcysteine has been used as an aerosol. It acts by breaking the sulfhydryl bonds of mucus glycoproteins, thereby reducing the viscosity of sputum. Its use has been limited, however, because of its offensive odor and tendency to cause bronchospasm.
- (D) The pregnancy need not be a planned one.** On the contrary, the pregnancy *should* be a planned one rather than an unexpected one. It is essential to optimize the overall condition of CF and other medical conditions of pregnant women to improve maternal and fetal outcomes. With improvement in survival of CF patients, issues like pregnancy and family planning carry more significance. Even though early outcomes of pregnancy were discouraging for CF patients, improvement in infection control, nutrition, and lung function continue to make possible better outcomes for both mother and baby.

Copyright 2001 by Turner White Communications Inc., Wayne, PA. All rights reserved.

#### PULMONARY DISEASE CERTIFICATION

A current list of certification and recertification exam dates and registration information is maintained on the American Board of Internal Medicine Web site, at <http://www.abim.org>.