

Gene Mutations



What is a mutation?



- **Mutation:** a permanent change in the DNA sequence of a gene.
- Mutations in a gene's DNA sequence can alter the amino acid sequence of the protein encoded by the gene.
- **Point mutation:** single nucleotide base changes in a gene's DNA sequence.

Point Mutations



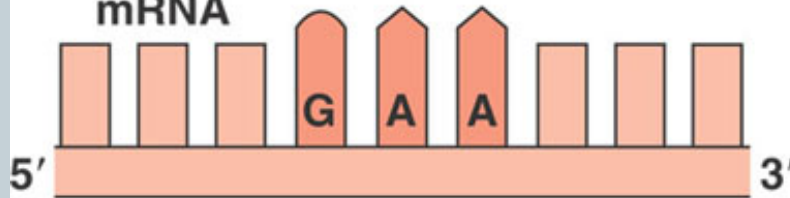
Wild-type hemoglobin DNA



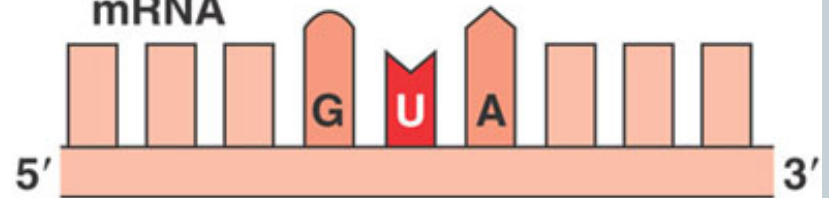
Mutant hemoglobin DNA



mRNA



mRNA



Normal hemoglobin

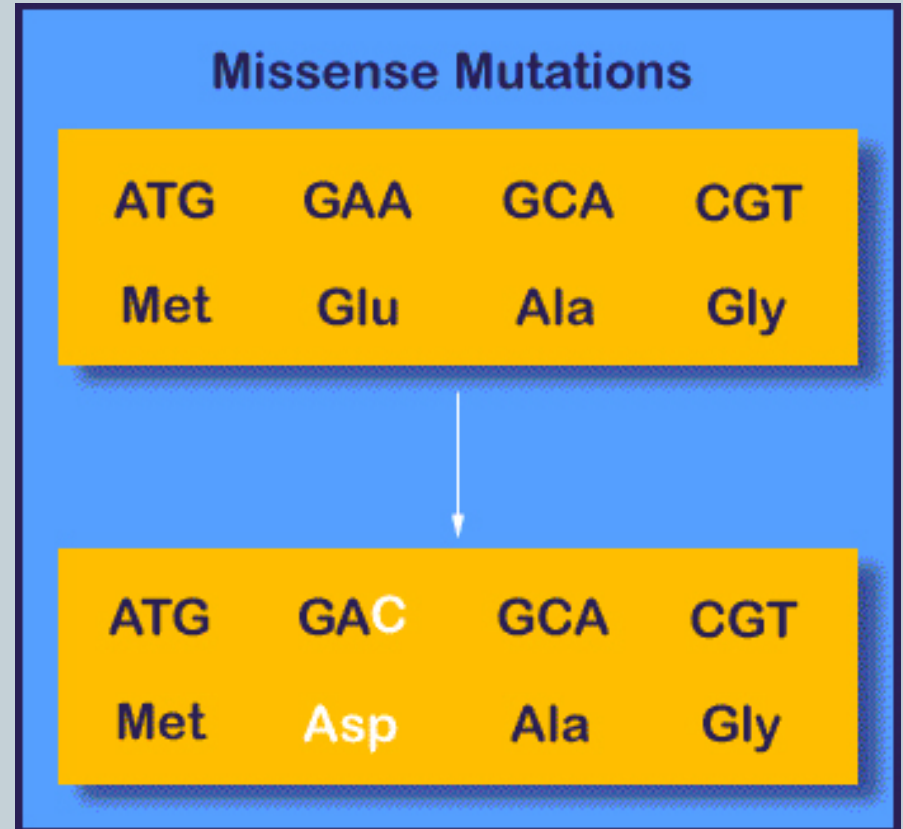


Sickle-cell hemoglobin



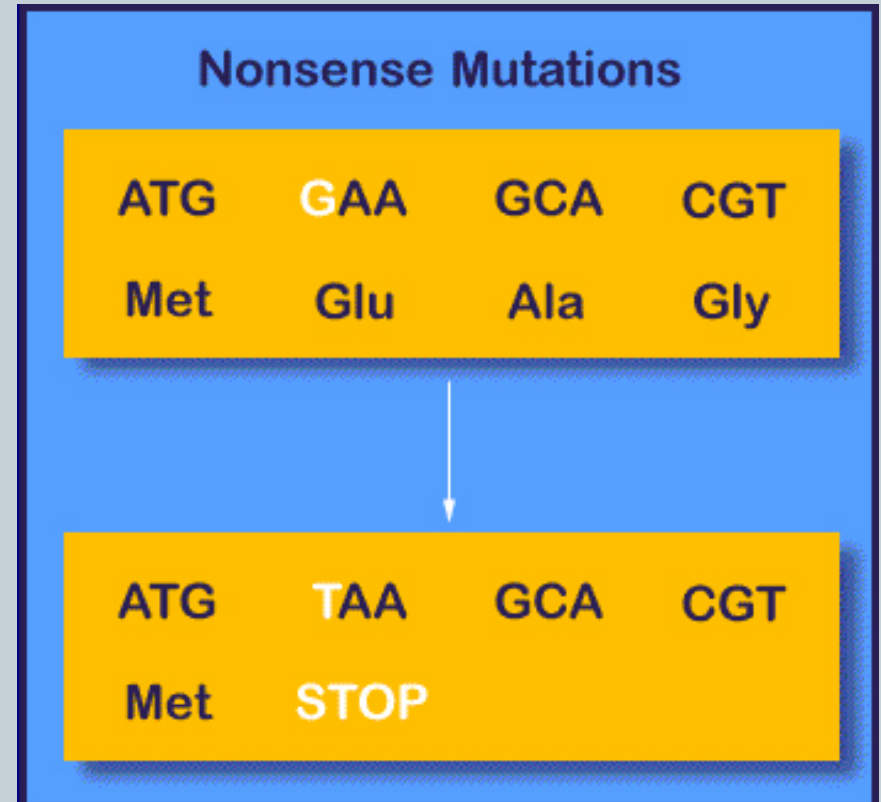
Missense mutations

- Point mutations that result in a single amino acid change within the protein.



Nonsense mutations

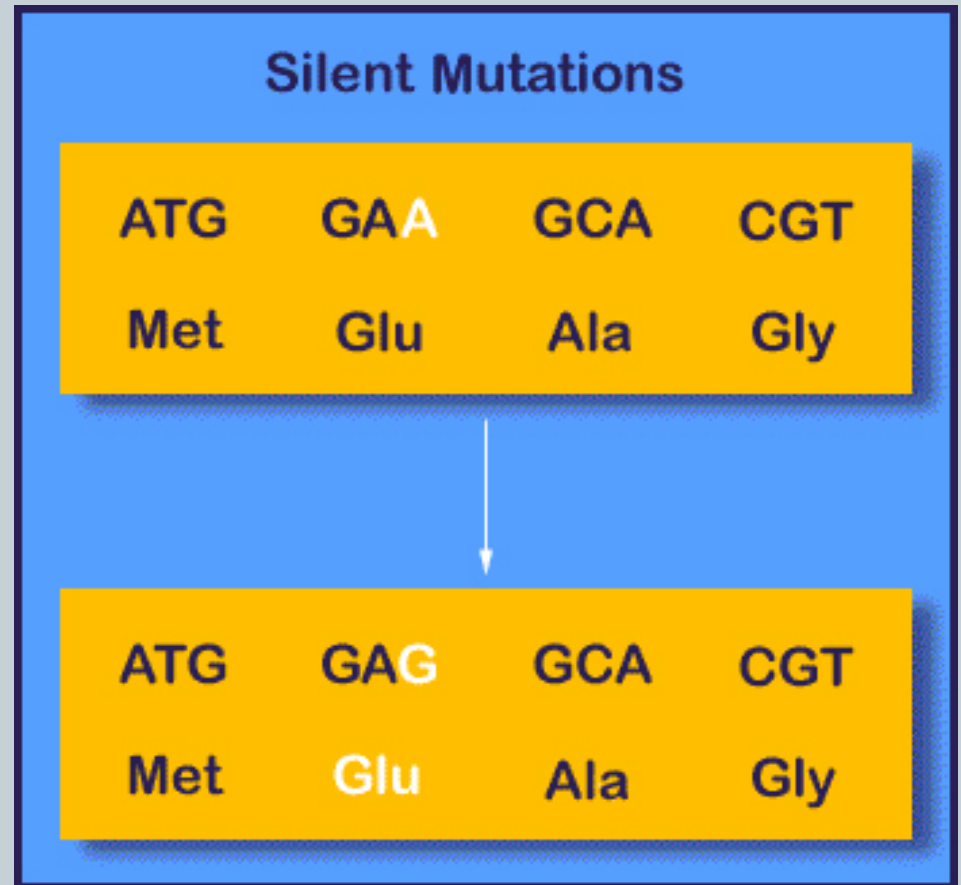
- Point mutations that create a premature "translation stop signal" (or "stop" codon), causing the protein to be shortened.



Silent Mutations



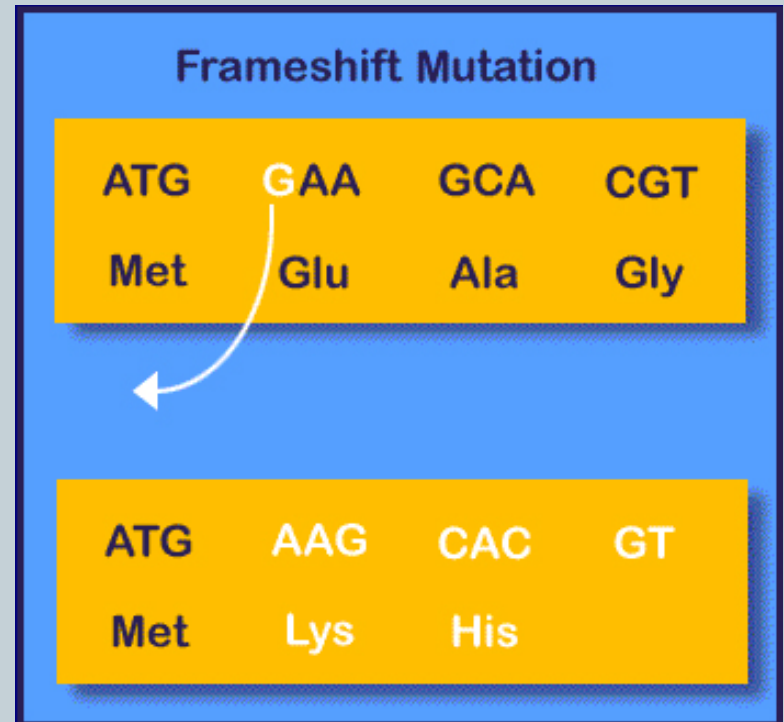
- Point mutations that do not cause amino acid changes within the protein.
- (Recall 64 codons for 20 Amino Acids)



Insertions and Deletions



- Add or remove one or more DNA bases.
- Insertion and deletion cause **frameshift mutations**, which change the grouping of nucleotide bases into codons. This results in a shift of "reading frame" during protein translation.



Mutation Rate



- Calculated based on probability that the DNA will mutate spontaneously during a specific interval
 - Measured as # DNA base pairs changed per unit time
 - Spontaneous mutations = DNA replication errors
- Non-spontaneous mutations:
 - Radiation: **ionizing** (X-Rays) & **Nonionizing** (UV light)
 - Chemicals: **alkylating agents** (methylate DNA), and **carcinogens** – act as alkylating agents.

