

MUTATIONS

POINT MUTATIONS [BASE SUBSTITUTIONS]

- Missence - one amino acid for another
- Conditional - environmentally dependent (**penetrance**)
- Nonsense - stop codon
- Silent - subtle or not expressed

FRAME SHIFT [SMALL REGIONS]

- Insertion
- Deletion

CHROMOSOMAL [LARGE REGIONS]

- Deficiencies - large deletions
- Translocations - from one chromosome to another
- Inversions - flipped over
- Duplications - hickups

ANEUPLODY [ENTIRE CHROMOSOME]

- Monosomy
- Trisomy