

## Point Mutations Activity

**Background:** There are two types of mutations, small-scale gene mutations and large-scale chromosomal mutations. In this activity you will be learning about gene mutations. There are two basic types of gene mutations: **base substitution** and **frameshift (insertions and deletions)**.

### Part 1: **Frameshift Mutations**

#### Example 1: **Insertion Frameshift**

DNA Sequence Sentence: THE FAT PIG ATE THE BIG BUN AND HOT DOG



Effect of Insertion: THE FAT XPI GAT ETH EBI GBU NAN DHO TDO



**Insert a nitrogen base**

#### Example 2: **Deletion Frameshift**

**Delete a nitrogen base**



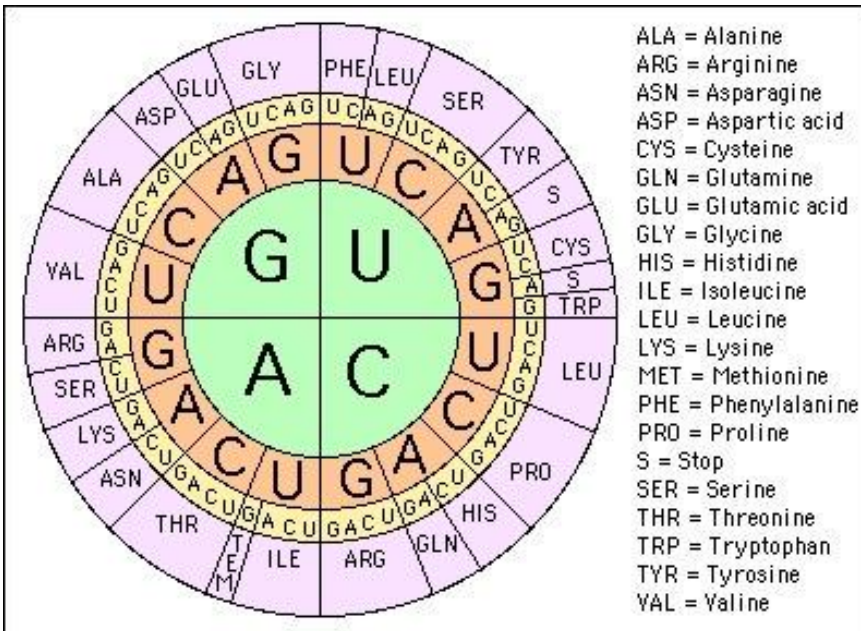
DNA Sequence Sentence: THE FAT PIG ATE ~~THE~~ BIG BUN AND HOT DOG



Effect of Deletion: THE FAT PIG ATE TEB IGB UNA NDH OTD

The insertion shifts the reading frame to the right. The deletion shifts the reading frame to the left. Complete the following lines for frameshift mutations.

1. Why did I only use 3 letter words? What do they represent? \_\_\_\_\_
2. What is the mutation's effect on the resulting protein? \_\_\_\_\_
3. Why are most frameshift mutations fatal? \_\_\_\_\_



**Part II:** Now use a real DNA sequence and transcribe and translate it into the correct amino acids. In the original DNA code put the insertion frameshift mutation in the **FOURTH CODON by inserting a G after the C in TCT**. Please use the mRNA codon chart below to find the corresponding amino acids.

Write each codon per line and **highlight** the mutated nitrogen base where the mutation took place and the amino acid(s) that are affected.

Original DNA	TAC	GGA	CGA	TCT	CAG	GAG	CCT	ATA	ATC
Insertion Mutation	_____	_____	_____	_____	_____	_____	_____	_____	_____
Mutated mRNA	_____	_____	_____	_____	_____	_____	_____	_____	_____
Mutated Amino Acids	_____	_____	_____	_____	_____	_____	_____	_____	_____
Original Amino Acid Sequence	Met	Pro	Ala	Arg	Val	Leu	Gly	Try	STOP

**Part III: Base Substitution Mutations**

Example 1: **Missense Point Mutation**

A missense mutation occurs when one nitrogen base is substituted for another nitrogen base causing a different amino acid than previously to occur in the protein sequence.

Use the DNA code below to demonstrate another type of *missense mutation*. All you have to do is change the DNA base in the **SECOND CODON from GCC to GAC**. Write each codon per line and **circle the mutated nitrogen base and affected amino acids**.

**\*This is the “boy in the bubble” syndrome (severe combined immunodeficiency)\***

Original DNA	AAT	GCC	AGT	GGT	TCG	CAC	CTG	ACC	ACT
Mutation	_____	_____	_____	_____	_____	_____	_____	_____	_____
Mutated mRNA	_____	_____	_____	_____	_____	_____	_____	_____	_____
Mutated Amino Acids	_____	_____	_____	_____	_____	_____	_____	_____	_____
Original Amino Acid Sequence	Leu	Arg	Ser	Pro	Ser	Val	Asp A	Trp	STOP



Example 2: **Silent Point Mutation** – 40% of all mutations are this type.

A *silent mutation* happens when one base in a codon is changed but both code for the same amino acid.

Use the DNA code below to demonstrate a *silent mutation*. All you have to do is change one DNA base in the **THIRD CODON from TCT to TCC**. Write each codon per line and **circle** the mutated nitrogen base and amino acid.

Original DNA		TAC	CAT	TCT	CGG	TGT	AAA	AGG	GCG	ATT
Silent Mutation	_____	_____	_____	_____	_____	_____	_____	_____	_____	_____
Mutated mRNA	_____	_____	_____	_____	_____	_____	_____	_____	_____	_____
Mutated Amino Acids	_____	_____	_____	_____	_____	_____	_____	_____	_____	_____
Original Amino Acid Sequence	Met	Val	Arg	Ala	Thr	Phe	Ser	Arg	STOP	

Example 3: **Nonsense Point Mutation**

A base mutation that creates a new stop codon in place of an amino acid causing a premature stopping of translation is called a *nonsense mutation*.

Use the DNA code below to demonstrate a nonsense mutation. All you have to do is change one DNA base to create a new amino acid sequence with a premature stop codon in the **FIFTH CODON by changing ATA to ATT**. Write each codon per line and **circle** the mutated nitrogen base and amino acid. **\*This is the type of mutation that occurs in Duchenne muscular dystrophy, hemophilia and thalassemia.\***

Original DNA		TAC	GGT	AAT	CAA	ATA	GAA	CCT	GAG	ACT
Nonsense Mutation	_____	_____	_____	_____	_____	_____	_____	_____	_____	_____
Mutated mRNA	_____	_____	_____	_____	_____	_____	_____	_____	_____	_____
Mutated Amino Acids	_____	_____	_____	_____	_____	_____	_____	_____	_____	_____
Original Amino Acid Sequence	Met	Pro	Leu	Val	Tyr	Leu	Gly	Leu	STOP	

**Analysis Questions. Put the answers to these in complete sentences in your notebook.**

1. How many amino acids were affected in the point mutations compared to the frameshift mutations?  
One, Lots, a few.....
2. Describe the difference between a frameshift mutation and a point mutation.
3. Which type of mutation, a frameshift or a point mutation, has more effect on the organism? Why?
4. Usually a frameshift mutation results in the synthesis of a nonfunctional protein. Why do you think mutated proteins might not be functional?
5. Which type of point mutation would be insignificant to the organism? Why?
6. Which type of point mutation would most likely cause major changes to physical structures of the organism?
7. In your notebooks, draw a diagram to shows the relationships between: transcription, translation, DNA, tRNA, mRNA, amino acids, and polypeptide (protein).