From Mendel to models of heredity

Genetics

- Genetics studies the **mechanisms of transmission** of characters from one generation to another.
- A **character** is a physical characteristic that can be observed in an individual.
- A **trait** is the particular form that a character can assume, and is said to be hereditary if it is transmitted from parents to children.
- **Gregor Mendel** carried out the first studies on heredity in the second half of the 19th century using pea plants.

Dominance (first law)

- Mendel performed cross-fertilization between a pure line of pea plants for opposite forms of the same character.
- The parental generation (P) produces the first filial generation (F_1) of hybrid individuals that manifest only one of the traits present in P (**dominant trait**); the other trait (**recessive trait**) does not appear in F_1.

Segregation (second law)

- The second filial generation (F_2) is obtained by self-pollination of F_1.
- In F_2, both the dominant and the recessive trait are manifested in a ratio of 3:1.
- The units responsible for the heredity of a character (**genes**) can exist in different versions (**alleles**).
- Each individual possesses two copies of each gene; these are separated during the formation of gametes, of which only one copy is inherited. An individual is **homozygous** for a gene when both alleles are the same, and is **heterozygous** if the two are different.
- The **genotype** is the entire set of alleles that determines a character. The **phenotype** is the observable characteristic determined by the genotype.
- Crossing with the recessive homozygote (**testcross**) reveals whether an individual of a dominant phenotype is homozygous or heterozygous.
- The **Punnett square** can be used to predict the allele combinations resulting from a cross.

Independent assortment (third law)

- The cross between two individuals that are heterozygous for two genes (**dihybrids**) generates four possible phenotypes, two of which are the same as the **parental** phenotypes and two of which are **recombinant**.
- Different genes segregate independently during the formation of gametes.

Genes and chromosomes

- A gene is a sequence of DNA located on a **locus** on a chromosome.
- Genes that are on the same chromosome make up an **associated group** and segregate independently only in case of crossing-over between sister chromatids during meiosis.
- Two nearby loci have less probability of undergoing **recombination** than distant loci.
- Based on the study of recombination frequencies, we obtain **genetic maps** that show the position of the genes on the chromosomes.
- The sex of many organisms is determined by a pair of **sex chromosomes** (in humans, XX for female and XY for male), while **autosomes** are present in two copies of the genome.
- Women produce only gametes with the X chromosome, while men produce half gametes with X and half with Y. Some genes found on the sex chromosome are inherited in a particular way.

Interactions between alleles

- Mutations result in the existence of different alleles. The **wild** allele is the one present in nature in most individuals. A gene is **polymorphous** if the wild allele is present in less than 99% of individuals.
- For many genes, there are more than two possible alleles (**multiple alleles**).
- The **pleiotropic** allele affects the phenotype of more than one character. The term **incomplete dominance** describes heterozygous individuals showing an intermediate phenotype between those of different homozygotes, and **codominance** describes heterozygous individuals both showing homozygous phenotypes.

Interactions between genes

- One gene can influence the phenotypic expression of another (**epistasis**) or cancel the expression of a mutated allele of another gene (**suppressor**).
- Mating between close relatives (**inbreeding**) can lead to homozygosity for some detrimental recessive alleles, while heterosis due to hybridization is often advantageous for the offspring.
- A character regulated by many genes is called **polygenic**. The simple characters studied by Mendel determine **qualitative** phenotypic differences. On the other hand, many complex characters determine **quantitative** phenotypic differences, which show continuous variability within the population and depend on interaction between genes and the environment.

AUDIO
Summing-up