* point mutation & frame shift mutation

→ point mutation : Changing single nucleotide base in coding region of mRNA called point mutation.

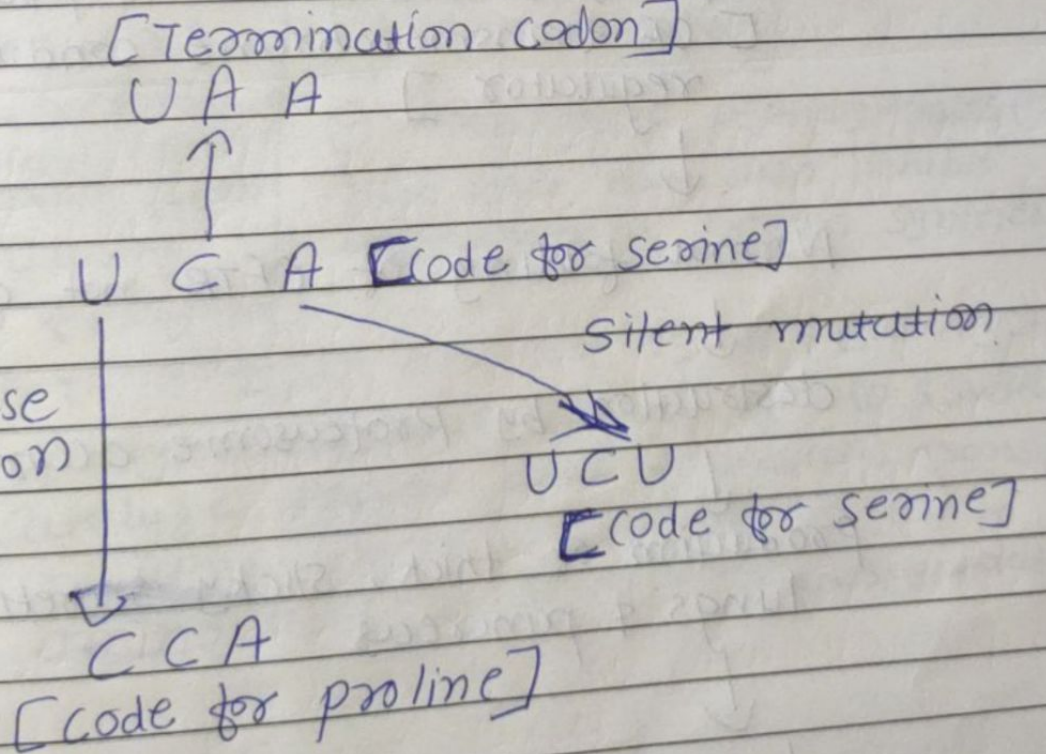
→ It causes following :-

1- Silent mutation
codon containing changed base may code for same amino acid.

2- Missense mutation
codon containing changed base may code for different amino acid.

3- Nonsense mutation
codon containing changed base may become termination codon.
- produce shortened protein

e.g.



→ frameshift mutation :-

If one or two nucleotides either deleted from or added to coding region of mRNA. → causes frameshift mutation

- Alter the reading frame.
- product with radically different amino acid sequence or truncated product due to eventual creation of termination codon.

- If 3 nucleotide are added, a new amino acid is added to peptide.
- If 3 nucleotide are deleted, an amino acid lost

- loss of 3 nucleotide - maintain reading frame
  - Results in serious pathology

e.g. cystic fibrosis (CF)
- chronic, progressive, inherited disease
- Affect pulmonary & digestive system
- loss of phenylalanine in CFTR protein
  [CF transmembrane conductance regulator]

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Normal folding of CFTR not occur

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destruction by proteasome occur.

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production of thick, sticky secretion in lungs & pancreas

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lung damage & digestive deficiency.