STRUCTURAL CHANGES

It includes changes in number, position, or chemistry of gene within the normal sits of chromosomes of an organism. These are known as Chromosomal Aberations. It is of two types →

(I) Intrachromosomal → Changes within the chromosomes. It is of following types →

a) Deletion → or Deficiency → Here a segment of chromosome is lost.

b) Duplication → A segment of chromosome is repeated or reduplicated.

c) Inversion → A segment of chromosome possessing a sit of genes rotates by 180° on its own axis. It ~~involves two cuts and two~~

(II) Interchromosomal :- These changes are between the chromosomes. i.e. It involves transfer of one segment of one chromosome to the other ~~one~~ chromosome. It is known as Translocation. It is of two types -

a) Reciprocal → Translocation → Both chromosome interchange their parts.

b) Non reciprocal → Translocation → One chromosome give parts to the other chromosome.

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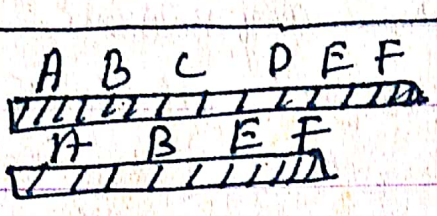
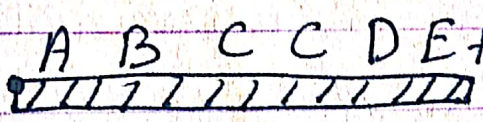
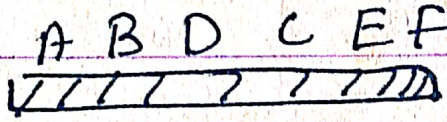
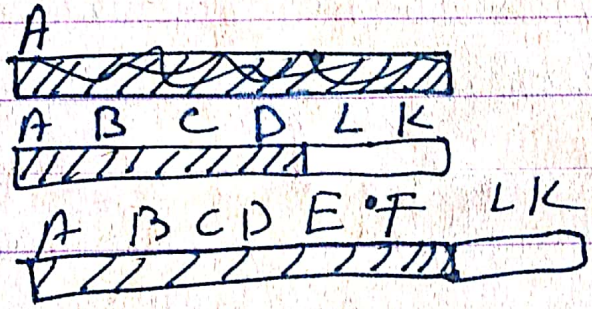
Types of Chromosomal aberrations:

Types	Non-Homologous Chromosomes
1. Deletion	
2. Duplication or Addition	
3. Inversion	
4. Translocation (Reciprocal) (Unilateral)	

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Gene Mutation or Point Mutation or Intragenic Mutation

The permanent alterations in the sequences of nucleotides (bases) in the nucleic acids, (which form the genetic material) There is a change in the normal base ~~se~~ sequence of the DNA molecule. The unit of gene mutation is known as Muton.

Defn:- The permanent alterations in the sequence of nucleotides (bases) in the nucleic acid (which forms the genetic material) leading to change in phenotype is known as Gene Mutation.

The unit of gene mutation is known as Muton. Gene mutation occurs due to mistakes in replication of the DNA, leading to a change in chemical make up of a gene. It occurs due to →

[A] Base pair Analogues &

[B] Tautomerism.

[A] Base-pair-Analogues → There are a number of chemicals which resemble the normal bases. They are known as Base pair analogues e.g (i) 5-Bromo uracil (~~5-BrU~~ (5 BU) or 5-Bromodeoxyuridine (BUdR) is an analogue of Base Thymine. Normally it pairs with Adenine, but it may pair with ~~G~~ Guanine by mistake.

(ii) Similarly 2-Aminopurine (AP) can pair with (T) Thymine or (C) Cytosine.

[B] Tautomerism:- ~~It is~~ The base pairs have an ability to exist in two forms. Normal form is known as Keto forms which show normal pairing. but the unusual form is known as Enol form in which they show abnormal pairing.

Normal → A = T ; C ≡ G.

Abnormal → A – C ; G – T

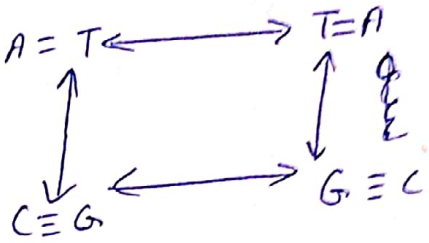
Gene Mutation may be classified as
(A) Substitution Mutation & (B) Frame-Shift Mutation

[A] Substitution Mutation :- It leads to two types of Gene mutations →

(a) Transition → Here purine is replaced by another purine or, Pyrimidine is replaced by another pyrimidine.

A = T ⟶ G ≡ C or C ≡ G ⟶ T = A.

(b) Transversion → Here purine is replaced by pyrimidine or Vice-versa.



These are included as under Substitution or Base pair substitution mutation or Silent mutation.

Inversion → It is another type of substitution mutation in which a segment of DNA is removed and reinserted in a reverse direction.

[B] Frameshift Mutations :- The other main type of Mutation is known as Frameshift Mutations. It includes a shift in the reading frame backward or forward by one or two nucleotides. It includes, ~~Addition~~, Deletion ; Insertion (Addition)

1) Deletion → It is removal of one or a few bases from a nucleotide. Even a single base removal can change the message beyond the point of deletion.

e.g. A T T C A G T C C A G (wild.

Deletion of A

A T T C G T C C A G Message out of frame.

2) Insertion :- It includes addition of one or a few bases, but the number of bases should not be multiple of three.

e.g. A T T G A G T C C A G.

Addition of A

A T T C A A G T C C A G.

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