Name: _____ Date: _____ Period: _____

Mutations that Affect Protein Production Wk #: _____

Directions: Read the description of each condition and answer the corresponding questions.

Cystic fibrosis is an inherited disease that affects the lungs and digestive system. A mutated mRNA sequence results in a defective gene and thus, protein, that cause the body to produce unusually thick, sticky mucus that clogs the lungs, obstructs the pancreas, and stops enzymes from helping the body break down and absorb food.

	Normal Sequence	Mutated Sequence
DNA	TAC GTC TCC AGC GGA GAC ATT	TAC GTC TCC CGC GGA GAC ATT
mRNA		
Amino Acids		

1. What type of mutation occurs that results in Cystic fibrosis? _____

2. Explain which codon is mutated and how that mutation affects the amino acid chain.

_____

_____

Hemophilia is a rare bleeding disorder in which the blood doesn't clot normally. A mutated DNA sequence results in a defective gene. Clotting factor is a protein needed for normal blood function. These proteins work with platelets to help the blood clot. Individuals with hemophilia have either no clotting factor or limited clotting factor.

	Normal Sequence	Mutated Sequence
DNA	ATG CTC TGC AAA GAA ATT	ATG CTC TCA AAA GAA ATT
mRNA		
Amino Acids		

3. What type of mutation occurs that results in Hemophilia? _____

4. Explain which codon is mutated and how that mutation affects the amino acid chain.

_____

_____

Name: _____ Date: _____ Period: _____

Researchers have recently discovered that everyone with blue eyes is descended from the same ancestor. This individual had a genetic mutation occur in a gene that controls eye color. This mutation turned off the mechanism which produces melanin in the iris of the eye. Without melanin to give eyes their brown color, they became blue.

	Normal Sequence	Mutated Sequence
DNA	ATG TGC TGG GAA GCC TGC TAA	ATG TGC TGG GTA GCC TGC TAA
mRNA		
Amino Acids		

5. What type of mutation occurs that results in blue eyes? _____

6. Explain which codon is mutated and how that mutation affects the amino acid chain.

_____

_____

Huntington's disease is a neurodegenerative genetic disorder in which nerve cells in certain parts of the brain waste away. This disease is caused by a genetic defect in DNA in which a specific nucleotide sequence, CAG, repeats itself many more times than it is supposed to.

	Normal Sequence	Mutated Sequence
DNA	ATG GTC CAG CAG CAG CAG CCG	ATG GTC CAG CAG CAG CAG CAG CAG CAG CCG
mRNA		
Amino Acids		

7. What type of mutation occurs that results Huntington's disease ? _____

8. Explain how Huntington's disease affects the number of nucleotides in a DNA sequence.

_____

Duchenne muscular dystrophy (DMD) is a fatal disorder, affecting 1 in every 3,500 live male births. Dystrophin is an important protein that acts as a kind of glue, holding muscles together by maintaining the structure of cells. Without dystrophin, muscles are unable to operate properly. According to researchers, "individuals with DMD display premature translational termination resulting in the total loss of protein function." Based on this description, what 3 codons could mistakenly appear in the middle of an mRNA sequence that could cause this disorder? What happens to the mRNA when one of these codons is translated?

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