**Cystic Fibrosis Case Study**

Brad and Joanne were worried when their newborn daughter Jada seemed to have trouble gaining weight. They had two older children who never had any problems with weight gain. Jada’s appetite was normal, but after meals, she seemed to be in pain. Their pediatrician suggested that it could be a result of acid reflux, but steps such as propping Jada up to sleep, which typically help with infant reflux, didn’t seem to make any difference.

At around 1 year of age, Jada started getting chronic respiratory infections. Her low body weight dropped even further, and her family physician recommended that she be admitted to the hospital for close monitoring. In the hospital, one of the doctors examining her noticed that she had nasal polyps, which are unusual in an infant; the combination of nasal polyps and gastrointestinal problems prompted one of her doctors to order a sweat test that came back positive for cystic fibrosis, a lethal genetic disorder.

Her parents were devastated and overwhelmed. The doctor told them that cystic fibrosis is relatively common among Caucasians, occurring in about 1 in 4,000 births. All forms of cystic fibrosis involve mutations in a gene called CFTR, which encodes a chloride ion channel. Joanne was shocked, because she had actually undergone prenatal testing that had failed to detect any genetic predisposition for cystic fibrosis in her own DNA.

After the doctor left the room, Brad and Joanne realized that they still had many unanswered questions about their daughter’s diagnosis. They did an online search and found a website for a nonprofit organization called the [Cystic Fibrosis Foundation.](http://www.cff.org/aboutcf/)

**Review Questions**

1. Assuming that nobody involved with Joanne’s prenatal testing made a mistake, what is a reason why her test might have failed to show an elevated risk for cystic fibrosis in her children?

2. The sweat test is the most common test to diagnose cystic fibrosis, and it simply works by measuring salt content in a patient’s sweat. Would you expect a cystic fibrosis patient’s sweat chloride levels to be high or low? Why? Sweat is secreted from exocrine glands in the skin; how do you think sodium, chloride, and water, the main components of sweat, get into the sweat glands?

3. The two main organ systems affected by cystic fibrosis are the respiratory and digestive systems. What is the common ground between these two seemingly dissimilar systems? Why are both affected in cystic fibrosis?

4.  Cystic fibrosis is treated by managing the symptoms rather than by treating the underlying disease. Most scientists believe that the only cure for cystic fibrosis will be gene therapy. Why do you think this is? Why can’t a pill “fix” cystic fibrosis?