

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:

...ACT CAG AAC...
...Thr Gln Asn...

...ATA CGA GCT...
...Ile Arg Ala...

...ACT CGG AAC...
...Thr Arg Asn...

...ATA TGA GCT...
...Ile Stop

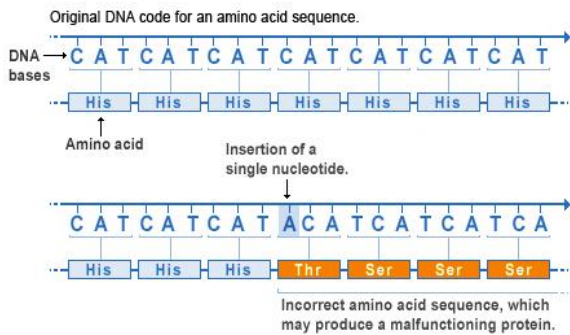
...and sometimes, because there is more than one codon for each amino acid, these mutations can be **SILENT**:

...TTC TGT AGT GGT...
...Phe Cys Ser Gly...

...TTC TGC AGT GGT...
...Phe Cys Ser Gly...

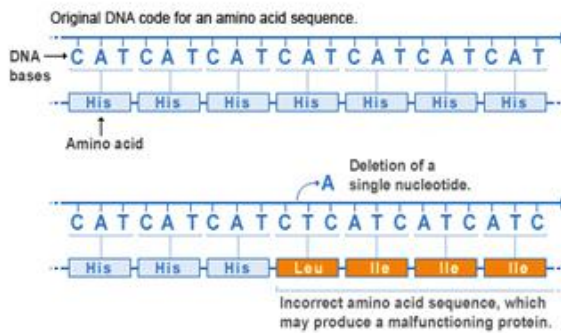
(b) Other types of mutations (**insertions** or **deletions** of nucleotide bases) cause **FRAMESHIFTS**:

Insertion mutation



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Deletion mutation



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

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

3. (a) Name one amino acid that has more than one codon. (b) Name an amino acid that has only one codon

4. Look at the following sequence: THE FAT CAT ATE THE RAT. (a) Delete the first H and regroup the letters in groups of three (write out the new groups of three). (b) Does the sentence still make sense? (c) What type of mutation is this?

5. You have a DNA sequence that codes for a protein and is 105 nucleotides long. A frameshift mutation occurs at the 85th base in the nucleotide sequence. How many amino acids will be still 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

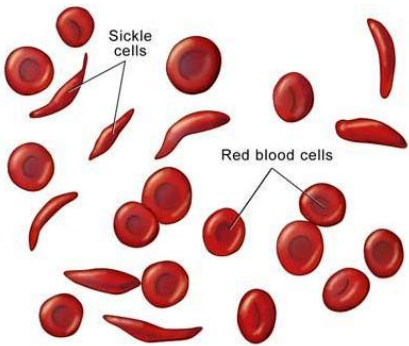
#2 UCG CUA GCG ACC AGU UCA

#3 AGC CUC GCC ACU CGU AGU

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



Normal gene: GGG CTT CTT TTT
Sickle gene: GGG CAT CTT TTT

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

mRNA codon sequence: normal:

sickle:

amino acids coded: normal:

sickle:

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

C. If the base sequence read GGG CTT CTT TTC instead... (a) would this result in functional hemoglobin? (b) Explain.

Chromosomal Mutations

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

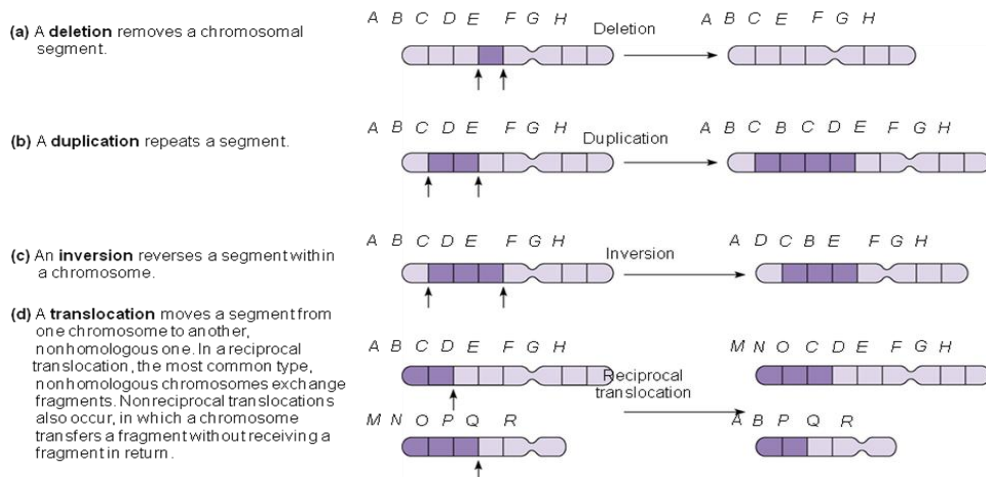


Figure 15.14a-d

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9. Match the chromosome mutation with its description.

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| <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 Wolf-Hirschhorn syndrome, 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 Charcot-Marie-Tooth disease type 1A which may be caused by duplication of the gene encoding peripheral myelin protein 22 (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?

mRNA Codon decoder grid:

| | U | C | A | G |
|---|------------------------------------------|--------------------------------------|-------------------------------------------|-----------------------------------------------|
| U | UUU } Phe UUC } UUA } Leu UUG } | UCU } UCC } Ser UCA } UCG } | UAU } Tyr UAC } UAA } Stop UAG } | UGU } Cys UGC } UGA } Stop UGG } Trp |
| C | CUU } CUC } Leu CUA } CUG } | CCU } CCC } Pro CCA } CCG } | CAU } His CAC } CAA } Gln CAG } | CGU } CGC } Arg CGA } CGG } |
| A | AUU } AUC } Ile AUA } AUG } Met | ACU } ACC } Thr ACA } ACG } | AAU } Asn AAC } AAA } Lys AAG } | AGU } Ser AGC } AGA } Arg AGG } |
| G | GUU } GUC } Val GUA } GUG } | GCU } GCC } Ala GCA } GCG } | GAU } Asp GAC } GAA } Glu GAG } | GGU } GGC } Gly GGA } GGG } |