(reference pgs. 239-240 in Modern Biology textbook)

 Name_____
 Date_____
 Per_____

Look at the diagrams, then answer the questions.

Gene Mutations affect a single gene by changing its base sequence, resulting in an incorrect, or nonfunctional, protein being made.

(a) A **SUBSTITUTION** mutation, occurs where one nucleotide base is replaced by another. These are often called "**point mutations**", because a **single base** is changed, at one **point** in the gene.

SOMETIMES, these base (A,T,C,G) substitutions lead to "Missense" or "Nonsense" mutations:

Missense mutation

Nonsense mutation

Original DNA code for an amino acid sequence.	DN ba	Original IA \rightarrow C A ses GI	DNA code fo	GCAG	id sequence.
Amino acid Replacement of a single nucleotide. ↓	T Amino acid Replacement of a single nucleotide.				ement of a nucleotide. ↓
CATCATCATCCTCATCAT HIS H HIS H HIS H Pros H HIS H HIS H HIS H.		C A	GCA n H Gin		TAGCAGCAGCAG Stop
Incorrect amino acid, which may produce a malfunctioning protein.		202	Protein	-	Incorrect seqence causes shortening of protein.
U.S. National Library of Medicine	U.	S. National L	ibrary of Med	licine	
1	Silent Mutations]
there is more than one codon	ATG	GAA	GCA	СGТ	
for each amino acid, these	Met	Glu	Ala	Gly	
mutations can be SILENT :					
				_	
	ATG	GAG	GCA	CGT	
	Met	Glu	Ala	Gly	

(b) Other types of mutations (insertions or deletions of nucleotide bases) cause FRAMESHIFTS:

Insertion mutation





U.S. National Library of Medicine

Deletion mutation

(reference pgs. 239-240 in Modern Biology textbook)

1. There are several types of gene mutations. List two. What do they have in common? How are they different?

2. A geneticist found that a particular mutation had no effect on the protein coded by a gene. What kind of mutation was this? Why?

- 3. Name one amino acid that has more than one codon. Name an amino acid that has only one codon
- 4. Look at the following sequence: THE FAT CAT ATE THE RAT. Delete the first H and regroup the letters in groups of three- write out the new groups of three. Does the sentence still make sense? What type of mutation is this an example of?
- 5. You have a DNA sequence that codes for a protein and is 105 nucleotides long. A frameshift mutation occurs at the 85th base- how many amino acids will be correct in this protein?
- 6. Given the following three mRNA sequences, TWO code for the same protein. Which two?

#1. AGU UUA GCA ACG AGA UCA#2 UCG CUA GCG ACC AGU UCA#3 AGC CUC GCC ACU CGU AGU

7. What kind of mutation is *more likely* to result in a nonfunctional protein, a **frameshift** (resulting from an insertion or deletion) or a **point mutation**?

Why?

(reference pgs. 239-240 in Modern Biology textbook)

8. Below is the DNA base sequence for the normal protein for normal hemoglobin and the base sequence for (abnormal) sickle cell hemoglobin:



Normal GGG CTT CTT TTT Sickle GGG CAT CTT TTT

A. *Transcribe* and *translate* the normal and sickle cell DNA sequences above.

mRNA codon sequence:

amino acids:

B. What kind of a gene mutation is this? Support your answer.

C. If the base sequence read GGG CTT CTT TTC instead, would this result in functional hemoglobin? Explain.

Chromosomal Mutations

- Alterations of chromosome number or structure cause some genetic disorders
- Large-scale chromosomal alterations often lead to spontaneous abortions or cause a variety of developmental disorders, or even cancers.
- Breakage of a chromosome can lead to four types of changes in chromosome structure:



Figure 15.14a-d

(reference pgs. 239-240 in Modern Biology textbook)

9. Match the chromosome mutation with its description.

	-
	A). A portion of the chromosome is missing or
	deleted. Known disorders in humans include Wolf-
1. Translocation	Hirschhorn syndrome, which is caused by partial
	deletion of the short arm of chromosome 4; Cri du
2. Inversion	chat syndrome is due to a partial deletion of the short
	arm of chromosome number 5.
3. Deletion	
	B). A portion of the chromosome is duplicated,
4. Duplication	resulting in extra genetic material. Known human
-	disorders include Charcot-Marie-Tooth disease type
	1A which may be caused by duplication of the gene
	encoding peripheral myelin protein 22 (PMP22) on
	chromosome 17.
	C). When a portion of one chromosome is transferred
	to another chromosome. Sometimes, parts of different
	chromosomes switch places (reciprocal exchange).
	D). A portion of the chromosome has broken off,
	turned upside down and reattached, therefore the
	genetic material is backward.

10. Why are chromosome mutations potentially more serious than gene mutations?

	U	с	A	G
U	UUU Phe UUC Phe UUA Leu UUG Leu	UCU UCC UCA UCG	UAU UAC UAA UAA UAG	UGUCys UGC UGA - Stop UGG - Trp
с	CUU CUC CUA CUG	CCU CCC CCA CCG	CAU His CAC His CAA Gin CAG	CGU CGC CGA CGG
A	AUU AUC AUA AUG — Met	ACU ACC ACA ACG	AAU AAC AAA AAA AAG	AGU Ser AGC AGA AGA Arg
G	GUU GUC GUA GUG	GCU GCC GCA GCG	GAU Asp GAC GAA GAA Glu GAG	GGU GGC GGA GGG

mRNA Codon decoder grid:

DNA Base triplet decoder grid:

	Т	С	Α	G	
т	Phe Phe Leu Leu	Ser Ser Ser Ser	Tyr Tyr *	Cys Cys * Trp	T C A G
с	Leu	Pro	His	Arg	T
	Leu	Pro	His	Arg	C
	Leu	Pro	GIn	Arg	A
	Leu	Pro	GIn	Arg	G
Α	lle	Thr	Asn	Ser	T
	lle	Thr	Asn	Ser	C
	lle	Thr	Lys	Arg	A
	Met	Thr	Lys	Arg	G
G	Val	Ala	Asp	Gly	T
	Val	Ala	Asp	Gly	C
	Val	Ala	Glu	Gly	A
	Val	Ala	Glu	Gly	G