MUTATION

Presentation by

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A heritable change in the structure, content and organization of the genetic material that can be passed down to the next generation is termed **mutation**. Mutation may occur in **one gene** when it is termed **point mutation** or may affect a number of genes on a part of chromosome when it is termed **chromosomal mutation**.

Chromosomal mutation

Involves a number of genes. It is of two types,

(1) Change in number of chromosomes and

(2) Change in structure of chromosomes.

The number of chromosomes in individuals of a species is fixed. For example humans have 2n = 46 chromosomes. But sometimes one or more chromosomes may be lost or added and such a change in number is termed **Aneuploidy** when 2n = 45 or 2n = 47 is found is an individual. Sometimes the whole set of chromosomes may be duplicated so that instead of 2n, an individual way possess 3n or 4n chromosomes. This is **polyploidy**.

CHROMOSOMAL ABERRATIONS

Chromosomal change in structure is also termed as **chromosomal aberration**. It is of four types

1. **Deletion**, in which a piece of a chromosome may be lost.

2. **Inversion**, a piece of a chromosome breaks off and rejoins in the reverse direction.

3. **Duplication** A part of the chromosome may get represented twice and

4. **Translocation** a piece from another chromosome may get attached.

Genes mutation or Point Mutation

A change which affects only one gene is called gene mutation or point mutation. You already know that gene is a segment of DNA and is made of a sequence of nucleotides. Whenever one nucleotide is changed within a gene, it may cause a change in the phenotype.

Gene mutation is of the following types :

1. Transition : When a purine base is replaced by another purine base or a pyrimidine base by another pyrimidine

ATGCATGC → A**G**GC ATGC

2. Transversion : When a purine base is replaced by pyrimidine base and similarly a pyrimidine base by a purine

ATG**C** ATGC → ATG**T** ATGC

3. Frameshift : Sometimes due to loss or gain of one nucleotide the reading frame of the genetic code for an entire protein changes when C gets lost after CAT

CAT ~~C~~AT CAT CAT → CAT ATC ATC ATC

4. Missense : A change in the genetic code due to replacement of a nucleotide (base) may give rise to a different protein e.g. sickle cell haemoglobin.

5. Nonsense : If a genetic code changes such that it becomes a stop codon mid way, no protein is formed e.g.

GAAGAAGAA → GAA**UAA**AA

synthesis stops as UAA in stop codon

6. Silent : When the changed nucleotide does not bring about any phenotypic change because it also codes for same amino acid.

- **Mutagens**

Agents that cause mutation in the genetic material are called **mutagens**. Mutagens belong to two categories

1. Radiations : x-ray, UV rays, α radiations.

2. Chemical : Mustard gas, Actinomycin D