

## Gene and Chromosome Mutation Worksheet

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

Name \_\_\_\_\_ Date \_\_\_\_\_ Per \_\_\_\_\_

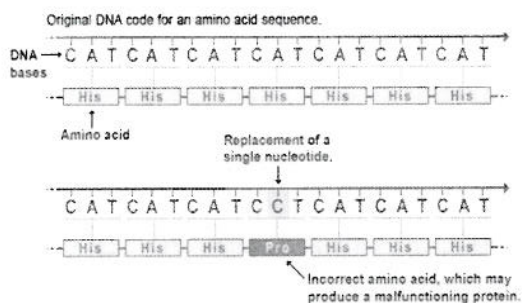
Look at the diagrams, then answer the questions.

**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.

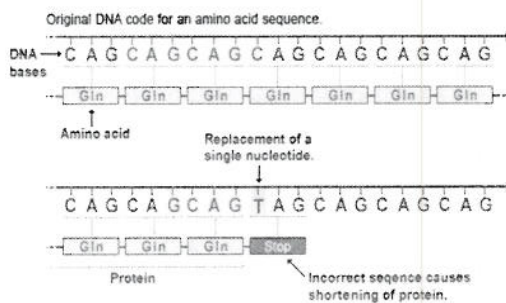
SOMETIMES, these base (A,T,C,G) substitutions lead to "**Missense**" or "**Nonsense**" mutations:

Missense mutation



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Nonsense mutation



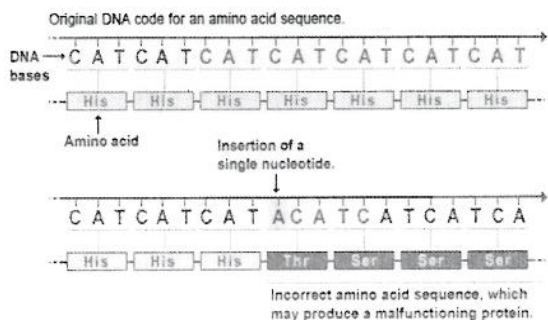
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...and sometimes, because there is more than one codon for each amino acid, these mutations can be **SILENT**:

Silent Mutations			
ATG	GAA	GCA	CGT
Met	Glu	Ala	Gly
↓			
ATG	GAG	GCA	CGT
Met	Glu	Ala	Gly

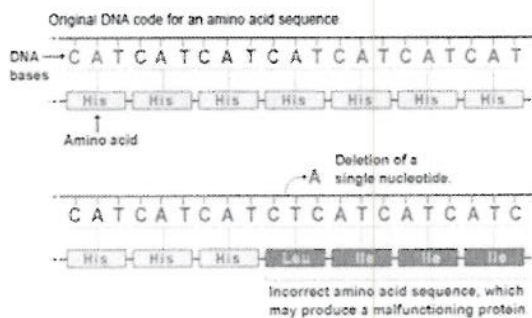
(b) Other types of mutations (insertions or deletions on nucleotide bases) cause **FRAMESHIFTS**:

Insertion mutation



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Deletion mutation



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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 do you think is the most likely type of mutation in this gene? 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 85<sup>th</sup> 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?

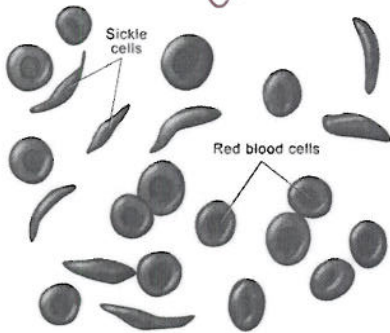
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8. Below is the DNA base sequence for the normal protein for normal hemoglobin and the base sequence for (abnormal) sickle cell hemoglobin:

Use Codon Chart

**Normal** GGG CTT CTT TTT  
**Sickle** GGG CAT CTT TTT



A. Transcribe and translate the normal and sickle cell DNA.

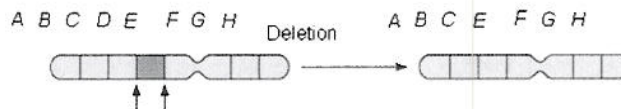
B. Identify this as a point or frameshift mutation. Explain.

C. If the base sequence read GGG CTT CTT AAA instead, would this result in sickle cell hemoglobin? Explain.

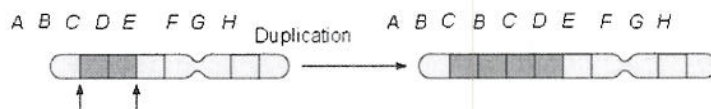
### Chromosomal Mutations

- 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:

(a) A **deletion** removes a chromosomal segment.



(b) A **duplication** repeats a segment.



(c) An **inversion** reverses a segment within a chromosome.



(d) A **translocation** moves a segment from one chromosome to another, nonhomologous one. In a reciprocal translocation, the most common type, nonhomologous chromosomes exchange fragments. Nonreciprocal translocations also occur, in which a chromosome transfers a fragment without receiving a fragment in return.

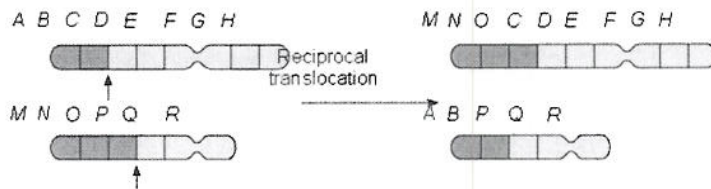


Figure 15.14a-d

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9. Match the chromosome mutation with its description.

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

### Codons in mRNA

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