**Delahunty/Biology Honors Mutations Worksheet Name** KEY

There are several types of mutation:

 **DELETION** (a base is lost)

 **INSERTION** (an extra base is inserted)

 Deletion and insertion may cause what’s called a **FRAMESHIFT**, meaning the reading “frame”

 changes, changing the amino acid sequence.

 **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.

**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 the red blood cells to become stiff and 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 and lungs, and other conditions.

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

1a.

Normal hemoglobin DNA **DNA: GGG CTT CTT TTT**

 **mRNA: CCC GAA GAA AAA**

Normal hemoglobin A.A. sequence PRO-GLU-GLU-LYS

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Sickle cell hemoglobin DNA **GGG CAT CTT TTT**

Sickle cell hemoglobin mRNA CCC GUA GAA AAA

Sickle cell hemoglobin A.A. sequence PRO-VAL-GLU-LYS

1b. This is a point mutation, it affect a single point through a substitution mutation.

1c. Not necessarily. This mutation would result in a different protein, so the potential health effects would need to be more specifically examined.



Substitution Substitutions affect a single amino acid, whereas deletion and

 Insertion mutations result in a frameshift mutation and affect all

Deletion amino acids after the change. All types involve a change in a

 Single nucleotide.

Insertion

TEF ATC ATE TET HER AT

This sentence no longer makes sense. This is an example of a deletion resulting in a frame shift.

Methionine and Tryptophan have only one codon. All other amino acids have more than one codon.

Substitution – silent mutation.

If there was no effect on the protein coded by the gene, then the amino acid sequence must have been the same as the original sequence. Therefore the mutation must have been a substitution resulting in a codon that encoded the same amino acid as the original amino acid.



(84 nucleotides/3 nucleotides for each amino acid=28 amino acids before the mutation.)

A frame shift mutation results from an insertion or a deletion, alters the sequence of bases in codons at the mutation and after the mutation. This changes the amino acid sequence and the resulting protein.

A point mutation could be a silent mutation, maintaining the original amino acid sequence and the resulting protein.

A frameshift mutation is more likely to result in a nonfunctional protein.

# 1 and # 3 have the same amino acid sequence, therefore they code for the same protein

The first 28 amino acids will be correct.

 Complete the boxes below. Classify each as either Deletion, Insertion, or Substitution **AND** as either

 frameshift, missense, silent or nonsense (hint: deletion or insertion will always be frameshift).

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| **Original DNA Sequence**: **T A C A C C T T G G C G A C G A C T****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) |

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| 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? A U G U A G A A C C G C U G C U G A (Circle the change) What will be the amino acid sequence? METHIONINE -(STOP) Will there likely be effects? YES What kind of mutation is this? SUBSTITUTION - NONSENSE |

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| 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 Will there likely be effects? YES What kind of mutation is this? INSERTION - FRAME SHIFT |

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| Mutated DNA Sequence #3: **T A C A C C T T A G C G A C G A C T** What’s the mRNA sequence? A U G U G G A A U C G C U G C U G A (Circle the change) What will be the amino acid sequence? METHIONINE-TRYPTOPHAN-ASPARAGINE- ARGININE- (STOP)Will there likely be effects? NO What kind of mutation is this? SUBSTITUTION – SILENT MUTATION |

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| 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- (STOP) Will there likely be effects? YES What kind of mutation is this? SUBSITUTION - NONSENSE |

**Original DNA Sequence**: **T A C A C C T T G G C G A C G A C T**

A U G U G G A A C C G C U G C U G A

**mRNA Sequence:**

METHIONINE-TRYPTOPHAN-ASPARAGINE-ARGININE-CYSTEINE- (STOP)

**Amino Acid Sequence:**

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| Mutated DNA Sequence #5: **T A C A C C T T G G G A C G A C T** What will be the corresponding mRNA sequence? A U G U G G A A C C C U G C U G A What will be the amino acid sequence? METHIONINE-TRYPTOPHAN-ASPARAGINE- PROLINE - ALANINE Will there likely be effects? YES What kind of mutation is this? DELETION – FRAME SHIFT |