1) What is a codon?
2) Given the following sentence, perform each indicated task. Remember that codons must be 3 letters each.

THE FAT CAT ATE THE RAT.

A) Substitute the "T" in CAT with an "R". Rewrite the sentence 3 letters at a time.

B) Insert an "R" after the A in FAT. Rewrite the sentence 3 letters at a time.

C) Delete the "A" in ATE. Rewrite the sentence 3 letters at a time.

3) Which change resulted in the most minimal effect? Justify your choice.
4) Which change resulted in the most dramatic effect? Justify your choice.

Biology Chapter 12 Section 4

Mutations

Objectives: Students will

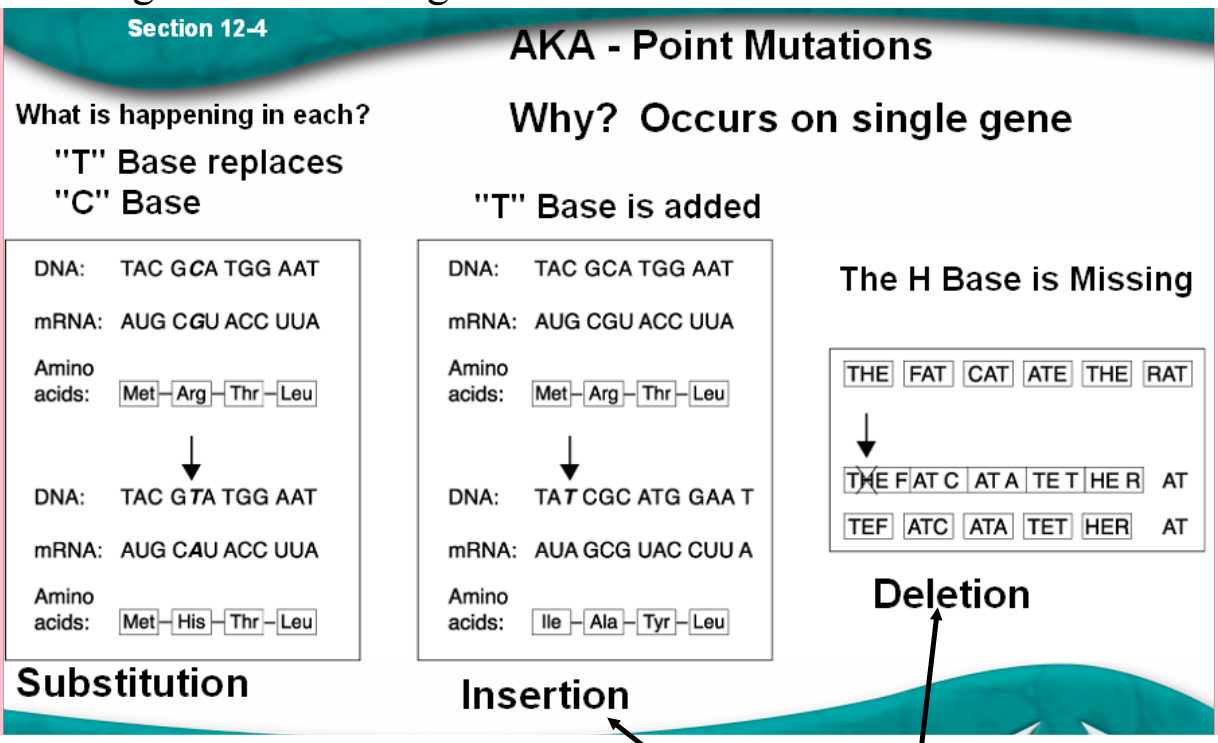
A) Define the three types of point mutations.

B) Define the four types of chromosomal mutations.

C) Explain the impact of mutations on the body.

OBJ. A) DEFINE THE 3 TYPES OF POINT MUTATIONS.

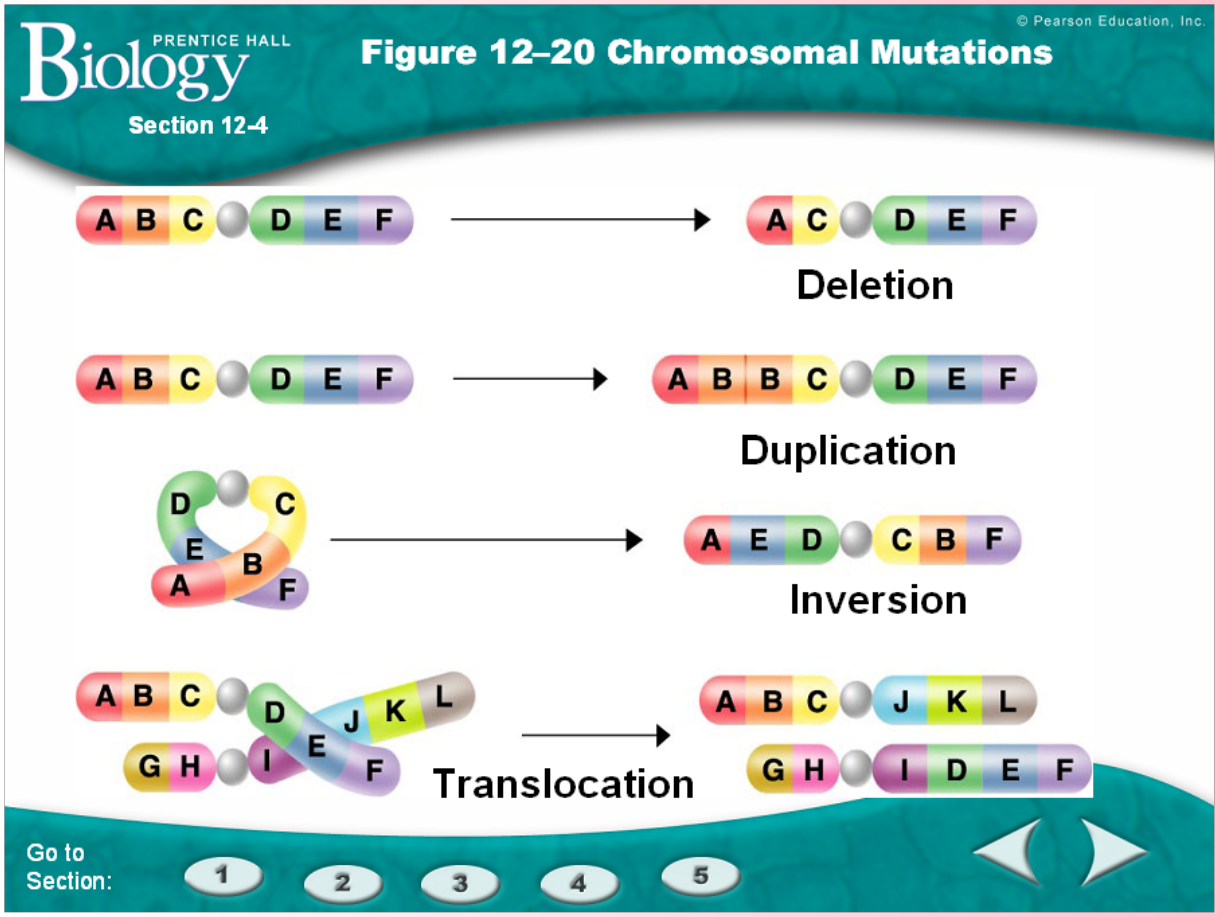
What is a point mutation?
1 nitrogen base is changed



SILENT MUTATION - RESULTS IN SAME AMINO ACID. EX: CGU = Arg CGG, CGC, CGA = also give Arg

FRAMESHIFT MUTATIONS

OBJ. B) DEFINE THE 4 TYPES OF CHROMOSOMAL MUTATIONS.



OBJ. C) EXPLAIN THE IMPACT OF MUTATIONS ON THE BODY.

Mutations are rare.

Humans inherit 3,000,000,000 base pairs from each parent.

Considering single base substitutions = 6 billion targets of substitutions
Single base substitutions = most apt during DNA copying of S phase

Even the most highly skilled typist will introduce errors when copying a manuscript. So does DNA even though it proofreads.

Mutations are estimated to occur at a rate of 1 in every 50 million nucleotides.

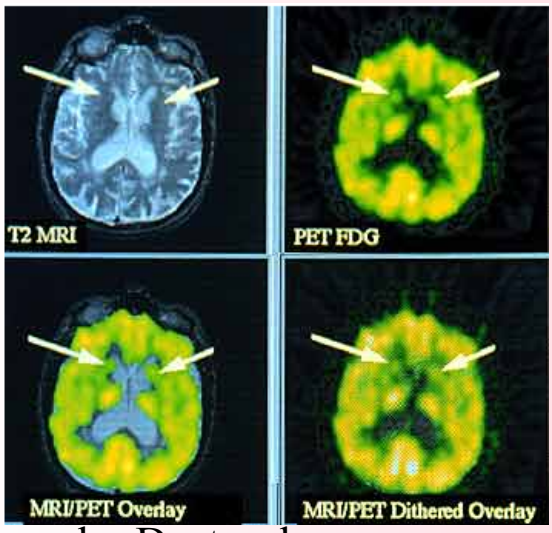
With 6×10^9 base pairs this means that each new cell contains some 120 new mutations.

Worried? Don't be
Most (as much as 97%) of our DNA does not encode anything.

Examples: Insertions

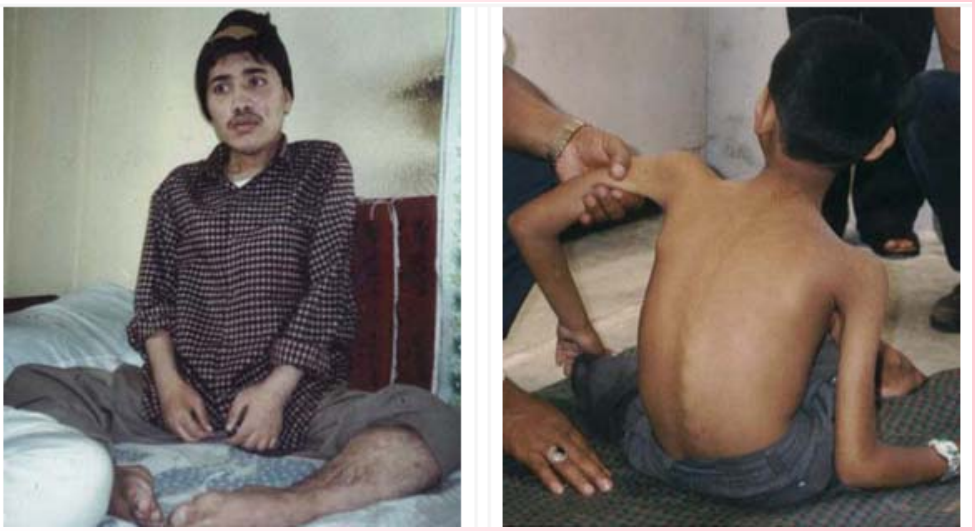
Huntington's Disease

Disorder in which trinucleotide CAG is repeated. Adds a string of glutamines (Gln) to the encoded protein. Increases p53 protein in the brain cells causing their death.



Muscular Dystrophy

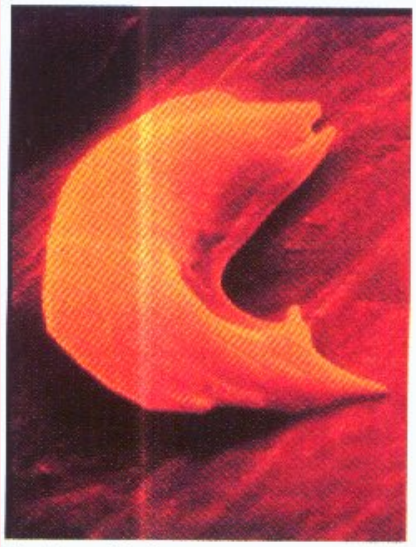
Caused by tri- or tetranucleotide repeats (CTG or CCTGs) These repeats may run into the thousands. The huge mRNA transcripts that result interfere with the splicing of other mRNAs in the nucleus. Result = loss of muscle and death by thirty. Muscle required to breath and pump the heart.



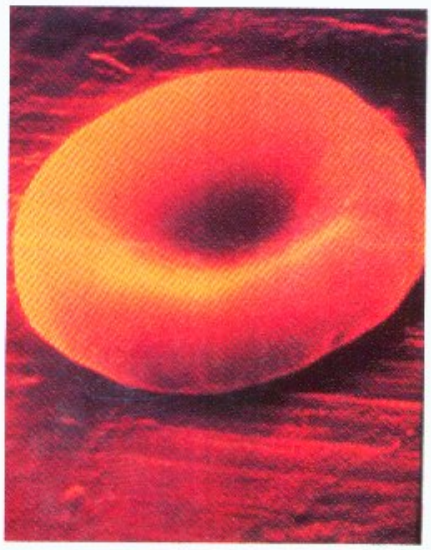
Examples: Single base substitutions
Sickle cell disease

EXAMPLE: **sickle-cell disease** The replacement of A by T at the 17th nucleotide of the gene for the beta chain of hemoglobin changes the codon GAG for glutamic acid to CTG which codes for valine. Thus the 6th amino acid in the chain becomes valine instead of glutamic acid.

You will complete a worksheet analyzing the 3 types of point mutations.



Sickle-cell phenotype



Normal phenotype

Biology Chapter 12-4 Mutations Practice Reference Sheet

There are three ways that DNA can be altered when a mutation (change in DNA sequence) occurs.

1. **Substitution** – one base-pairs is replaced by another:
   a. Example: G mutated to **C** or A mutated to **G**
      C becomes **G** or U becomes **C**
2. **Insertion**: one or more base pairs is added to a sequence:
   a. Example: CGA UGG mutates to CGA **A**UG G
      Complementary bases GCU ACC become GCU **U**AC C
3. **Deletion** – one or more base pairs is lost from a sequence:
   a. Example: C**G**A UGG mutates to CAU GG
      Complementary bases G**C**U ACC become GUA CC

There are five **possible results** of a mutation.

1. **Silent mutation** : When a base pair is substituted but the change still codes for the same amino acid in the sequence:
   Example: UC**U** and UC**C** both code for the amino acid Serine.
2. **Substitution**: When a base pair is substituted and the new codon codes for a different amino acid:
   Example: **U**CU codes for Serine and **C**CU codes for Proline.
3. **Premature Stop**: When a substitution results in the formation of a STOP codon before all of the codons have been read and translated by the ribosome.
   Example: GUG GUC UG**G** AAC ACC – GUG GUC UG**A** CGA AAC ACC
   Val - Val –Trp –Asn -Thr Val – Val - STOP
4. **Codon Deletion or Insertion**: A whole new amino acid is added, or one is missing as a result of the mutant base.

DIRECTIONS: Transcribe and translate the original DNA sequence. Then, do the same for each mutated DNA sequence. Then, determine the consequence, if any, for each mutation, by circling your choice for each question. **You will need a Genetic Code Chart.**

Original DNA sequence:	**TAC ACC TTG GCG ACG ACT**
mRNA transcript:	
amino acids:	

Mutated DNA sequence #1:	**TAC ATC TTG GCG ACG ACT**		
mRNA transcript: ***(Circle any changes)***			
amino acids:			
Type of mutation (Circle one.)	Point ⇨	Substitution	
How did the mutation affect the amino acid sequence (protein)? (Circle one.)	No change	1 amino acid changed	Prematu stop sign

Mutated DNA sequence #2:	**TAC GAC CTT**
mRNA transcript: ***(Circle any changes)***	
amino acids:	

Codons Found in Messenger RNA

Second Base

First Base	U	C	A	G	*Third Base*
U	Phe	Ser	Tyr	Cys	U
	Phe	Ser	Tyr	Cys	C
	Leu	Ser	Stop	Stop	A
	Leu	Ser	Stop	Trp	G
C	Leu	Pro	His	Arg	U
	Leu	Pro	His	Arg	C
	Leu	Pro	Gln	Arg	A
	Leu	Pro	Gln	Arg	G
A	Ile	Thr	Asn	Ser	U
	Ile	Thr	Asn	Ser	C
	Ile	Thr	Lys	Arg	A
	Met	Thr	Lys	Arg	G
G	Val	Ala	Asp	Gly	U
	Val	Ala	Asp	Gly	C
	Val	Ala	Glu	Gly	A
	Val	Ala	Glu	Gly	G