CHROMOSOMAL ABERRATIONS

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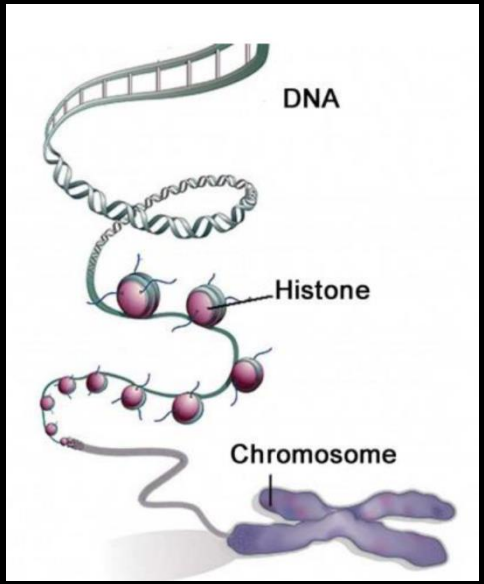
- The arrangement and presence of many genes on a single chromosome provides a change in genetic information not only through change in chromosome number but also by a change in chromosome structure.
- The change in chromosome is due to alteration in genetic material through loss, gain or rearrangement of a particular segment. Such changes are called **chromosomal aberrations**.
- The modification brings about chromosomal mutations.
- Chromosomal mutations are very rare in nature but can be created artificially by 'X' rays, atomic radiation and chemicals, etc.
- The structural changes in chromosomes are due to breaks in chromosome, or in its cell division subunit, i.e., chromatid.



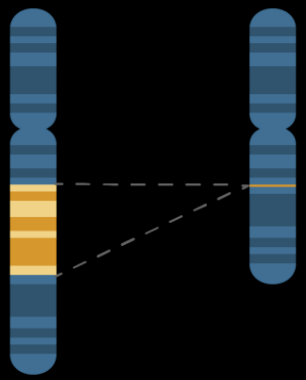
- Each break produces 2 ends which may then follow three different paths.
  - They may reunite, leading to eventual loss of that chromosomal segment which does not contain the centromere.
  - Immediate reunion or reconstitution of the same broken ends may occur, leading to reconstitution of the original structure.
  - One or both ends of one particular break may join those produced by a different break causing an exchange, or non reconstitutional union.

TYPES

- Chromosomal aberrations are of 4 major types
  - Deletion
  - Duplication
  - Translocation and
  - Inversion

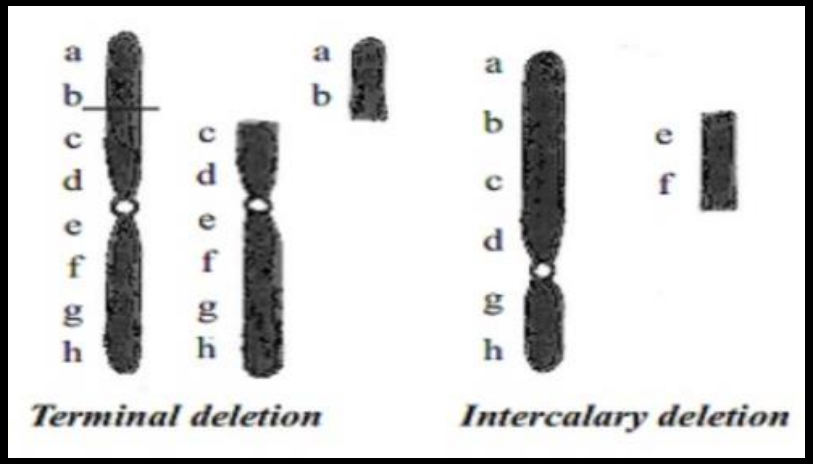
DELETION

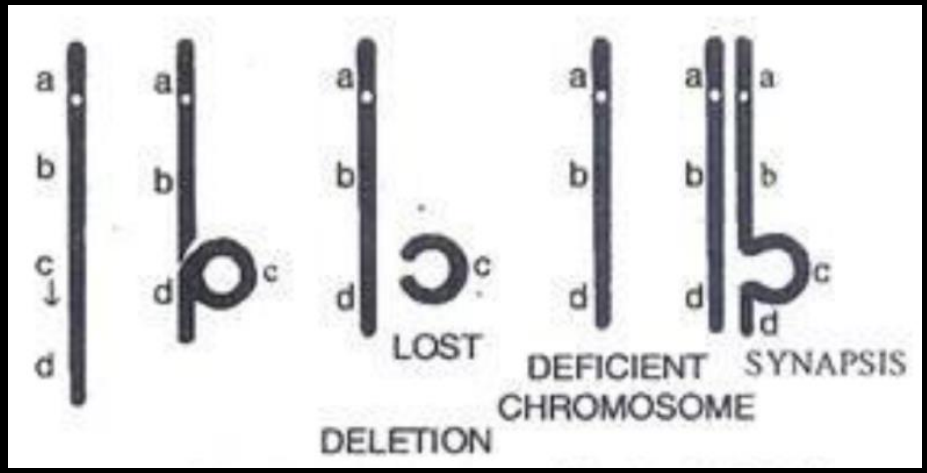
- A missing chromosome segment is referred to either as a deletion or as a deficiency.
- Large deletions can be detected cytologically by studying the banding patterns in stained chromosomes, but small ones cannot.
- In a diploid organism, the deletion of a chromosome segment makes part of the genome hypoploid.
- This hypoploidy may be associated with a phenotypic effect, especially if the deletion is large.



TYPES OF DELETION

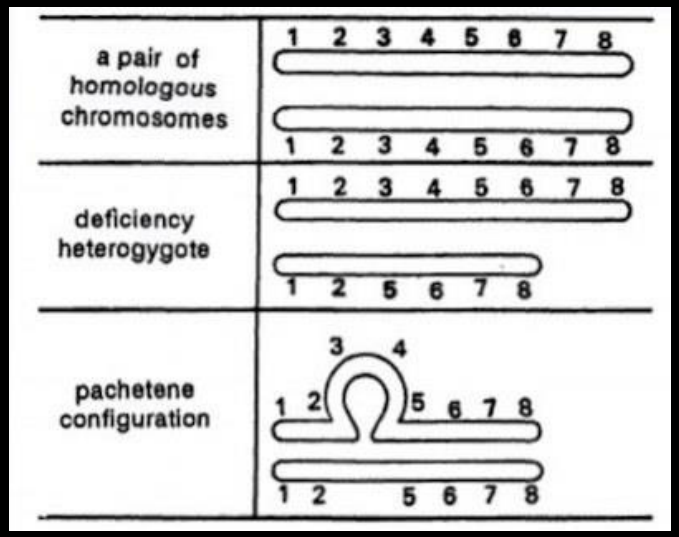
- Two types of deletions are found:
  - Terminal deletion:
    - A single break near the end of the chromosome.
    - Described in maize but otherwise not common.
  - Interstitial deletion:
    - Chromosome breaks and reunites but the part is lost from in between.
    - Deletions are detected at the time of homologous pairing.





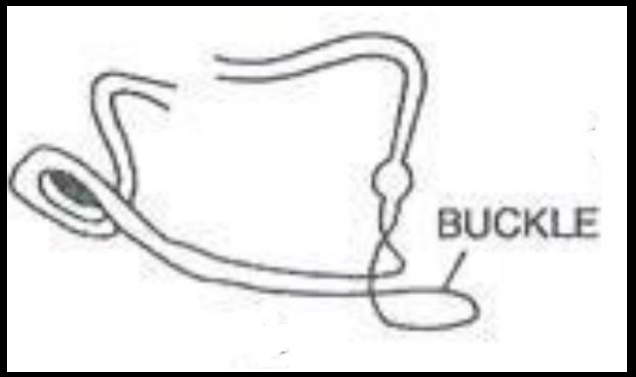
MEIOTIC BEHAVIOUR

- If a part of chromosome is missing then the other chromosome also has to omit it in the form of bulging in order to make synapse.
- If the missing segment is of physiological importance the individual will not survive.
- If dominant gene 'A' is missing the recessive allele 'a' may express itself. It is called pseudo dominance.



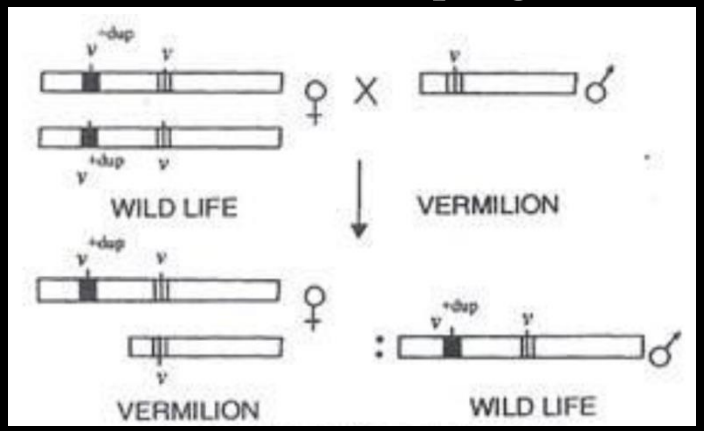
EFFECT OF DELETION

- In human, deletion of chromosome 5 results in cri-du-chat syndrome, children cry like cat, they have small head and are mentally retarded.
- Partial deletion of 18th chromosome results in a syndrome with large ears and long fingers.
- In corn the deficiency is restricted to pollen sterility. The male haploid gametophyte shows deficiency while female of it may receive metabolites from maternal tissue supplementing the deficiency. The omitted segment forms buckles.



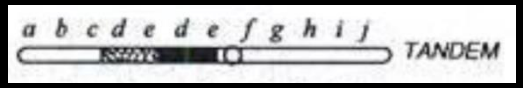
DUPLICATION

- Here a segment of chromosome is repeated twice, i.e., duplicated.
- Duplication was discovered in Drosophila 'X' chromosome for the first time carrying wild type allele for vermilion (v^+) and has been transposed to an 'X' chromosome carrying the mutant vermilion allele (v).
- Bridges found that due to the fact that 'X' chromosome was carrying allele v and v^+ both it was wild type instead of vermilion.
- Equal properties of v and v+ produced wild type effect.
- Such 'duplication females' when crossed with non-duplicated vermilion males all female progeny was vermilion and all male progeny, i.e., y was wild type.



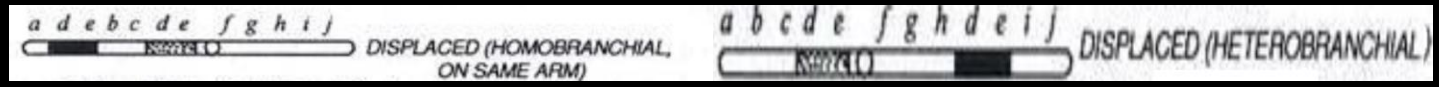
TYPES OF DUPLICATION

- Duplication is of various types.
- Tandem duplication:



  - When the duplicating segment is near the centromeres e.g., the sequence on chromosome is abcdefghi the centromere is present between e and f the segment d e is repeated immediately after its normal position.
- Reverse tandem:

  - When the segment is reversed in duplication, e.g., it is d e segment that is duplicated it will be duplicated as d e e d instead of d e d e.
- Displaced tandem:



  - The segment is repeated somewhere away from its original location but on the same arm (homo-brachial displacement) or on the other arm (hetero-brachial displacement).
- Transposition:

  - When the segment is duplicated on the non homologous chromosome it is called transposition.
- Extra chromosomal:
  - Duplication involves centromere it is called extra chromosomal.

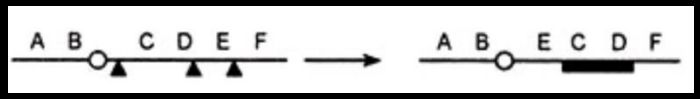
  - In salivary gland chromosome duplications are common either as buckling in the duplication heterozygote or as cross pairing between sections of different chromosomes.

TRANSLOCATION

- Transfer of a section of one chromosome to non homologous chromosome is known as translocation.
- When there is exchange of segments on two non homologous chromosomes it is called reciprocal translocation.
- It also includes exchange of segments between non homologous parts of a pair of chromosomes, e.g., 'X' or 'Y' chromosomes. The segment is neither lost or added it is just exchanged.

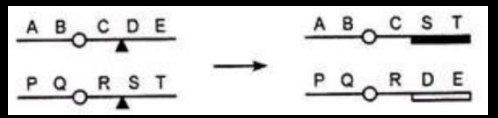
TYPES OF TRANSLOCATION

- Simple translocation: One break
  - A single break in the chromosome and it is transferred onto the end of the other.
- Shift or intercalary translocation: Three breaks
  - Common type of translocation involving 3 breaks so that a two break section of one chromosome is inserted within the break produced in a non homologous chromosome.

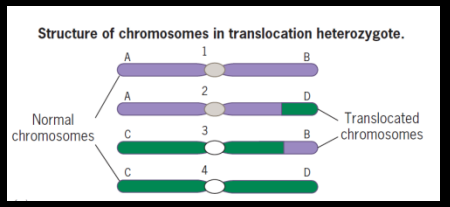


- Reciprocal translocation or interchange:
  - Exchange of segments between two or more non-homologous chromosomes is called reciprocal translocation or interchange.
  - It is of two types: asymmetrical or aneucentric and symmetrical or eucentric.

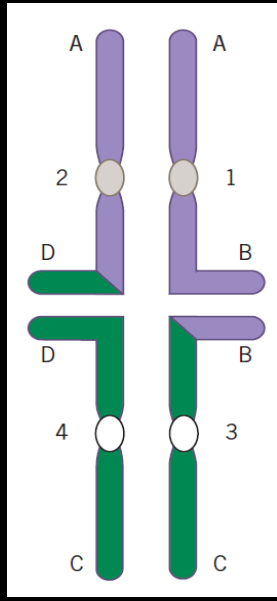
  - Asymmetrical or aneucentric translocation: After breakage, the broken acentric segments fuse to form a trans-located acentric chromosome, while the two chromosomes with centromeres fuse to produce a trans-located chromosome with two centromeres (dicentric). The dicentric chromosome will produce bridge at anaphase if the two centromeres move to opposite poles
  - Symmetrical or eucentric translocation: Broken segments are exchanged between the two non-homologous chromosomes so that both the chromosomes involved in translocation possess only one centromere each (mono-centric)



TYPES OF TRANSLOCATION….

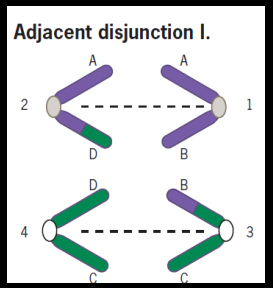


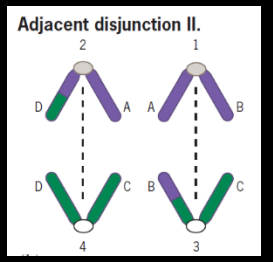
- Reciprocal translocation….
  - Pieces of two non-homologous chromosomes are interchanged without any net loss of genetic material.
  - These chromosomes have interchanged pieces of their right arms.
  - During meiosis, these translocated chromosomes would be expected to pair with their untranslocated homologues in a cruciform, or crosslike, pattern.
  - The two translocated chromosomes face each other opposite the center of the cross, and the two untranslocated chromosomes do likewise; to maximize pairing, the translocated and untranslocated chromosomes alternate with each other, forming the arms of the cross.
  - This pairing configuration is diagnostic of a translocation heterozygote.



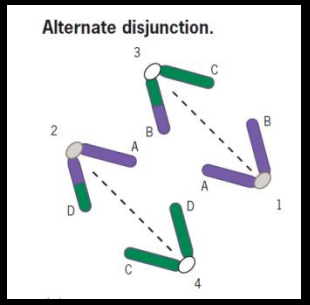
MEIOTIC BEHAVIOUR

- Because cruciform pairing involves four centromeres, which may or may not be coordinately distributed to opposite poles in the first meiotic division, chromosome disjunction in translocation heterozygotes is a somewhat uncertain process, prone to produce aneuploid gametes.
- Altogether there are three possible disjunctional events:
  - Adjacent disjunction I:
    - When the centromeres that move to the same pole are from different chromosomes (that is, they are heterologous), the disjunction is referred to as adjacent I
    - If centromeres 2 and 4 move to the same pole, forcing 1 and 3 to the opposite pole, all the resulting gametes will be aneuploid—because some chromosome segments will be deficient for genes, and others will be duplicated.
  - Adjacent disjunction II:
    - When the centromeres that move to the same pole are from the same chromosome (that is, they are homologous), the disjunction is referred to as adjacent II.
    - If centromeres 1 and 2 move to one pole and 3 and 4 to the other, only aneuploid gametes will be produced).
  - Each of these cases is referred to as adjacent disjunction because centromeres that were next to each other in the cruciform pattern moved to the same pole.





- Alternate disjunction:
  - Centromeres 1 and 4 move to the same pole, forcing 2 and 3 to the opposite pole.
  - This case, called alternate disjunction, produces only euploid gametes, although half of them will carry only translocated chromosomes.

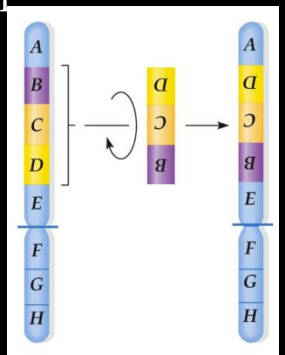


RESULT

- The production of aneuploid gametes by adjacent disjunction explains why translocation heterozygotes have reduced fertility.
- When such gametes fertilize a euploid gamete, the resulting zygote will be genetically unbalanced and therefore will be unlikely to survive.
- In plants, aneuploid gametes are themselves often inviable, especially on the male side, and fewer zygotes are produced.
- Translocation heterozygotes are therefore characterized by low fertility.

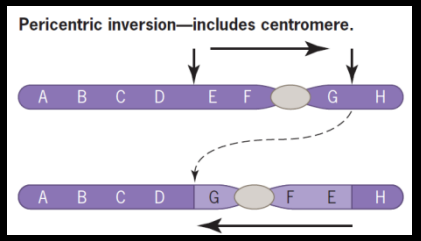
INVERSIONS

- An inversion occurs when a chromosome segment is detached, flipped around 180^0, and reattached to the rest of the chromosome; as a result, the order of the segment's genes is reversed.
- Such rearrangements can be induced in the laboratory by X-irradiation, which breaks chromosomes into pieces.
- Sometimes the pieces reattach, but in the process a segment gets turned around and an inversion occurs.
- There is also evidence that inversions are produced naturally through the activity of transposable elements—DNA sequences capable of moving from one chromosomal position to another. Sometimes, in the course of moving, these elements break a chromosome into pieces and the pieces reattach in an aberrant way, producing an inversion.

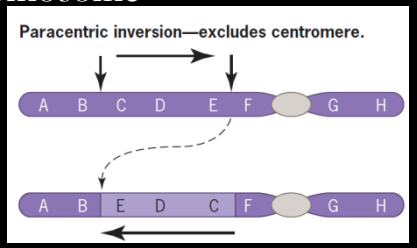


TYPES OF INVERTION

- Cytogeneticists distinguish between two types of inversions based on whether or not the inverted segment includes the chromosome's centromere.



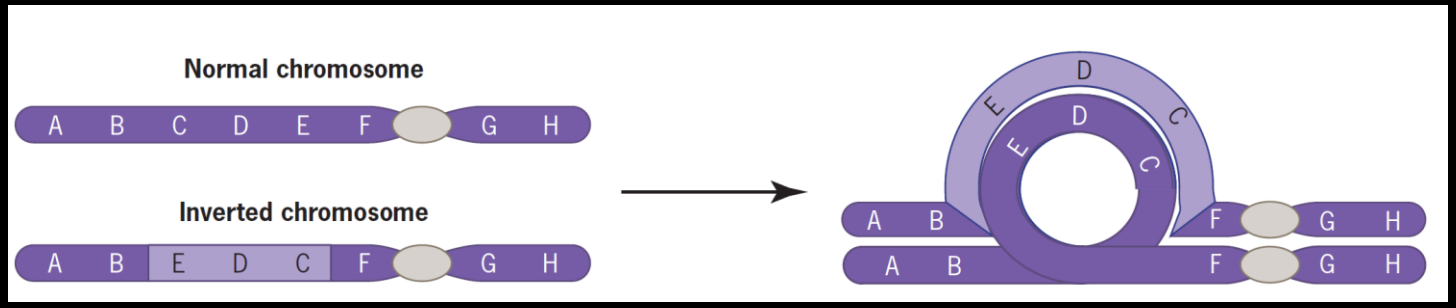
- **Pericentric inversions:**
  - It include the centromere,
  - A pericentric inversion may change the relative lengths of the two arms of the chromosome



- **Paracentric inversions:**
  - do not.
  - A paracentric inversion has no such effect.
- Thus, if an acrocentric chromosome acquires an inversion with a breakpoint in each of the chromosome's arms (that is, a pericentric inversion), it can be transformed into a metacentric chromosome.
- However, if an acrocentric chromosome acquires an inversion in which both of the breaks are in the chromosome's long arm (that is, a paracentric inversion), the morphology of the chromosome will not be changed.
- Hence, with the use of standard cytological methods, pericentric inversions are much easier to detect than paracentric inversions.

MEIOTIC BEHAVIOUR

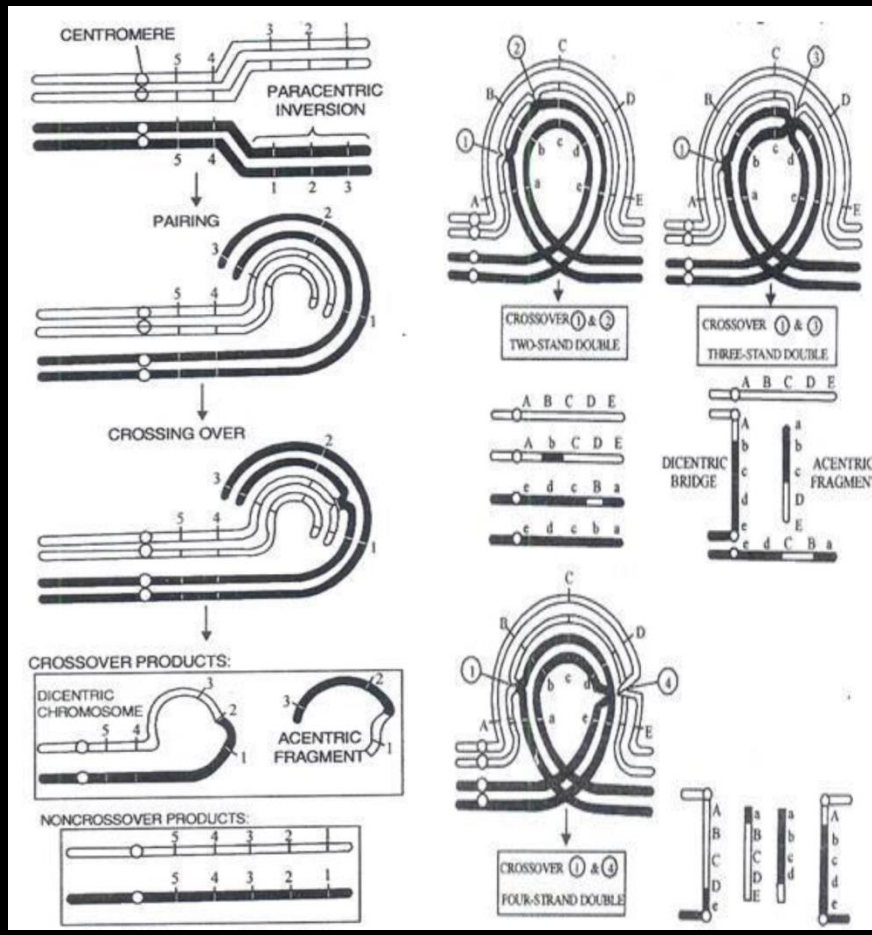
- Inversion heterozygote: An individual in which one chromosome is inverted but its homologue is not.
- During meiosis, the inverted and non-inverted chromosomes pair point-for-point along their length.
- However, because of the inversion, the chromosomes must form a loop to allow for pairing in the region where their genes are in reversed order.
- Breaks may occur at the point of intersection of the loops.
- Reunion of the broken ends takes place in a new combination, and inverts.
- Inversion heterozygotes are formed by loops and bulges in pairs.



Pairing between normal and inverted chromosomes

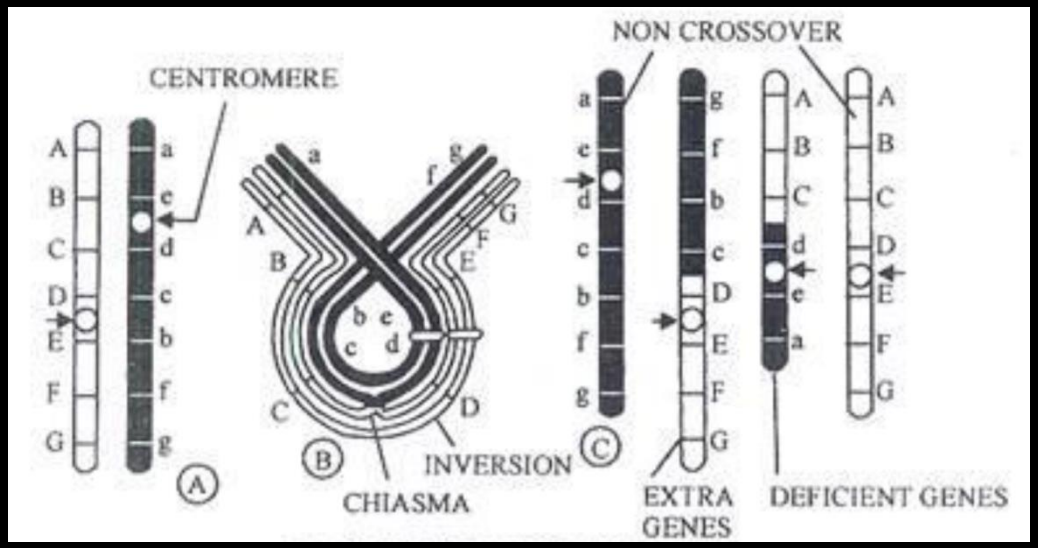
PARACENTRIC

- A single crossing over inverted region will result:
  - Into formation of a dicentric chromosome (with 2 centromeres) and an acentric chromosome (with no centromere).
  - Of the remaining 2 chromatids one will be normal and the other will carry inversion.
  - The dicentric chromatid and acentric chromatid will be observed at anaphase I in the form of a bridge and a fragment.
- Double crossover shows deficiencies and duplication giving rise to variations in anaphase I configurations.



PERICENTRIC INVERSION

- In pericentric inversion centromere is in the inverted segments.
- In pachytene stage 2 of the 4 chromatids resulting after meiosis will have deficiencies and duplications.
- No dicentric bridge or acentric fragment will be observed.
- In pericentric inversion, if two breaks are not situated equidistant from the centromere, a change in shape of chromosome results.
- A metacentric chromosome may become sub-metacentric and vice versa.



Thank You