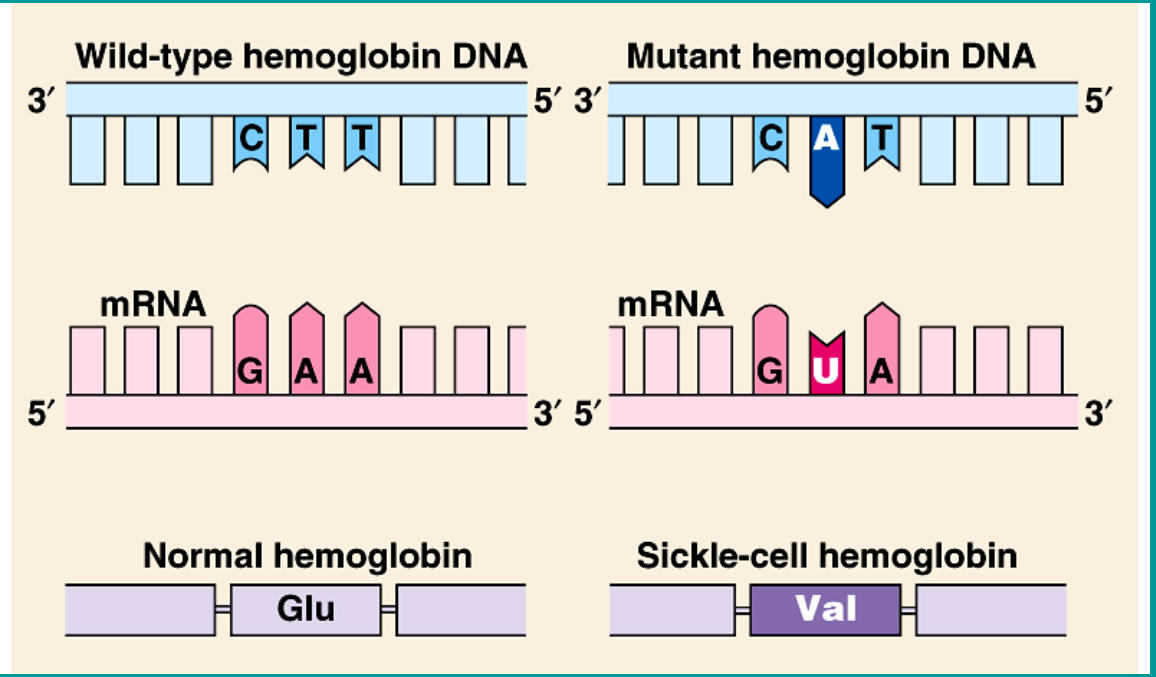
Mutations in DNA

- **Mutations** are changes in the genetic material of a cell (or virus).
- A chemical change in just one base pair of a gene causes a **point mutation**.

- For example, sickle-cell disease is caused by a mutation of a single base pair in the gene that codes for one of the polypeptides of hemoglobin.
  - A change in a single nucleotide from T to A in the DNA template leads to an abnormal protein.

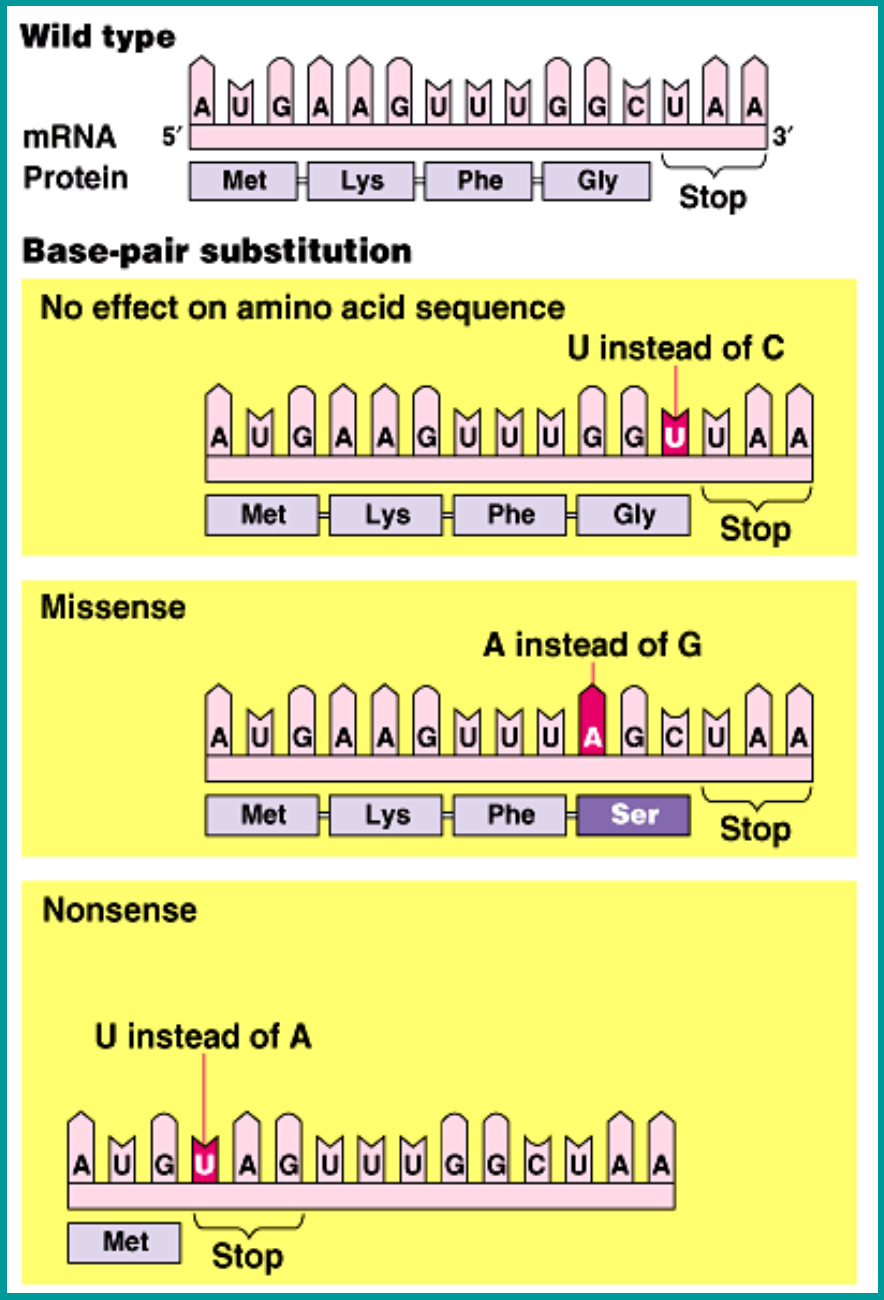


- A point mutation that results in replacement of a pair of complimentary nucleotides with another nucleotide pair is called a **base-pair substitution**.

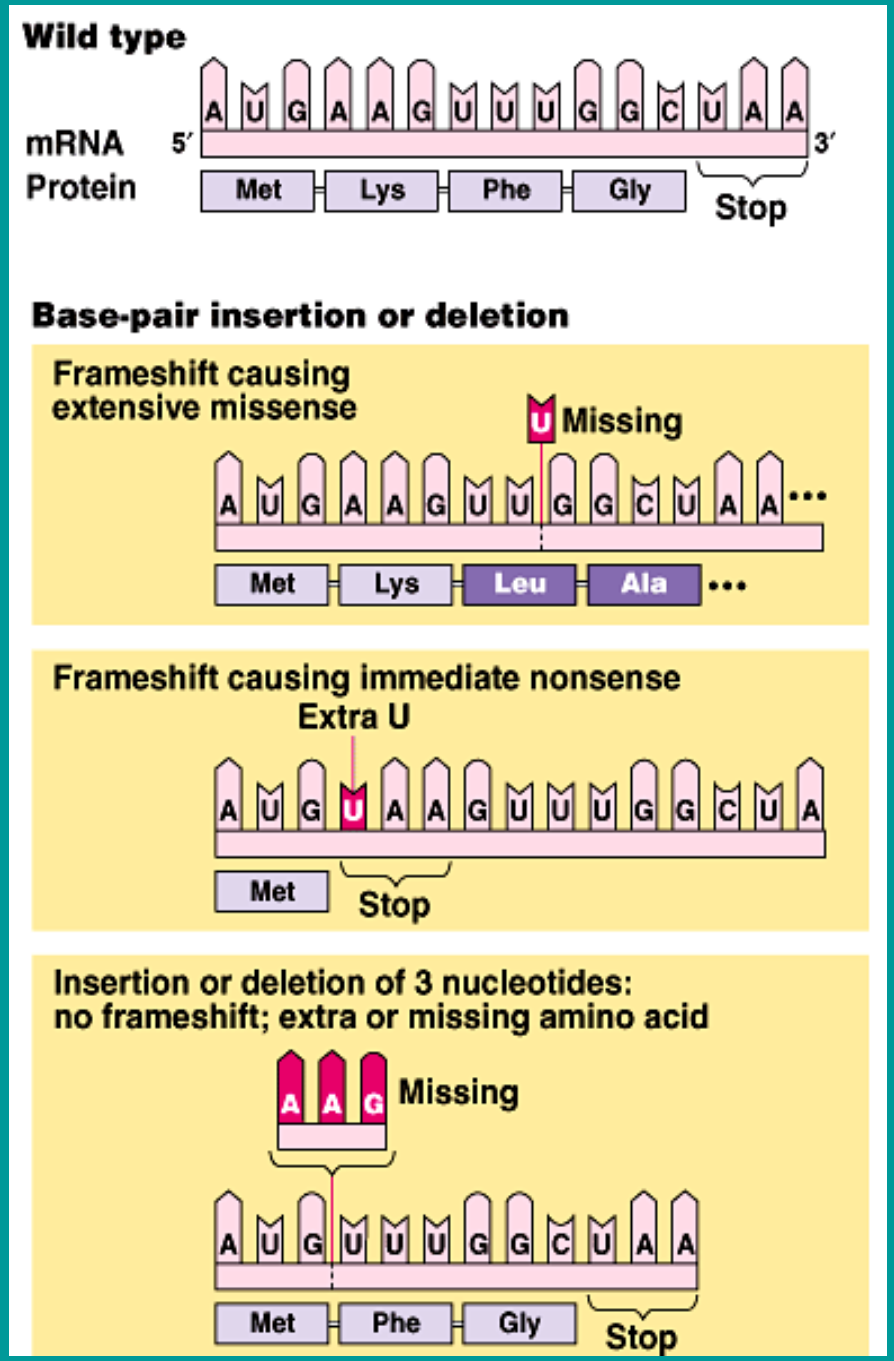
Normal “THE BIG FAT CAT ATE THE WET RAT”

- **Missense mutations** are those that still code for an amino acid but change the indicated amino acid.
- “THE BIZ FAT CAT ATE THE WET RAT”

- **Nonsense mutations** change an amino acid codon into a stop codon, nearly always leading to a nonfunctional protein.
- “THE BIG RAT”


Wild type
mRNA
Protein
5′
3′
A U G A A G U U U G G C U A A
Met Lys Phe Gly
Stop
Base-pair substitution
No effect on amino acid sequence
U instead of C
A U G A A G U U U G G U U A A
Met Lys Phe Gly
Stop
Missense
A instead of G
A U G A A G U U U A G C U A A
Met Lys Phe Ser
Stop
Nonsense
U instead of A
A U G U A G U U U G G C U A A
Met
Stop

- **Insertions** and **deletions** are additions or losses of nucleotide pairs in a gene.
- Unless these mutations occur in multiples of three, they cause a **frameshift mutation**.
- Insertions “THE BIG ZFA TCA TAT ETH EWE TRA”
- Deletions “THF IGF ATC ATA TET HEW ETR AT”


Wild type
mRNA
Protein
5′
A U G A A G U U U G G C U A A
3′
Met
Lys
Phe
Gly
Stop
Base-pair insertion or deletion
Frameshift causing extensive missense
U Missing
A U G A A G U U G G C U A A
Met
Lys
Leu
Ala
Frameshift causing immediate nonsense
Extra U
A U G U A A G U U U G G C U A
Met
Stop
Insertion or deletion of 3 nucleotides: no frameshift; extra or missing amino acid
A A G Missing
A U G U U U G G C U A A
Met
Phe
Gly
Stop

- **Mutagens** are chemical or physical agents that interact with DNA to cause mutations.