**Jennifer’s Story: Cystic Fibrosis**

*A compilation of a journal entry February 12, 2015 by Stephen Shannon on the Cystic Fibrosis Today Website and the HAPSI Website on Cystic Fibrosis*

Text Mark Up Directions:

1. Preview and number the paragraphs.
2. Read once and circle the vocabulary terms; then look them up and define or write a synonym in the margin next to the word.

<http://www.personal.psu.edu/users/j/n/jnb5091/Images/CFTR%20Protein.jpg>

1. Re-read a second time and highlight the main points (per paragraph) and annotate (paraphrase) in the margin.

My little sister, Jennifer, was a beautiful woman who, like myself, fought Cystic Fibrosis throughout her life and on Saturday, February 7, she passed away at twenty-three after her body rejected the set of lungs she had received by transplantation the previous October in Denver, Colorado.

The day before Jennifer’s death I rushed to the airport after I received a call that she would likely not make it more than a day. My tears began to course as I entered her room and saw her strain every muscle in her chest with each breath she took, almost shaking the bed with her effort. That night, before her death, I spent my final moments alone with her, and as I sat on her bed and held her hand I wanted her to know that her death was not her fault. I didn’t want her to die thinking that she didn’t do enough to fight the disease. The reality is that she was sick because of a genetic mutation that was arbitrarily given to her and she died because the lungs she was given were not compatible with her body.

Cystic Fibrosis is a common genetic disease caused by a DNA mutation in a gene called the cystic fibrosis transmembrane conductance regulator (CFTR) gene. The CFTR gene is located on chromosome 7 and has the recipe to create the CFTR protein. The CFTR protein is a channel protein that regulates how salts and water move through the cell membranes of epithelial cells of the skin, respiratory, and digestive tracts.

When the CFTR protein does not function correctly, chloride (Cl-) is unable to pass through the center channel and sodium (Na+) is also unable to pass through the cell membrane. When the sodium and chloride are imbalanced, mucus that normally lubricates the epithelial cells is unable to move into the tissues and becomes extremely sticky and thick. This thick mucous resulted in Jennifer’s persistent cough, shortness of breath and frequent respiratory infections due to bacteria getting trapped in the mucus. Her lungs finally filled with fluid and her blood grew toxic with built up carbon dioxide ending her life.

All of this pain was due to a simple typo in her DNA recipe to make the CFTR protein that would have kept her mucus at a normal thickness. Think of these recipes where your body adds a bit too much flour or an extra teaspoon of sugar. Other gene mutations are more destructive; they make your body leave out an entire ingredient, or delete half the recipe. Each of us carries two copies of each gene recipe and often as long as one of them is good your body can make enough of the needed protein and you live a normal life. Cystic Fibrosis works this way in that it is recessive, meaning that you have to have two bad copies of the gene to get the disease. Jennifer’s parents were carriers, each with one bad copy but no symptoms of the disease. They unknowingly each passed Jennifer their bad copies and as with all recessive diseases a person with two bad copies cannot make the needed protein and develops the disease. When Cystic Fibrosis was first discovered, few sufferers lived past 6 years old, but due to medical advances the age of survival has increased to 37 years old. Jennifer died at age 23.

**Jennifer’s Story Questions**

1. Who was Jennifer and why did she die? What was done to attempt to save her life?
2. What causes Cystic Fibrosis? How is the disease related to proteins?
3. What are the symptoms of Cystic Fibrosis? Why do patients frequently have lung infections?
4. How could Jennifer’s parents not have the disease but Jennifer did?