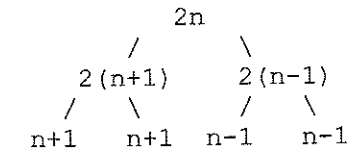
Nondisjunction

Non-disjunction ("not coming apart") is the failure of chromosome pairs to separate properly during meiosis stage 1 or stage 2, specifically in the anaphase. This could arise from a failure of homologous chromosomes to separate in meiosis I, or the failure of sister chromatids to separate during meiosis II or mitosis. The result of this error is a cell with an imbalance of chromosomes. Such a cell is said to be aneuploid. Loss of a single chromosome (2n-1), in which the daughter cell(s) with the defect will have one chromosome missing from one of its pairs, is referred to as a monosomy. Gaining a single chromosome, in which the daughter cell(s) with the defect will have one chromosome in addition to its pairs is referred to as a trisomy.

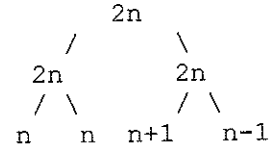
In the event that an aneuploidic gamete is fertilized, a number of syndromes might result. The only known survivable monosomy is Turner syndrome, where the individual is monosomic for the X chromosome. Examples of trisomies include Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), and Patau syndrome (trisomy 13).

The following diagram shows the two possible types of nondisjunction in meiosis:



Schematic of nondisjunction in meiosis I. Duplicated chromosomes in diploid cell (2n).

All gametes are affected by nondisjunction in meiosis I. Two gametes have a single extra chromosome; two gametes are missing a single chromosome.



Schematic of nondisjunction in meiosis II. Duplicated chromosomes in diploid cell (2n).

Half of the gametes are affected by nondisjunction in meiosis II. One gamete has a single extra chromosome; one gamete is missing a single chromosome

Insertion (genetics)

In genetics, an **insertion** (also called an **insertion mutation**) is the addition of one or more nucleotide base pairs into a DNA sequence. This can often happen in microsatellite regions due to the DNA polymerase slipping. Insertions can be anywhere in size from one base pair incorrectly inserted into a DNA sequence to a section of one chromosome inserted into another.

On a chromosome level, an *insertion* refers to the insertion of a larger sequence into a chromosome. This can happen due to unequal crossover during meiosis.

Insertions can be particularly hazardous if they occur in an exon, the amino acid coding region of a gene. A frameshift mutation, an alteration in the normal reading frame of a gene, results if the number of inserted nucleotides is not divisible by three, i.e., the number of nucleotides per codon. Frameshift mutations will alter all the amino acids encoded by the gene following the mutation. Usually, insertions and the subsequent frameshift mutation will cause the active translation of the gene to encounter a premature stop codon, resulting in an end to translation and the production of a truncated protein. These truncated proteins frequently are unable to function properly or at all and can possibly result in any number of genetic disorders depending on the gene in which the insertion occurs.

Chromosomal translocation

In genetics, a **chromosome translocation** is a chromosome abnormality caused by rearrangement of parts between nonhomologous chromosomes. A gene fusion may be created when the translocation joins two otherwise separated genes, the occurrence of which is common in cancer. It is detected on cytogenetics or a karyotype of affected cells. There are two main types, **reciprocal** (also known as non-Robertsonian) and **Robertsonian**. Also, translocations can be **balanced** (in an even exchange of material with no genetic information extra or missing, and ideally full functionality) or **unbalanced** (where the exchange of chromosome material is unequal resulting in extra or missing genes).

Chromosomal inversion

An **inversion** is a chromosome rearrangement in which a segment of a chromosome is reversed end to end. An inversion occurs when a single chromosome undergoes breakage and rearrangement within itself. I

Inversions usually do not cause any abnormalities in carriers as long as the rearrangement is balanced with no extra or missing DNA. However, in individuals which are heterozygous for an inversion, there is an increased production of abnormal chromatids (this occurs when crossing-over occurs within the span of the inversion). This leads to lowered fertility due to production of unbalanced gametes.

The most common inversion seen in humans is on chromosome 9, at inv(9)(p12q13). This inversion is generally considered to have no deleterious or harmful effects, but there is some suspicion it could lead to an increased risk for miscarriage or infertility for some affected individuals.[*citation needed*]

An inversion does not involve a loss of genetic information, but simply rearranges the linear gene sequence.