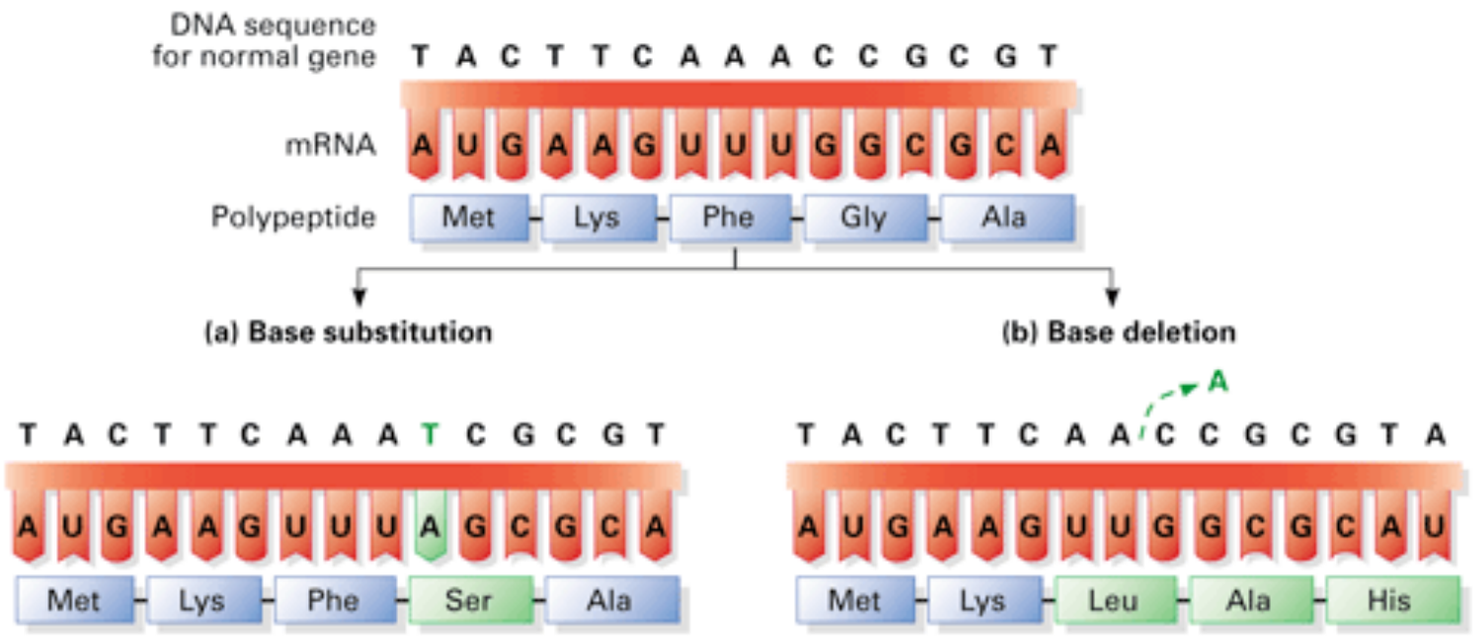
Mutation Notes

Mutations- changes to genetic material.

Two types: **Gene Mutations** and **Chromosomal Mutations**

Gene Mutations (a.k.a. point mutations)- changes to a single gene

Point mutations- changes in one or a few nucleotides at a single point in the DNA sequence.
ex. substitutions, insertions, deletions.



Insertions and deletions are ***Frameshift Mutations*** and tend to be more problematic than substitutions. Substitutions tend to result in one wrong amino acid in the chain. Frameshift Mutations alter the codons read from that point on, therefore the entire sequence of amino acids is incorrect. This results in an altered protein that is unable to perform its normal function.

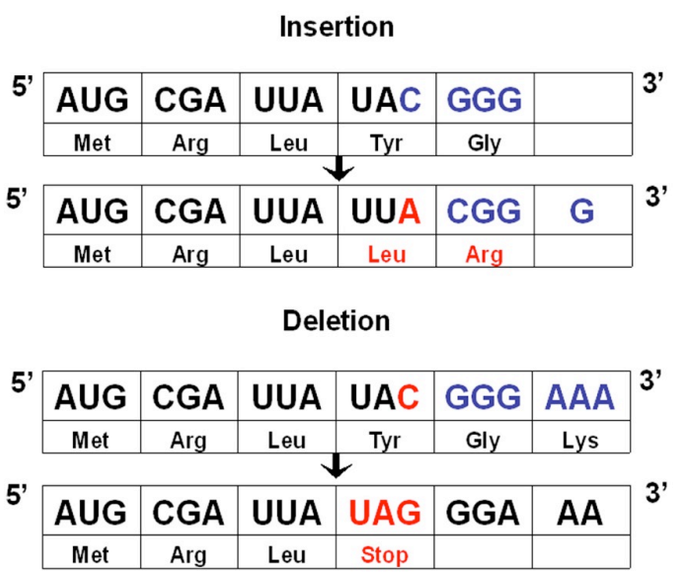


Figure 2. Schematic representation of nucleotide insertion and deletion

Mutations: good or bad?

Many, if not most, mutations are neutral to gene expression.

Mutations that cause dramatic changes in protein structure or gene activity often disrupt normal biological activities.

ex. sickle-cell anemia, cystic fibrosis.

Beneficial mutations produce genetic variation which can be beneficial to populations. New proteins or altered activity can be useful in a changing environment. ex. HIV resistance

Chromosomal Mutations

involve changes in the number or structure of chromosomes.

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