**Activity 2: Mutations and Coding Practice**

**(Topic 1: DNA and Protein Synthesis)**

Complete the following with Mrs. Perkins:

In your own words, what happens in the following mutations?

a) Insertion

b) Deletion

c) Substitution

d) “Silent”

Practice:

*Directions: Identify the type of mutation present*

1. Normal Strand: AUCGACTTAACT

Transcribed:

Translated:

Protein Amino Acid Sequence:

2. Mutated Strand: AUCGACTTACT

Transcribed:

Translated:

AA Sequence:

Type of Mutation:

3. Mutated Strand: AUCGCCTTAACT

Transcribed:

Translated:

AA Sequence:  
Type of Mutation:

4: Mutated Strand: AUCGAACTTAACT

Transcribed:

Translated:

AA Sequence:

Type of Mutation:

How the Gene for Sickle Cell Hemoglobin Results in Sickle Cell Anemia

Different versions of the same gene are called different alleles. These different alleles share the same general sequence of nucleotides, but they differ in at least one nucleotide in the sequence. Different alleles can result in different characteristics as follows:

Different nucleotide sequence in the different alleles of a gene different nucleotide sequence in messenger RNA (mRNA)

transcription

 different amino acid sequence in a protein translation

 different structure and function of the protein (e.g. normal enzyme vs. defective enzyme)

different characteristics (e.g. normal skin and hair color vs. albinism)

In this section, you will learn about another example of how different alleles produce different characteristics. To begin, you will work with your partner to understand how a difference between the alleles for normal and sickle cell hemoglobin results in two different types of hemoglobin protein.

22. In the table below, compare the nucleotide sequence in the DNA for the Beginning of the Normal Hemoglobin Gene vs. the Beginning of the Sickle Cell Hemoglobin Gene. What is the only difference?

23. Complete this table. (Use the table on page 5 to help with translation.)

  

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Beginning of Normal Hemoglobin Gene | | CACGTAGACTGAGGACTC | | | | | | |
| Transcription produces: | | codon1 | codon 2 | codon 3 | codon 4 | codon 5 | codon 6 | |
| Beginning of Normal Hemoglobin mRNA | |  |  |  |  |  |  | |
| Translation produces: | | amino acid 1 | amino acid 2 | amino acid 3 | amino acid 4 | amino acid 5 | amino acid 6 | |
| Beginning of Normal Hemoglobin Protein | |  |  |  |  |  |  | |
|  |  | | | | | | |  |
| Beginning of Sickle Cell Hemoglobin Gene | | CACGTAGACTGAGGACAC | | | | | | |
| Transcription produces: | | codon 1 | codon 2 | codon 3 | codon 4 | codon 5 | codon 6 | |
| Beginning of Sickle Cell Hemoglobin mRNA | |  |  |  |  |  |  | |
| Translation produces: | | amino acid 1 | amino acid 2 | amino acid 3 | amino acid 4 | amino acid 5 | amino acid 6 | |
| Beginning of Sickle Cell Hemoglobin Protein | |  |  |  |  |  |  | |

24. What is the difference in the amino acid sequence of the beginning of the hemoglobin molecules synthesized by translating the sickle cell vs. normal hemoglobin mRNA molecules?

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Each complete hemoglobin protein has more than 100 amino acids. Sickle cell hemoglobin and normal hemoglobin differ in only a single amino acid. This difference in a single amino acid results in the very different properties of sickle cell hemoglobin, compared to normal hemoglobin.

If a person inherits two copies of the sickle cell hemoglobin allele and produces only sickle cell hemoglobin, then the sickle cell hemoglobin molecules tend to clump together in long rods. These rods can change the shape of the red blood cells from their normal disk shape to a sickle shape. Sickle- shaped red blood cells can block blood flow in the small blood vessels. This causes pain and damage to body organs. In addition, sickle-shaped red blood cells do not last nearly as long as normal red blood cells, so the body cannot produce enough replacement red blood cells and the person develops anemia (not enough red blood cells).

25. Circle the arrows in the chart that represent transcription + translation.

In summary, the sickle cell allele results in production of the sickle cell hemoglobin protein, which results in the health problems observed in sickle cell anemia. This is a dramatic example of the importance of the nucleotide sequence in a gene, which determines the amino acid sequence in a protein, which in turn influences the characteristics of an individual.

26. Considering that we are all made up of the same 4 nucleotides in our DNA, the same 4 nucleotides in our RNA, and the same 20 amino acids in our proteins, why are we so different from each other? For example, why do some people have sickle cell anemia and others don't?

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Genotype (genes) |  | Protein |  | Phenotype (characteristics) |
| 2 copies of the allele that codes for normal hemoglobin (SS) |  | Normal hemoglobin dissolves in the cytosol of red blood cells. |  | Disk-shaped red blood cells can squeeze through the small blood vessels  normal health |
| 2 copies of the allele that codes for sickle cell hemoglobin (ss) |  | Sickle cell hemoglobin can clump in long rods in red blood cells. |  | When sickle cell hemoglobin clumps in long rods  sickle-shaped red blood cells   clogged small blood vessels + fragile red blood cells  pain, damage to body organs  + anemia = sickle cell anemia |

