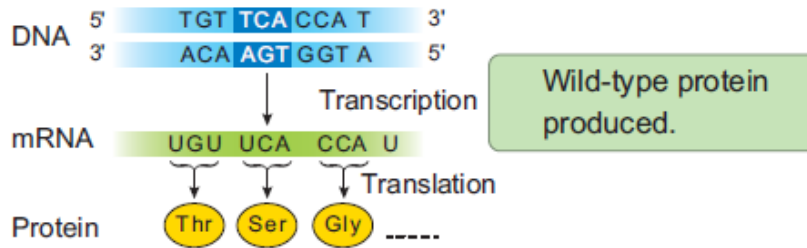# Types of point mutations

Point mutation in DNA are categorised into two main types. They are base pair substitutions and base pair insertions or deletions. Base substitutions are mutations in which there is a change in the DNA such that one base pair is replaced by another. It can be divided into two subtypes: transitions and transversions. Addition or deletion mutations are actually additions or deletions of nucleotide pairs and also called base pair addition or deletions. Collectively, they are termed **indel mutations** (for <u>in</u>sertion-<u>del</u>etion).

Substitution mutations or indel mutations affect translation. Based on these different types of mutations are given below.

The mutation that changes one codon for an amino acid into another codon for that same amino acid are called **Synonymous or silent mutations.** The mutation where the codon for one amino acid is changed into a codon for another amino acid is called **Missense or non-synonymous mutations.**

The mutations where codon for one amino acid is changed into a termination or stop codon is called **Nonsense mutation.** Mutations that result in the addition or deletion of a single base pair of DNA that changes the reading frame for the translation process as a result of which there is complete loss of normal protein structure and function are called **Frameshift mutations.**

DNA
5' TGT TCA CCA T 3'
3' ACA AGT GGT A 5'

Transcription

mRNA UGU UCA CCA U

Translation

Protein Thr Ser Gly -----

Wild-type protein produced.

b) Transition mutation

5' TGT TCG CCA T 3'
3' ACA AGC GGT A 5'

changes a purine nucleotide to another purine or pyrimidine to another pyrimidine

c) Transversion mutation

5' TGT TCT CCA T 3'
3' ACA TGA GGT A 5'
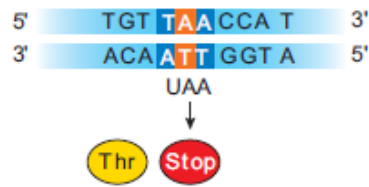
Single purine changed to pyrimidine or vice versa.

d) Missense mutation

5' TGT TTA CCA T 3'
3' ACA AAT GGT A 5'

Thr Leu Gly -----
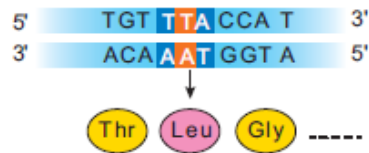
The new codon encodes a different amino acid due to transtion mutation

e) Non-Sense mutation

5' TGT TAA CCA T 3'
3' ACA ATT GGT A 5'

UAA

Thr Stop

The new codon is a stop codon (UAA) due to transition mutation, it leads to premature termination translation.

f) Silent mutation

5' TGT TCG CCA T 3'
3' ACA AGC GGT A 5'

Thr Ser Gly -----
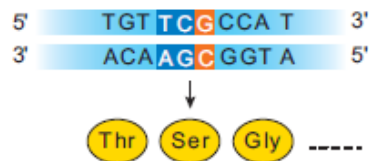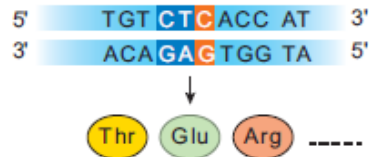
The new codon encodes the same amino acid after transition mutation

g) Frame shift mutation

i) Addition mutation

5' TGT CTC ACC AT 3'
3' ACA GAG TGG TA 5'

Thr Glu Arg -----

Addition or Deletion of one nucleotide, change the reading frame results completely different translation

ii) Deletion mutation

5' TGT CAC CAT 3'
3' ACA GTG GTA 5'

Thr Val Val -----