

Major types of mutations

S.No	Basis of classification	Major types of mutations	Major features
1.	Origin	Spontaneous	Occurs in the absence of known mutagen
		Induced	Occurs in the presence of known mutagen
2.	Cell type	Somatic	Occurs in non-reproductive cells
		Germ-line	Occurs in reproductive cells
3.	Effect on function	Loss-of-function (knockout, null)	Eliminates normal function
		Hypomorphic(leaky)	Reduces normal function
		Hypermorphic	Increases normal function
		Gain-of-function (ectopic expression)	Expressed at incorrect time or inappropriate cells
4.	Molecular change	Nucleotide substitution	A base pair in DNA duplex is replaced with a different base pair
		• Transition	Purine to purine(A→G)or pyrimidine to pyrimidine(T→C)
		• Transversion	Purine to pyrimidine(A→T) or pyrimidine to purine(C→G)
		• Insertion	One or more extra nucleotides are present
		• Deletion	One or more nucleotides are missing
5.	Effect on translation	• Silent (synonymous)	No change in amino acid encoded
		• Missense (non-synonymous)	Change in amino acid encoded
		• Nonsense(termination)	Creates translational termination codon (UAA, UAG, or UGA)
		• Frameshift	Shifts triplet reading of codons out of correct phase