1. **The maternal and paternal chromosomes from your parents are "independently assorted", meaning that chromosomes from the same source do not have to end up in the same gamete.**

   https://biologydictionary.net/law-of-independent-assortment/

2. **During homologous recombination, the chromosomes can be "cut" at random points after which they are combined with another copy of a homologous chromosome that has been cut at the same point.**

   https://biologydictionary.net/linked-genes/

3. **Although homologous recombination happens numerous times, these linked Genes are inherited together most of the time because the chances that the DNA coding for these two genes is split up are very low. (Gene Map: If the percentage of the offspring having combined parental genes instead of parental genes is very low, e.g. 4%, then the genes sit very close together on the chromosome.)**

   https://biologydictionary.net/linked-genes/

4. **Ramdom fertilization: egg x sperm**

5. **In early development, or in the production of gametes however, the mutations of a gene into different alleles can be passed onto entire organisms, which can then reproduce the allele to its full benefit. Other mutations, known as deleterious mutations, cause a disruption of cellular function.**

   https://biologydictionary.net/allele/

   Substitution mutations: single nucleotide is changed – nonsense(stop coden), missense (different coden) or silent (same amino acid) mutation.

   Insertions and **deletions**: addition or removal of short stretches of nucleotide sequences, they can cause frame shift mutations (among the most deleterious changes to the coding sequence of a protein), altering the entire amino acid sequence downstream of the mutation site.

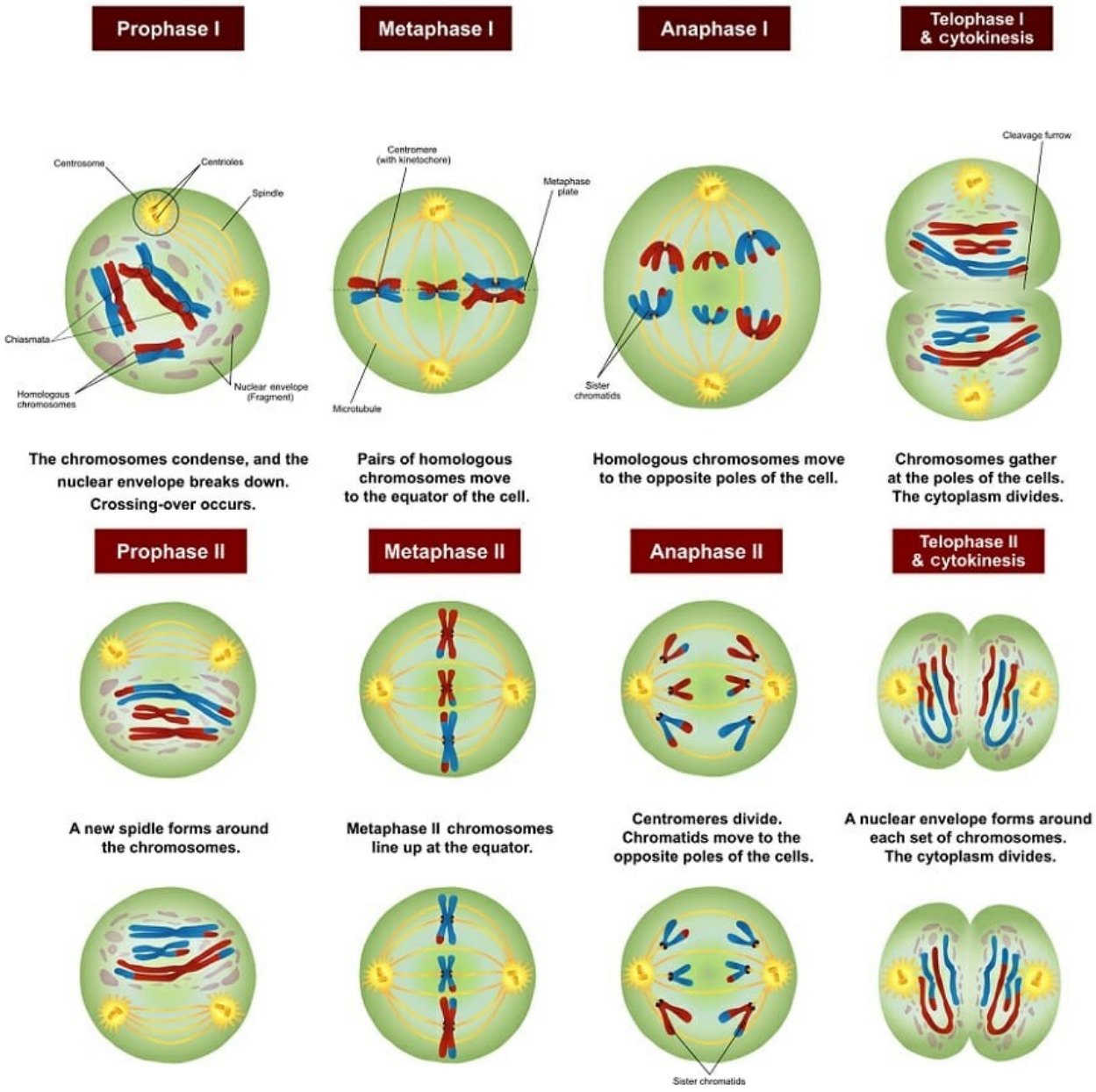
   Amplifications, where segments of genetic material are present **in multiple copies.**

   Translocations : translocated to a different chromosome, **or reinserted** into the same position, but in an **inverted** orientation (what you meant invertion).

   https://biologydictionary.net/mutation/

6. ***Alternative RNA splicing: Constitutive splicing, exon skipping, intron retention, mutually exclusive exons, alternative 5' splice site, alternative 3' splice site and trans splicing.***

   https://biologydictionary.net/alternative-splicing/



Meiosis Stages

Law of Independent Assortment Definition

The Law of Independent Assortment states that different genes and their alleles are inherited independently within sexually reproducing organisms. During meiosis, chromosomes are separated into multiple gametes. Genes linked on a chromosome can rearrange themselves through the process of crossing-over. Therefore, each gene is inherited independently.

The maternal and paternal chromosomes from your parents are "independently assorted", meaning that chromosomes from the same source do not have to end up in the same gamete.

In effect, the Law of Independent Assortment creates a large amount of variety based on different combinations of genes which have not previously occurred.

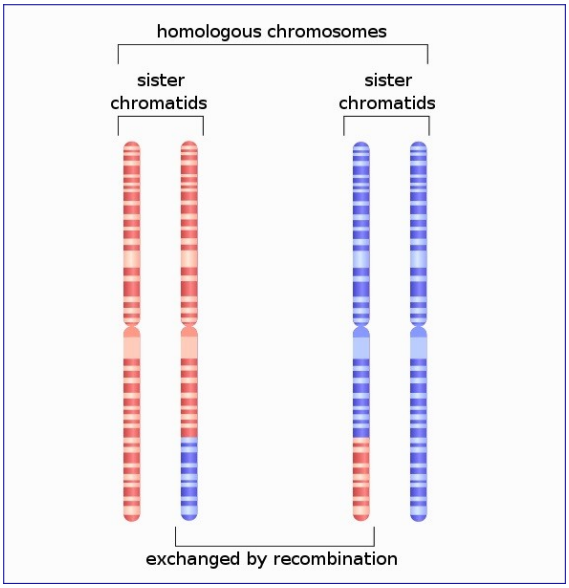
In one instance, genes cannot be assorted completely randomly. This occurs with linked genes, or genes which share the same chromosome. However, the process of crossing-over during meiosis ensures that even these genes get rearranged. During crossing-over, homologous parts of maternal and paternal chromosomes can be exchanged. This ensures that even linked genes get independently assorted.

https://biologydictionary.net/law-of-independent-assortment/

Homologous Recombination

Homologous chromosomes are two pieces of DNA within a diploid organism which carry the same genes, one from each parental source.

Homologous chromosomes take part in a process known as *homologous recombination* during the formation of gametes. This process is also known as "crossing over", because parts of the homologous chromosomes are exchanged when they come into close contact. The chromosomes contain the same genes, which are generally the same length and size. These sections can easily be transferred between chromosomes. The image below shows recombination:



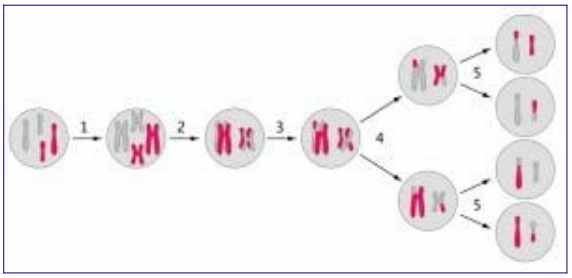
In this image, each chromosome has already been replicated in preparation for meiosis. However, two of the chromatids have exchanged genetic material. This process is extremely important for the creation and maintenance of variety within a population. For instance, if red is the paternal chromosome and blue is maternal, the genes they carry will no longer be linked. Just because your father had blue eyes and black hair does not mean you will automatically inherit these traits. Homologous recombination ensures that traits are randomly mixed together, from both parental sources.

https://biologydictionary.net/homologous-chromosomes/

Homologous Recombination

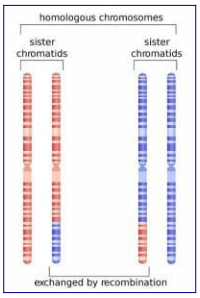
Chromosomes are recombined during meiosis. **During homologous recombination, the chromosomes can be "cut" at random points after which they are combined with another copy of a homologous chromosome that has been cut at the same point.**

The following figure shows how homologous recombination occurs during meiosis:



In this figure, the cell on the left originally has two different copies (gray and pink) of two different chromosomes (large and small). Each copy has the same genes as the other copy but is very likely to have different alleles; that is why they are shown in different colors.

Recombination takes place in the fourth panel of the figure: the DNA of two homologous sister chromatids are cut and rejoined. This step is also illustrated and can be seen more clearly here:



One chunk of the DNA has been swapped between homologous chromosomes, resulting in a new combination of alleles. Importantly, the site at which this DNA cut occurs is mostly random. Furthermore, meiosis occurs every time new gametes are formed; therefore, the cut is in a different location along the DNA, leading to numerous possible combinations.

Recombination of **Linked Genes**

Although homologous recombination happens numerous times, **these two features are inherited together most of the time because the chances that the DNA coding for these two genes is split up are very low**, consequently leading to the genes being inherited together most of the time.

Gene Maps

If two different genes are on two separate chromosomes, then the offspring will inherit the four alleles (two alleles for each gene) in equal percentages: 25% will inherit allele A of chromosome 1 and allele A of chromosome 2, 25% will inherit allele B of chromosome 1 and allele A of chromosome 2, 25% will inherit allele A of chromosome 1 and allele B of chromosome 2, and 25% will inherit allele B of chromosome 1 and allele B of chromosome 2. The way it is usually calculated is in terms of the alleles shared with the parent organisms. In this case, 50% would inherit the parental genes, and the other 50% would inherit a combination of the parental genes.

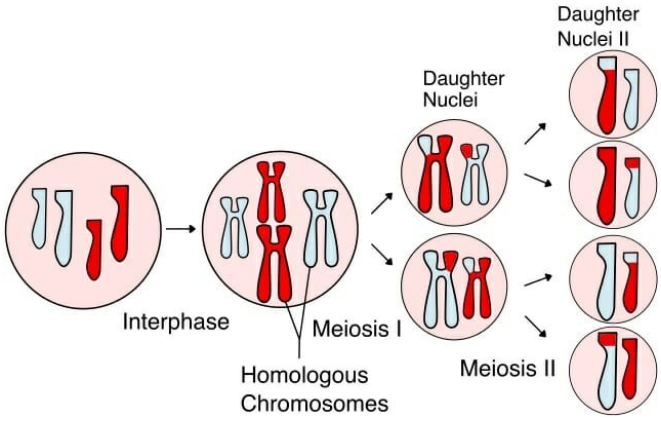
If, on the other hand, the two different genes sit on the same chromosome, then the offspring will inherit the four alleles in different percentages. The percentage of offspring inheriting the parental genes will be larger than 50%, whereas the percentage of offspring inheriting a combination of the parental genes will be lower than 50%. If the percentage is lower but still close to the expected 50% of combined parental genes, then the genes are on the same chromosome but far apart, perhaps one on each side of the chromosome so that it is very likely that the DNA in between them will be cut during recombination. **If the percentage of the offspring having combined parental genes instead of parental genes is very low, e.g. 4%, then the genes sit very close together on the chromosome.**

https://biologydictionary.net/linked-genes/

Functions of Crossing Over

Organisms that divide only asexually without the chance of such recombination suffer from a condition called Muller's Ratchet. That is, each generation of that species contains at least as many genetic mutations as the previous generation, if not more. In other words, when all the progeny are genetically identical to one another, there is no scope for genetic errors to be corrected, or for new and beneficial combinations to arise.

Crossing over increases the variability of a population and prevents the accumulation of deleterious combinations of alleles, while also allowing some parental combinations to be passed on to the offspring. This way, there is a balance between maintaining potentially useful allelic combinations as well as providing the opportunity for variation and change.



Meiosis Overview

https://biologydictionary.net/crossing-over/

Allele Definition

An allele is specific variation of a gene. Bacteria, because they have a single ring of DNA, have one allele per gene per organism. In sexually reproducing organisms, each parent gives an allele for each gene, giving the offspring two alleles per gene. Because alleles are just variants of specific genes, different alleles are found on the same locations on the chromosomes of different individuals. This is important because it gives organisms to be incredibly varied in the functions of their various alleles, while at the same time being able to reproduce. This create variety caused by mutations in specific genes gives rise to a wide number of alleles for any trait in a given population.

Sometimes, the polymerase makes a mistake, and the wrong base pairs are put together. Other enzymes are designed to "check" the DNA after it has been synthesized to find these errors. The enzyme runs along the DNA, checking for bumps that signify two base pairs are not properly bonded. If all these

mechanisms fail to catch the mutation, it will be replicated the next time the cell divides. In bacteria this can give rise to whole colonies that have novel mutations and can be easily studied.

In sexually reproducing organisms, a beneficial mutation is only valuable if it happens early in development or in the production of gametes. A mutation in a single skin cell, for instance, will not be able to help the organism in a large way. The cell may give rise to a few thousand "good" skin cells, but compared to the trillions on your body, they wouldn't matter. **In early development, or in the production of gametes however, the mutations of a gene into different alleles can be passed onto entire organisms, which can then reproduce the allele to its full benefit.**

Other mutations, known as **deleterious mutations, cause a disruption of cellular function.** These mutations cause non-functional alleles to arise, as is the case in cancers. Some cancers are caused by mutations in tumor-suppressing genes, which regulate the size, shape, and growth of individual cells. A non-functional allele at this gene means the cell will continue to grow and divide, regardless of the signals it receives. As part of a functioning body of trillions of cells, this can cause a terrible amount of damage if the cancerous cells are in a sensitive or vital area.

https://biologydictionary.net/allele/

Mutation

These structural changes can be classified as substitutions, deletions, insertions, amplifications, or translocations.

Substitution mutations are situations where a single nucleotide is changed into another.
In highly conserved regions, both in the coding and regulatory stretches of DNA, mutations often lead to **deleterious effects**. Other, more variable stretches are more accommodating. In the promoter region or in other regulatory parts of the genome, a substitution mutation may change gene expression or the response of the gene to stimulus. Within the coding region, a substitution in the third or wobble position of a codon is called a silent mutation since there is no change to the amino acid sequence. When a substitution mutation results in a new amino acid but with similar properties – it is a neutral or a conserved mutation. For instance, if aspartic acid is substituted with glutamic acid, there is a reasonable chance that there would be very few changes to the biochemistry of the protein.
Lastly, the most drastic substitution mutation is one that results in the premature termination of amino acid elongation because of the sudden appearance of a **stop codon** in the middle of the coding sequence. It will likely lead to an extremely short, possibly non-functional protein.

Insertions and deletions refer to the addition or removal of short stretches of nucleotide sequences. These types of mutations are **usually more deleterious** than substitutions since they can cause frame shift mutations, altering the entire amino acid sequence downstream of the mutation site. They can lead to a change in polypeptide length, either creating abnormally long proteins that cause aggregates or truncated polypeptides that are non-functional and can clog the translation machinery of the cell.

Insertions and deletions in the regulatory regions of a polypeptide coding sequence or in genes coding for non-coding RNA are less obviously harmful. Here again, **the position of the mutation matters** – in highly conserved regions, the mutation is more likely to result in negative effects.

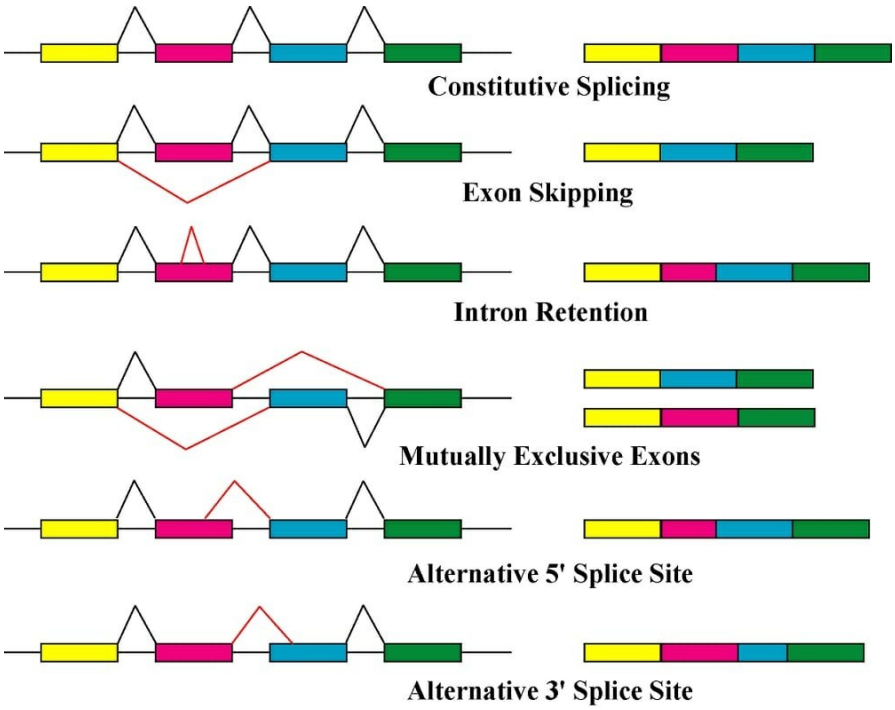
These kinds of mutations include **amplifications**, where segments of genetic material are present **in multiple copies**, and **deletions**, where a large chunk of genetic material is removed. Occasionally, some parts of the genome are translocated to a different chromosome, or **reinserted** into the same position, but in an **inverted orientation**. **Translocations** and deletions can bring together genes that are normally placed far apart from each other, either leading to the formation of mosaic polypeptides, or to the differential regulation of the genes within the segment.

https://biologydictionary.net/mutation/

Alternative Splicing Definition

Alternative splicing is a method cells use to create many proteins from the same strand of DNA. It is also called *alternative RNA splicing*. In regular *DNA translation*, specialized proteins create *messenger RNA* (mRNA) from the DNA template. This mRNA then finds its way to a *ribosome*, where the RNA code is *translated* into the structure of a new protein. In alternative splicing, interactions between different proteins, the cell, and the environment can cause different segments of the original DNA to be omitted from the mRNA. When this happens, the alternate mRNA is translated into an entirely different protein.

Constitutive splicing, exon skipping, intron retention, mutually exclusive exons, alternative 5’ splice site, alternative 3’ splice site and trans splicing.



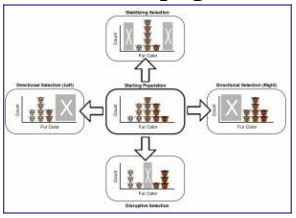
There is another form of alternative splicing, known as ***trans splicing*, in which exons from two different genes get assembled together by a spliceosome**. This genetic process has only been observed in a few single-celled organisms, but could help explain their genetic diversity without sexual reproduction. While sexual reproducing organisms must breed to mix their genetics and produce new varieties, these organisms can do it much faster. This form of alternative splicing can easily create entirely new functions in these organisms, which may prove to be beneficial.

https://biologydictionary.net/alternative-splicing/

Directional selection, stabilizing selection and disruptive selection are three types of natural selection. They are also examples of adaptive evolution. Natural selection is the mechanism of evolution which favors organisms that are better adapted to their environments. Such organisms tend to survive longer and produce more offspring. Selection pressures act against organisms that do not have favorable traits and they are removed from the population. As a result, natural selection plays a major role in the creation of new species over time.

Comparison Chart

	Directional Selection	Stabilizing Selection	Disruptive Selection
Decreases genetic variance in a population	No	Yes	No
Expresses extreme traits	Yes	No	Yes
Number of traits favored	1	1	2
Most common mechanism of natural selection	No	Yes	No
Type of selection mechanism effecting the beak size of Galapagos finches	No	No	Yes



The image above shows the three patterns of natural selection using an allele that determines fur color.

https://biologydictionary.net/directional-selection-stabilizing-directional-disruptive-selection/

Compiled by Dr. rer. nat. Qun Göthel
Bremerhaven, 5th December 2019