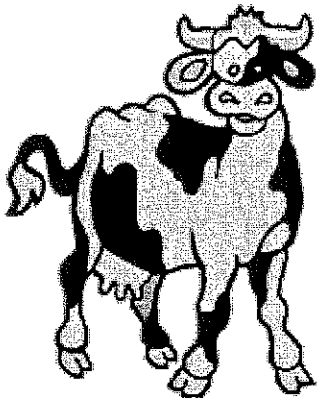
Name: _____

nvestigation: DNA, Proteins, and Mutations



Below are two partial sequences of DNA bases (shown for only one strand of DNA) Sequence 1 is from a human and sequence 2 is from a cow. In both humans and cows, this sequence is part of a set of instructions for controlling a bodily function. In this case, the sequence contains the gene to make the protein insulin. Insulin is necessary for the uptake of sugar from the blood. Without insulin, a person cannot use/digest sugars the same way others can, and they have a disease called diabetes.

Instructions:

1 Using the DNA sequence, make a complimentary RNA strand from both the human and the cow. Write the RNA directly below the DNA strand (remember to substitute U's for T's in RNA)

2. Use the codon table in your book to determine what amino acids are assembled to make the insulin protein in both the cow and the human.

**Note: This is not the real sequence for insulin, which actually contains 51 amino acids. Uniprot provides full sequencing information on insulin and known variants. http://www.uniprot.org/uniprot/P01308*

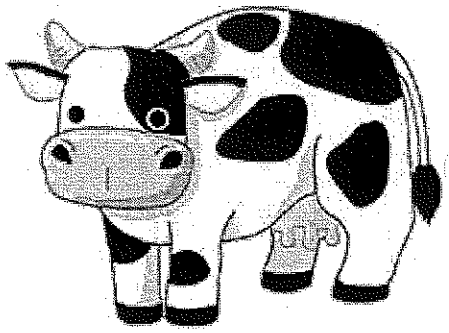
HUMAN DNA									
DNA	CCA	TAG	CAC	GTT	ACA	ACG	TGA	AGG	AAA
RNA									
Amino Acid									

COW DNA									
DNA	CCA	TAG	CAT	GTT	ACA	ACG	CGA	AGG	GAC
RNA									
Amino Acid									

Analysis

1. Compare the DNA Sequence and circle any single base that is different in the cow and human sequences. How many bases are different? _____

2. Examine the amino acids produced. Highlight in yellow any amino acids that are different in the two sequences. How many are there? _____



3. Could two humans (or two cows) have some differences in their DNA sequences for insulin, yet still make the exact same insulin proteins? Explain.

4. Examine the codon chart and list all of the codons that code for the amino acid leucine. List them:

MUTATIONS: Changes in DNA

Diabetes is a disease characterized by the inability to break down sugars. Often a person with diabetes has a defective DNA sequence that codes for the making of the insulin protein. This mutation is called a **POINT MUTATION** because only one base is affected.

When the amino acid in a sequence has been changed, the shape of the protein changes and can become less functional (or not function at all.)

5.. Suppose a person has a mutation in their DNA, and the first triplet for the gene coding for insulin is T A T (instead of C C A). Determine what amino acid the new DNA triplet codes for. Will this person be diabetic? Explain

6. **A SILENT MUTATION** occurs when the nucleotide changes, but the resulting amino acid is the same. The protein that is made from this new DNA will have no functional difference from the original.

What if a mutation occurred in the human insulin gene and the first triplet was changed to C C G? Is this a silent mutation? Explain how you know.

A **FRAMESHIFT MUTATION** occurs when a base is added (or removed) from a DNA/RNA sequence.

7. Determine the amino acid chain coded for by the following sequence. Suppose a mutation occurs where another A is added after the first codon. What would the new sequence of amino acids be?

Normal DNA: T G G A G T C G A G G T

Normal RNA:

Amino Acids:

Mutant DNA: T G G A A G T C G A G G T

Mutant RNA:

Amino Acids.

Why are frameshift mutations likely to cause more problems than a point mutation?

8. A **NONSENSE MUTATION** occurs when a codon is changed to a STOP codon. Many proteins are thousands of amino acids in length. At the end of the gene, the cell reaches the stop codon. If a codon is changed to STOP, the building of the amino acid terminates and the protein is incomplete.

Why would a Nonsense mutation be less damaging if it occurred at the end of the gene rather than the beginning?

Gene Sequences Across Species

9. DNA sequences are often used to determine relationships between organisms. DNA sequences that code for a particular gene can vary widely. Organisms that are closely related will have sequences that are similar.

Below is a list of sequences for a few organisms:

Human: C C A T A G C A C C T A	Chimpanzee: C C A T A A C A C C T A
Pig: C C A T G T A A A C G A	Cricket: C C T A A A G G G A C G

Based on these sequences, which two organisms are most closely related? _____

10. An unknown organism is found in the forest and the gene is sequenced as follows:

Unknown: C C A T G G A A T C G A .....

What kind of an animal do you think this is? _____

SYNTHESIS

Answer each of the following using a single summary sentence.

11. What is the relationship between DNA, codons, and proteins?

12. How does the shape of a protein relate to its function?

Use this table to compare the nucleotide sequence in the "Beginning of Allele for Normal Hemoglobin" vs. the "Beginning of Allele for Sickle Cell Hemoglobin". What is the only difference?

Beginning of **Allele** for **Normal** Hemoglobin	**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 **Allele** for **Sickle Cell** Hemoglobin	**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**						

Complete the above table. Use the table below to help with translation.

mRNA codon	ACU	CAU	CCU	CUG	GAG	GUG
Corresponding **amino acid**	Thr (Threonine)	His (Histidine)	Pro (Proline)	Leu (Leucine)	Glu (Glutamic acid)	Val (Valine)

Compare the amino acid sequence for the beginning of sickle cell hemoglobin vs. the beginning of normal hemoglobin. What difference do you observe?

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

1st base

2nd base	U		C		A		G		3rd base
U	UUU	Phenylalanine	UCU	Serine	UAU	Tyrosine	UGU	Cysteine	U
	UUC	Phenylalanine	UCC	Serine	UAC	Tyrosine	UGC	Cysteine	C
	UUA	Leucine	UCA	Serine	UAA	Stop	UGA	Stop	A
	UUG	Leucine	UCG	Serine	UAG	Stop	UGG	Tryptophan	G
C	CUU	Leucine	CCU	Proline	CAU	Histidine	CGU	Arginine	U
	CUC	Leucine	CCC	Proline	CAC	Histidine	CGC	Arginine	C
	CUA	Leucine	CCA	Proline	CAA	Glutamine	CGA	Arginine	A
	CUG	Leucine	CCG	Proline	CAG	Glutamine	CGG	Arginine	G
A	AUU	Isoleucine	ACU	Threonine	AAU	Asparagine	AGU	Serine	U
	AUC	Isoleucine	ACC	Threonine	AAC	Asparagine	AGC	Serine	C
	AUA	Isoleucine	ACA	Threonine	AAA	Lysine	AGA	Arginine	A
	AUG	Methionine (Start)	ACG	Threonine	AAG	Lysine	AGG	Arginine	G
G	GUU	Valine	GCU	Alanine	GAU	Aspartic Acid	GGU	Glycine	U
	GUC	Valine	GCC	Alanine	GAC	Aspartic Acid	GGC	Glycine	C
	GUA	Valine	GCA	Alanine	GAA	Glutamic Acid	GGA	Glycine	A
	GUG	Valine	GCG	Alanine	GAG	Glutamic Acid	GGG	Glycine	G