# Unit 5. Genetics

Genetics is the study of how hereditary characters are transferred from one individual to another. Let's review some basic concepts which are fundamental for a better understanding of the unit.

## NUCLEIC ACIDS

Nucleic acids are macromolecules composed of monomers called **nucleotides.** (Composed of C, H, O, N and P) There are two types of nucleic acids: **ribonucleic acid (RNA)** and **deoxyribonucleic acid (DNA)**. All living organisms contain both types of nucleic acids. Viruses (which as you should remember are not considered to be living organisms) only contain one type of nucleic acid.

**Nucleotides:** A nucleotide is composed of three parts:

1. A **phosphate group** (P)
2. A pentose (a five-carbon monosaccharide), which can be: **ribose (R)** or **deoxyribose (D).** Nucleotides which contain a ribose are called **ribonucleotides**, and those which contain a deoxyribose are called **deoxyribonucleotides.**
3. A nitrogenous base: **adenine (A), guanine (G), cytosine (C), thymine (T) and uracil (U).** The first three bases are common to both types of nucleotides. In ribonucleotides, the fourth base is always uracil (never thymine); while in deoxyrybonucleotides the fourth base is always thymine (never uracil). The bond between the sugar molecule and the nitrogenous base is called a **nucleoside.**

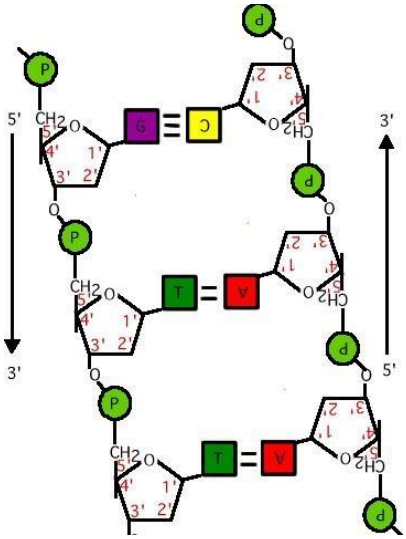
Within cells there are other types of nucleotides which do not make up nucleic acids. Among these types of nucleotides ATP (adenosine-triphosphate) stands out.

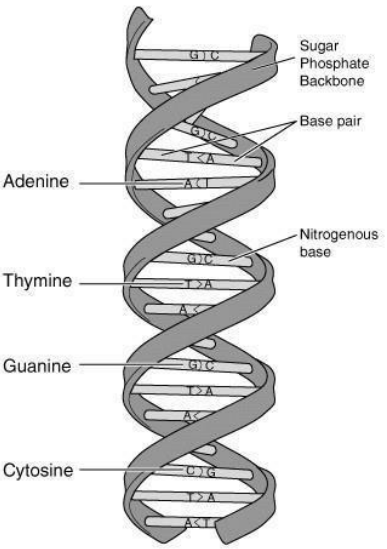
Two nucleotides joined together gives rise to a dinucleotide. Many nucleotides joined together form a **polynucleotide**. Nucleic acids are polynucleotides. RNA is a polynucleotide made of ribonucleotides, whilst DNA is a polynucleotide made of deoxyribonucleotides.

## DNA

Deoxyribonucleic acid is found in the nucleus of eukaryotic cells, and in the cytoplasm of prokaryotic cells (we will see this later on). It consists of two polynucleotide chains or strands, as they are called, that spiral around an imaginary axis to form a double helix (a spiral staircase shape). The two strands of the double helix are **complementary** to

each other. Only certain bases are compatible with each other: Adenine will only fit next to Thymine and Cytosine will only fit next to Guanine. Therefore, if in one of the strands there is an adenine, in the other strand there will be a thymine, and vice versa. In the same way, when in one strand there is cytosine, in the other there will be guanine, and vice versa.




(Scheme taken from [3] http://www.ageds.iastate.edu/meat/images/dna2.gif)

Additionally, the two strands are **anti parallel** to each other; one is inverted with respect to the other. One of them goes from 5' to 3' while the other from 3' to 5'. This double helix structure described by Watson and Crick is very stable due to the hydrogen bonds between the nitrogenous bases: 2 hydrogen bonds between A and T, and 3 hydrogen bonds between G and C.

**Function**: DNA is the most important molecule in living organisms since it contains the genetic material of the organism, which will also be transferred to its descendants. DNA is the main component of chromosomes which are long coils of double-stranded DNA, which contain the genes. A gene is a portion of DNA- of chromosome- responsible for a particular hereditary character (the unit of hereditary information); or, as we will see later on, responsible for protein synthesis. Each gene determines the synthesis of a particular protein.

## RNA

Ribonucleic acid is a single-stranded polynucleotide molecule. There are three types of RNA in living organisms: ribosomal RNA (rRNA), transfer RNA (tRNA) and messenger RNA (mRNA).
**Function**: the function of RNA is to aid in protein synthesis, and therefore in the expression of the genetic information encoded in DNA.

## The genetic information

As we have already said, the genetic information is found in the DNA. But, how? This information is encoded in a particular **sequence** or order of its nucleotides. You could compare it to the order of the letters in a sentence. Every living organism is characterized and defined by a determined order of such nucleotides.

The information encoded in DNA is organized in small units called **genes**. A gene is a portion of DNA that carries the information for a particular character of an individual (e.g. eye colour). Genes are located one after another along the filaments of DNA. Each DNA filament is a **chromosome.** In the nucleus of each and every cell of an organism there are a determined number of chromosomes. Each species of organisms has its own number of chromosomes, which will be the same in all of its cells, with the exception of sex cells or gametes. (Gametes contain half the number of chromosomes than the rest of the cells). For example, we humans have 46 chromosomes inside each of our cells (23 pairs). A chimpanzee has 48 chromosomes (24 pairs) inside each of its cells etc. Therefore, all genes a species contains are found in the chromosomes (the entire set of genes of an organism is its **genome**). In humans, each cell contains about sixty thousand genes (60 000), which are located throughout the 23 pairs of chromosomes. The genetic information of those genes is exactly the same in each cell of the individual. However, not all cells in our body are the same nor do they carry out the same functions. That is because not all of these genes are used by any one cell at any one time. Just a few genes will be 'switched on' in any one cell at any one time and the rest of the genes remain 'switched off'. For example, a nerve cell will express a set of genes that will not be expressed in a muscle cell and vice versa.

## How is the genetic information expressed?

We have said that each gene – portion of DNA- contains the information of a particular character, such as eye color. Now, how can a piece of DNA, with its particular nucleotide sequence, have information to determine that a person will have, for

example green eyes? **The answer lays in proteins**; which are the molecules that carry out cellular functions. What genes really have are chemical instructions for protein synthesis. Each gene determines the synthesis of a particular protein. As we will see later on, the synthesized protein will be responsible for the expression of that particular character, such as the green-colored eyes (refer to protein synthesis).
As we have seen, in order for the genetic information contained in genes (in DNA) to be expressed, such information must be passed down to proteins. Proteins are synthesized in the ribosomes, which are in the cytoplasm of the cell, so the information must somehow get to them. This process, known as protein synthesis, is carried out with the aid of other molecules (mRNA, rRNA and tRNA), and it has **two steps (phases).**

1st **Transcription**: the information contained in the DNA –in the gene- is transferred to a molecule of mRNA (transcription takes place in the nucleus of the cell). The mRNA leaves the nucleus taking the information to the cytoplasm of the cell.

2nd **Translation**: Once in the cytoplasm the mRNA joins with the organelles known as ribosomes, which will translate the information into a protein. There will be a different protein for each gene and each protein will be responsible for a particular character of the individual- green colored eyes-.

This is the first property of DNA,- transcription and translation- and it is the process by which the genetic code is expressed.

Transcription

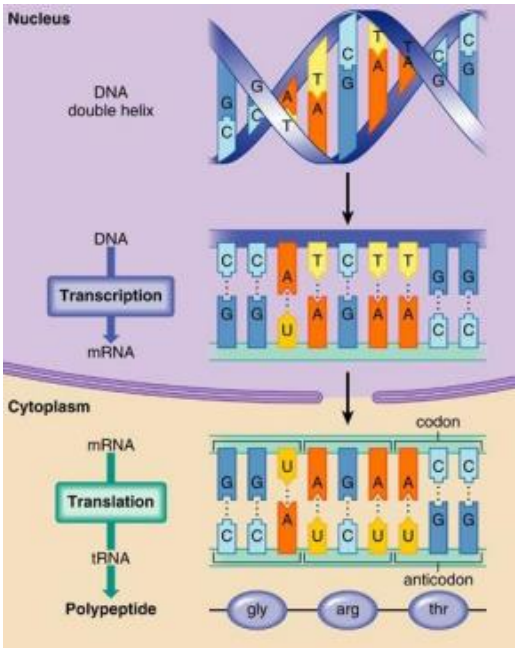
Translation

DNA ⟶ mRNA ⟶ PROTEIN

Replication ↓

2 DNA

Additionally, DNA presents another property only present in living organisms; it can make copies of itself: **replication.** During replication, a double-stranded DNA molecule is copied to produce two identical DNA molecules. Replication is essential; thanks to this property a cell can divide and make two daughter cells that are identical to each other and identical to the mother cell with the same genetic information. Sometimes, when DNA replicates **mutations** or alterations of the information take place. These mutations are responsible for the introduction of new characters in organisms, which with time, will lead to the evolution of the species.



**WHAT IS A GENE?**

A gene is a section of DNA that codes for a specific protein. It is the basic unit of hereditary. Genes hold the information needed to build and maintain an organisms cells. This determines the traits of the organism. These genetic traits can then be passed on to offspring.

**MUTATIONS**

A genetic mutation is a permanent change in the DNA sequence. If it occurs in somatic (normal body) cells then it is not passed on to offspring. However, if a mutation occurs in the DNA of a gamete (egg or sperm) it may be passed on to offspring. Fot the most part, genetic differences between organisms come from some kind of mutation. Every unique gene began as a mutation of an existing gene.

Mutations occur naturally as accidental changes to DNA or chromosomes during the cell cycle. There are enzymes that repair and check DNA during replication but these enzymes are not always 100% effective so changed or damaged DNA can be replicated.

The rate of mutation can be increased by some environmental factors. These factors are called mutagens and include things like UV light, radiation, X rays and some chemicals..

The effect of mutations depends on where and when the mutation occurs. Because of the way DNA is translated, a change in the DNA code may affect just one amino acid in a protein or cause the entire protein to be translated incorrectly… Most mutations have no visible effect on the organism. For mutations where there is an effect the result is usually bad. An incorrect protein works less effectively or doesn't work at all and this decreases the fitness of the organism. In very rare cases a mutation might result in a protein that works better or does something new that provides an advantage to the organism. This is what allows species to evolve and adapt to new conditions over time.

**Types of mutations**

There are many different ways that DNA can be changed, resulting in different types of mutation. Here is a quick summary of a few of these:.

1. Substitution: a mutation that exchanges one base for another. This one is also known as a missense mutation.

2. Insertion: a mutation in which extra base pairs are inserted into a new place in the DNA.

3. Deletion: a mutation in which a section of DNA is lost, or deleted.

**Frameshift**: Since protein-coding DNA is divided into codons three bases long, insertions and deletions can alter a gene so that its message is no longer correctly parsed.

There are other types of mutations as well, but this short list should give you an idea of the possibilities.

## THE NEW GENETIC OR DNA TECHNOLOGY

Since the 70's there have been great advances in the knowledge of nucleotides, as well as some major developments in DNA technology (known as **recombinant DNA technology or genetic engineering**) that allows the manipulation of genetic material. DNA technology has launched an industrial revolution in biotechnology: specific genes can be isolated, large amounts of pure DNA can be obtained, DNA can be spliced at specific locations, genes from one organism can be transferred to other living organisms where it can be replicated and expressed (**transgenic organisms**), the synthesis of artificial genes etc.
The application of DNA technology is of great importance in different fields such as the medical field, in the diagnosis of diseases and a possible cure of such diseases; in the forensic field, identifying dead people or possible criminals; in the pharmaceutical industry, in agriculture etc. The old mysteries of Genetics have now become clearer, but the manipulation of the genetic material opens up new unknowns.

## GENETIC ENGINEERING

What is genetic engineering?
**It is a group of techniques which allow the manipulation of the genome of a living organism.**

Such genetic manipulation basically consists of the following:

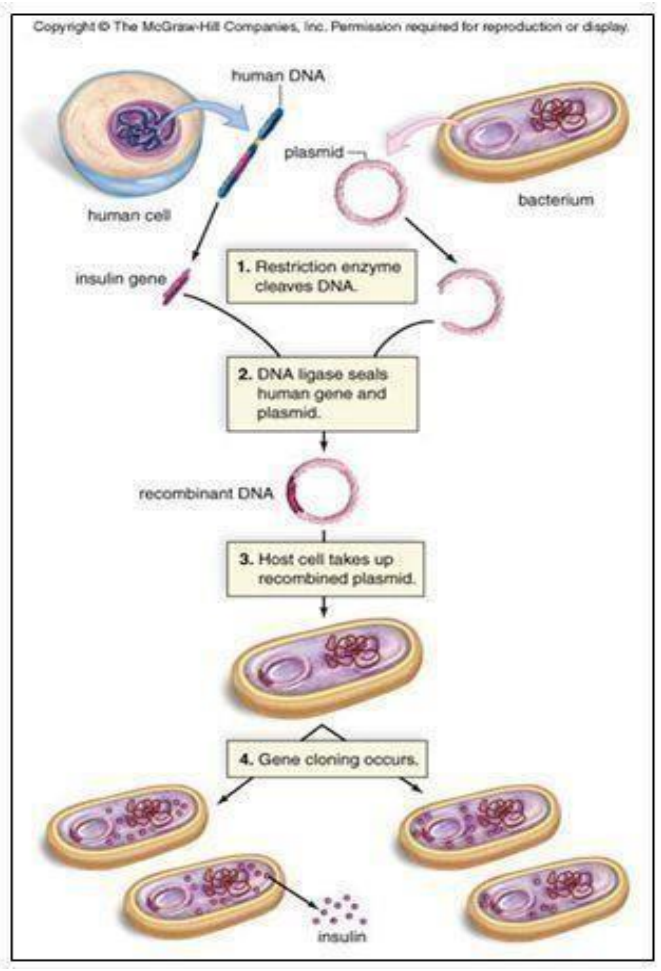
a) Insertion of new genes into a genome (transferring genes).
b) Eliminating existing genes.
c) Modifying the information of a particular gene.

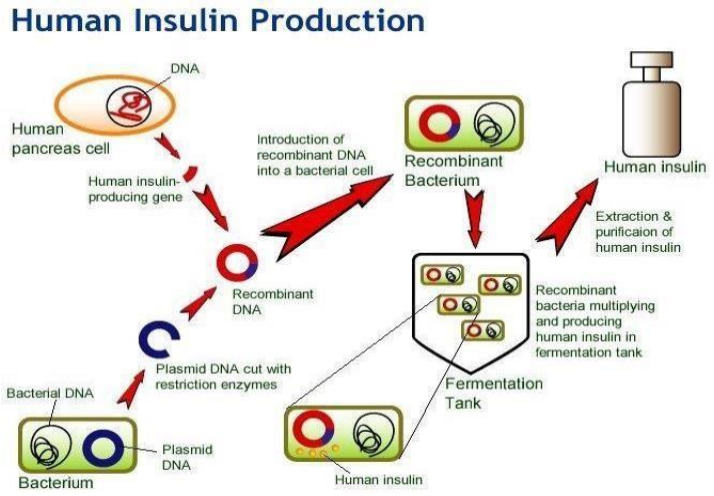
The advances achieved in the field of molecular Genetics, (in reference to the genetic material its replication and transcription), have been the basis for a group of techniques used for manipulating and analysing DNA used in genetic engineering and known as **recombinant DNA technology.**

The use of genetic engineering in research has allowed the production of valuable products by manipulating the genetic material of microorganisms such as: insulin and the growth hormone, interferons, vaccines, enzymes for industrial use, etc. Additionally, manipulation of the genome of some organisms has allowed the development and production of a number of transgenic organisms (animals and plants) for potential agricultural use.

**PRACTICAL APPLICATIONS OF GENETIC ENGINEERING**

1. **Gene therapy**. Genetic engineering has the potential to actually correct some genetic disorders in individuals. In the near future, many therapeutic treatments will not be providing the individual with the molecule that he/she is not able to synthesize (insulin, growth hormone etc), but rather supplement the defective gene with a functional, normal gene, so that the individual can again synthesize its own molecule.
2. **Making antibiotics.** Traditionally specific strains of microorganisms were used for the production of antibiotics. With genetic engineering, the genes responsible for the production of the particular antibiotic are cloned in microorganisms. Modified antibiotics can also be obtained.
3. **Producing mammalian proteins.** Before genetic engineering a number of proteins of medical interests (insulin, growth hormone, several proteins of the immune system – such as molecules called interferons) were directly obtained from tissues. Nowadays, these are made by recombinant DNA procedures. The genes in charge of a particular protein are cloned in microorganisms for use in treating human patients E.g. Human insulin is produced in bacteria or yeast – *Saccharomyces cerevisiae.*



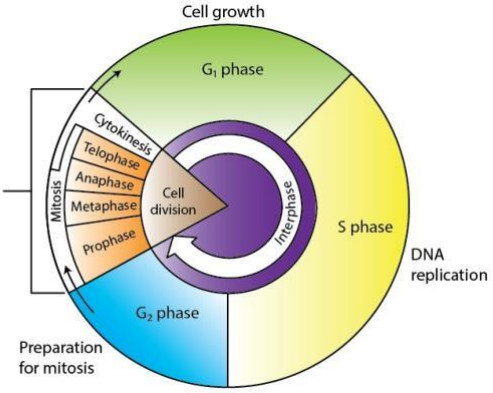



4. **Vaccines**. Traditional vaccines for viral diseases are of 2 types: particles of a virulent virus that have been inactivated by a chemical or physical means, and active virus particles of an attenuated (non-pathogenic) viral strain. In both cases some risks are taken as not all the micoorganism is completely inactivated. With genetic engineering, since most **antigens** are proteins, the genes of interest are cloned. (An antigen is a foreign substance that triggers an immune response; some antigens include molecules of viruses, bacteria, fungi, etc). Recombinant DNA techniques can generate large amounts of a specific protein molecule from the protein coat of a particular disease- causing virus, bacterium or other microbe. This avoids the risks mentioned earlier.

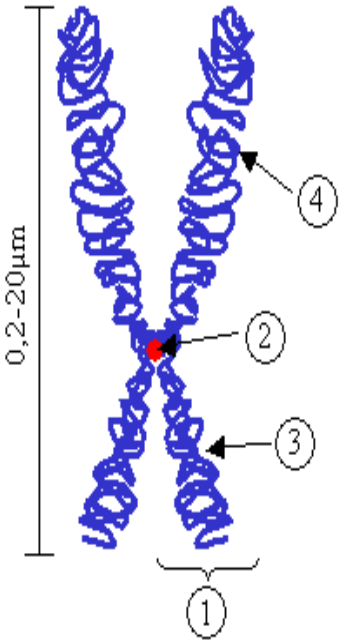
5. **Production of transgenic organisms (animal and plants).** It is possible to produce living organisms which genome has been artificially modified by the insertion of genes. Transgenic animals are produced by injecting foreign DNA into the nuclei of egg cells or early embryos. And transgenic plants are usually obtained by using DNA vectors to move genes from one organism to another in cell cultures.

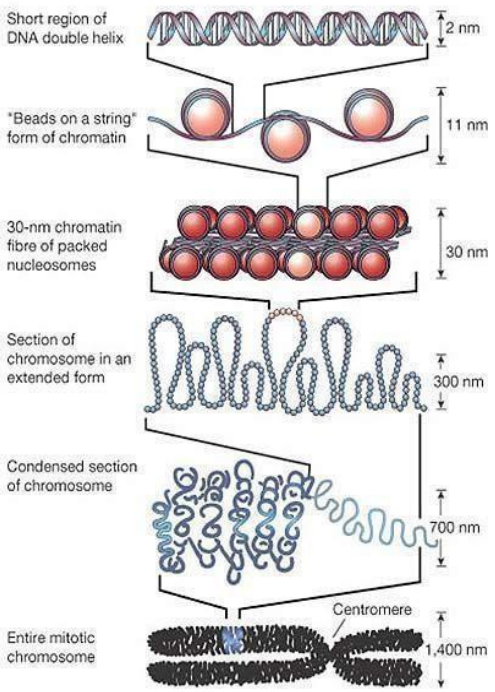
## Chromosomes and the cell cycle

The cell cycle is the period that goes from the moment the cell has just been formed (from the division of the mother cell into two), to the point where the same cell divides in two daughter cells. The period when the cell is not dividing is called **interphase**. Therefore, the cell cycle consists of two successive **mitotic divisions** alternated with a much longer **interphase** or growth period. Interphase consists of three periods of growth, the



first period is called **G1 (gap 1)**. This is when the cell grows in size and organelles are being duplicated. The cell starts performing its functions. This is followed by the **S (synthesis)** phase, during which the chromosomes duplicate. Then during the **G2 (gap 2)** phase the cell starts to prepare for Mitosis. Mitosis divides the nucleus and its chromosomes, and cytokinesis divides the cytoplasm, producing two daughter cells.




(Scheme taken from http://bhs.smuhsd.org/bhsnew/academicprog/science/vaughn/Student%20Projects/P aul%20&%20Marcus/cycle.jpg)

**Figure 1:** Chromosome. (1) Chromatid. One of the two identical parts of the chromosome after S phase. (2) Centromere. The point where the two chromatids touch, and where the microtubules attach. (3) Short arm. (4) Long arm.

(Schemes taken from http://www.biocrawler.com/encyclopedia/Chromosome and http://home.planet.nl/~gkorthof/images/chromosome_structure.jpg

During the interphase the long filaments of DNA-protein complex called **chromatin** are spread through the nucleus. The proteins associated with the DNA are called **histones.** During all three phases of the interphase the chromatin is transcribing itself (therefore synthesizing RNA, mRNA, tRNA and rRNA). It is an active period. However, when the

cell is getting ready to divide, the chromatin condenses into compactly folded and coiled **chromosomes**. These chromosomes now do not transcribe. The cell copies its entire genome by duplicating its chromosomes during the S phase, after replication each chromosome consists of two **sister chromatids** (each a collection of the same genes present in single copy prior to replication). The sister chromatids are held together by a structure called the **centromere**. The end parts of a chromosome are called the **telomeres**.

Once cellular division has come to an end, each daughter cell has one sister chromatid from every chromosome (see mitosis later on) so both daughter cells carry identical information. As the interphase starts in each of the new daughter cells, chromosomes start to uncoil going back to the thin fiber chromatin. The chromatin is so thin it can not be seen very well. It starts again to transcribe, then during the S phase it replicates to be ready for the next mitotic division.

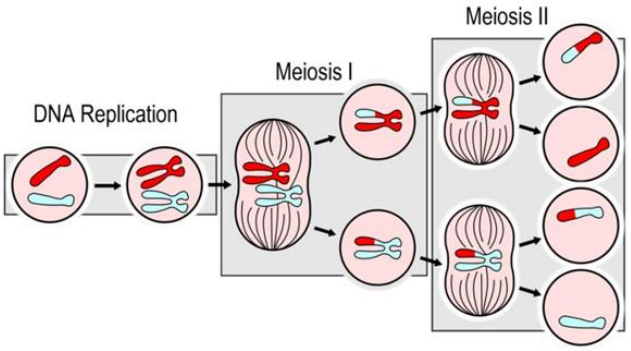
The majority of the cells of an organism are **diploid (2n)**; they have two sets of chromosomes. Chromosomes go in pairs (**homologous**), of the same size and shape, and have the same information for the same characters but they are not identical. In a particular place of a chromosome (the same place for both of the pair) we will find the information for hair colour, but in one chromosome we could have the allele for black hair and on the homologous chromosome the allele for blond hair. Each homologous chromosome comes from one of the parents. To summarize, we can say that a diploid cell has **2n chromosomes**, where n indicates the number of one set of chromosomes. This number is characteristic of each species.

Let's use humans as an example. In all normal body cells (somatic cells) we have 2n chromosomes as these cells are diploid (2n is 46, therefore, n is 23). However, the number of chromosomes in a **gamete** – sperm and ovum- is the **haploid number n.** Cells that are haploid only have one set of chromosomes. When fertilization takes place during sexual reproduction, two haploid gametes fuse together to restore the diploid number in the zygote (46 in humans).

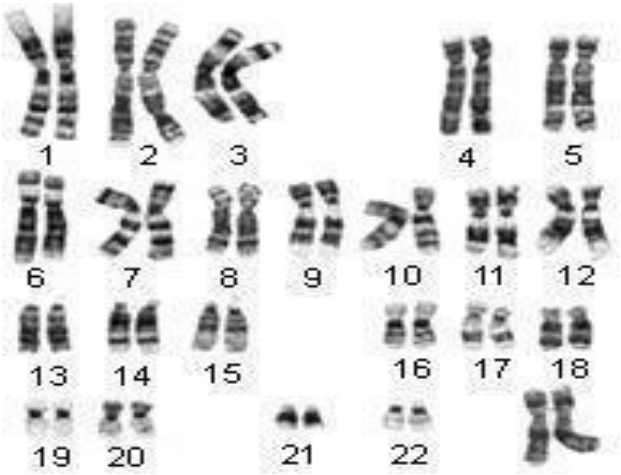
When normal body cells – somatic cells- divide, they do it through a process known as **mitosis**. First of all, cells will replicate their DNA, their chromosomes. Therefore, they will have 4n chromosomes (92 in humans) and then 23 homologous pairs will go to each new daughter cell. Therefore, daughter cells are identical to each other and to the mother cell (all diploid cells).

However, **sex cells, or gametes**, divide through a process known as **meiosis**. Again, they will first replicate their DNA, having 4n chromosomes, but they undergo two divisions.

From the first division 2 cells are formed each having 2n chromosomes, which again divide resulting in a total of four haploid cells (n), each different from the other and different from the mother cell. When gametes fuse during fertilization, the characteristic 2n of the species is restored.

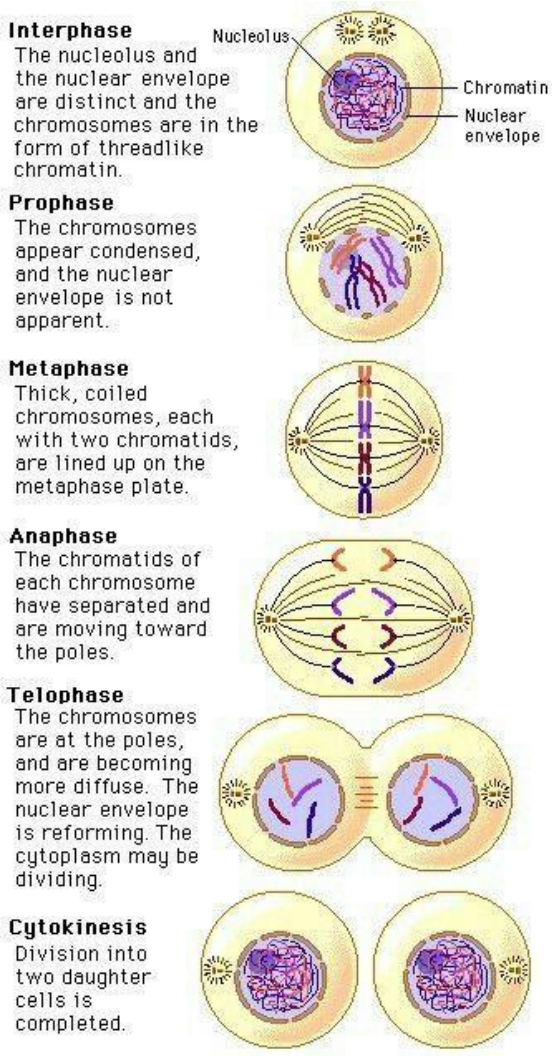



In mammals and other living organisms there is a chromosome pair that determines the sexual characteristic of the individual. These are called **sex chromosomes X and Y.** Females have two X chromosomes and males have an X and a Y chromosome. The rest of the 44 chromosomes (in humans) are called **autosomes (autosomal chromosomes).** The group of chromosomes of a cell is the karyotype. Chromosomes can be classified by their homologous pair, and ordered by size and shape. The order obtained is called an **idiogram**. Using specific staining techniques, **banding** of chromosomes is obtained which allows us to pair the homologous chromosomes, as well as to group them by decreasing size. This also allows us to determine some genetic diseases.



### Cellular division in eukaryotic cells: Mitosis

Cellular division in eukaryotic cells consists of two phases: first the nucleus divides (**mitosis**) and then the cytoplasm divides (**cytokinesis**). The scheme on the right shows the stages of mitotic cell division in an animal cell.




Mitosis is a cell reproduction process by which multicellular organisms regenerate lost or damaged cells, or simply makes new cells. In the case of unicellular organisms it can be considered as asexual reproduction. **It does not generate genetic variability,** as the new daughter cells are identical to each other and to the mother cell. This is how all somatic cells divide (epithelial cells, liver cells, etc. All but sex cells).

## Meiosis

Concept: it is a type of cellular division **needed** in organisms with sexual reproduction. In sexual reproduction, there is **fertilization**, which is the fusion of haploid gametes to restore the diploid number in the zygote. The zygote, by successive mitotic divisions gives rise to the multi-cellular organism, which cells are therefore diploid, all containing identical genetic information.

If the gametes were also produced by mitosis, these would be diploid and genetically identical. During fertilization, the fusion of these diploid gametes would produce a tetraploid (4n) zygote, which would give rise to 4n organisms. In the same line, these

4n organisms would produce 4n gametes…. and so on. In each generation the number of chromosomes of the species would be doubled. Therefore a mechanism of cell

division is needed where the number of chromosomes is reduced (halved): going from 2n cells to n cells. This process is meiosis.

Besides being necessary, meiosis is very beneficial, as it generates **genetic variability**: the daughter cells are different from each other and also different from the mother cell.

Process: meiosis (like mitosis) is preceded by the replication of chromosomes or DNA. However, this single replication is followed by **two** consecutive cell divisions, called meiosis I and meiosis II. These divisions result in four daughter cells (rather than the two daughter cells of mitosis), each with only half as many chromosomes as the parent.

Meiosis I: consists of prophase I, metaphase I, anaphase I and telophase I.

During **prophase I** homologous chromosomes, each made up of two chromatids, come together as pairs (forming a tetrad, a complex of four chromatids). At numerous places along their length, nonsister chromatids (chromatids belonging to homologous chromosomes, in contrast to sister chromatids belonging to the same chromosome) are **criss-crossed** and **recombined**. As a result of these crossings, mixed
chromatids are formed with fragments from the mother and the father chromosomes (this is the first source of variability in meiosis)

During **metaphase I**, the homologous pairs are randomly arranged on the metaphase plate: sometimes the paternal chromosome is on the right and the maternal on the left and it could also be the other way around. This way many different and diverse combinations can happen (this is the second variability source of meiosis: for example, a new daughter cell could have chromosomes 1, 2', 3, 4, 5' etc, while the other would have
1', 2, 3', 4', 5 etc).

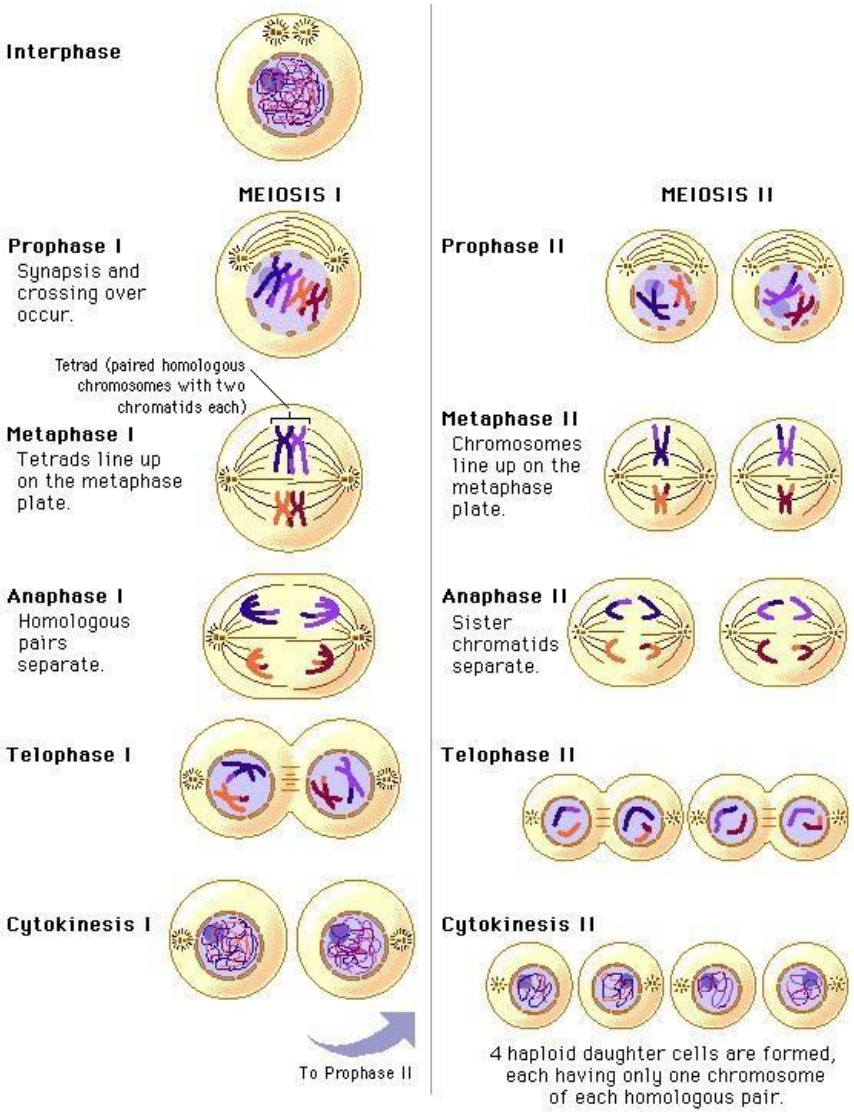
In **anaphase I** and **telophase I** the homologous chromosomes migrate toward the opposite poles of the cells. Segregation of chromosomes (each pole now has a haploid chromosome set, but each chromosome still has two chromatids) and finally cytokinesis, usually occurring simultaneously with telophase I forms two daughter cells each with only one of the homologous chromosomes.

There is no further replication of the genetic material prior to the second division of meiosis II.

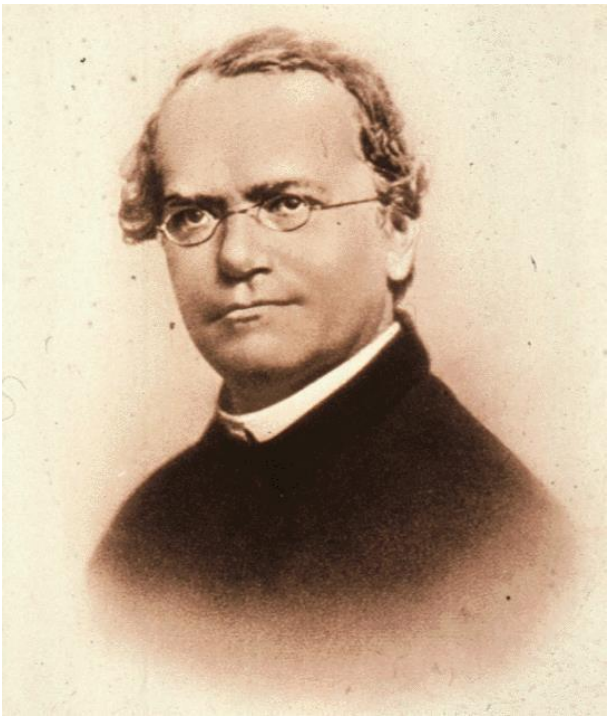
**During the 2nd meiotic division,** the two chromatids of each chromosome separate into the daughter cells in a very similar way as mitosis. At the end of meiosis II there will be

four daughter cells, each with the haploid number (n) of chromosomes and genetically different from one another and from the mother cell (genetic variation).

SUMMARY: Meiosis is a cellular division necessary for organisms with sexual reproduction. Meiosis reduces the number of chromosomes by half (which will be restored during fertilization) and allows for genetic variation (the daughter cells are genetically different from each other, and also from the mother cell).



## MENDELIAN GENETICS



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Gregor Mendel, the father of genetics, was an Austrian monk that lived in the XIX century. He worked on the transmission of the characters of pea plants through successive generations. Gregor Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments. Mendel spoke of *heritable factors.*

The concepts of gene and chromosome came later. Walter Sutton and Theodore Boveri in 1903 proposed the *chromosome theory of inheritance.* And thanks to the works of Watson and Crick about the nature of DNA, it was proven that the heritable factors (the genes) were in fact DNA fragments.

These are the main concepts that you should know:

GENE: there are different ways to define a gene. If we are talking about classic or Mendelian genetics, we will define a gene as a sequence of nucleotides (usually a DNA fragment, but in viruses composed of RNA it is an RNA fragment) that contains information for a particular character, e.g. the gene that determines eye colour. (Geneticists use the term character for a heritable feature, such as eye colour, that varies among individuals. Each variant for a character, such as brown or blue, is called a trait).

A more complete definition of a gene is:

**A sequence of nucleotides to which a specific function can be assigned**. Examples of such functions: a) control of the expression of a particular character; b) coding of a specific polypeptide chain (the transcription of itself into mRNA and the following translation into a polypeptide chain in the ribosomal unit); that it transcribes to tRNA; d) transcription to rRNA.

Each gene occupies a specific position on a chromosome called its **locus** (loci –plural). Genes are passed down from parents to offspring (via the gametes) during sexual reproduction. Each gamete has one set of chromosomes (they are haploid). Fertilization between a male and a female gamete gives rise to a zygote, with two sets of chromosomes (diploid). The individual from that zygote will also have two sets of

chromosomes (also diploid). E.g. Humans have 23 chromosomes in their gametes or sex cells (ovum and sperm). The rest of the cells of our body have 23 pairs (46) of chromosomes.

**HOMOLOGOUS CHROMOSOMES** are a pair of chromosomes having the same structural features. Each member of the pair of chromosomes has the same number and pattern of genes. The genes found in the same locus on both chromosomes carry information for the same character, but may have different alleles.

**ALLELE (OR ALLELOMORPH):** An allele is an alternative form that a gene can have due to successive mutations. They are found in the same locus in each homologous and carry information for the same character, e.g. the gene that determines straight hair and that which determines curly hair. A gene can have multiple alleles, but diploid organisms can only have two of those, as we have our chromosomes in pairs.

**HOMOZYGOUS (TRUE-BREED):** An organism which has a pair of identical alleles for a certain character is said to be homozygous for that character. Normally genes are represented by letters, so a homozygous organism will have the same letters (AA, aa, BB, etc)

**HETEROZYGOUS (HYBRID):** Organisms having different alleles for a character. E.g. Aa, Bb, etc.

**COMPLETE DOMINANCE (DOMINANT HEREDITY):** In all hybrid individuals an allele (the dominant allele) for a particular character, is fully expressed, and the other allele or recessive allele has no noticeable effect on the organisms appearance.

**CODOMINACE**: Neither allele is dominant or recessive. Instead both alleles are separately (equally) manifested in the phenotype. E.g. human blood type group AB.

**INCOMPLETE DOMINANCE**: It is characterized by an intermediate phenotype. For example the pink flowers of snapdragon hybrids.

**GENOTYPE:** It is the genetic information of a particular organism as specified by its alleles. The genotype is hereditary, as progenitors pass it down to their offspring, e.g. AaBb, etc.

**PHENOTYPE**: It is the observable characteristics of an organism produced by the interaction of its genes and the environment which surrounds its development. E.g. Black hair, blue eyes, etc.

## SEX CHROMOSOMES AND SEX-LINKED INHERITANCE

Generally, males produce small motile gametes, and females produce large non-motile gametes. Although the anatomical and physiological differences between male and females are numerous, the chromosomal basis of sex is rather simple. Most frequently, in determining the sex of an individual there are two chromosomes involved. These are known as **heterochromosomes** or **sex chromosomes** (we have already seen that chromosomes which do not determine the sex are called autosomes) In humans and many other species, there are two varieties of sex chromosomes, designated the X and the Y chromosomes. Females have two X chromosomes (genotype XX), and males have an X chromosome and a smaller Y chromosome (genotype XY). When meiosis occurs in the gonads (male testes and female ovaries), the two sex chromosomes segregate, and each gamete receives one. Each ovum contains one X chromosome. In contrast, half the sperm cells contain an X chromosome, and half contain a Y chromosome. If a sperm cell bearing an X chromosome happens to fertilize an ovum, the zygote is XX, a female; if a sperm cell containing a Y chromosome fertilizes an ovum, the zygote is XY, a male. Sex is a matter of chance; a 50% probability for each sex.

In addition to the role of sex chromosomes in determining sex, these chromosomes, especially the X chromosome, have genes for many characters unrelated to sex.
Therefore, the heredity of such characters is **sex-linked**. In humans the term sex-linked usually refers to X-linked characters. Fathers pass X-linked alleles to all their daughters, but to none of their sons. On the other hand, mothers can pass sex-linked alleles to both sons and daughters. There are very few but yet some characters which are Y-linked. Fathers pass Y-linked alleles to all their sons. If a sex-linked (X-linked) trait is due to a recessive allele, a female will express the phenotype only if she is a homozygote. Any males receiving the recessive allele from his mother will express the trait. A heterozygote female for a sex-linked disorder will show a normal phenotype but as she is carrying the recessive allele is called a carrier. Some sex-linked (X-linked) disorders are the following:

**Colour blindness (daltonism)** is a mild disorder due to a recessive allele (d), where the person with the disorder can not distinguish red from green. The dominant allele (D) determines normal vision. The different genotypes and corresponding phenotypes possibilities are: $X^DX^D$ (normal female); $X^DX^d$ (normal female but carrier of the disorder); $X^dX^d$ (colour blind female); $X^DY$ (normal male) and $X^dY$ (colour blind male).

**Hemophilia** is a sex-linked recessive trait where the dominant (H) allele determines a protein required for blood clotting. The recessive allele (h) determines hemophilia; the most seriously afflicted individuals may bleed to death after relatively minor cuts, etc. $X^HX^H$ (normal female); $X^HX^h$ (normal but carrier female); $X^hX^h$ (hemophiliac female); $X^HY$ (normal male) and $X^hY$ (hemophiliac male).