Mutations Worksheet - Deletion, Insertion & Substitution

There are several types of mutations:

- > **DELETION** (a base is lost/deleted)
- > INSERTION (an extra base is added/inserted)
 - --- Deletion & insertion may cause what's called a FRAMESHIFT mutation, meaning the reading "frame" changes, thus changing the amino acid sequence from this point forward

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- > SUBSTITUTION (one base is substituted for another)
 - --- If a substitution changes the amino acid, it's called a MISSENSE mutation
 - --- If a substitution does not change the amino acid, it's called a SILENT mutation
 - --- If a substitution changes the amino acid to a "stop," it's called a NONSENSE mutation



Complete the boxes below. Classify each as **Deletion**, **Insertion** or **Substitution AND** as either **frameshift**, **missense**, **silent** or **nonsense** (**Hint**: Deletion & Insertion will always be frameshift).

Original DNA Sequence: TACACCTTGGGCGACGACT...

mRNA Sequence: A U G/U G G/A A C/C G C/U G C/U G A

Amino Acid Sequence: Methionine-Tryptophan-Asparagine-Arganine-Cysteine-Stop

Mutated DNA Sequence #1 T A C A T C T T G G C G A C G A C T ...

What's the **mRNA** sequence? $\mathbf{A} \ \mathbf{U} \ \mathbf{G} / \mathbf{U} \ \mathbf{A} \ \mathbf{G} / \mathbf{A} \ \mathbf{A} \ \mathbf{C} / \mathbf{C} \ \mathbf{G} \ \mathbf{C} / \mathbf{U} \ \mathbf{G} \ \mathbf{A} \ (Circle the change)$

What will be the **amino acid** sequence? Methionine-Stop

Will there likely be effects? Yes! What type of mutation is this? Nonsense

Mutated DNA Sequence #2 T A C G A C C T T G G C G A C G A C T ...

What's the **mRNA** sequence? A U G (C) U G / G A A / C C G / C U G / C U G / A (Circle the change)

What will be the amino acid sequence? Methionine-Leucine-Glutamic Acid-Proline-Leucine-Leucine-

Will there likely be effects? Yes! What type of mutation is this? Insertion

Mutated DNA Sequence #3 TACACCTTAGCCGACGACT...

What will be the amino acid sequence? Methionine-Tryptophan-Asparagine-Arganine-Cysteine-Stop

Will there likely be effects? No! What type of mutation is this? Silent

Mutated DNA Sequence #4 T A C A C C T T G G C G A C T A C T ...

What's the **mRNA** sequence? A U G/U G G/A A C/C G C/U G(A)U G A (Circle the change)

What will be the amino acid sequence? Methionine-Tryptophan-Asparagine-Arganine-Stop

Will there likely be effects? Yes! What type of mutation is this? Nonsense

Original DNA Sequence: TACACCTTGGGCGACGACT...

mRNA Sequence: A U G/U G G/A A C/C G C/U G C/U G A

Amino Acid Sequence: Methionine-Tryptophan-Asparagine-Arginine-Cysteine-Stop

Mutated DNA Sequence #5 T A C A C C T T G G G A C G A C T ...

What's the mRNA sequence? A U G/U G G/A A C/C(C)U/G C U/G A (Circle the change)

What will be the amino acid sequence? Methionine-Tryptophan-Asparagine-Proline-Alanine-

Will there likely be effects? Yes! What type of mutation is this? Deletion

- 1. Which type of mutation is responsible for **new variations** of a trait? Missense Mutations
- 2. Which type of mutation does **not** result in an abnormal amino acid sequence? Silent Mutation
- 3. Which type of mutation stops the **translation** of an mRNA molecule? Nonsense Mutation

Sickle Cell Anemia



Sickle cell anemia is the result of a type of mutation in the gene that codes for part of the **hemoglobin** molecule. Recall that hemoglobin carries **oxygen** in your **red bloods cells**. The mutation causes these red blood cells to become stiff & sickle-shaped when they release their oxygen. The sickled cells tend to get stuck in blood vessels, causing pain and increased risk of stroke, blindness, damage to the heart & lungs, and other conditions.

--- Analyze the DNA strands below to determine what amino acid is changed AND what type of mutation occurred

Normal hemoglobin DNA CACGTAGACTGAGGACTC ...

Normal hemoglobin mRNA GUG/CAU/CUG/ACU/CCU/GAG

Normal hemoglobin AA sequence Valine-Histidine-Leucine-Threonine-Proline-Glutamic Acid

Sickle cell hemoglobin DNA CACGTAGACTGAGGACAC ...

Sickle cell hemoglobin mRNA GUG/CAU/CUG/ACU/CCU/GUG

Sickle cell hemoglobin AA sequence Valine-Histidine-Leucine-Threonine-Proline-Valine

4. What type of mutation is this? Please explain why.

This is a substitution-missense mutation because one base changes and it changes the amino acid sequence.