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	Mutations Powerpo	oint Guide	
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Mutations

Introduction:

Mutations are caused by changes in DNA. Knowing a few basic types of mutations can help you understand why some mutations have major effects and some may have no effect at all. The following are some of the types of mutations that can occur. There are 4 main types of mutations.

1. Substitution (aka-point mutation)

A **substitution (point)** is a **mutation** that exchanges one **base** for another (i.e., a change in a single "chemical letter" such as switching an A to a G). Such a substitution could:

- Change a codon to one that encodes a different amino acid and cause a small change in the
 protein produced. For example, sickle cell anemia is caused by a substitution in the betahemoglobin gene, which alters a single amino acid in the protein produced. These are called
 missense mutations.
- Change a codon to one that encodes the same amino acid and causes no change in the protein produced. These are called **silent mutations**.
- Change an amino-acid-coding codon to a single "stop" codon and cause an incomplete protein. This can have serious effects since the incomplete **protein** probably won't function. These are called **nonsense mutations**.

CTGGAG CTGGGG

2. Insertion

Insertions are mutations in which 2+ extra nucleotides are inserted into a new place in the DNA.



3. Deletion

Deletions are mutations in which a section of DNA is lost, or deleted (i.e-2+ nucleotides are deleted).

CTAGAC

4. Frameshift

Since protein-coding DNA is divided into **codons** that are 3 bases long, insertions and deletions can alter a gene (DNA) so that its message (codons) is no longer read correctly. These changes are called **frameshifts mutations**.

- For example, consider the sentence, "The fat cat sat."
 - o Each word represents a codon.
 - o If we delete the first letter and read the sentence in the same way (in 3 letter words), it doesn't make sense because the "reading frame" has shifted.
 - Ex: "The fat cat sat" becomes "hef atc ats at" because the 1st "T" is removed and all the letters shift down one place.
 - o In frameshifts, a similar error occurs at the DNA level, causing the codons to be divided up and read incorrectly. This usually generates proteins that are as useless as "hef atc ats at" is uninformative.

The fat cat sat hef atc ats at