Name _____ **Cystic Fibrosis**

Cystic fibrosis is an inherited chronic disease that affects the lungs and digestive system of about 30,000 children and adults in the United States (70,000 worldwide). A defective gene and its protein product cause the body to produce unusually thick, sticky mucus that:

-clogs the lungs and leads to life-threatening lung infections; and

-obstructs the pancreas and stops natural enzymes from helping the body break down and absorb food.

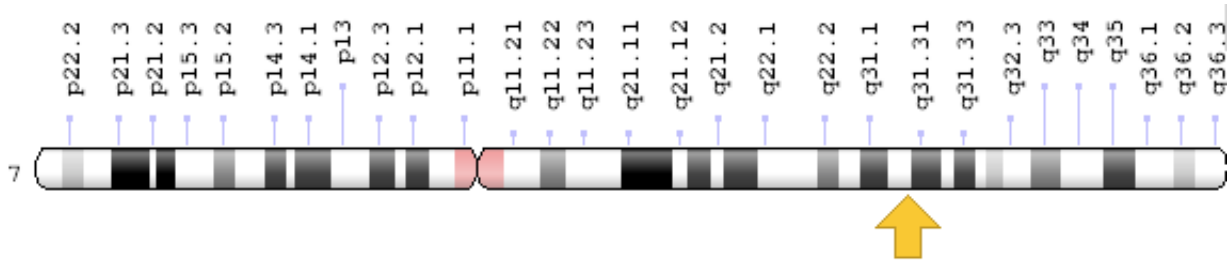
In the 1950s, few children with cystic fibrosis lived to attend elementary school. Today, advances in research and medical treatments have further enhanced and extended life for children and adults with CF. Many people with the disease can now expect to live into their 30s, 40s and beyond.

Statistics

- About 1,000 new cases of cystic fibrosis are diagnosed each year.
- More than 70% of patients are diagnosed by age two.
- More than 40% of the CF patient population is age 18 or older.
- In 2006, the predicted median age of survival was 37 years.[1]

Much has been learned since then about the function of the gene's protein, named CFTR (for CF transmembrane conductance regulator). It appears to work like a two-way pump, channeling vital compounds in and out of a cell. When it functions normally, the protein helps regulate the transfer of sodium across cell membranes and serves as a chloride channel. But in CF this process fails, and the chloride channel stays closed. The sodium, which does not move freely, builds up in the lungs and disables a natural antibiotic that would otherwise guard against a wide range of lung infections. Bacteria then thrive in the thick, sticky mucus.

The gene for the CFTR protein is on the long arm of chromosome 7. It runs from base pairs 117,478,367 to 117, 668, 665.



There are several mutations that lead to this disease. This isn't surprising since there are 190,298 bases involved in this gene. Some don't change the protein enough to cause disease but others do. The following mutations are some of the most common that do cause the disease. Determine what the mutations are. First figure out the mRNA sequence, then the amino acid sequence to figure out what kind of mutation can cause cystic fibrosis.

Mutation #1

Unaffected DNA – TTTCTTTTATAGTAGAAACCACAAAGGATACTACTT

mRNA –

amino acids -

Affected DNA TTTCTTTTATAGTAGCCACAAAGGATACTACTTATA

mRNA –

amino acids -

What kind of mutation is #1 and how does it affect the protein CFTR?

Mutation #2

Unaffected DNA - AAACCACAAAGGATACCACTTATATCTATGTCTTCG

mRNA –

amino acids -

Affected DNA - AAACCACAAAGGATACTACTTATATCTATGTCTTCG

mRNA –

amino acids -

What kind of mutation is #2 and how does it affect the protein CFTR?

Mutation #3

Unaffected DNA - GAAACCACAAAGGATACCAGTTATATGTATGTCTTC

mRNA –

amino acids -

Affected DNA - GAAACCACAAAGGATACTAGTTATATGTATGTCTTC

mRNA –

amino acids -

What kind of mutation is #3 and how does it affect the protein CFTR?

The most common mutation that causes cystic fibrosis is a deletion mutation that deletes the amino acid phenylalanine. Which mutation is that?