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Topic: Chromosome aberrations

Chromosome aberrations

A chromosomal aberration, anomaly or mutation is a missing, extra, or irregular portion of chromosomal DNA. It can be from a typical number of chromosomes or a structural abnormality in one or more chromosomes.

Chromosome mutation was formerly used in a strict sense to mean a change in a chromosomal segment, involving more than one gene.

The term "karyotype" refers to the full set of chromosomes from an individual; this can be compared to a "normal" karyotype for the species via genetic testing. A chromosome anomaly may be detected or confirmed in this manner.

Chromosome anomalies usually occur when there is an error in cell division following meiosis or mitosis.

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.

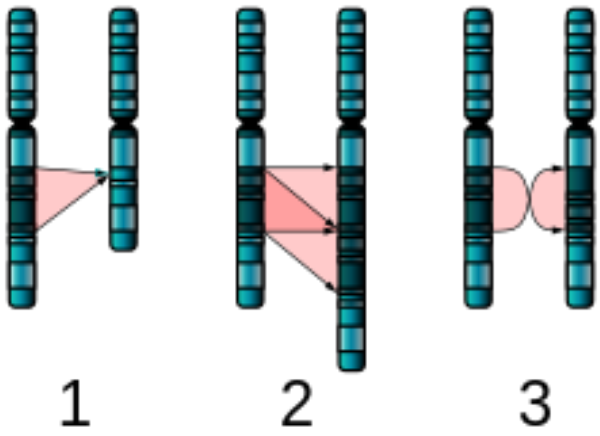
The structural changes in chromosomes are due to breaks in chromosome, or in its cell division subunit, i.e., chromatid.

Each break produces 2 ends which may then follow three different paths:

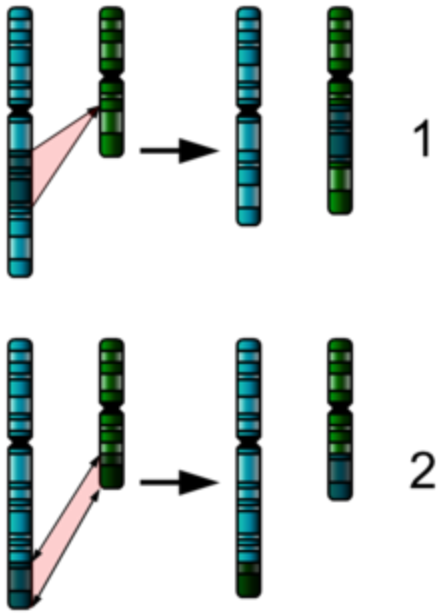
(a) They may reunite, leading to eventual loss of that chromosomal segment which does not contain the centromere.

(b) Immediate reunion or reconstitution of the same broken ends may occur, leading to reconstitution of the original structure.

(c) One or both ends of one particular break may join those produced by a different break causing an exchange, or non reconstitutional union.



The three major single-chromosome mutations: deletion (1), duplication (2) and inversion (3).



The two major two-chromosome mutations: insertion (1) and Translocation (2).

There are many types of chromosome anomalies. They can be organized into two basic groups, numerical and structural anomalies.

Structural abnormalities:

When the chromosome's structure is altered, this can take several forms:

Deletions:

The simplest, but perhaps most damaging, structural change is a deletion—the complete loss of a part of one chromosome. In a haploid cell this is lethal, because part of the essential genome is lost.

However, even in diploid cells deletions are generally lethal or have other serious consequences. In a diploid a heterozygous deletion results in a cell that has one normal chromosome set and another set that contains a truncated chromosome.

Such cells show genomic imbalance, which increases in severity with the size of the deletion. Another potential source of damage is that any recessive, deleterious, or lethal alleles that is in the normal counterpart of the deleted region will be expressed in the phenotype.

In humans, cri-du-chat syndrome is caused by a heterozygous deletion at the tip of the short arm of chromosome 5. Infants are born with this condition as the result of a deletion arising in parental germinal tissues or even in sex cells.

The manifestations of this deletion, in addition to the "cat cry" that gives the syndrome its name, include severe intellectual disability and an abnormally small head.

Duplications:

A heterozygous duplication (an extra copy of some chromosome region) also results in a genomic imbalance with deleterious consequences. Small duplications within a gene can arise spontaneously.

Larger duplications can be caused by crossovers following asymmetrical chromosome pairing or by meiotic irregularities resulting from other types of altered chromosome structures.

If duplication becomes homozygous, it can provide the organism with an opportunity to acquire new genetic functions through mutations within the duplicate copy.

Inversions:

An inversion occurs when a chromosome breaks in two places and the region between the break rotates 180° before rejoining with the two end fragments.

If the inverted segment contains the centromere (i.e., the point where the two chromatids are joined), the inversion is said to be **pericentric;** if not, it is called **paracentric.**

Inversions do not result in a gain or loss of genetic material, and they have deleterious effects only if one of the chromosomal breaks occurs within an essential gene or if the function of a gene is altered by its relocation to a new chromosomal neighbourhood (called the position effect).

However, individuals who are heterozygous for inversions produce aberrant meiotic products along with normal products. The only way uninverted and inverted segments can pair is by forming an inversion loop.

If no crossovers occur in the loop, half of the gametes will be normal and the other half will contain an inverted chromosome.

If a crossover does occur within the loop of a **paracentric inversion**, a chromosome bridge and an acentric chromosome (i.e., a chromosome without a centromere) will be formed, and this will give rise to abnormal meiotic products carrying deletions, which are inviable.

In a **pericentric inversion**, a crossover within the loop does not result in a bridge or an acentric chromosome, but inviable products are produced carrying duplication and a deletion.

Translocations:

If a chromosome break occurs in each of two nonhomologous chromosomes and the two breaks rejoin in a new arrangement, the new segment is called a translocation.

A cell bearing a heterozygous translocation has a full set of genes and will be viable unless one of the breaks causes damage within a gene or if there is a position effect on gene function.

However, once again the pairing properties of the chromosomes at meiosis result in aberrant meiotic products. Specifically, half of the products are deleted for one of the chromosome regions that changed positions and half of the products are duplicated for the other.

These duplications and deletions usually result in inviability, so translocation heterozygotes are generally semisterile ("half-sterile").

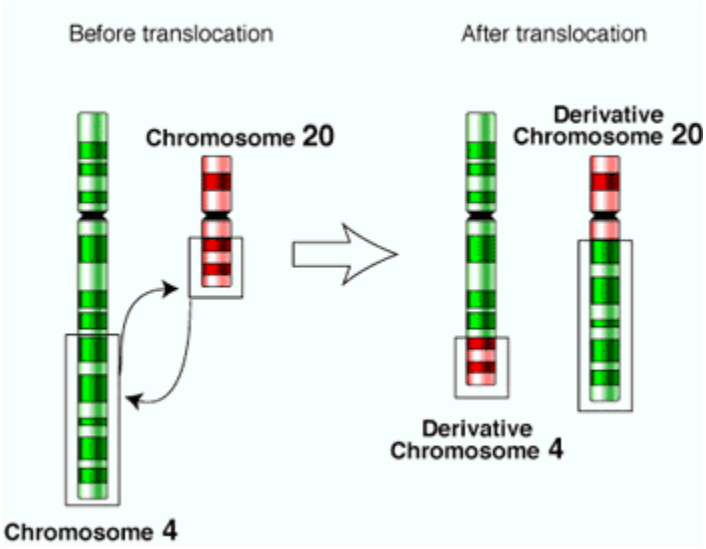


Fig: Translocation

Changes in chromosome number:

Two types of changes in chromosome numbers can be distinguished:

a change in the number of whole chromosome sets (polyploidy) and a change in chromosomes within a set **(aneuploidy**).

Polyploids:

An individual with additional chromosome sets is called a polyploid. Individuals with three sets of chromosomes (triploids, 3*n*) or four sets of chromosomes (tetraploids, 4*n*) are polyploid derivatives of the basic diploid (2*n*) constitution.

Polyploids with odd numbers of sets (e.g., triploids) are sterile, because homologous chromosomes pair only two by two, and the extra chromosome moves randomly to a cell pole, resulting in highly unbalanced, nonfunctional meiotic products.

It is for this reason that triploid watermelons are seedless. However, polyploids with even numbers of chromosome sets can be fertile if orderly two-by-two chromosome pairing occurs.

Though two organisms from closely related species frequently hybridize, the chromosomes of the fusing partners are different enough that the two sets do not pair at meiosis, resulting in sterile offspring.

However, if by chance the number of chromosome sets in the hybrid accidentally duplicates, a pairing partner for each chromosome will be produced, and the hybrid will be fertile. These chromosomally doubled hybrids are called allotetraploids.

Bread wheat, which is hexaploid (6*n*) due to several natural spontaneous hybridizations, is an example of an allotetraploid.

Some polyploid plants are able to produce seeds through an asexual type of reproduction called apomixis; in such cases, all progeny are identical to the parent. Polyploidy does arise spontaneously in humans, but all polyploids either abort in utero or die shortly after birth.

Aneuploids:

Cells have an abnormal number of chromosomes that is not a whole multiple of the haploid number. This condition is called **aneuploidy**. Most aneuploids arise by nondisjunction, a failure of homologous chromosomes to separate at meiosis.

When a gamete of this type is fertilized by a normal gamete, the zygotes formed will have an unequal distribution of chromosomes. Such genomic imbalance results in severe abnormalities or death. Only aneuploids involving small chromosomes tend to survive and even then only with an aberrant phenotype.

The most common form of aneuploidy in humans results in Down syndrome, a suite of specific disorders in individuals possessing an extra chromosome 21 (trisomy 21).

The symptoms of Down syndrome include intellectual disability, severe disorders of internal organs such as the heart and kidneys, up-slanted eyes, an enlarged tongue, and abnormal dermal ridge patterns on the fingers, palms, and soles.

Other forms of aneuploidy in humans result from abnormal numbers of sex chromosomes. Turner syndrome is a condition in which females have only one X chromosome.

Symptoms may include short stature, webbed neck, kidney or heart malformations, underdeveloped sex characteristics, or sterility.

Klinefelter syndrome is a condition in which males have one extra female sex chromosome, resulting in an XXY pattern. (Other, less frequent, chromosomal patterns include XXXY, XXXXY, XXYY, and XXXYY.)

Symptoms of Klinefelter syndrome may include sterility, a tall physique, lack of secondary sex characteristics, breast development ,and learning disabilities.