GENETICS: BASIC CONCEPTS AND GENERALITIES

Gustavo Forero Acosta

INTERDISCIPLINARY GROUP OF SECTORAL STUDIES (GIES IN SPANISH)

School of Agricultural, Livestock and Environmental Sciences

Sello Editorial
Universidad Nacional
Abierta y a Distancia

GENETICS: BASIC CONCEPTS AND GENERALITIES

Gustavo Forero Acosta

Research group:
Interdisciplinary Group of Sectoral Studies (GIES in Spanish)

575.1 Forero Acosta, Gustavo
F715 Genetics, Basic Concepts and Generalities / Gustavo Forero Acosta -- [1.a. ed.]. Bogotá: Sello Editorial UNAD/2024. (Research Group: Interdisciplinary Group of Sectoral Studies - GIES – School of Agricultural, Livestock, and Environmental Sciences - ECAPMA.)

e-ISBN: 978-628-7786-25-7

1. Genetics 2. Evolution (Biology) 3. Inheritance I. Forero Acosta, Gustavo.

GENETICS: BASIC CONCEPTS AND GENERALITIES

Author:
Gustavo Forero Acosta

Research group: Interdisciplinary Group of Sectoral Studies - GIES

e-ISBN: 978-628-7786-25-7
School of Agricultural, Livestock and Environmental Sciences

Edition: Hipertexto - Netizen
Translated by: Smart Translators SAS

©Publisher
UNAD Publishing House
Universidad Nacional Abierta y a Distancia
Calle 14 Sur No. 14-23
Bogotá, D.C.

November 2024

How to cite this book: Forero Acosta, G., (2024). *Genetics: basic concepts and generalities*. Sello Editorial, Universidad Nacional Abierta y a Distancia - UNAD. https://doi.org/10.22490/UNAD.9786287786257

BOOK REVIEW

This material is an important resource within the guiding documents for the academic, pedagogical and didactic activities of students of an introductory genetics course. It includes concepts, fundamentals and applications of basic genetics in agricultural sciences and serves as a pedagogical tool for students in the School of Agricultural, Livestock and Environmental Sciences. Students take this course as a common requirement in their programs or as an elective course in other programs where it is not included. This material focuses specifically on basic genetics, including the concepts of genetics, cell, cell cycle, DNA, RNA and genes. It also describes and provides examples of Mendelian and non-Mendelian genetic models and introduces basic statistical concepts used in basic genetics. Other topics such as mutations, chromosomal aberrations, linkage or genetic mapping are not covered, since, according to the internal coherence of the courses offered at the University, these are part of the advanced course on plant breeding, where they are explored in greater detail. The material includes several exercises and review questions to help students apply the content covered.

AUTHOR'S PROFILE

Gustavo Forero Acosta holds a Bachelor's degree in Chemistry and Biology, a Master's degree in Biology with an emphasis in Genetics and Molecular Biology, doctoral studies in Sustainable Development, Economics, Society and Environment, (non-graduate), and doctoral studies in Project Design and Evaluation (non-graduate). He has served as a professor and researcher at several private and public universities in the country and has extensive experience in curriculum design, course creation and materials adapted for virtual and distance education systems, as well as in advising on entrepreneurship and research projects of different kinds.

He is currently part of the career teaching staff (ranked as Associate Professor) at the School of Agricultural, Livestock and Environmental Sciences at the Universidad Nacional Abierta y a Distancia – UNAD currently, a doctoral student in Project Studies..

CONTENTS

II. TEACHING UNIT
MODELS OF MENDELIAN GENETICS

Chapter 4

Chapter 5

Chapter 6

III. TEACHING UNIT
NON-MENDELIAN GENETICS MODELS

Chapter 7

Chapter 8

LIST OF FIGURES

LIST OF TABLES

PRESENTATION

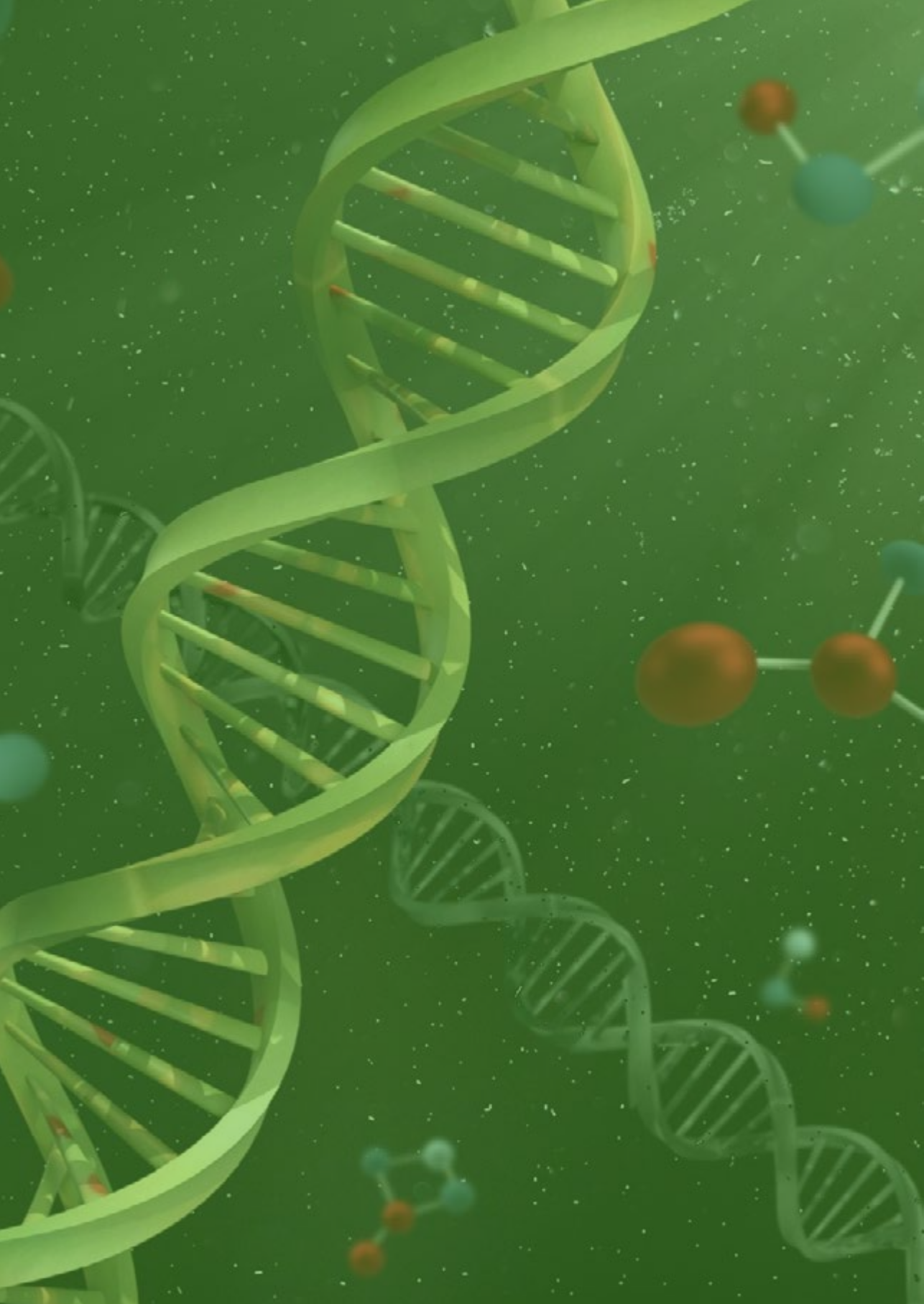
This text is divided into teaching units. The first unit presents fundamental concepts such as cell, cell cycle, DNA, RNA and genes. The second one covers concepts related to non-Mendelian genetic models, some basic notions of probability, phenotypic proportion testing and types of crosses. The third unit explores non-Mendelian genetic models in depth.

This material is dedicated to basic genetics and will allow students to become familiar with genetic terms such as segregation, independent assortment, complete dominance, codominance, recessive alleles, dominant alleles, and multiple alleles, among others. In addition, clarifying processes such as cell division and chromosome segregation will help students understand the universal laws governing reproductive processes and the principles of molecular genetics, applying these concepts through a variety of problems and application questions.

Students will develop the ability to understand, relate and apply fundamental genetic concepts and Mendelian inheritance mechanisms. They will optimize their professional performance as zootechnicians, agronomists, forestry engineers, etc. by analyzing how the variability of organisms depends on the genetic endowment of each individual and its various manifestations.

Students will acquire knowledge by consulting texts or online resources about the topics covered, as well as through practice with workshops to be developed in virtual classroom groups. To this end, workshops have been designed based on supplementary readings, article reviews and application exercises that will guarantee a deeper and better assimilation and understanding of the studied topics.

To successfully complete the course, students will consult the proposed bibliography to access all necessary information and meet academic requirements. It is important to note that once learning is completed, students will be able to relate and understand articles or experimental works related to Mendelian genetics and independently interpret any natural process involving phenotypic variation of qualitative traits.



INTRODUCTION

Genetics, as a branch of biology studying the composition and transmission of genetic material in living organisms, is essential to any study of animal or plant life.

By studying genetics, general concepts about biological processes of inheritance are acquired. In addition, Mendelian laws for monohybrid, dihybrid and polyhybrid crosses are checked, illustrating the segregation and distribution of genetic material. By the end of the course, students will connect Mendelian inheritance mechanisms with qualitative traits to explain phenotypic variability in living organisms.

Since the 20th century, genetics has become an essential component in strengthening other fields of knowledge. It is highly valuable in medicine and in the social field, and is also used in the directed transformation of animal breeds and plant varieties.

TEACHING UNIT

INTRODUCTION TO THE STUDY OF GENETICS

Gustavo Forero Acosta

CHAPTER 1
CELL THEORY

1.1 INTRODUCTION TO THE STUDY OF THE CELLS

When reviewing the main concepts associated with the term "cell" today, we must trace back to its initial use by scientist and researcher Robert Hooke in 1655. Hooke used it to describe his observations of a slice of cork examined under a magnifying lens. However, the cell theory was formally proposed in 1839 by scientists Matthias Schleiden and Theodor Schwann. They concluded that all living organisms are made up of fundamental units called cells. This marked the development of cytology (the science that studies cells), an important branch of biology (Curtis et al., 2013).

Defining the concept of life remains a complex and challenging task, especially considering the diversity observed among living organisms and the wide range of molecules involved in forming life, many of which are seen in isolation. The existence of complex interactions among molecules establishes a functional unit represented in the cell structure. Since life is based on these remarkable interactions, it is difficult to determine the actual meaning of *life*.

All cells have the same fundamental structure that allows them to solve similar survival problems (e.g., searching for energy, reproducing), but each cell has to face specific problems depending on its environment. This is why there are several types of cells, each with variants relative to the base model (Curtis et al., 2013).

Cells in multicellular organisms specialized and shared tasks to ensure better functioning of the individual. Thus, very different cells are found within the same organism, performing functions such as transmitting signals, making movements, transporting oxygen or producing substances for other cells. Unlike unicellular organisms, none of these cells can live independently and all are essential for the survival of the individual (Forero G., & Bernal, L. 2013a).

1.2 HISTORY OF THE CELL

For many centuries, the concept of the *cell* could not be elucidated. Only with the invention of the microscope, further investigation into cells became possible, leading to discoveries about cell structure throughout the 17th century. These events marked a true scientific revolution and gave rise to modern biology. The discovery of cells is generally credited to Robert Hooke, who, through his observations of slices of cork with a homemade microscope, named the compartments he observed as *cells* because they reminded him of the cells inhabited by monks in a monastery. Actually, Hooke had observed the empty walls of dead plant tissue, walls originally produced by the living cells that surrounded them.

Meanwhile, Anton Van Leeuwenhoek, a Dutchman who sold fabric and buttons, spent his leisure time carving lenses and building microscopes of remarkable quality. For 50 years, he sent letters to the Royal Society of London describing his microscopic observations, along with a vague discourse about his daily habits and health. Leeuwenhoek was the first to examine a drop of pond water and observe the abundant number of microscopic "critters" moving about. He was also the first to describe the initial forms of bacteria from water in which he had soaked pepper and from material scraped from his own teeth.

His early letters to the Royal Society, describing this previously unseen world, aroused such skepticism that the Royal Society sent its guardian, Hooke, to confirm the observations. Soon, Leeuwenhoek became a worldwide celebrity and was visited in Holland by Peter the Great of Russia and the Queen of England (Karp, 2019).

It was not until the 1830s that the great importance of cells was confirmed. In 1838, Matthias Schleiden, a German lawyer turned botanist, concluded that, despite differences in the structure of various types, plants were made up of cells and that the plant embryo originated from a single cell. In 1839, Theodor Schwann, a German zoologist and colleague of Schleiden, published a very thorough work on the cellular basis of animal life and concluded that animal and plant cells were similar. These scientists proposed the dogmas of cell theory:

- All organisms are made up of one or more cells.
- The cell is the structural unit of life.

In 1855, Rudolf Virchow, a German pathologist, proposed a convincing hypothesis for the third dogma of cell theory:

- Cells can only originate from the division of a pre-existing cell.

All living organisms are made up of cells and it is widely accepted that no organism is considered a living being if it does not consist of at least one cell. Some microscopic organisms, such as bacteria and protozoa, are single cells, whereas animals and plants are made up of several million cells organized into tissues and organs (Karp, 2019).

1.3 THE CELL THEORY

In general, modern cell theory can be summarized in three postulates:

- The cell is the basic structural unit of all living organisms; all organisms are made up of cells.
- The cell is the functional unit of all organisms. The entire functioning of the organism depends on the processes that occur inside the cell: respiration, reproduction, digestion and growth, among others.
- All cells originate from the division of pre-existing cells (in other words, through reproduction). Each cell contains genetic material that is passed on during this process (Karp, 2019).

1.4 BASIC PROPERTIES OF THE CELLS

A living being is characterized by fundamental properties such as having its own metabolism, replicating genetic material, renewing structures at the molecular level and interacting with the environment. Cells are the units capable of fulfilling these properties.

Cells have a highly organized structure and have the capacity to self-regulate, respond to different stimuli, and perform functions such as respiration, movement, digestion, reproduction, communication and reaction to stimuli.

In unicellular organisms such as protozoa and bacteria, the cell is autonomous and performs all functions. In contrast, in multicellular organisms such as plants and animals, which are made up of thousands of cells organized into tissues and organs, cells perform specific functions (Karp, 2019).

a. Structure. All cells have three fundamental parts: a membrane system, cytoplasm and a nuclear region that houses the genetic material.

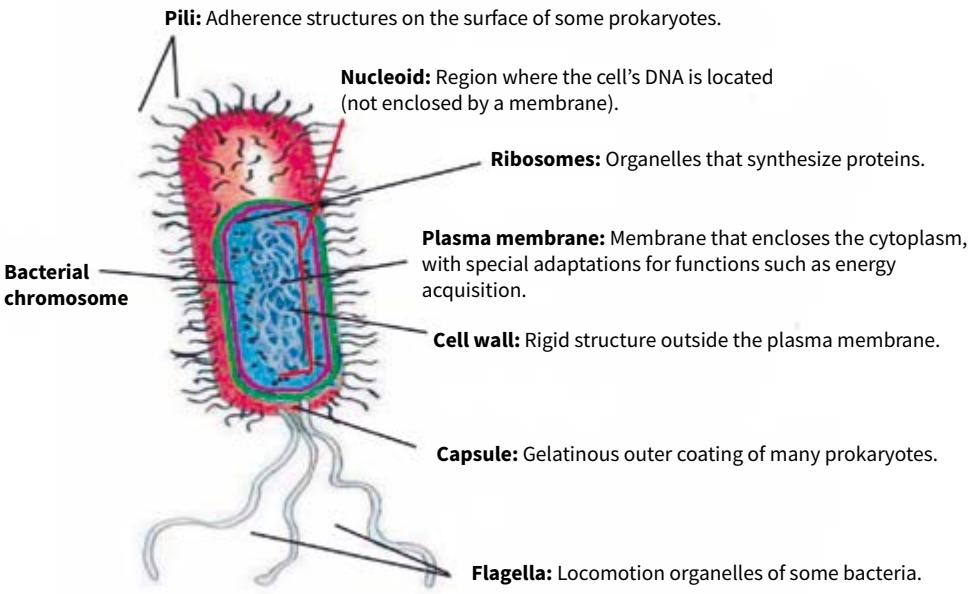
b. Cell Types. There are two basic types of cells based on their organizational complexity: prokaryotic cells and eukaryotic cells.

1.5 PROKARYOTIC CELLS

Prokaryotic cells lack a nucleus, meaning their genetic material is not enclosed by a membrane. Examples of prokaryotic cells include bacteria (Figure 1).

Prokaryotic Cell Organelles. A prokaryotic cell is generally made up of a cell wall, a plasma membrane, ribosomes and genetic material (chromosome).

FIGURE 1. *Diagram of a Prokaryotic Cell*



Source: Mosso, (2010a)

- **Cell Wall.** The cell wall provides osmotic protection to the bacteria. It is essential for cell division, gives the cell its shape and contains antigenic determinants that serve as virulence factors, as well as for serological classification.

- **Plasma Membrane.** It is different from that of eukaryotic cells because of the absence of sterols. The plasma membrane performs the following functions:

    - Selective permeability and solute transport.

    - Electron transport and oxidative phosphorylation in aerobic species. This occurs in invaginations of the membrane called mesosomes, which function similarly to mitochondria.

    - Excretion of hydrolytic exoenzymes to degrade polymers into subunits that penetrate the cytoplasmic membrane and serve as nutrients. Many pathogenic bacteria release exoenzymes such as proteases and toxins, which are important virulence factors.

    - Biosynthetic functions Contains sites for the deposition of enzymes needed for phospholipid synthesis and cell wall compounds. It also houses enzymes required for DNA replication, located at the site where DNA attaches, presumably in the septum mesosomes.

- **Cytoplasm.** It is a gel-like solution containing insoluble granules that serve as reserve material. It includes:

    - Ribosomes: Made up of ribosomal RNA and proteins. They differ from the ribosomes of eukaryotic cells in their sedimentation coefficients.

    - Proteins: Most of them are enzymes involved in cell metabolism.

- **Genetic material.** The genetic material is made up of a single circular DNA strand, referred to as the chromosome of the bacteria.

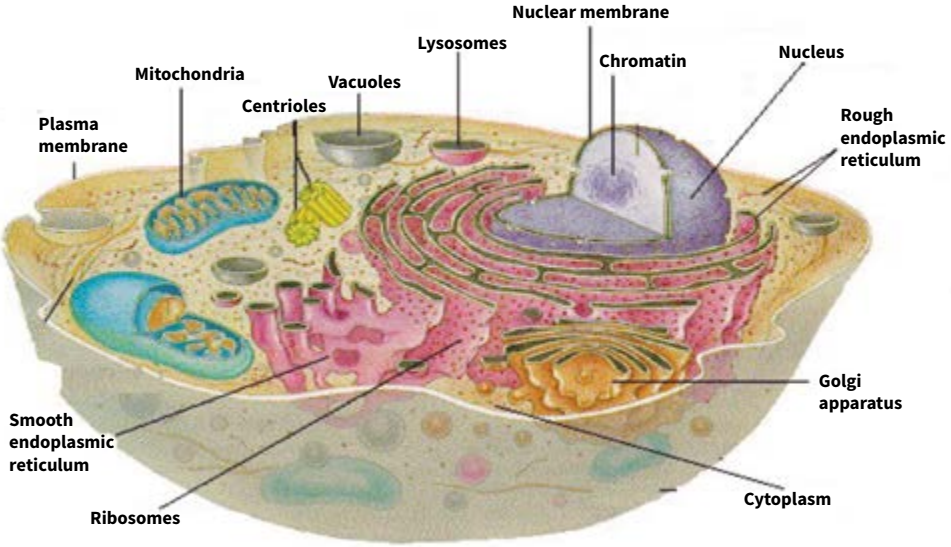
1.6 EUKARYOTIC CELLS

In general, eukaryotic cells are characterized by having a nuclear membrane that surrounds and protects the genetic material, forming the nucleus. They have many organelles that function interdependently and perform vital functions (Figure 2).

Eukaryotic cells can be of two types: animal and plant cells. They differ in the presence of some cellular organelles such as cell walls, chloroplasts and plastids.

- **Organelles of Eucharistic Cells.** A typical eukaryotic cell is made up of a membrane system. It includes the cell wall (in plant cells), the plasma membrane, smooth endoplasmic reticulum, rough endoplasmic reticulum, Golgi apparatus, the cytoplasm containing cellular organelles such as mitochondria, the nucleus, ribosomes, centrioles (in animal cells), vacuoles (more predominant in plant cells than in animal cells), chloroplasts (characteristic of plant cells) and DNA, which is usually found in the form of chromatin (Figure 3).

FIGURE 2. *Eukaryotic cell*



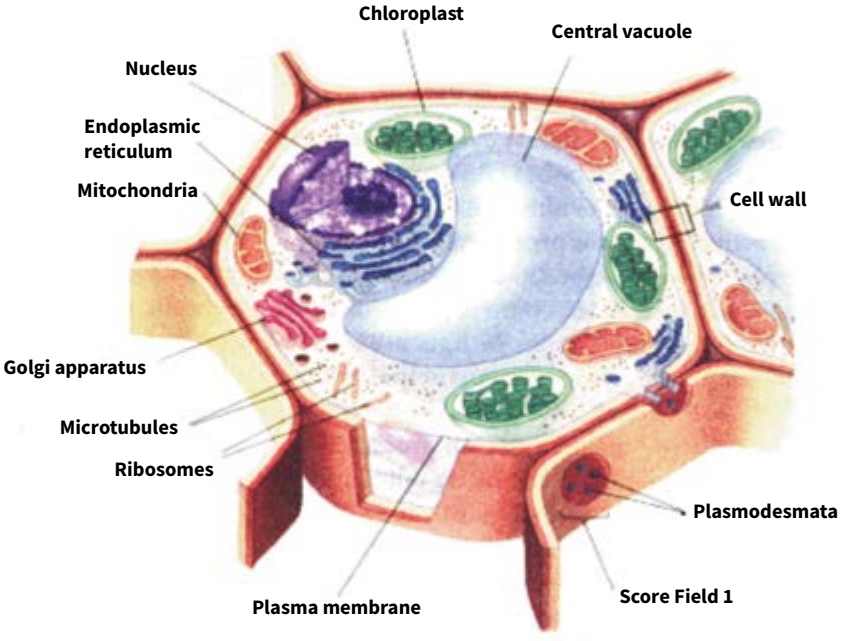
Membrane Systems.
The membrane system is made up of:

Cell Wall. It is composed of cellulose fibers within pectin aggregates, which provide high resistance and offer significant protection to the cell.

Plasma Membrane. Found in both animal and plant cells, this membrane is typically semipermeable or has differential permeability and is described by the fluid mosaic model (Figure 4). It is composed of a phospholipid bilayer along with carbohydrates and proteins. Some proteins are firmly embedded in the lipid bilayer and are known

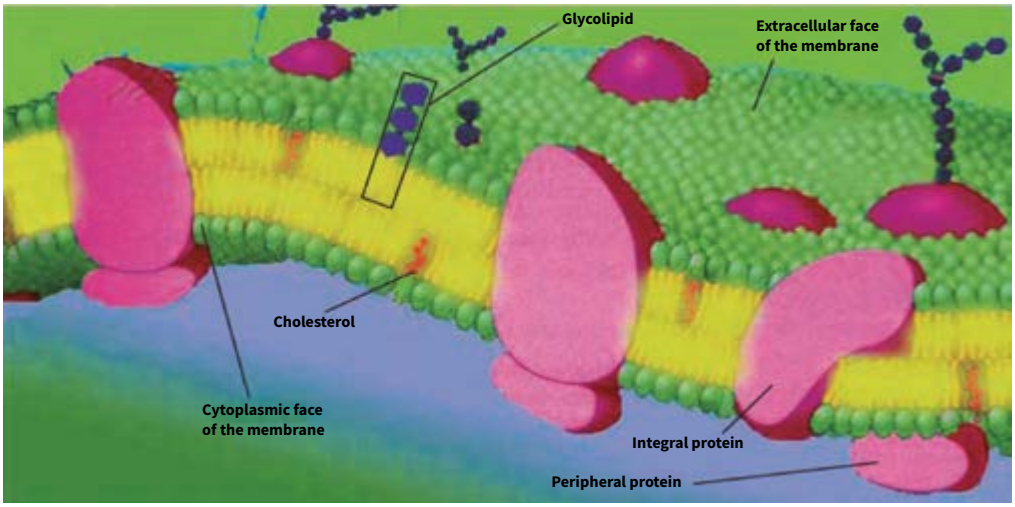
as *integral proteins.* Among these, some span the entire lipid bilayer and are called *transmembrane proteins.* Other proteins are loosely attached to the inner or outer surface of the membrane and are referred to as *peripheral proteins.*

FIGURE 3. *Parts of a Plant Eukaryotic Cell*



This membrane regulates the passage of substances into and out of the cell through different processes and mechanisms. It separates the cell from its extracellular environment but allows interaction due to its structure and composition (Karp, 2019).

FIGURE 4. *Fluid Mosaic Model of the Plasma Membrane*



Source: Mosso, (2010b)

The cytoplasmic membrane isolates the cytoplasm from the external environment and selectively allows specific substances to enter or exit the cell. Proteins are used for the transport of molecules, as receptors or ligands, and for cell communication.

The membrane is selectively permeable and regulates the movement of materials in and out of the cell. Likewise, it regulates water content within the cell due to the biochemical characteristics of its molecular components, thus making it semipermeable.

Cytoplasm and Cytosol. The cytoplasm comprises the entire volume of the cell, except for the nucleus. It includes numerous specialized structures and organelles.

The concentrated aqueous solution in which organelles are suspended is called the cytosol. It is a water-based gel containing a large number of large and small molecules. In most cells, the cytosol is by far the largest compartment (in bacteria, it is the only intracellular compartment). Many of the most important cell maintenance functions occur in the cytosol, such as the early stages of breakdown of nutrient molecules and the synthesis of many large molecules constituting the cell.

Although many cytosol molecules are in true solution and move rapidly through free diffusion, others are rigorously organized. These ordered structures give the cytosol internal organization, acting as a framework for the production and breakdown of large molecules and channeling many cellular chemical reactions along restricted pathways (Karp, 2019).

Cytoskeleton. The cytoskeleton is a network of protein filaments in the cytosol that occupies the interior of all animal and plant cells. It is particularly important in animals, which lack a rigid cell wall, as it maintains the cell's shape and structure. It acts as a frame for organizing the cell and anchoring organelles and enzymes. It is also responsible for many cell movements. In many cells, the cytoskeleton is not a permanent structure, but is endlessly dismantled and rebuilt. It consists of three main types of protein filaments: microtubules, actin filaments, and intermediate filaments, linked to each other and other cellular structures by various proteins.

Eukaryotic cell movements are often mediated by actin filaments or microtubules. Many cells have flexible hair-like structures on the surface called cilia or flagella, which contain a nucleus formed by a bundle of microtubules capable of developing regular flexing movements requiring energy. For example, sperm swim with the help of flagella, and cells lining the intestines and other vertebrate body ducts have numerous cilia on their surfaces that move fluids and particles in a specific direction. Large beams of actin filaments are found in muscle cells where, together with a protein called myosin, they generate powerful contractions. Movements associated with cell division in animals and plants depend on actin filaments and microtubules, which distribute chromosomes and other cellular components between the two daughter cells during segregation. Animal and plant cells perform many other movements to acquire a certain shape or to preserve their complex internal structure (Karp, 2019).

Nucleus. It is surrounded by a membrane, is spherical and measures about 5 µm in diameter. Inside the nucleus, DNA molecules and proteins are organized into chromosomes, which usually appear in identical pairs. Chromosomes are highly condensed and difficult to identify separately. Just before cell division, they condense to the maximum and become thick enough to be detectable as independent structures. Each chromosome contains a single long, coiled DNA molecule with linear sequences of genes. These genes contain coded instructions for building the proteins and RNA molecules needed to produce a functional copy of the cell.

Chromatin and Chromosomes. Chromatin is the network or membrane observed during interphase through the optical or electron microscope as very thin and twisted filaments. It is made up of DNA, proteins and nucleic acids. During cell division, chromatin organizes into individual structures that are much more visible under the light microscope; these structures are known as chromosomes.

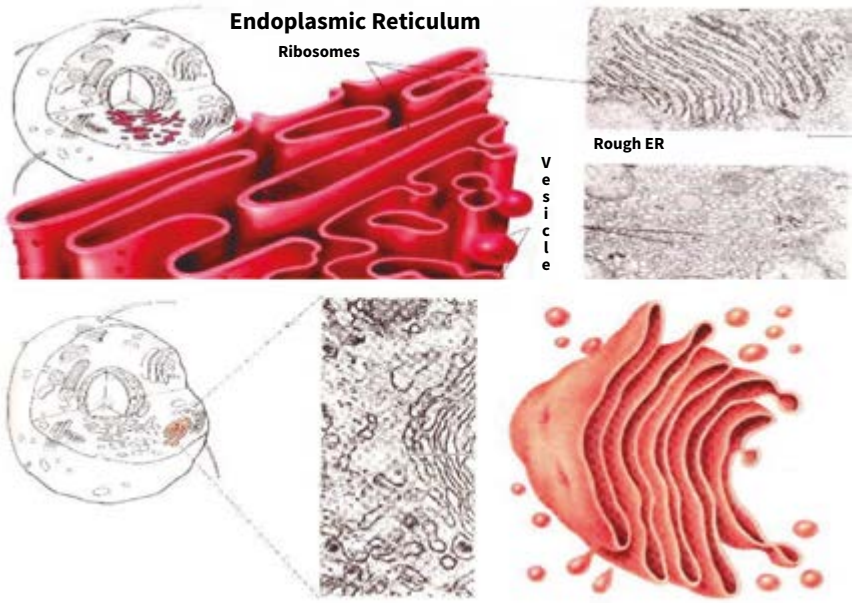
A chromosome is a highly condensed structure containing a very long DNA molecule and is made up of a series of genes. Each chromosome is made up of two chromatids, each containing a folded and identical DNA nucleofilament, joined at a central point

called the centromere. Each chromatid also has a kinetochore, which is the microtubule organizing center that forms during mitosis and helps attach chromosomes to the mitotic spindle (Karp, 2019).

Endoplasmic Reticulum (ER). It is a membranous system (Figure 5) containing very small particles called *ribosomes* that are responsible for protein synthesis. The presence of ribosomes gives a rough appearance; hence, it is known as *rough endoplasmic reticulum*. In contrast, the ribosome-devoid membrane, or *smooth endoplasmic reticulum*, contains enzymes responsible for lipid synthesis.

Golgi Apparatus. It is a system of membranous sacs (Figure 5) that stores, modifies and packages macromolecules synthesized in the endoplasmic reticulum for secretion or delivery to various organelles.

FIGURE 5. *Rough and Smooth Endoplasmic Reticulum. Golgi Apparatus.*

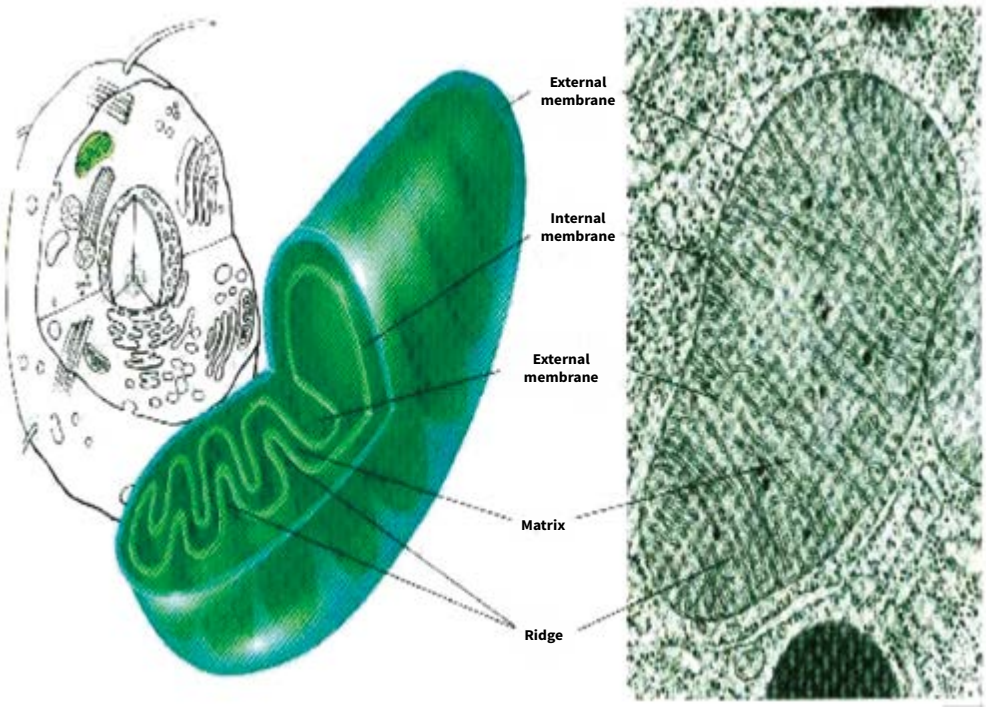


Source: Adapted from Sadava and Purves (2009)

Mitochondria. The mitochondria (Figure 6) are surrounded by a double membrane and have cristae, matrix and DNA. They carry out a set of reactions in which pyruvic acid is broken down into carbon dioxide, water and ATP, the end product of the

metabolism of carbohydrates, lipids and proteins. In addition, the mitochondrion performs the process of cellular respiration. Figure 6 shows the typical structures of a mitochondrion (Audesirk., & Byers, 2003).

FIGURE 6. *Main structures present in the mitochondria*



Source: Audesirk., & Byers, (2003)

TABLE 1. *Differences between prokaryotic and eukaryotic cells*

PROKARYOTIC CELL	EUKARYOTIC CELL (Animal)
Simple structure Size: 1 to 5 µm*	Complex structure Size: 10 to 30 µm
They have no nucleus or nucleolus	They have nucleus and nucleolus
No presents system of membranes	Presents system of membranes
No organelles	Presents organelles with defined functions
Cell wall	Cell membrane
Double circular DNA	Helical double DNA
DNA with few genes	DNA with many genes
In most cases the Genes have no introns	Genes have introns and exons
DNA is packaged forming a circular structure	DNA is packaged forming chromosomes
Simple division process	Mitosis division process
For the most part, they are asexual organisms. They do not have mechanisms for training gametes or true fertilization.	They perform division by meiosis, which allows the formation of gametes.
Structure cell phone typical of Bacteria	Structure cell phone typicalof protists, mushrooms plants and animals.
Simple locomotion processes	Processes of locomotion complex

Source: Own-made (2020)

1.7 IMPLEMENTATION QUESTIONS

1. Graphically describe the characteristics and differences between a prokaryotic cell and a eukaryotic cell.

2. In a diagram of ideas or a mind map describe the characteristics and differences between an animal cell and a plant cell.

3. How important are cells in sustaining life? Briefly explain your answer.

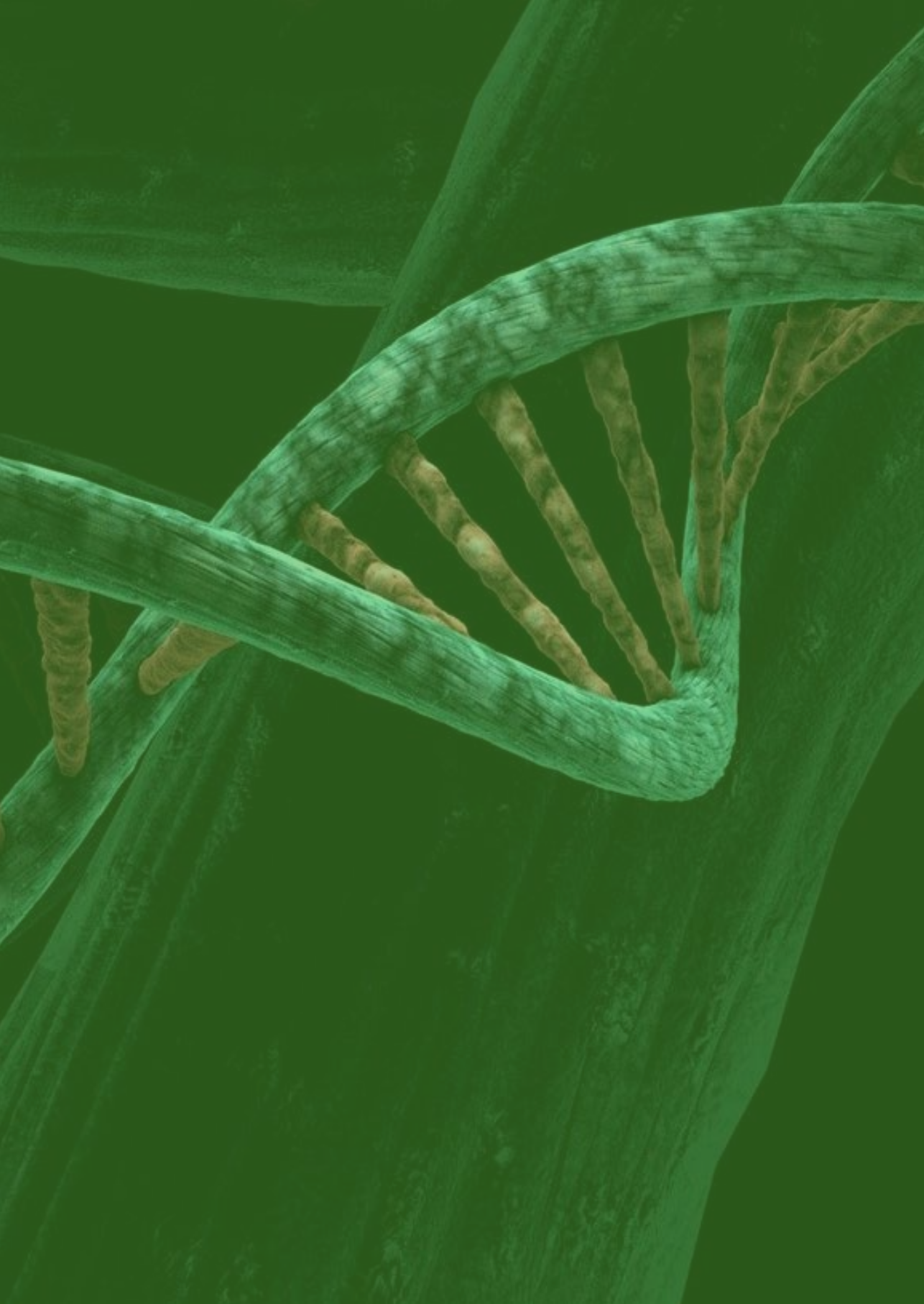
4. Why is it said that prokaryotic cells gave rise to eukaryotic cells? Briefly explain your answer.

5. The cell is said to be the fundamental, structural and origin unit of every living being. Why? Briefly explain your answer.

6. In a timeline describe the main scientists who have contributed to the study of the cell.

7. In a table or chart, describe the functions of each organelle in a eukaryotic cell.

8. Why is it said that the nucleus is perhaps one of the most important organelles in a eukaryotic cell? Briefly explain your answer.



CHAPTER 2
DNA CONDENSATION

2.1 CELL CYCLE

The cell cycle is the period of time that cell takes to perform a division until the onset of the next one; this process consists of four phases identified as G0, G1, S, G2, and M phase.

In G0 phase the cell prepares to perform division; small growth is evidenced at the nuclear and cytoplasmic level.

In the G1 phase the genetic material (DNA) begins to condense; migration of unnecessary cellular organelles occurs during division.

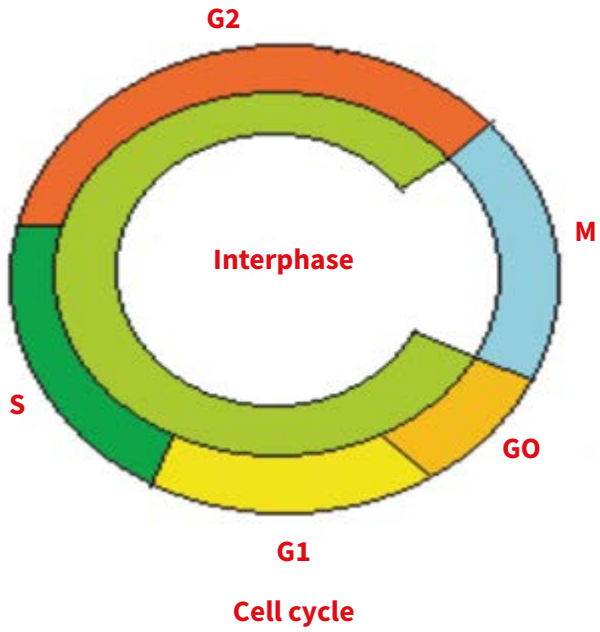
In the S phase the synthesis of genetic material occurs; that is, new DNA fibers are formed, that is, chromosomes that were previously seen as unit filaments now appear duplicated (composed of two chromatids).

In G2 phase a maximum condensation of genetic material occurs; cell growth at the level of nucleus and cytoplasm continues to be evidenced. The cell is ready to initiate cell division.

In the M phase the final division of the cell occurs. In general, it can be concluded that the cell cycle contemplates two phases: the interphase, which contemplates the G1, S and G2 stages and the M phase or mitosis.

Two of the processes of cell division that are related to this cycle are explained below: mitosis and meiosis.

FIGURE 7. *Stages of the cell cycle*



Source: Author's own creation, (2018)

2.2 MITOSIS

It is a process of cell division characteristic of both haploid and diploid organisms and ensures that each of the cellular products (two) receive exactly the same amount of genetic information from the cell from which they precede, that is, from the progenitor cell; this process is carried out in a series of stages that are part of the cell cycle and include: interphase, prophase, metaphase, anaphase, telophase and cytokinesis.

2.3 MEIOSIS

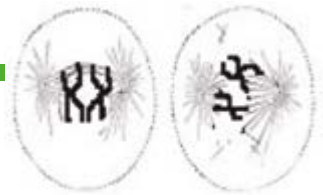
It is a process of cell division by which both male and female gametes or sex cells are obtained; it is characterized by presenting two continuous divisions: in the first division (meiosis I) a reduction of genetic material is presented, for which it is known as a reductional type division; the second division (meiosis II) preserves the genetic material, for which it is known as a conservative type division. By this process four haploid cells are obtained for each diploid cell that enters the division.

FIGURE 8. *Stages of mitotic cell division*

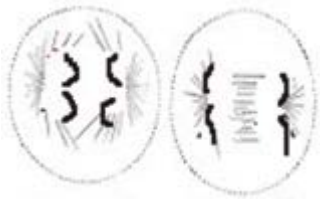
At the interface the cell duplicates its genetic material, grows and prepares the structures and proteins needed to carry out the division.

In the metaphase the chromosomes are directed to the equatorial plane of the cell and the achromatic spindle appears, which originates from each centriole and is fixed to the centromeres of each chromosome; in this same stage the centromeric division is carried out.

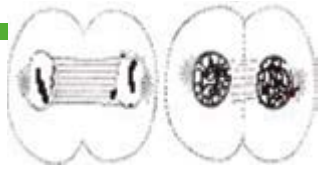
In the prophase the centriole of the cell duplicated and each one is directed to one of the poles of the cell; the nuclear membrane disintegrates and the chromosomes condense and make their double structures visible.



In the anaphase, given the centromeric division of the previous stage, the chromatids that form each chromosome are separated and directed to each cell pole; this happens by condensation of the fibers of cytoplasmic use.



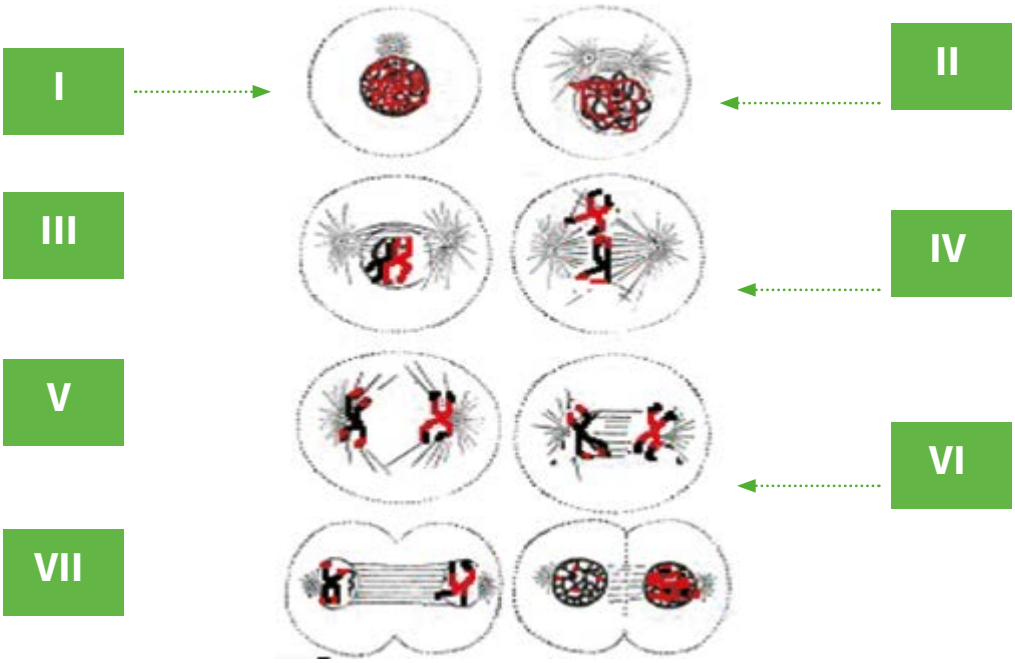
In the telophase a new nuclear membrane begins to form around each chromosomal complement and cytokinesis begins.



In the cytokinesis, which is the last stage, the cell's cytoplasm is divided and two new products are obtained that are genetically identical, that is, with the same amount of DNA. This process ensures that each somatic cell has exactly the same number of chromosomes.

Source: Author's own creation, (2018)

FIGURE 9. *Stages of the meiotic division process*



Source: Author's own creation, (2018)

The stages that happen in meiosis are:

I. **Interphase.** Before starting the division process, the cell enters a premeiotic stage; it is the longest of the process. In this phase the cell duplicates its genetic material, grows and prepares the structures and proteins necessary to carry out the division.

II. During the **prophase I** the centriole of the cell is duplicated and each one is directed to one of the poles of the cell; also the nuclear membrane disintegrates and the chromosomes condense and make visible their double structures.

III. In the same **prophase I** the homologous chromosomes synapse (in all their thickness) to form tetrads; crosslinking occurs (genetic material is exchanged) and the nuclear membrane degrades; at this same stage the homologous chromosomes once crosslinked and exchanged genetic material, separate from their counterparts and travel through the cytoplasm.

IV. In the **metaphase I** the tetrads of chromosomes align in the equatorial plane of the cell and no centromeric division occurs, resulting in the non-division of the chromosome.

V. y VI. and VI. In the **anaphase I** the homologous chromosomes are separated and moved to opposite poles of the cell; this process occurs thanks to the condensation of the fibers of cytoplasmic use. Note that the sister chromatids remain joined by their centromeres.

VII. In the **telophase I,** cell cytokinesis begins and an equatorial plaque begins to form around each chromosomal complement.

INTERKINESIS. In this small lapse (not to be confused with an interphase) each cell product does not duplicate its DNA and a small decondensation of the chromosomes occurs; before each cell completes its nuclear membrane the second meiotic division begins, which begins with a short prophase II in which centrioles reappear and migrate to opposite poles of the cell; at this stage there is no longer exchange of genetic material.

VIII. In **metaphase II** the chromosomes are directed towards the equatorial plane of the cell; the achromatic spindle appears, originating from each centriole and fixed to the centromeres of each chromosome; in this same stage the centromeric division is carried out.

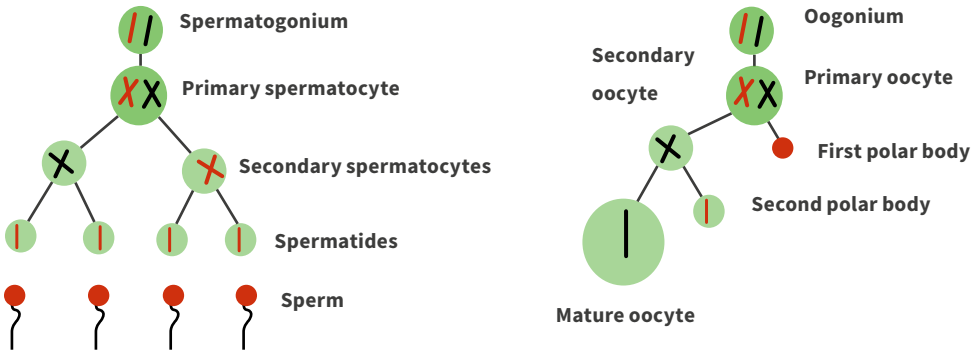
IX. In **anaphase II**, given the centromeric division of the previous stage, the chromatids that form each chromosome are separated and directed to each cell pole; this happens by the condensation of the fibers of cytoplasmic use.

X. In **telophase II**, cell cytokinesis is carried out and two new nucleus are formed from each cell; this gives rise at the end of the process to four completely haploid and genetically different gametes or spores (which is due to the chromosomal exchange carried out in prophase I of the first meiotic division).

2.4 GAMETOGENESIS

Gametogenesis is the process of cell division by which highly specialized cells are produced for fertilization. This type of cell is known as gametes and can be of two types: male gametes, which are produced by spermatogenesis, and female gametes, which are produced by means of ovogenesis. Below are two schematics that illustrate these two processes.

FIGURE 10. *Spermatogenesis and Oogenesis*



Source: Author's own creation , (2018)

2.5 QUESTIONS AND APPLICATION ISSUES

1. Make a comparative table where you list the main characteristics and differences between:

- **a.** Mitosis and meiosis
- **b.** Spermatogenesis and Ovogenesis

2. Research some genetic diseases or disorders that may be caused by mitosis and meiosis malfunction.

3. An animal cell possessing 50 chromosomes at one stage of the G1 cell cycle wishes to perform the gametogenic process known as spermatogenesis.

- **a.** How many chromosomes will the primary spermatocyte have?
- **b.** How many chromosomes will the secondary spermatocyte have?
- **c.** How many chromosomes will the formed sperm have?
- **d.** How many chromatids will the primary spermatocyte have?
- **e.** How many chromatids will the secondary spermatocyte have?
- **f.** How many chromatids will the sperm have formed?

4. Suppose that by some error produced in the environment a cell does not perform recombination between homologous chromosomes. What conclusions could you mention regarding this fact?

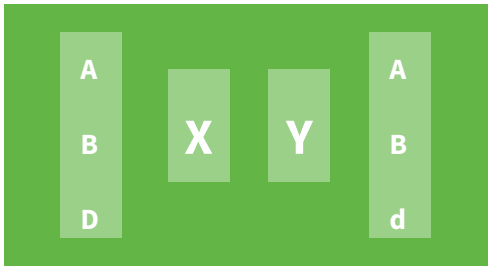
5. Suppose that 200 spermatogonies initiate the spermatogenic process.

- **a.** How many secondary sperm would be obtained at the end of the first meiotic division?
- **b.** How many sperm would be obtained at the end of the process?

6. Explain through schematics the following types of cell reproduction or division in plants:

- **a.** Sporulation
- **b.** Gemation
- **c.** Alternating generations

7. Suppose you have the following cell from an amacho at meiotic interphase with four genetically labeled chromosomes with the following genes:



Note. Please note the following:

- Homologous chromosomes are those that can exchange genes (synapse)
- Sex chromosomes do not exchange genes. Gene exchange is linear
- The chromosome exchanges genes in all their thickness

a. Outline all possible meiotic products that would be obtained at the end of the normal process.

b. Suppose that for some unknown cause the prophase I synapse is not performed. What would be the possible meiotic products that would be obtained at the end of the process?

8. Describe in a table the main characteristics and differences between mitosis and meiosis.

9. Describe in a table the main characteristics and differences between oogenesis and spermatogenesis.

10. Describe at least 10 diseases or genetic problems related to malfunctioning cell division and briefly explain what each of them consists of.

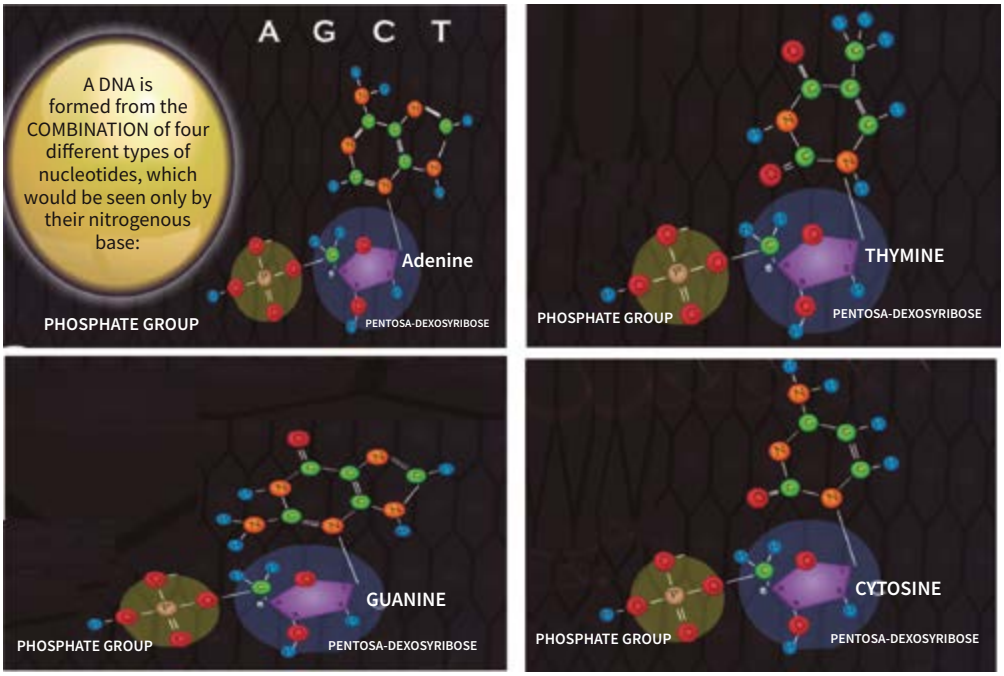
CHAPTER 3

BASIC CONCEPTUALIZATION OF DNA, RNA AND GENE

3.1 DNA

Deoxyribonucleic acid is considered as the chemical molecule responsible for inheritance in living beings; this complex chemical molecule is constituted by other molecules known as nucleotides, which, in turn, are constituted by other small chemical molecules called nitrogenous bases (adenine A, guanine G, cytokine C and Thymine T), phosphate group and a sugar, which for this case is deoxyribose.

FIGURE 11. *Molecular structure of DNA nitrogenous bases*



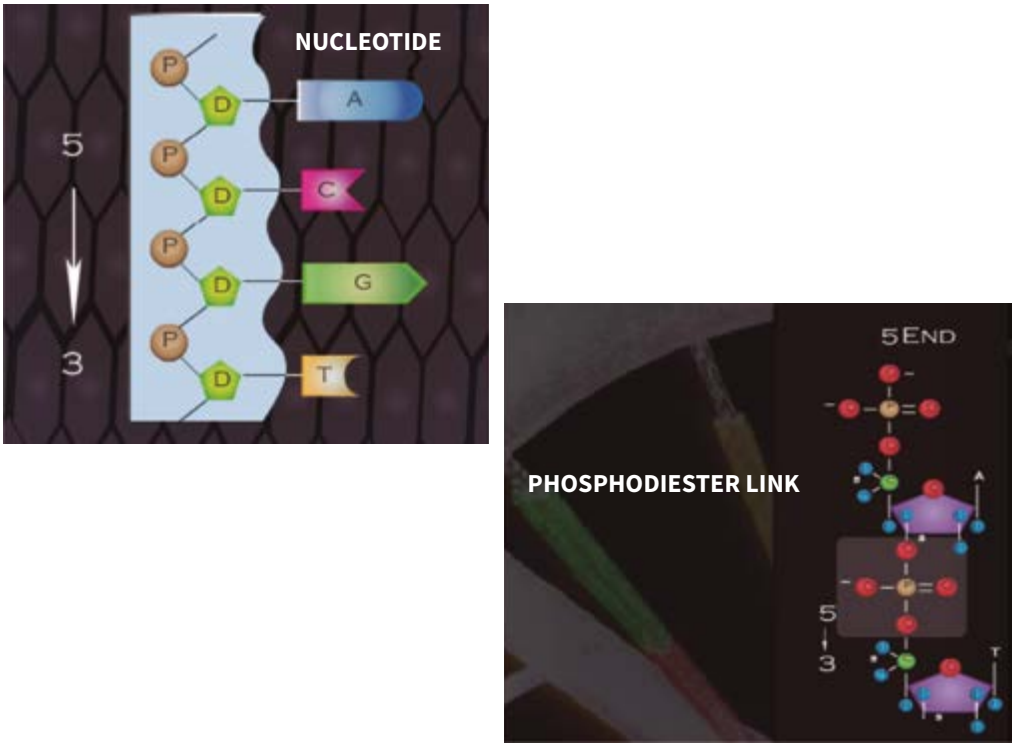
Source: Author's own creation, (2020)

DNA, considered the genetic material of all living organisms and a vast range of viruses, is one of the most complex biomolecules known; its nucleotide sequence contains the information necessary for a living being to monitor, control and carry out metabolism.

DNA carries the information needed to direct protein synthesis and replication. In almost all cellular organisms, it is organized in the form of chromosomes, located in the nucleus of the cell.

DNA is formed by the binding of many deoxyribonucleotides. The vast majority of DNA molecules have two chains that are wound like a spiral staircase; these chains are called antiparallel chains (one goes in the 5'-3' direction and the other in the 3' 5' direction), contain nitrogenous bases, phosphate groups and sugar and are joined together by hydrogen bonds or bridges (Burriel, 2018).

FIGURE 12. *DNA chain 3' 5'*

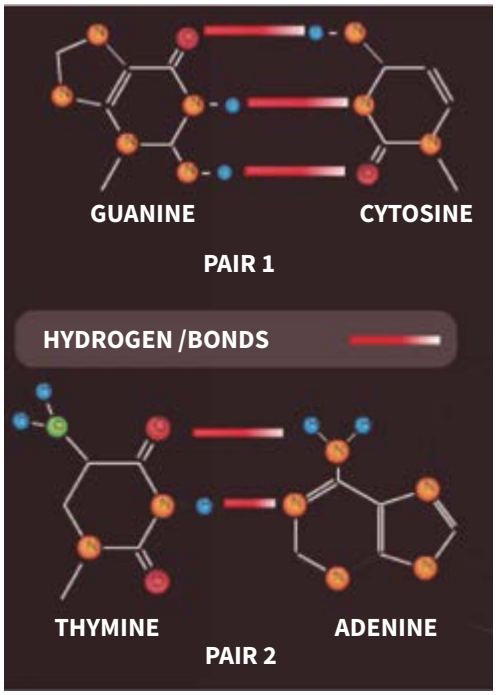


Source: Author's own creation, (2020)

Adenine is linked to thymine by means of two hydrogen bonds or bridges while guanine is linked to cytosine by means of three hydrogen bonds.

The study of its structure can be evidenced in different levels of organization, the primary, secondary, tertiary and quaternary structure, as well as in different levels of higher order packaging.

FIGURE 13. *Binding of nitrogenous bases by means of hydrogen bridges in the DNA molecule*



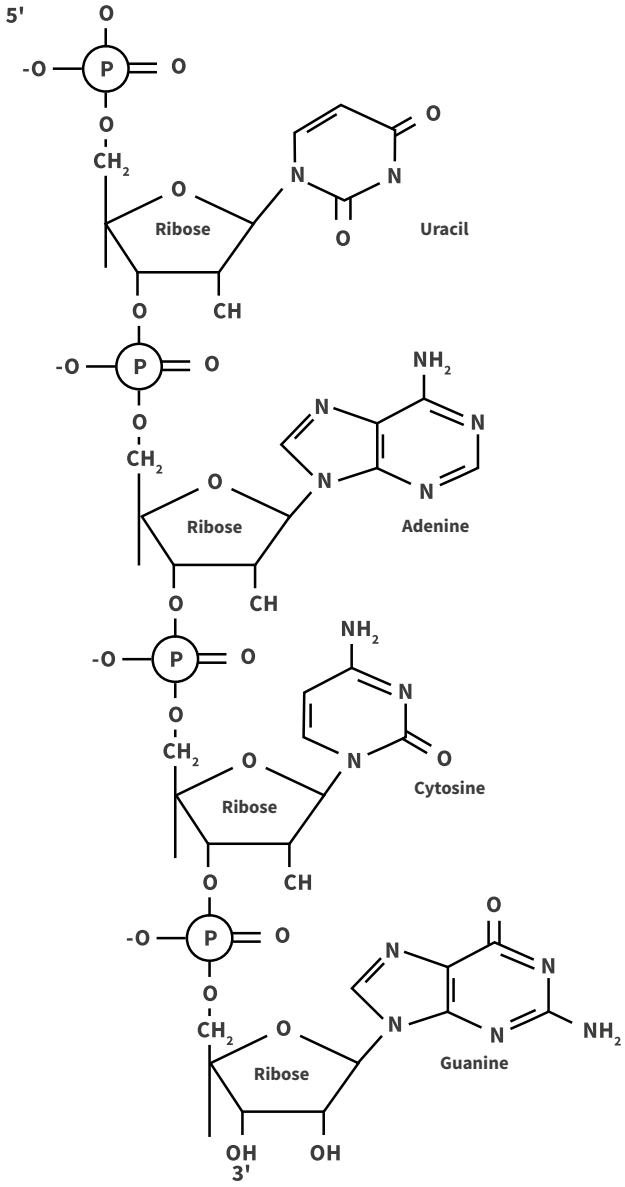
Source: Author's own creation, (2020)

Primary structure

This structure is done to one of the deoxyribonucleotide chains that form the structure of DNA. Genetic information is contained in the exact order in which nucleotides are found.

The nitrogenous bases that are found forming the DNA nucleotides are adenine, thymine, guanine and cytosine. The nucleotides are joined to each other by means of the phosphate group of the second nucleotide, which serves as a bridge between the 5' carbon of the first nucleotide and the 3' carbon of the next nucleotide. Since the first nucleotide has 5' carbon free and the next nucleotide has 3' carbon free, the nucleotide sequence is said to be ordered from 5' to 3' (5' → 3') (Burriel, 2018).

FIGURE 14. *Primary structure of DNA*



Source: Coll, V. B. (2007)

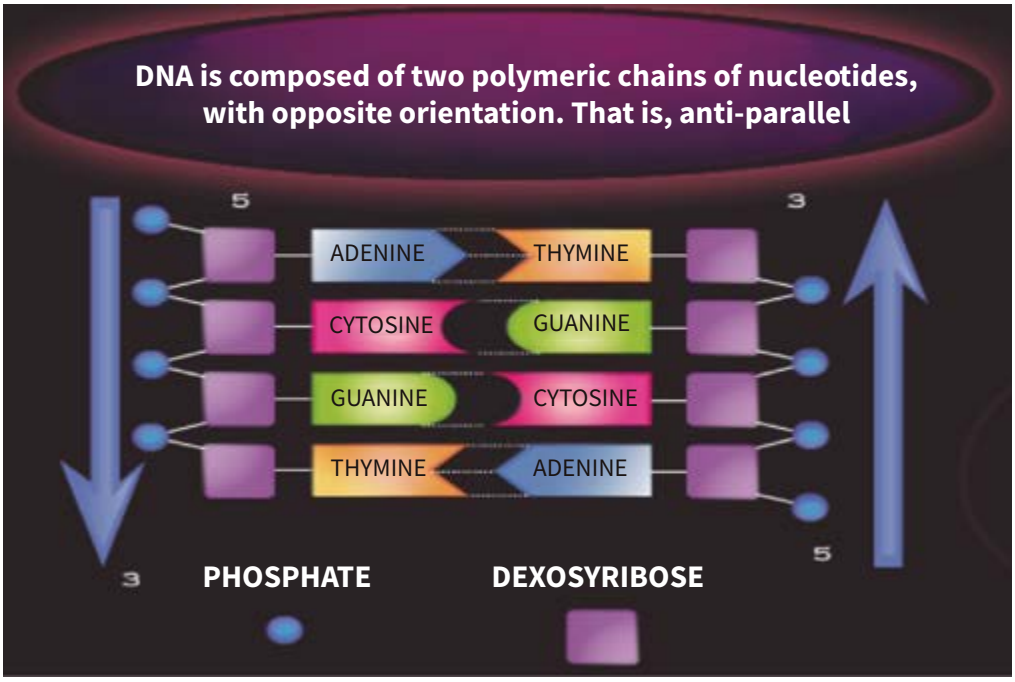
Secondary structure

It corresponds to a double helix structure that allows explaining the storage of genetic information in the DNA molecule. This structure was postulated by scientists James Watson and Francis Crick in 1953 and also the mechanism of DNA duplication.

It is a double strand, dextrogyric or levogyric, depending on the type of DNA. Both chains are complementary; in this case the adenine of one of the chains binds with the thymine of the other chain and the cytosine of one of the chains binds with the guanine of the other chain. It must be remembered that this union occurs through bridges or hydrogen bonds. Both chains are antiparallel as the 3' end of one chain faces the 5' end of the other chain.

The two strands are wrapped around an imaginary axis that rotates counter-clockwise. The structure allows strands that are formed by DNA duplication to be complementary copies of each of the existing strands.

FIGURE 15. *Complementarity of nitrogenous bases in the formation of DNA in its primary structure*



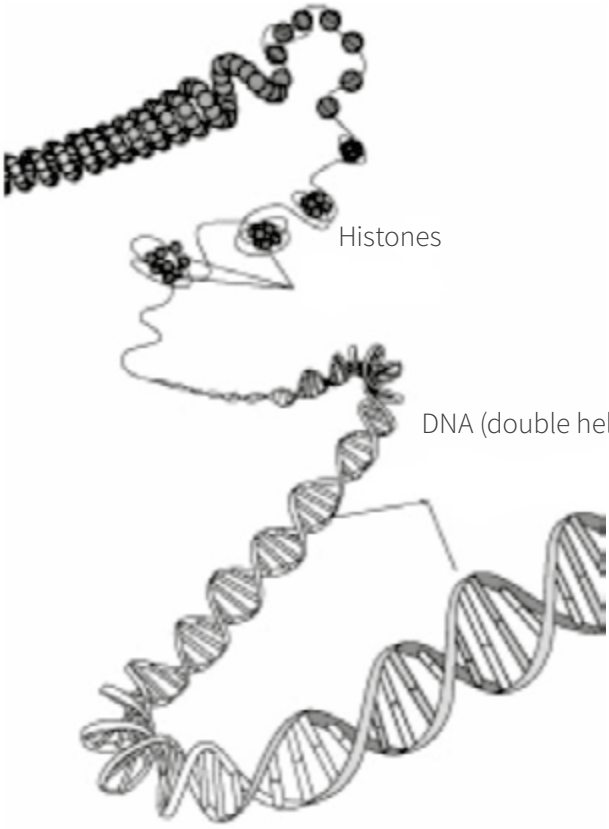
Author's own creation (2020)

Tertiary structure

The tertiary structure of DNA is that the 20 Å fiber, which is twisted on itself, forms a kind of superhelix. This structure is commonly known as supercoiled DNA and is due to the action of enzymes called topoisomerases-II. This coiling of DNA gives stability to the molecule and allows its packaging because it reduces its length.

This newly formed structure has a repetitive appearance similar to the way a pearl necklace is constructed, where the pearls would be the nucleosomes, joined by the linker DNA.

FIGURE 16. *Tertiary structure of DNA*



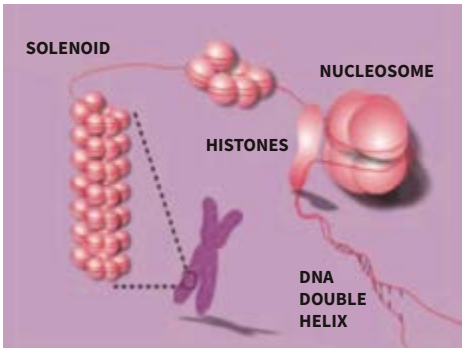
Source: Coll, V. B. (2007)

Quaternary structure

In this structure, the chromatin within the core has a thickness of 300Å. The 100Å chromatin fiber is packaged forming a 300Å chromatin fiber. The coiling that undergoes the nucleosome set is called the Solenoid.

Solenoids begin to coil into a new nuclear structure called chromatin, which remains almost invisible until the cell decides to initiate cell division. When the cell begins the process of cell division, DNA undergoes its maximum condensation or compaction, forming the chromosomes, which are visible under the optical microscope with the help of specific dyes. (Burriel, 2018).

FIGURE 17. *Quaternary structure of DNA*

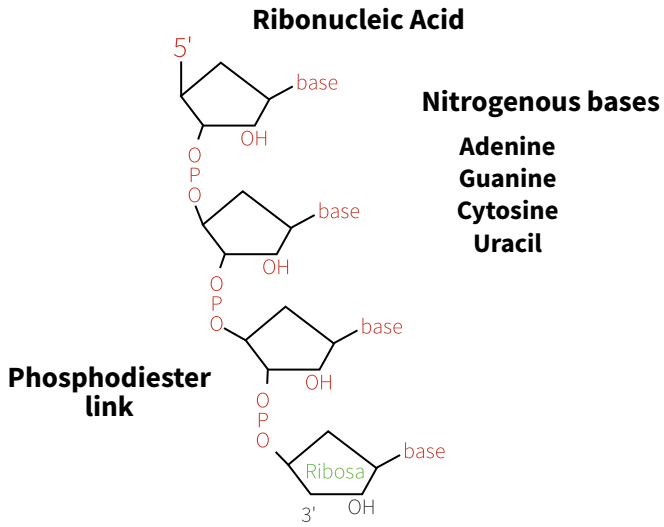


Source: Coll, V. B. (2007)

3.2 RNA

RNA is known as ribonucleic acid and is made up of nucleotides, which are made up of nitrogenous bases (adenine, uracil, guanine, and cytosine). It is simple or single stranded and copied from one of the strands of DNA during the transcription process; it is important to note that this molecule is the one that carries the message during translation in the complex process called protein synthesis. RNA does not contain heritable genetic information and its importance lies in being the carrier molecule of the message that finally translates into proteins, whose process we will not address in this text (Burriel, 2018).

FIGURE 18. *Structure of RNA*



Source: Coll, V. B. (2007)

There are different types of RNA; the most well-known include the following:

- mRNA, known as messenger RNA, is a type of RNA that transmits DNA-coding information by serving as a guideline for protein synthesis.
- tRNA, known as transfer RNA, is a type of RNA that carries amino acids for protein synthesis
- rRNA, or ribosomal RNA, which as the name suggests is located in ribosomes and helps read mRNAs and catalyze protein synthesis.

Some of the differences between DNA and RNA have already been mentioned; for example, that DNA is double-stranded and RNA is single-stranded. Other differences are:

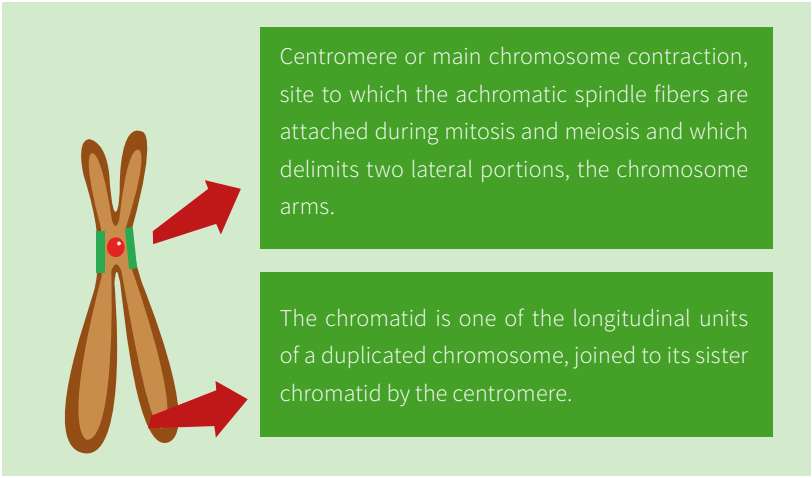
- The sugar in them is different. For DNA it is deoxyribose and for RNA it is ribose.
- In the nitrogenous bases of RNA the nitrogenous base thymine is replaced by uracil, with adenine, guanine, cytosine and uracil being then; in this case, adenine binds with uracil and guanine with cytosine.
- The molecular weight of RNA is less than that of DNA.

3.3 GENE

In classical genetic terms, the gene is defined as the elemental unit of inheritance, the physical and functional region that controls a particular hereditary characteristic, the carrier of genetic information from one generation to the next, and the one that ultimately governs the characteristics of a particular trait.

Genes are normally organized into cellular structures visible under the optical microscope called chromosomes, which are structurally formed by two chromatids (supercoiled DNA); these genes are arranged along both chromatids, occupying on the chromosome a certain position or place called the locus. The total set of genes carried by an individual on each and every chromosome in the cell nucleus is called the genome (EcuRed, 2018).

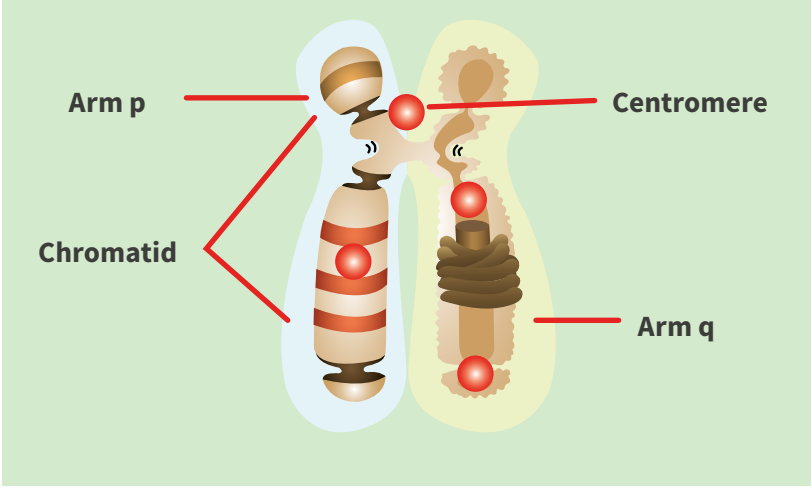
FIGURE 19. *Chromatid*



Source: Author's own creation, (2020)

Chromosomes containing all heritable genetic information are given different names depending on the location of the centromere (point of union of the two chromatids); likewise, both the p arm of the chromosome and the q arm can change in length depending on the site where their centromere is located, so we can find the following types:

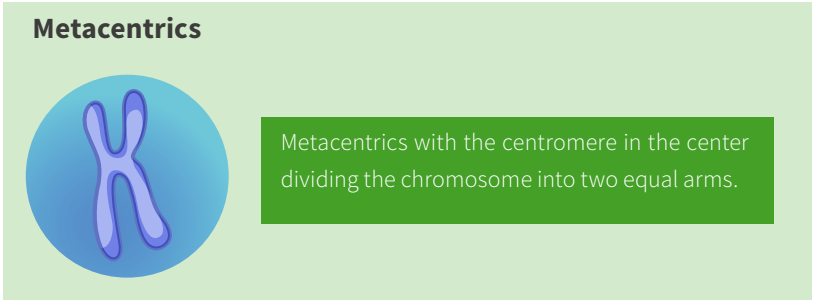
FIGURE 20. *Structure of a chromosome*



Source: Author's own creation, (2020)

Metacentric chromosomes. They are those whose centromere is located just in the middle of the two chromatids; in this case both the p arm of the chromosome and the q arm have the same dimensions.

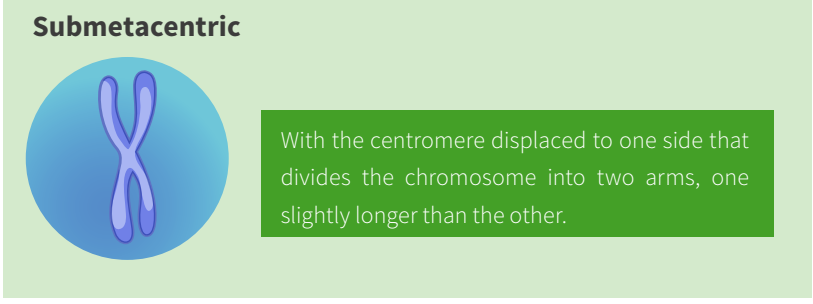
FIGURE 21. *Structure of a metacentric chromosome*



Source: Author's own creation, (2020)

Submetacentric chromosomes. They are those whose centromere is located more towards the end of the p-arm of the chromosome; hence the p-arm is shorter than the q-arm.

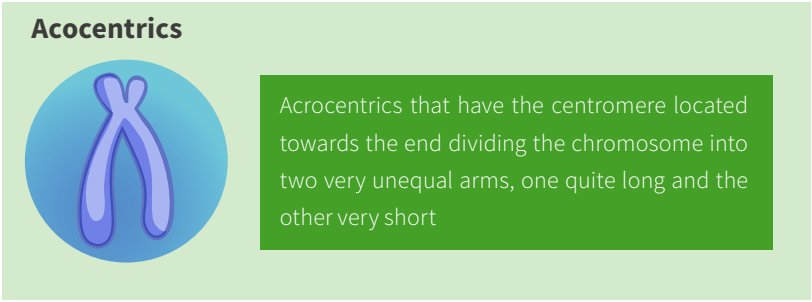
FIGURE 22. *Structure of a submetacentric chromosome*



Source: Author's own creation, (2020)

Acrocentric chromosomes. They are those whose centromere is located more towards the end of the p-arm of the chromosome (there is almost no p-arm); therefore the p-arm is much shorter than the q-arm.

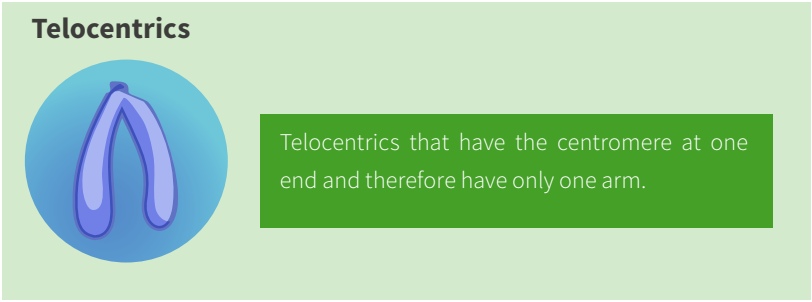
FIGURE 23. *Structure of an acrocentric chromosome*



Source: Author's own creation, (2020)

Telocentric chromosomes. They are those whose centromere is located entirely at the p-end of the chromosome; that is, on this type of chromosome there is no p-arm, only q-arm.

FIGURE 24. *Structure of a telocentric chromosome*



Source: Author's own creation, (2020)

3.4 APPLICATION QUESTIONS

The following questions aim to assess the knowledge acquired through the application of concepts to solve specific situations.

1. On 28 June 2000, several newspapers announced the first report on the Human Genome project. The news raised a big question: What now? What does this mean? The news at the time contained expressions such as: "deciphering the code of life", "sequencing the 3.1 trillion letters of the genetic code", "completing the draft of the work, with about 97% of the genome", "known the language with which God

created life". Do you consider that the search for meaning in this information requires the prior construction of knowledge about the structure and function of the gene and nucleic acids? Support your answer.

2. In the film Dinosaur Park a scientist creates new generations of dinosaurs in the laboratory, extinct 65 million years ago, by means of blood preserved in mosquitoes that would have bitten them and remained fossilized in amber. With the blood it was possible to determine the DNA of the dinosaurs, thus arriving at the formula to recover the species. Consider the possibility that the DNA obtained is not of optimal quality and only the RNA molecule is in optimal condition. Would it be possible to recover the species? Justify your response.

3. People with cystic fibrosis inherit faulty genetic information and cannot produce normal CFTR proteins. Scientists have used gene therapy to introduce normal segments of DNA encoding the missing CFTR protein into the lung cells of people with cystic fibrosis. It is correct to say that a result that is NOT typical of such therapy is:

a. Altered lung cells can make the normal CFTR protein.
b. The altered lung cells can divide to produce other lung cells with the normal CFTR gene.
c. The normal CFTR gene can be expressed in altered lung cells.
d. The offspring of someone who has altered lung cells will inherit the normal CFTR gene.

Support your answer.

4. Among individuals in a population there is variability in their characteristics, leading to differences in the ability to survive and reproduce. Why is this? Explain your answer.

5. Three of the following five statements about DNA and RNA are true.

a. DNA is composed of deoxyribonucleotides and ribonucleotide RNA.
b. Both DNA and transfer RNA have a double chain.
c. DNA is usually synthesized from DNA and RNA is transcribed from DNA.
d. DNA duplication and RNA transcription involve the same enzyme polymerases.
e. Hydrogen bonding with complementary bases is important for good duplication, transcription and translation.

What are the three correct statements? Justify your response.

Select the correct answer and support your choice for each of the following questions:

6. Nucleic acids are of paramount importance in the storage and transfer of information used in the synthesis of proteins and other molecules. These biological molecules are constituted as follows:

a. Inorganic reserve and transfer molecules
b. Energy storage molecules
c. Non-metabolizable energy molecules
d. Different molecules for transferring genetic information

7. DNA has indications for protein synthesis. If an individual carries a modification in the part of the DNA that encodes the enzyme lactase, responsible for dividing lactose into two monosaccharides, in the individual it may occur that:

a. Unable to properly absorb milk
b. Unable to properly assimilate meats
c. Improve your digestion by drinking milk
d. Unable to perform the Krebs cycle

8. When a cell divides, DNA molecules must replicate; this means that:

a. The replication process is based on the specificity of the combination between the nitrogenous bases
b. The process starts with a separation of the chains
c. They must make exact copies of themselves
d. Each chain acts as a mold to synthesize another complementary chain

9. Genetic information stored in DNA, which is translated into an amino acid sequence and then proteins, is known as genetic code. Its presence in all organisms allows to affirm that these probably:

a. Share a common ancestor
b. Have cells with internal membranes
c. Produce the same type of protein
d. Reproduce sexually

10. A mutation is the change of one or more DNA nucleotides in an individual. If the mutation is expressed in the change of a phenotypic characteristic of the individual it can be said that:

a. Changed chromosome number
b. Haploid cell formation
c. No protein synthesis occurred
d. A different protein was synthesized than expected

DIDACTIC UNIT

MODELS OF MENDELIAN GENETICS

Gustavo Forero Acosta

CHAPTER 4
MENDEL'S GENETICS

Gregor Mendel was born in 1822 in Silesia. After finishing his secondary education, he suffered physical and economic hardship and decided to enter a profession "that would rid him of the bitter necessities of life" (as he wrote himself), so that at 21 he became a monk.

He was assigned an interim professor assignment and retained that category because he failed the exams to achieve a definitive position. It has been said that the encouragement that prompted Mendel to start his experiments was a discussion with one of his botany professors in the last of the exams he held without success.

4.1 INTRODUCTION TO GENETICS

Since the beginning of mankind, man has always been concerned with knowing the phenomena that affect the transmission of the characteristics that identify descendants from one generation to another such as eye color, hair, skin, size, the transmission of hereditary diseases, the conformation of different races and their crosses in both humans and plants and animals, the development of pure lines, the spread of plants and mutations and improvement. These aspects, among others, were not considered as such from the beginning, only until 1860 when Mendel (Augustinian monk), discovered the inheritance patterns around seven characteristics that appeared in seven different varieties of the pea; he observed that these characters were inherited independently and determined that each parent has pairs of units but that only one contributes to each couple of their offspring (Forero, G. 2005)

Genetics has become one of the emerging scientific disciplines that has managed to gain a preponderant place in the scientific community not only internationally but also nationally. He has obtained recognition as a young science dedicated to investigating organized genetic material thanks to the construction of theoretical-practical models founded from elements of the development of other basic and instrumental sciences that give permanent support to the development of this science.

Research in genetics has been permanently nourished by the results obtained from the work carried out by cellular and molecular biology, biochemistry and the contributions of molecular genetics, which has generated a systematic and rigorous dynamic of formulation and construction of research models rather than genetic theories. In this sense it is important to highlight that the work model in genetics is considered as an organized structure that describes, explains and, depending on its degree of maturity, predicts different options and realities in the field of inheritance and animal developing in a source of hypotheses contrastable with practice.

In general, the function of this material is to provide both the tutor and the student with the basic tools necessary for the understanding, interpretation and application of the basic concepts of genetics and to highlight the importance that this area represents in the programs of selection, crossing, improvement, production and conservation. In the same way, it is expected that with the development of this course the professional will take ownership of the genetic concepts in a real and effective way and propose alternatives of solution to the serious problem that from the agricultural and livestock perspective faces the country today (Forero, 2005).

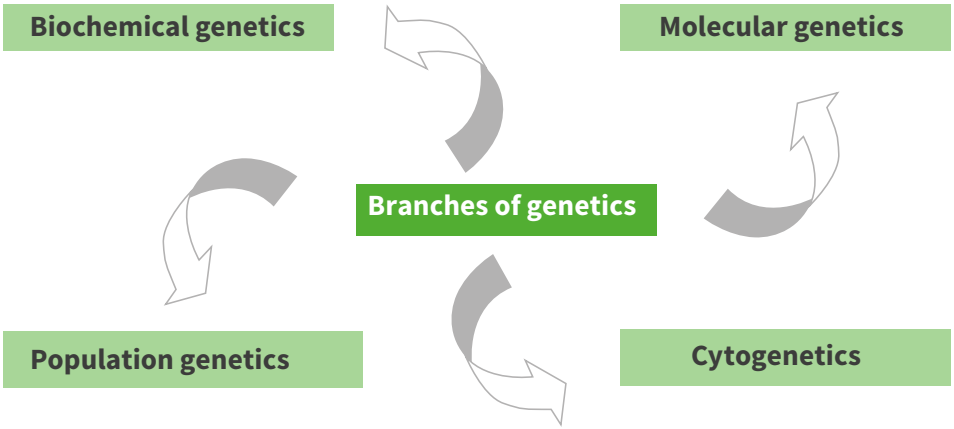
4.2 GENETICS BEFORE AND AFTER MENDEL. CONCEPT OF GENETICS

Genetics is considered to be the science that studies the inheritance, transformation, and variation of genetic material; in other words, it is the science that studies inheritance in animals, plants, and all living things.

Branches of genetics

Genetics basically comprises at a general level four important branches which are: biochemical genetics, molecular genetics, population genetics and cytogenetics.

FIGURE 25. *Branches of genetics*



Biochemical genetics

It treats metabolic disorders due to inherited chemical or enzyme defects. It was founded by the English sage Archibald Garrod in 1909 and has clarified a large number of metabolic conditions such as phenylketonuria, alkaptonuria, galactosemia, certain hemolytic anemias, etc.

Cytogenetics

Emerging from the convergence of cytology with genetics, it is the branch of genetics that deals with the study of chromosomes and their aberrations in case of somatic and sexual anomalies.

Molecular genetics

Through the methods provided by molecular biology and recombinant DNA technology, it enables the study of molecules containing biological information and the chemical processes of their transmission and manifestation. This allows genetic counseling to plan the reproductive future and in some cases prenatal detection of possible genetic alterations or conditions.

Population genetics

It covers the study of inheritance mechanisms, the frequency of certain genes, the rate of mutation or change of certain genes, the relative fertilization of individuals with certain genes, and the establishment of genetic chaining.

Modern genetics owes its origin to the discoveries made by Mendel through his experiments with plants, published in 1886, and which currently constitute the universal laws of inheritance. He found that biological characteristics passed down from parent to child were determined by hereditary units that were passed down from generation to generation in a uniform and predictable manner. Their valuable discoveries had to wait for 34 years until three researchers (Hugo de Vries, Carl Correns and Erich Von Tschermark), through individual efforts, confirmed in their experiences the dimension of these. These three discoverers are known as the rediscoverers of the laws of inheritance. Next, we will take a look and describe in a very synthesized way the facts, discoveries and contributions that have been given around genetics and molecular biology over the years; these are:

- In 1000 B.C the Babylonians celebrate with religious rites the pollination of palm trees.
- In 323 B.C Aristotle speculates on the nature of reproduction and inheritance.
- Metaphorical texts on the nature of human reproduction are written in India in 100-300 AD.
- In 1676 sexual reproduction in plants was confirmed.
- In 1677, animal sperm was viewed under a microscope.
- In 1838 It is discovered that all living organisms are composed of cells.
- In 1859 Darwin published his theory on the evolution of species.
- In 1866 Mendel described in the peas the fundamental units of inheritance (which later will be called genes).
- In 1871, DNA was isolated in the nucleus of a cell.
- In 1883 Francis Galton coined the term eugenics.
- In 1887 it was discovered that reproductive cells constitute a continuous lineage different from the other cells of the body.
- In 1908 mathematical models of gene frequencies in Mendelian populations were established.
- In 1909 the fundamental units of biological inheritance were named genes.
- In 1924, the U.S. Immigration Act limited entry to the U.S. on the basis of racial or ethnic origin.
- In 1925 it was discovered that the activity of the gene is related to its position on the chromosome.
- In 1927 it was discovered that X-rays cause genetic mutations.
- In 1931 thirty U.S. states implemented compulsory sterilization laws.
- In 1933 Nazi Germany sterilized 56,244 “hereditary defective subjects”.

- In the years 1933-45, the Nazi Holocaust exterminated six million Jews through its eugenics policy. 1943: DNA is identified as the genetic molecule.
- Between 1940 and 1950 it was discovered that each gene encodes a single protein.
- In 1953 the double helix structure of DNA was proposed.
- In 1956, 23 pairs of chromosomes were identified in cells of the human body.
- In 1966 the complete genetic code of DNA was deciphered.
- In 1972, the first recombinant DNA molecule was created in the laboratory.
- In 1973 the first genetic engineering experiments took place in which genes of one species were introduced into organisms of another species and functioned correctly.
- In 1975, the Asilomar conference evaluated the biological risks of recombinant DNA technologies and approved a moratorium on experiments with these technologies; in the same year, the hybridomas that produce monoclonal antibodies were obtained for the first time.
- In 1976 it was founded in the United States. Genetech, the first genetic engineering company.
- In 1977, a human hormone was successfully manufactured in a bacterium using genetic engineering techniques; in the same year scientists developed the first techniques for rapidly sequencing the chemical messages of DNA molecules.
- In 1978, the human insulin gene was cloned.
- In 1980, the U.S. Supreme Court ruled that genetically engineered microbes can be patented.
- In 1981, the first prenatal diagnosis of a human disease was made by DNA analysis.
- In 1982 the first transgenic mouse (the “super mouse”) was created by inserting the rat growth hormone gene into fertilized mouse eggs; in the same year insulin was produced using recombinant DNA techniques.
- In 1983, the PCR technique was invented, which allows specific genes to be replicated (copied) very quickly.
- In 1984, the first transgenic plants were created.
- In 1985, the use of interferons in the treatment of viral diseases began.
- In 1985 the “genetic fingerprint” was first used in a judicial inquiry in Great Britain.
- Clinical trials of the genetically engineered hepatitis B vaccine were authorized in 1986.

- In 1987, a commercial proposal was made to establish the complete sequence of the human genome (Genome project), consisting of approximately 100,000 genes; in the same year, the first monoclonal antibody for therapeutic use was marketed.
- In 1988, the first patent for an organism produced by genetic engineering was granted.
- In 1989, the first automatic DNA sequencing machines were commercialized.
- In 1990, the first successful gene therapy treatment was given to children with immune disorders ("bubble children") and numerous experimental gene therapy protocols were put in place to try to cure cancer and metabolic diseases.
- In 1993 the first campus in Britain for the study of the human genome is opened.
- In 1994, the first genetically modified vegetable (a tomato) was marketed in California and the reproduction of the first transgenic bull was authorized in the Netherlands.
- In 1995, the first complete genome sequences of organisms were completed: the bacteria Hemophilus influenzae and Mycoplasma genitalium.
- In 1996, the genome sequence of a eukaryotic organism, the brewing yeast "Saccharomyces cerevisiae", was completed for the first time. Moreover, the catalog of human genes that Victor McKusick and his colleagues at John Hopkins University update each week already contains more than 5,000 known genes. The Genome project, coordinated by HUGO (Human Genome Organization) is progressing well.
- In 1997 the first mammal, a sheep named "Dolly", was cloned.
- In 1998, the Human Genome project was evaluated and 2003 was set as the completion date. Venter founded the company Celera Genomics Inc., whose goal is to complete the decoding of the human genome by the end of 2001.
- In 1999, the complete genetic code of human chromosome number 22 was published.
- In 2000 Celera announced that it had 90% of the first draft of the complete human genome ready.
- In 2003, the human genome sequence was completed.
- On December 23, 2005, scientists are now scrutinizing it, as well as the result of another international effort, the largest map to date of single-letter variations in the human genetic sequence, in the hope of being able to see more clearly the history of our species' evolution.

Since 2005, numerous studies have been carried out in molecular genetics, population genetics, cytogenetics, the genetics of viruses and bacteria, animal and plant transgenesis, animal and plant developing and some little progress in terms of immunogenetics and pharmacogenetics, which has allowed countless researches focused on the animal, plant, microorganism and human world to help understand its functioning, its diseases and possible treatments from the study of its various genomes (Forero and Bernal, 2018).

This brief historical review is obviously presented as a panoramic look at both the development of this discipline and the temporal location of the basic discoveries that constitute it.

4.3 THE BIRTH OF MODERN GENETICS

Modern genetics owes its principles to Mendel. His monastery was devoted to the teaching of science and scientific research, so that he was sent to the University of Vienna in order to obtain his teaching degree. However, he failed the examinations and returned to the monastery of Brunn. There he enrolled in a research program on plant hybridization that led him posthumously to be recognized as the founder of genetics. Their findings remain valid today, although exceptions and variants have been found.

Mendel formulated two major principles: the principle of *segregation*, according to which hereditary characteristics are determined by genes, which are presented in pairs, a member of each pair inherited from each parent, and the principle of *independent distribution*, according to which the alleles of one gene segregate independently from the alleles of another gene. Mendel also did not know the biological structures responsible for inheritance; they were known years later when the chromosomal theory of inheritance came to affirm the responsibility of genes in the transmission of hereditary characters and the location of such genes inside the chromosomes of the cell nucleus (Forero, G., & Bernal, L. 2013b).

4.4. THE MENDEL EXPERIMENTS

Mendel studied garden peas (Pisum sativun) as they were cheap and easy to obtain on the market, occupied little space and had a relatively short generation time, produced many descendants, there were different varieties that showed different color, shape, size, etc., so they presented genetic variability; they were also considered as an autogamous species, that is, it is self-pollinated (the pollen of the anthers of a flower falls on the stigma of the same flower); thus, it was easy to cross

between different varieties and it was possible to avoid or prevent autopollination by castrating the flowers of a plant (eliminating the anthers) (Forero, G., & Bernal, L. 20 3b).

These properties contributed to the success of Mendel's work, who, given his purely experimental work, selected seven characters of this pea, which were always going to manifest themselves in his generations or descendants. These are: tall or dwarf plants with green or yellow pods; axial and other terminal flowering plants; green and other yellow seed plants; smooth and other rough seed peas; plants with gray or white pigments; and white or violet flowering plants. Table 2 lists several of the results obtained by Mendel in the crossing of individuals that varied in one character (Forero and Bernal, 2013b).

TABLE 2. *Summary of Mendel's experiments with garden peas (Pisum sativun)*

Parental phenotype (crossing)	F1	F2	F2 ratio
Smooth x rough seed	All smooth	5474 smooth; 1850 rough	2.96:1
Yellow x green seed	All yellow	6022 yellow; 2001 green	3.01:1
Purple x white petals	All purple	705 purples; 224 white	3.15:1
Smooth x rough pod	All smooth	882 smooth; 299 rough	2.95:1
Green x yellow pod	All green	428 green; 152 yellow	2.82:1
Axial x terminal flowers	All axial	651 axial; 207 terminals	3.14:1
High plant x low	All high	787 discharges; 277 casualties	2.84:1

Source: Adapted from Griffiths A., (2002)

4.5 MENDEL'S LAWS

According to all the results obtained experimentally by Gregorio Mendel, his works can be summarized in a very general way in some basic postulates; which are part of what we very commonly know as the basic and fundamental laws of inheritance for the study of genetics: law of uniformity of the hybrids of the first generation, law of segregation and law of independent recombination or independence of non-antagonistic characters.

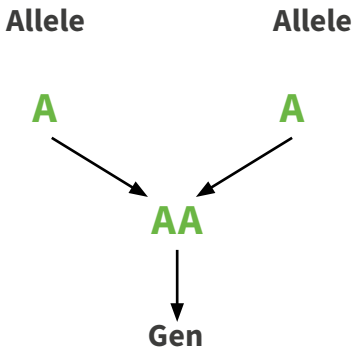
First generation hybrids uniformity law

When two varieties of individuals of pure race or strain (both homozygous) are crossed for a specific character, all the individuals obtained in the first generation are equal, that is, they manifest the same characteristic of one of their parents.

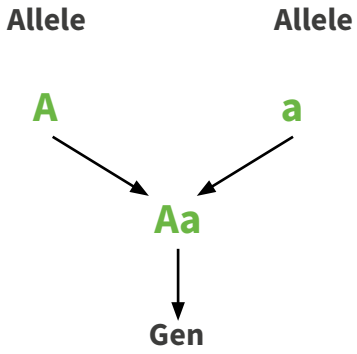
Mendel managed to reach this conclusion by crossing pure yellow seed varieties with pure green seed varieties and always obtained from this crossing the same result; that is, yellow seeds.

Equal Segregation Act

Mendel stated regarding his observations obtained that "the two members (alleles) of a gene pair are distributed separately (segregated) between the gametes; thus, half of the gametes contain one member of the pair, and the other half contain the other member". To understand this principle, we will briefly review some concepts such as gene, allele, phenotype, genotype, heterozygous, homozygous, carrier genotype and pure genotype, which are basic and essential for its understanding.



Gene. It is a portion or fragment of DNA that possesses heritable genetic information; the gene is written in a Mendelian way with the letters of the alphabet; each of them represents one of the alternative forms of the gene called the allele; for our case the Mendelian-type gene is composed of two alleles (Figure 26).

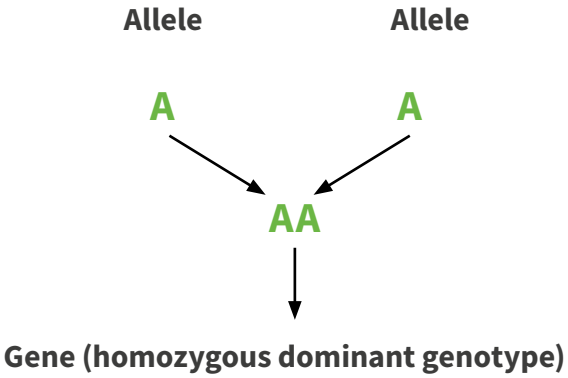


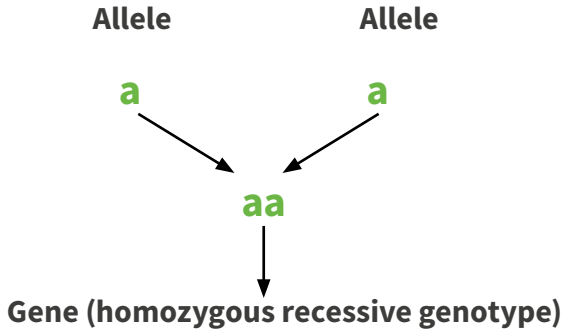
Allele. It is each of the alternate forms that make up the gene and is denoted by the letters of the alphabet.

Phenotype. Generally speaking it can be said that the phenotype is the physical manifestation of the genotype; in other words, they are the measurable, observable and visible (in most cases) characteristics of an individual; for example, when we refer to an individual being green, blue, brown, black eye color, etc., we are referring to the phenotype for eye color. These characteristics or phenotypes are perceptible to the naked eye, while there are other phenotypes that require some specific tests to determine them, such as the case of blood type or Rh factor in humans, etc.

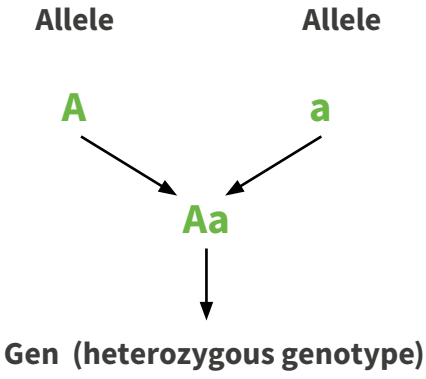
Genotype. It can be defined as the set of genes that a gene possesses throughout its genome, and its manifestation is evidenced through the phenotype.

Homozygous genotype. It is when the gene that the individual carries is constituted by two equal alleles; that is, the letter of each of the alleles that make up the gene, are equal. If the two letters that make up the gene are capital letters the genotype is homozygous dominant and if, on the contrary, the gene is composed of the two lower letters the homozygous is recessive.

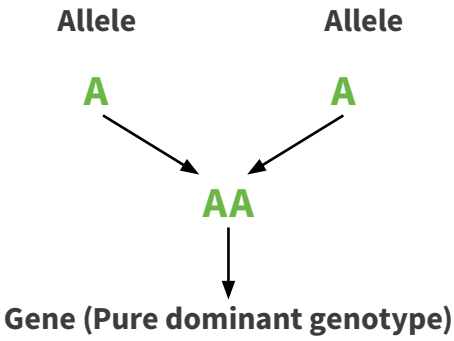


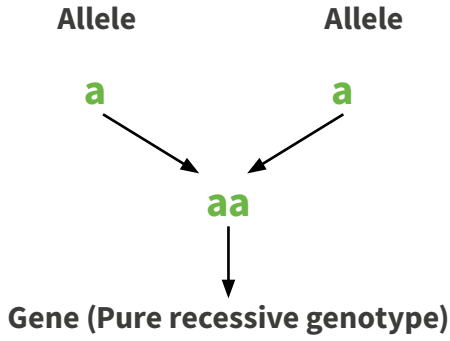


Heterozygous genotype. It is when the gene that the individual carries is constituted by two alleles denoted with different letters, that is, the letter of each of the alleles that make up the gene, are different. According to Mendel, the heterozygous genotype has phenotypic manifestation equal to the homozygous dominant genotype.

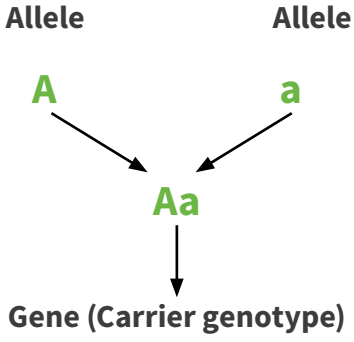


Pure genotype. It is when the gene carried by the individual is constituted by two equal alleles; that is, the letter of each of the alleles that make up the gene are equal. These letters can capital letter (pure dominant genotype) or lower case (pure recessive genotype).

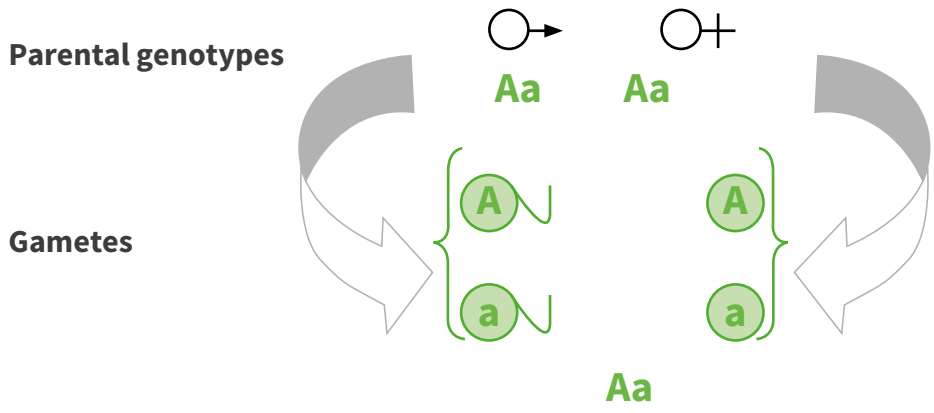




Carrier genotype. It is when the gene that the individual carries is constituted by two alleles denoted with different letters, that is, the letter of each of the alleles that make up the gene, are different.



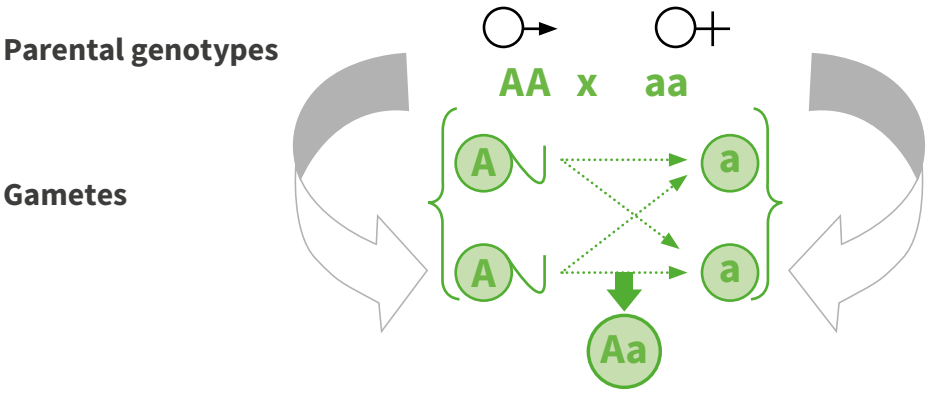
Let's assume that we have a couple of heterozygous genotypes of parental type like this: Aa and Aa; if we cross a couple of individuals with that type of genotypes with each other, that is heterozygous individuals Aa, the gametes that each individual would provide at the time of fertilization would be:



If we interpret one of the results obtained by Mendel reported in table 1 and follow the basic principle of segregation like this: if we cross plants with pure-line yellow seeds (homozygous) with pure-line green plants (also homozygous), all F1 is of yellow phenotype. If we self-cross F1 to produce F2, F2 consists of 6022 plants with yellow seeds and 2001 plants with green seed.

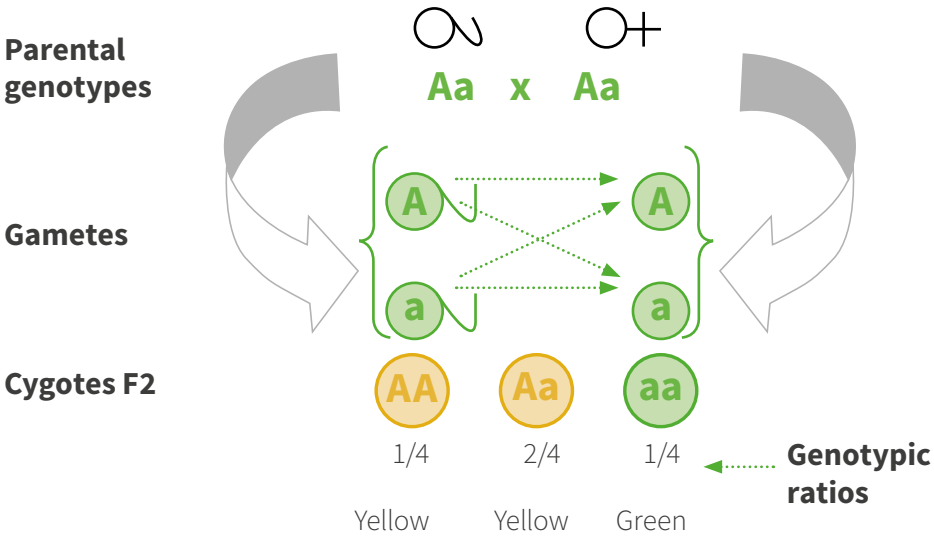
Let's assume that the yellow phenotype is conditioned by the dominant genotypes AA and Aa and the green phenotype by the recessive genotype aa; then:

To obtain F1 we cross a plant of genotype AA (yellow seeds) with another of genotype aa (green seeds) and obtain:



Zygotes (F1 generation): All **Aa** type

Crossing the entire F1 together we have:



In short, we have: 3/4 yellow and 1/4 green Phenotypic ratio

Phenotypic relation: 3:1

If we take into account the values of table 1 and relate them to the phenotypic ratio 3:1 for this crossing we have:

Total plants 8023

We take this value, multiply it by 3 and divide it by 4 like this: 8023 x 3/4 = 6017 plants with yellow seeds. We also do with the other fraction: 8023 x 1/4 = 2006 plants with green seeds.

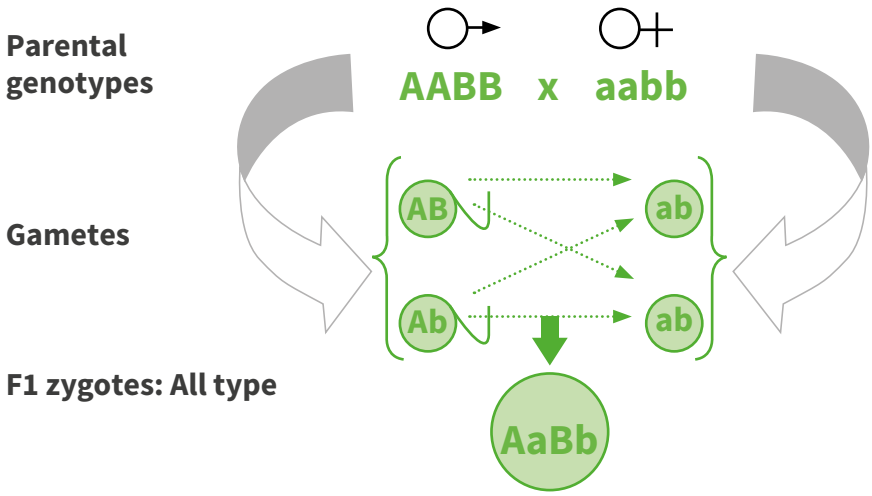
If we look at the value in table 1, we realize that it meets the phenotypic ratio 3:1 since the theoretically calculated values are very close to the values obtained experimentally by Mendel; the same procedure can be used to check the other crosses.

Law of Independent Recombination

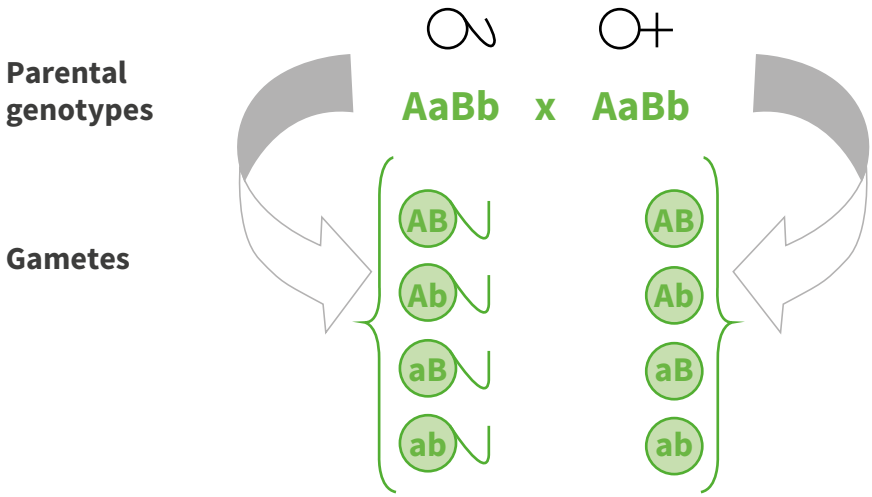
Mendel concluded that “members (alleles) of distinct genes segregate independently during the formation of gametes”. To understand this principle we will assume that plants that differ in two characteristics are interbred; that is, a dihybrid mating is performed.

Let's assume that the yellow phenotype is conditioned by the dominant genotypes AA and Aa and the green phenotype by the recessive genotype aa; the smooth-shaped phenotype is conditioned by the dominant genotypes BB or Bb and the rough seed shape by the genotype bb then:

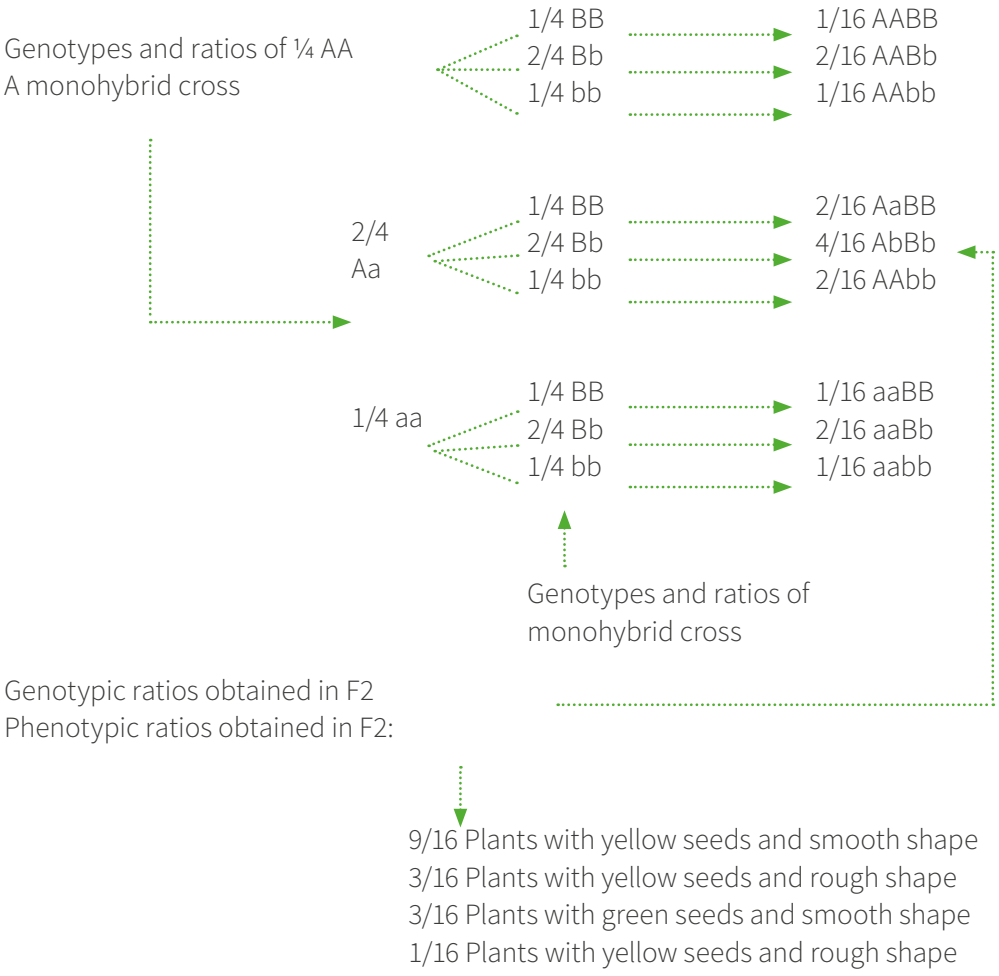
To obtain the F1 we cross a plant of genotype AABB (yellow seeds, smooth form) with another of genotype aabb (green seeds, rough form) and obtain:



Crossing the entire F1 together we have:



In order to determine the zygotes generated in F2, we can use the gametic tree as follows:



The phenotypic ratio translates to: 9:3:3:1

Table 3 summarizes the total number of gametes and zygotes that can be generated by a carrier (heterozygous) individual for one, two, three, and four characteristics.

TABLE 3. *Gametes and zygotes produced by carrier individuals for one, two, three, and four characteristics*

Number of features	Total number of gametes	Total number of zygotes
One (monohybrid)	2	4
Two (dihybrid)	4	16
Three (trihybrid)	8	64
Four (tetrahybrid)	16	256

Source: Author's own creation, (2021)

The same procedure can be used to solve trihybrid, tetrahybrid crosses, etc., types of crossings that will be addressed in more detail in the next chapter. The method of dates is rather complicated and the person who has no experience in it can easily be mistaken.

Nowadays, thanks to these impressive discoveries, it is possible to know the genome of many organisms and to combine genes from cells belonging to different species in the laboratory. This allows organisms to be designed according to the specific needs of humans. To be sure, the possibility of studying and manipulating genes represents a revolution in science that will change the way we see the world and live in the coming years, since its application in medicine, industry and food will bring benefits that are unthinkable until now.

In the next chapter we will address in detail the type of crosses of the Mendelian order, which will most likely be the main basis for understanding, comprehension and applying the basic principles of genetics in conservation, improving and production programs.

4.6 APPLICATION QUESTIONS

1. Explain through a conceptual map the characteristics and main contributions that have made to the development of science each of the disciplines or branches of genetics worldwide.

2. With the help of a map of ideas, briefly explain what each of the experiments carried out by Mendel consisted of and their main results.

3. In a table, describe the three laws of Mendel and highlight the main contributions that each of these postulates gave to the study of modern genetics.

4. In a table, describe 20 phenotypes and their respective genotypes in animals, plants, microorganisms and humans.

5. Briefly define the concept of genotype, phenotype and genome.

6. With the help of a mind map, make a brief bibliographical review of Mendel in which he highlights the main contributions he gave to the study of genetics.

7. Research who Punnett was and what he contributed to the study of genetics.

8. Research which biological models, other than the peas used by Mendel, can be used in the study of genetics.

CHAPTER 5

PROBABILITY AND PROOF OF PHENOTYPIC PROPORTIONS

Probability can be defined as the way or form (relative frequency) in which a given event or event can occur in a given experiment. Relative frequency refers to the number of times that event occurs out of a theoretical total N. If the theoretical frequency of an event is "a" times a total of N times, the probability of the event occurring is P = a/N. The relative frequency of any event varies between 0 and 1.

5.1. LAWS OF PROBABILITY. SUM OF PROBABILITIES

Read the following cases: the color character of the pea seed can not be at the same time yellow and green; a cow in a calving can not give at the same time a veal and a calf. These events are called mutually exclusive.

The probability of one or other of the mutually exclusive events occurring is calculated by adding together the probabilities of each of the events.

That is, if: p is the probability that an event occurs and q is the probability that it does not occur on the same occasion, p + q = 1, where p = 1 − q and q = 1 − p. The equation of p +q =1 constitutes the first law of probability.

Examples:

1. Suppose that in the experiment a coin is flipped into the air; the coin has two possibilities, the first possibility is that the coin will fall heads and the second is that the coin will fall tails; that is, there are two probabilities and each of them equals 50% or ½.

When applying the formula p + q = 1; being p, the probability of the coin falling heads and q the probability of the coin falling tails we have:

½ + ½ = 1.0

2. Suppose we now flip the coin on air twice, which generates the following data:

Flip 1	Flip 2	Summarazing
Heads ½	Heads ½	p^2 ¼
Heads ½	Tails ½	pq ¼
Tails ½	Heads ½	qp ¼
Tails ½	Tails ½	q^2 ¼

When replacing the values that were obtained in the previous example, we have the following: Adding we have $(p+q)^2$ =¼ + ¼ + ¼ + ¼ = 4/4 that is 1.0

Product of probabilities

The birth of a male calf in a cow's calving is independent of the sex of the next calf that the female is to produce. The probability of the same cow having two male calves in consecutive calves is calculated by multiplying the individual probabilities of the occurrence of each event independently. This constitutes the second law of probability.

Example:

1. The probability of a cow having three males in 3 consecutive calving is: ½ x ½ x ½ = 1/8

2. The probability of a female having two male calves and one female in three calving is: ½ x ½ x ½ = 1/8

In the next section we will address some other applications of probability, as well as some statisticians commonly used in the study of genetics.

5.2 PROBABILITY AND SEGREGATION OF GENES IN GAMETES

When crossing individuals of different sex it is important to determine the proportions of the genes that are segregated and would be obtained at the end of the gametogenic

process; this is indispensable to be able to determine the probabilities that would be expected in the offspring. For example, if a red male individual of RR genotype is to be crossed with a red female of Rr genotype, the probability that the male of RR genotype produces a gamete carrying the R gene, is one while the probability that it produces gametes with the r gene is zero. The situation in the case of female genotype Rr is different. The probability of producing gametes with the R gene is ½ and the probability of producing gametes with the r gene is also ½. The reason for this is the segregation of the members of the gene pairs into the gametes.

Applying now the law of probabilities for two independent events to the gametes and using the characters related to the color and shape of the seed we have for example that:

The yellow phenotype of the seed may be conditioned by the genotype (A-), which is dominant over the green (aa) phenotype and the phenotype for the smooth seed shape (L-) is dominant over the rough seed phenotype (ll).

The genes for seed color are found on a pair of homologous chromosomes, and the genes for seed shape are found on a different pair of homologous chromosomes.

Let us consider the probabilities that individuals carry the genes (AaLl), i.e., they are heterozygous. The following would be the case:

Allele	Probability that a gamete carries this allele
A	½
a	½
L	½
l	½

The probability of several combinations of these two different pairs of alleles occurring together would be:

Possible gene combinations in gamete formation	Probability that a gamete carries these two genes
AL	½ x ½ = ¼
Al	½ x ½ = ¼
aL	½ x ½ = ¼
al	½ x ½ = ¼

5.3 PROBABILITY AND GENE RECOMBINATION IN THE ZYGOTE

The concept of probability and the combination of probabilities can be extended to the union of genes in the zygote. It is assumed that the different pairs of alleles segregate and recombine independently. Using a pair of genes (yellow or green for seed color), the probability that the gametes of parents of the three genotypes carry one of each allele would be:

	Parent genotype		
	AA	Aa	aa
Probability of A in a gamete	1	½	0
Probability of a in a gamete	0	½	1

We can now calculate the probability of various combinations of gametes in the offspring of parents who are both heterozygous (Aa).

Offspring Genotype	Probability of these genotypes
AA	½ x ½ = ¼
Aa	½ x ½ = ¼
aA	½ x ½ = ¼
aa	½ x ½ = ¼

Equal to the 1:2:1 genotype ratio in the offspring of the cross of heterozygous individuals.

5.4 BINOMIAL EXPANSION

Based on the concepts and examples related to probability analyzed above, this section will include the binomial expansion statistic to understand and interpret numerical data with powers higher than the formula $(a+b)^2$.

In previous examples, we were introduced to this term, but from this point on, we will review the binomial expansion applied to genetic studies; this statistic allows us to analyze numerical data of up to two phenotypes in a population of n individuals, which is precisely its limitation in genetic studies.

Example 1:

Based on the example discussed in Chapter Four that described the following:

Suppose we flip a coin twice, which generates the following data:

Flip 1	Flip 2	In summary
Heads ½	Heads ½	P^2 1/4
Heads ½	Tails ½	pq 1/4
Tails ½	Heads ½	qp 1/4
Heads ½	Tails ½	q^2 1/4

Adding up, we have ¼ + ¼ + ¼ + ¼ = 4/4, which is 1.0

Analyzing the values obtained, we can summarize them in the mathematical formula of the perfect square binomial $(a+b)^2$, which reads as: the first variable, i.e., has raised to the maximum power; in this case, it is two a^2, plus twice the first variable times the second (2a.b), plus the second variable b square b^2. This is summarized as: $(a+b)^2 = a^2 + 2ab + b^2$. Thus, we can say that the probability of two or more independent events occurring together follows the mathematical formula of the binomial expansion $(a+b)^n$.

Example 2:
Suppose we now flip the coin three times, which generates the following data:

Flip 1	Flip 2	Flip 3	Summarizing
Heads ½	Heads ½	Heads ½	p^3 1/8
Heads ½	Heads ½	Tails ½	p^2q 1/8
Heads ½	Tails ½	Heads ½	p^2q 1/8
Tails ½	Heads ½	Heads ½	p^2q 1/8
Tails ½	Tails ½	Heads ½	pq^2 1/8
Tails ½	Heads ½	Tails ½	pq^2 1/8
Heads ½	Tails ½	Tails ½	pq^2 1/8
Tails ½	Tails ½	Tails ½	q^3 1/8

When observing the results, we obtain an expression known as the perfect square trinomial, which is generally summarized as follows:

$$(p+q)^3 = p^3 + 3p^2q + 3pq^2 + q^3$$

When replacing the values obtained in the previous example and adding up, we have that $(p+q)^3 = 1/8 + 3/8 + 3/8 + 1/8 = 8/8$, which is 1.0

Next, we will expand the binomial up to the eighth power.

Binomial Expanded to the Eighth Power

$(a + b)^2 = a^2 + 2ab + b^2$
$(a + b)^3 = a^3 + 3a^2b + 3ab^2 + b^3$
$(a + b)^4 = a^4 + 4a^3b + 6a^2b^2 + 4ab^3 + b^4$

$(a + b)^5 = a^5 + 5a^4b + 10a^3b^2 + 10a^2b^3 + 5ab^4 + b^5$
$(a + b)^6 = a^6 + 6a^5b + 15a^4b^2 + 20a^3b^3 + 15a^2b^4 + 6ab^5 + b^6$
$(a + b)^7 = a^7 + 7a^6b + 21a^5b^2 + 35a^4b^3 + 35a^3b^4 + 21a^2b^5 + 7ab^6 + b^7$
$(a + b)^8 = a^8 + 8a^7b + 28a^6b^2 + 6a^5b^3 + 70a^4b^4 + 56a^3b^5 + 28a^2b^6 + 8ab^7 + b^8$

Finding the numerical coefficients that accompany each of the terms of the given binomial is not an easy task. However, for this, we can use what is commonly known as Pascal's triangle, which is constructed as follows: the triangle is built from top to bottom, starting with the number one, which must go at the beginning and the end of each level; this number one represents the numerical coefficient that will accompany the first and last term in the given binomial expression. On the second level, the numbers one, two, and once again one are placed; from the third level onward, the sum of the value of the previous term with the following term is calculated, and so on; it should be noted that the first and last terms of the binomial expression are always one. Let us look at the expression expanded to the eighth power.

1
$(a + b)^2$ = 1 2 1
$(a + b)^3$ = 1 3 3 1
$(a + b)^4$ = 1 4 6 4 1 *
$(a + b)^5$ = 1 5 10 10 5 1
$(a + b)^6$ = 1 6 15 20 15 6 1
$(a + b)^7$ = 1 7 21 35 35 21 7 1
$(a + b)^8$ = 1 8 28 56 70 56 28 8 1

If we analyze the expression $(a+b)^4$ * the mathematical expansion would be as follows:

$(a + b)^4 = 1a^4$	$+ \ 4a^3b$	$+ \ 6a^2b^2$	$+ \ 4ab^3$	$+ \ 1b^4$
1^{er} término	2^{do} término	3^{er} término	4^{to} término	5^{to} término

It is worth noting that, in binomial resolution, the variables increase and decrease in value; that is, while one variable decreases the value of its exponent by one unit, the second variable increases its value by one unit from the second term of the expression. The binomial expansion can be developed up to **n** power.

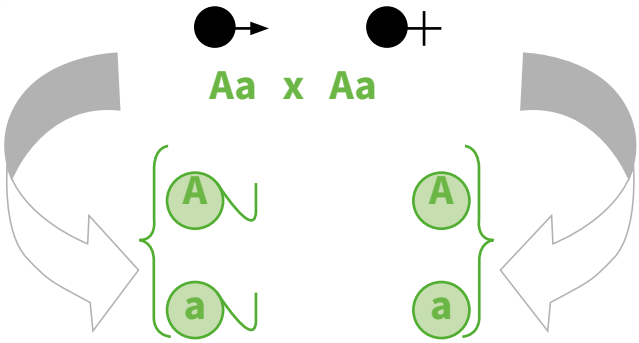
5.5 PRACTICAL APPLICATION OF THE BINOMIAL

If heterozygous black dogs are crossed with each other, what is the probability that three out of seven offspring will be black and four will be white?

Suppose the letter (a) represents black individuals and (b) represents white individuals. The probability of producing a black individual is ¾, and the probability of producing a white individual is ¼, as shown below:

Black male - Black female

Parental genotypes



Gametes

By organizing the gametes provided by each parent at the end of the meiotic process into a gametic square (Punnett square), we have:

Gametes	A	A
A	**AA Zygote**	**Aa Zygote**
A	**Aa Zygote**	aa Zygote

From this table, we analyze the genotypic and phenotypic ratios obtained in generation or filial F1 as follows:

Genotypic ratios: ¼ homozygous dominant AA, 2/4 heterozygous Aa, and ¼ homozygous recessive aa.

Phenotypic ratios: ¾ black dogs and ¼ white dogs; these would be the ratios for a single individual, either black or white.

Then to find the probability that out of the seven puppies, three are black and four are white, we must use the binomial expression $(a+b)^7$ and only consider the fifth term of the expression and replace the values of the variables by the respective ratios as follows:

$35a^3b^4 = 35(3/4)^3(1/4)^4 * 100 = 5.8\ \%$ This would be the probability that out of seven offspring, four will be black, and three will be white

Note. This type of procedure is used to calculate probabilities for n number of individuals for a maximum of two given traits or phenotypes; when analyzing more than two traits, another statistic known as the multibinomial expansion should be used, which we will discuss later, along with the chi-square test. These statistics will be applied once the main types of variants or variations to Mendelian inheritance principles are known, which will be developed below.

Once we have reviewed the concepts and laws governing the principles of probability, we will explore the Mendelian cross types in the next chapter.

5.6 APPLICATION PROBLEMS

1. Determine the probability of a die roll landing on the number six.

2. Determine the probability of rolling 2 dice where both land on the number three.

3. Determine the probability of flipping three coins, getting two tails and one heads, two heads and one tails, and getting all three tails.

4. Determine the probability of drawing the ace of spades in a 52-card deck of cards.

5. Determine the probability that in a litter of three rabbits resulting from the cross between heterozygous black individuals, two are black and one is white.

6. Determine the probability that in a cross between pure black and white individuals in the F2 generation, three black male individuals and two white female individuals are obtained.

7. In a cross between tall-stemmed plants and short-stemmed plants, the following phenotypic ratios were obtained: 5 tall-stemmed plants and 5 short-stemmed plants.

a. Explain with an example how these results were obtained and what result were expected.
b. The results obtained are the same as the expected results. Explain.

8. In a birth of a cross between black rats NN and yellow rats nn, all offspring in the F1 generation are black; if these F1 rats are interbred, what is the probability of having 3 black rats and 2 yellow rats in the F2? Explain.

CHAPTER 6
MENDELIAN CROSSES

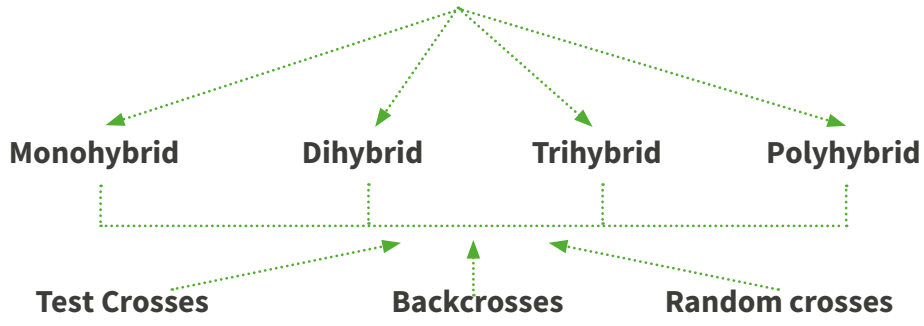
Once the fundamental principles that describe each of the Mendelian laws are understood and analyzed, this chapter will apply the basic concepts addressed in the previous chapter through various problems that will encourage and motivate students to comprehend, understand, and apply the statements promulgated by Mendel.

In this chapter, the definition of a Mendelian crosses will be provided, and the basic principles of probability, types of Mendelian crosses, examples of the Mendelian cross types, and applications of these type of crosses through the resolution of problems of varying complexity will be presented.

6.1 DEFINITION OF CROSSING OVER

Crosses refer to mating between two individuals, breeds, or species of different sexes. Following the principles of Mendelian inheritance in detail, we can find a series of regular crosses in nature, among which the following can be enunciated: monohybrid crosses, dihybrid crosses, trihybrid crosses, and polyhybrid crosses. In addition, other types of crosses or matings, such as the test cross, the backcross, and the random cross, can be applied, as shown in the following figure:

FIGURE 26. *Types of Mendelian Crosses*



Source: Author's own creation, (2021)

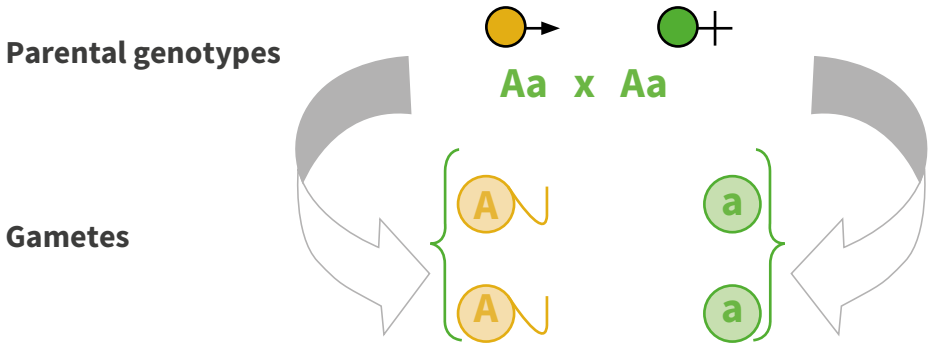
6.2 MONOHYBRID CROSSES

A monohybrid cross can be defined as the mating between two individuals of different sexes that differ by a single characteristic or trait (phenotype). An example from Mendel's work is the cross between a pure plant that produces yellow seeds and a pure plant that produces green seeds. It was observed that all individuals produced in the first generation or filial (offspring) manifested the yellow phenotype. Subsequently, Mendel self-crossed these individuals (offspring) with each other, obtaining in the second-generation plants that produced yellow and green seeds, which occurred in a phenotypic ratio of 3:1; that is, for every three yellow seeds, approximately one green seed was obtained. Let us analyze the results using the following model:

Suppose the yellow seeds have the following genotype: AA and Aa, and the green seeds have the genotype: aa.

When crossing yellow-seed plants with green-seed plants, it is obtained:

Plants with yellow seeds X Plants with green seeds



By organizing the gametes that each of the parents provides at the end of the meiotic process in a gametic square (Punnett square) we have:

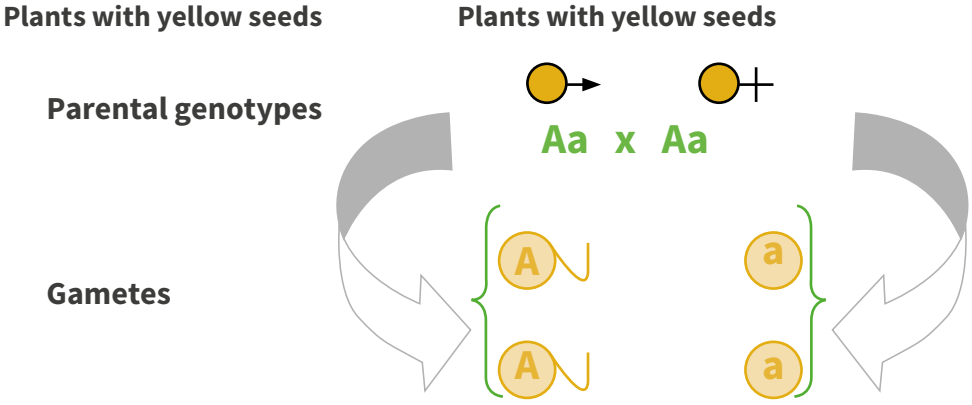
Gametes	a	a
A	Aa Zygote	Aa Zygote
A	Aa Zygote	Aa Zygote

From this table, we analyze the genotypic and phenotypic ratios obtained in the first generation or filial F1 as follows:

Genotype ratios: 4/4, i.e., all zygotes are heterozygous Aa.

Phenotypic ratios: 4/4, i.e., all plants have yellow seeds.

When self-crossing F1 individuals with each other, we obtain an F2 or second generation as follows:



By organizing the gametes that each of the parents provides at the end of the meiotic process in a gametic square (Punnett square) we have:

Gametes	A	a
A	AA Zygote	Aa Zygote
a	Aa Zygote	Aa Zygote

From this table, we analyze both genotypic and phenotypic ratios obtained in the second generation or filial F2, as follows:

Genotypic ratios: ¼ homozygous dominant AA, 2/4 heterozygous Aa, and ¼ homozygous recessive aa.

Phenotypic ratios: ¾ plants with yellow seeds and ¼ plants with green seeds, which would give us a phenotypic ratio of 3:1.

Considering the values of Table 1 and relating them to the 3:1 phenotypic ratio for this cross, we have:

Total plants 8023

Multiplying this value by 3 and dividing it by 4 gives: 8023 x 3/4 = 6017 plants with yellow seeds

Similarly, the other fraction: 8023 x 1/4 = 2006 plants with green seeds.

Through this same explanation, we can analyze all the traits and ratios Mendel determined in his experiments (Table 2). This model can also be applied to all animal and plant species with this type of genetics.

Types of Monohybrid Crosses

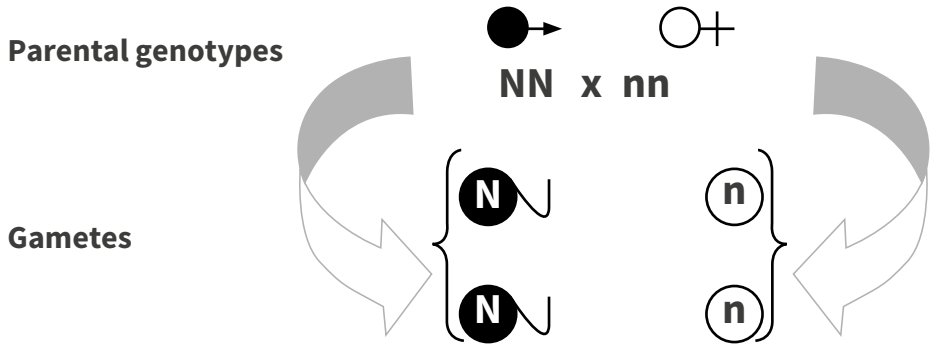
There are basically six types of monohybrid crosses listed below. You will explain each and determine the genotypic and phenotypic ratios using the same model as before; you can use any of the traits determined by Mendel.

1. Cross between pure dominant individuals, i.e., AA X AA.
2. Cross between pure dominant individuals with heterozygous, i.e., AA X Aa.
3. Cross between pure dominant individuals and pure recessive individuals, i.e., AA X aa.
4. Cross between heterozygous individuals, i.e., Aa X Aa.
5. Cross between heterozygous individuals and pure recessive individuals, i.e., Aa X aa.
6. Cross between pure recessive individuals with heterozygous, i.e., aa x aa.

Let us consider some other applications of monohybrid crosses:

Cross a pure black dog with a white female. Determine the phenotypic and genotypic ratios obtained in F1 and F2. Suppose that the female involved in the F2 cross has eight puppies. How many of these puppies would you expect to be black and how many white?

The black color is dominant and is conditioned by two types of genes: NN (for pure individuals) and Nn (for carrier individuals). White is conditioned by the recessive genotype nn.



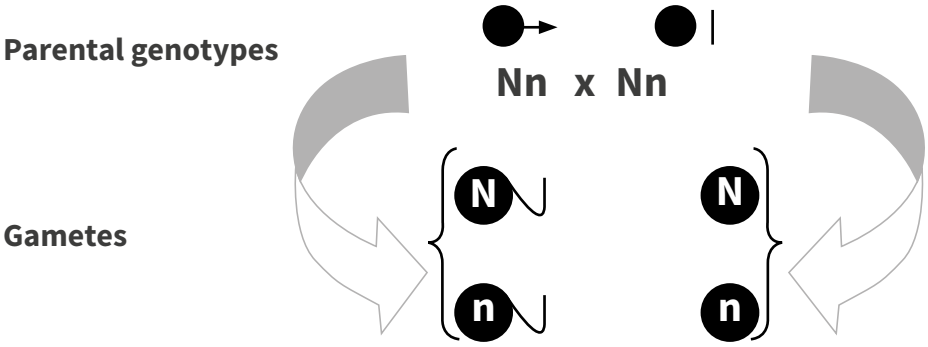
By organizing the gametes that each of the parents provides at the end of the meiotic process in a gametic square (Punnett square) we have:

Gametes	N	n
N	Nn Zygote	Nn Zygote
N	Nn Zygote	Nn Zygote

From this table, we analyze the genotypic and phenotypic ratios obtained in the first generation or filial F1, as follows:

Genotypic ratios: 4/4; i.e., all zygotes are heterozygous Nn.
Phenotypic ratios: 4/4; i.e., all dogs are black.

When self-crossing F1 individuals with each other, we obtain an F2, or second generation as follows:



By organizing the gametes that each of the parents provides at the end of the meiotic process in a gametic square (Punnett square) we have:

Gametes	N	n
N	NN Zygote	Nn Zygote
n	Nn Zygote	nn Zygote

From this table, we analyze the genotypic and phenotypic ratios obtained in the second generation or F2 filial as follows:

Genotypic ratios: ¼ homozygous dominant NN, 2/4 heterozygous Nn, and ¼ homozygous recessive nn.

Phenotypic ratios: ¾ black dogs and ¼ white dogs, which would give us a phenotypic ratio of 3:1.

Based on these phenotypic ratios, we can determine how many of the eight puppies will be black and how many will be white as follows:

¾ X 8 = 6. This would be the expected number of black dogs in the F2.

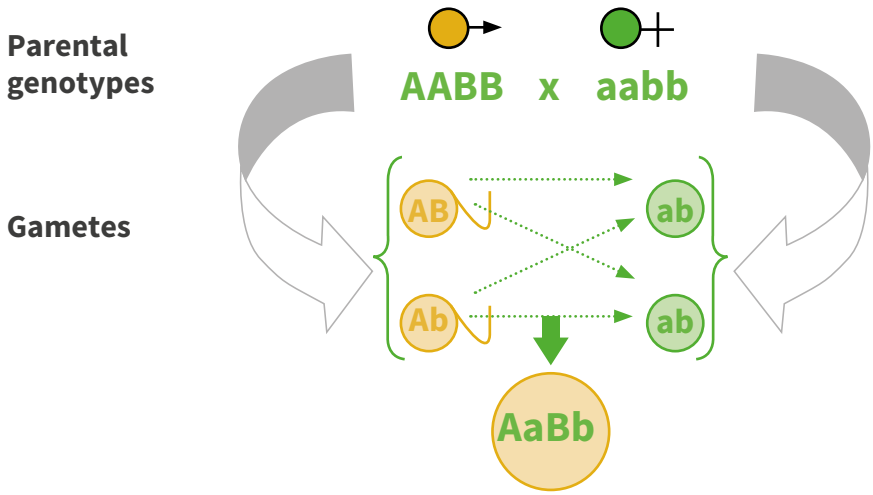
¼ X 8 = 2. This would be the expected number of white dogs in the F2.

6.3 DIHYBRID CROSSES

A dihybrid cross can be defined as the mating between two individuals of different sexes that differ in two characteristics or traits (phenotype). When analyzing Mendel's work, specifically the one that led to the formulation of the law of independent assortment, we can take as an example the cross between a pure plant that produces yellow, smooth seeds and a pure plant that produces green, rough seeds.

Let us assume that the yellow color phenotype is determined by the dominant genotypes AA and Aa, and the green color phenotype by the recessive genotype aa, and that the smooth seed phenotype is determined by the dominant genotypes BB or Bb, and the rough seed phenotype by the genotype bb. Then:

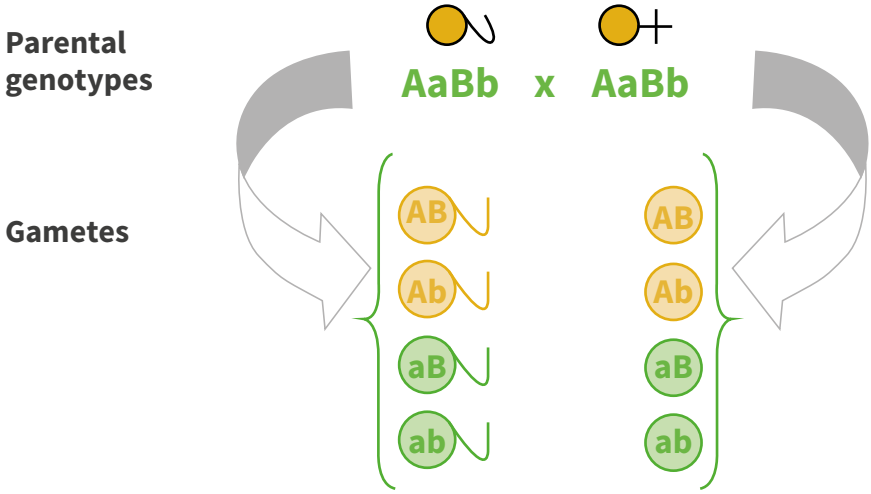
To obtain the F1, we cross a plant with the genotype AABB (yellow, smooth seeds) with another plant with the genotype aabb (green, rough seeds), and we obtain, using as a model, the gametic distribution of the Punnett square (method 1):



F1 Genotype: All of type AaBb

F1 Phenotype: All plants produce yellow, smooth seeds

When crossing the entire F1 with each other, we have F1 x F1:

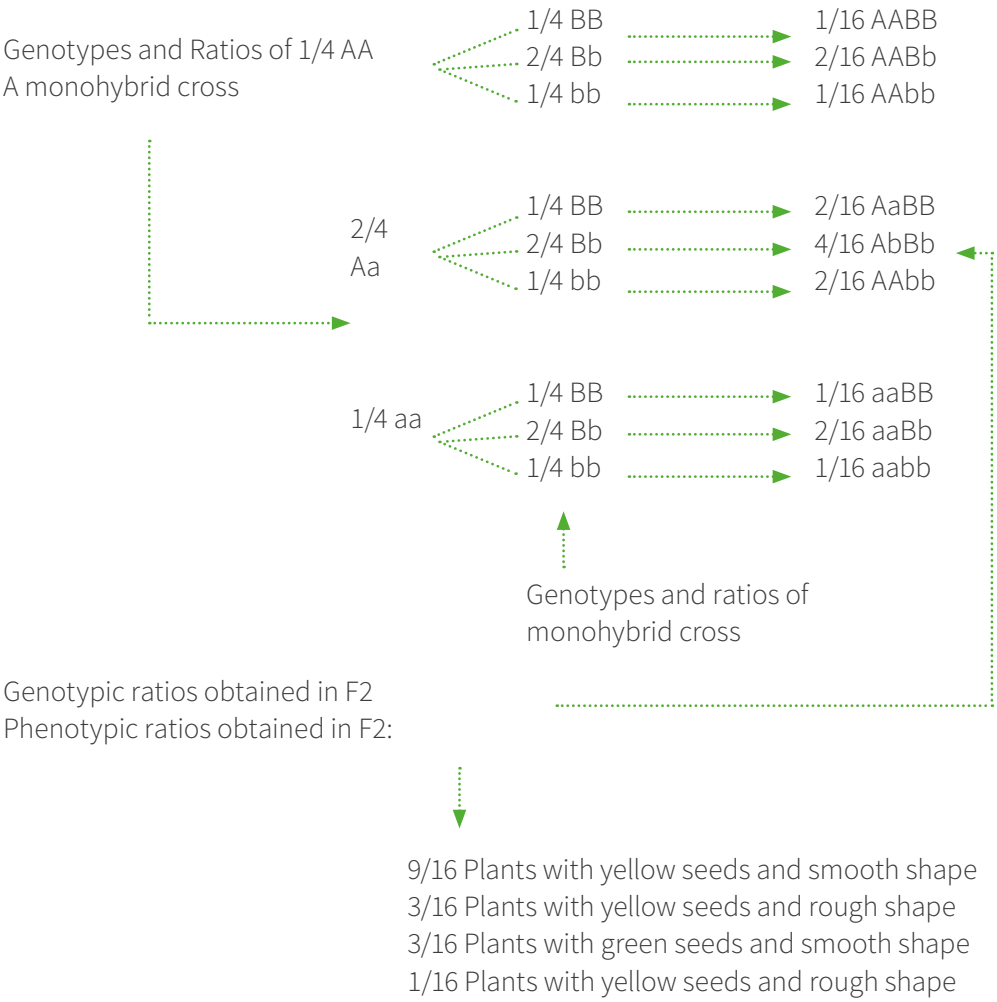


Placing the gametes produced by both the paternal parent and the maternal parent in a Punnett square, we have:

Gametes	AB	Ab	aB	ab
AB	Yellow AABB - smooth	Yellow AABb - smooth	Yellow AbBB - smooth	Yellow AaBb - smooth
Ab	Yellow AABb - smooth	Yellow AAbb - rough	Yellow AaBb - smooth	Yellow Aabb - rough
aB	Yellow AaBB - smooth	Yellow AaBb - smooth	Green aaBB - smooth	Green aaBb - smooth
ab	Yellow AaBb - smooth	Yellow Aabb - rough	Green aaBb - smooth	Green aabb - rough

Method 1 (Punnett square)

This type of procedure can be simplified and sped up by using the gametic combination tree (method 2), in which the ratios of each homozygous dominant genotype, which will always be equivalent ¼, heterozygous 2/4, and homozygous recessive ¼ are placed (this is the same for each trait or characteristic).



Method 2

The phenotypic ratio translates to: 9:3:3:1

Note. When comparing the data obtained using the Punnett square as those obtained in the gamete combination tree, we notice that they are the same; on the other hand, it is noteworthy that a dihybrid cross is essentially the product of two monohybrid crosses between heterozygous individuals.

The following table summarizes the total number of gametes and zygotes that a carrier individual (heterozygous) can produce for one, two, three, and four traits.

TABLE 4. *Gametes and Zygotes Produced by Carrier Individuals (Heterozygous) for One, Two, Three and Four Traits*

Total Number of Traits	Total Number of Gametes (2^n)	Total Number of Zygotes (n^2)
One (Monohybrid)	$2^1 = 2$	$2^2 = 4$
Two (Dihybrid)	$2^2 = 4$	$4^2 = 16$
Three (Trihybrid)	$2^3 = 8$	$8^2 = 64$
Four (Tetrahybrid)	$2^4 = 16$	$16^2 = 256$

Source: Author's own creation, (2021)

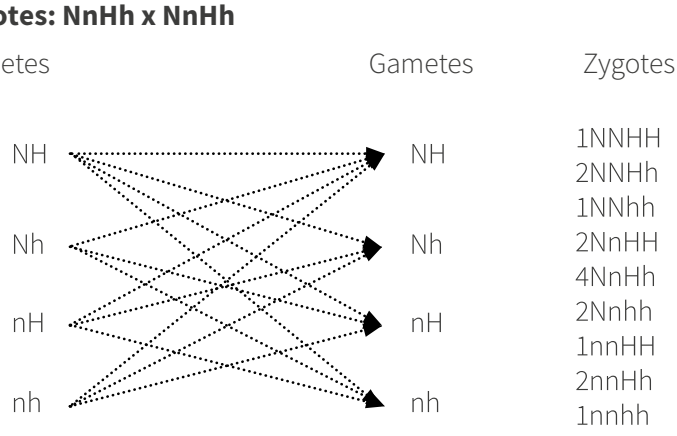
The same procedure can be used to solve trihybrid, tetrahybrid, etc., crosses.

Let us look at another example where this Mendelian principle is demonstrated: In cattle, the Angus is a carrier of the black coat and polled alleles, which are dominant over the Hereford's red coat and horned alleles. The white head of the Hereford is an independent dominant allele. From the cross between an Angus (black, polled) and a Hereford (red, horned), the F1 individuals are black and polled (heterozygous).

Parents	**Angus**	**x**	**Hereford**
Phenotype:	(black, polled)		(red, horned)
Genotype:	NN HH		nn hh
F1		**Nn Hh**	
(black, polled)		Heterozygous	

When heterozygous animals are mated, the genotypic results would be:

Zygotes: NnHh x NnHh



Method 3

In this case, the phenotypic ratios are: 9/16 black polled (individuals carrying N and H), 3/16 black horned (individuals with N and hh), 3/16 red polled (individuals carrying nn and H), and 1/16 red horned (individuals carrying nn and hh).

The method of dates is rather complicated, and someone without experience can easily make mistakes. Students can use any of these three methods to solve problems, depending on their understanding and expertise.

Another mathematical procedure that can be used is to determine the genotypic and phenotypic frequencies or ratios without the student necessarily having to fully work out the problem. This is quite helpful when working on problems involving tetrahybrids, pentahybrids, hexahybrids, etc.

Example:

Suppose two pure plant varieties differing in 3 traits are crossed as follows: Variety A produces red fruit, a thick stem, and purple flowers (all pure dominant traits); Variety B produces white fruit, a thin stem, and white flowers (all recessive). Determine the ratio of individuals resulting from the cross between (AaBbDd X AaBbDd) that are:

1. Individuals with genotype AaBBDd
2. Individuals with genotype AABBDd
3. Plants with red fruit, thin stems, and white flowers
4. Plants with white fruit, tall stems, and purple flowers
5. Etc.

When crossing these types of individuals, we expect the following genotype possibilities* in the offspring:

27/64	A_B_D_ (1)
9/64	A_B_dd (2)
9/64	A_bbD_ (3)
9/64	aaB_D_ (4)
3/64	A_bbdd (5)
3/64	aabbD_ (6)
3/64	aaB_dd (7)
1/64	aabbdd (8)
64 zygotes	

These would be the expected phenotypic ratios in the offspring; it should be noted that using the formula described in Table 2, we would have a total of $n^2 = 64$ zygotes.

Note. The line accompanying each letter (allele) indicates that the gene can be homozygous or heterozygous, knowing that by Mendelian inheritance, the dominant homozygous or the heterozygous determines the same phenotype.

Another easy way to determine the phenotypic relationships (ratios) of crosses between carrier individuals (heterozygous) can be done with the following procedure:

TYPE OF CROSS BETWEEN CARRIER INDIVIDUALS (HETEROZYGOUS)	PHENOTYPIC RATIOS	POSSIBLE TYPES OF GAMETES
MONOHYBRID AA X AA	3:1	3 A_ AND 1 AA
DIHYBRID AABB X AABB (RATIOS OF TWO MONOHYBRID CROSSES)	3:1 3:1 9:3:3:1	9 A_B_ 3 A_BB 3 AAB_ 1 AABB
TRIHYBRID AABBDD X AABBDD (RATIOS OF THREE MONOHYBRID CROSSES)	9:3:3:1 3:1 27:9:9:9:3:3:3:1	27/64 A_B_D_ 9/64 A _B_DD 9/64 A_BBD_ 9/64 AAB_D_ 3/64 A_BBDD 3/64 AABBD_ 3/64 AAB_DD 1/64 AABBDD

In answering the questions, we must consider that:

1. A homozygous genotype (dominant or recessive) in the zygote has a ratio of ¼ while the heterozygous has 2/4, as mentioned above; knowing this principle and replacing the values in the genotypes obtained in *

¼ X ¼ x 2/4 = 4/64. This would be the number of zygotes with the genotype AaBBDd.

2. ¼ X ¼ X 2/4 = 2/64. This would be the number of zygotes with the genotype AABBDd.

3. Analyzing the data obtained in *, we can calculate this value based on individuals carrying the following genes: A_bbdd. This would give us:

(¼ + 2/4)* X 1/4 X ¼ = 3/64. This would be the expected phenotypic ratio

*This is considering that the AA genotype is equivalent to ¼ and the heterozygous to 2/4; this implies adding the two fractions and multiplying with the ratios of the other genotypes.

4. ¼ X (1/4 + 2/4) x (1/4 + 2/4) = 6/64. This would be the expected phenotypic ratio.

6.4 BACKCROSS AND TEST CROSS

Backcross

This is the cross of F1 progeny with some of its parents (or individuals exhibiting a genotype identical to one of its parents); for example: a black female guinea pig homozygous is mated with a white male. One F1 offspring is backcrossed with its mother. Determine the genotypic and phenotypic ratios of this offspring.
We take as parental genotypes NN for the female and nn for the male.

NN X nn

F1: All offspring will be Nn, i.e., black; if we cross these individuals with the mother we have:

Nn x NN

F2: Half of the offspring are NN, and the other half are Nn; i.e., all offspring are black.

The Test Cross

Since a homozygous dominant genotype has the same phenotype as the heterozygous genotype (according to Mendelian inheritance), a test cross is required to distinguish them genotypically. The parent in the test cross must always be homozygous recessive for all genes under consideration. The purpose of performing a test cross is to determine how many different types of gametes are produced by

an individual whose genotype is unknown. A homozygous dominant individual (for a trait) will produce only one type of gametes, while a heterozygous individual (for a trait) will produce two types of gametes with equal frequency.

Example: Suppose a test cross is performed on a black female rat, and only black offspring are produced. What would be the genotype of the female?

We take the female's genotype as N_. The underscore indicates that we do not know the other allele for that gene since she could be either homozygous or heterozygous; as we do not know her genotype, we must necessarily resort to a test cross as follows:

N- x nn
F1: Nn y _n

This indicates that since all the offspring must be black, the only possible option is that the female is homozygous NN and produces only N-type gametes; if she were heterozygous, she would produce two types of gametes, N and n, meaning that half of her offspring would be black, and the other half would be white.

6.5 APPLICATION PROBLEMS

The questions and problems are intended to exercise thinking in genetics. The student who learns to solve these questions and problems will have mastered the genetics principles they refer to.

1. Mendel crossed yellow-seeded peas with green-seeded peas. All the first generation were yellow, and in the F2 of many crosses, he obtained 705 yellow and 224 green.

   a. Propose a hypothesis to explain these results.
   b. Outline the cross and compare the observed results with the expected ones.

2. In some dog breeds, black fur color is dominant over brown. If a homozygous black female dog is crossed with a brown male dog, what color will the F1 generation dogs be?

3. If a heterozygous male dog is crossed with a heterozygous female dog, and they have 12 puppies, how many are expected to be black? How many are expected to be brown?

4. Short fur in rabbits is caused by a dominant gene over long fur, which is recessive. A cross between a short-furred male and a short-furred female produces 11 short-furred rabbits and 1 long-furred rabbit.

a. What is the genotype of the parents?
b. What phenotypic ratio was expected in the offspring?

5. In pumpkin, white fruit color is determined by a dominant allele, and yellow fruit color is recessive. What phenotypic and genotypic ratios can be expected from the following pairings?

a. A white homozygous individual x another with the same phenotype but heterozygous
b. Two heterozygous individuals crossed together
c. A heterozygous individual and one with yellow fruit

6. Black heterozygous guinea pigs (Mm) are mated with white recessive homozygous (mm). Predict the genotypic and phenotypic ratios expected from the "backcross" of the black F1 progeny with:

a. The black parent
b. The white parent

7. Uniform color (S) in cattle is dominant over white spots (s); (LW), which causes less white in spotted animals, is dominant over its allele (lw), which allows spotted animals to have higher amounts of white. A dihybrid bull was mated with cows of genotype Sslwlw. What will be the phenotypic and genotypic ratios of this cross?

8. In peas, flowers that grow on the stem axis, called axial, are determined by a dominant gene, while flowers that are developed at the tip of the stems, called terminal, are determined by a recessive gene. Colored flowers are produced by another gene located on a different chromosome and are dominant over white flowers. Two dihybrid plants were crossed with each other. What phenotypic and genotypic ratios are to be expected in the F1?

9. In dogs, gene A is responsible for normal hearing, while gene a causes deafness. Forward-folded ears (F) are dominant over erect ears (f). Black fur (N) is dominant over brown fur (n). If a deaf, erect-eared, brown-furred male dog is crossed with female dogs that have normal hearing, forward-folded ears, and black fur for all three pairs of homozygous genes:

a. What will the phenotype and genotype of the F1 puppies be?
b. What will be the phenotypic and genotypic ratios of the F2?

10. A certain homozygosis gene causes a certain genetic disease. In this type of disease, known as sickle cell anemia, erythrocytes are sickle-shaped and unable to carry oxygen properly. Affected individuals usually die before reaching adulthood. Carrier individuals do not suffer from the disease, although their erythrocytes become sickle-shaped under low oxygen concentration conditions. A woman whose brother suffers from the disease seeks genetic counseling before marrying and having children. Blood tests show that her erythrocytes become sickle-shaped under low oxygen concentrations. The future husband has normal erythrocytes. Write a report (from a medical-scientific perspective) about the future children of this couple. What are this woman's parents like regarding the studied trait? What advice would you give the couple? Argument.

11. A pure line of the peas used by Mendel, which are dominant for the seven independently distributed gene pairs, undergoes a test cross.

a. How many different classes of gametes could each parents produce?
b. How many gametes could the F1 produce?
c. If the F1 is test-crossed, how many phenotypes and in what ratios could be expected in the offspring?
d. How many individuals out of a total of 1200 would be expected for each phenotypic and genotypic class from the backcross (with the paternal parent AABBCCDDEEFFGG) of a heterozygous individual for the seven traits studied by Mendel?

TEACHING UNIT

NON-MENDELIAN GENETICS MODELS

Gustavo Forero Acosta

CHAPTER 7
NON-MENDELIAN CROSSES I

One of the factors that contributed to the success of Mendel's work was that the traits he selected were regulated by genes that followed a consistent pattern of dominance and recessiveness. In addition, he never considered other genetic factors that were also heritable because his biological material of study did not express them in any of the generations. These factors, consequently, represent modifications to his Mendelian principles or laws and include: codominance or incomplete dominance, lethal genes, multiple alleles, and epistasis, among others.

7.1 CODOMINANCE OR INCOMPLETE DOMINANCE

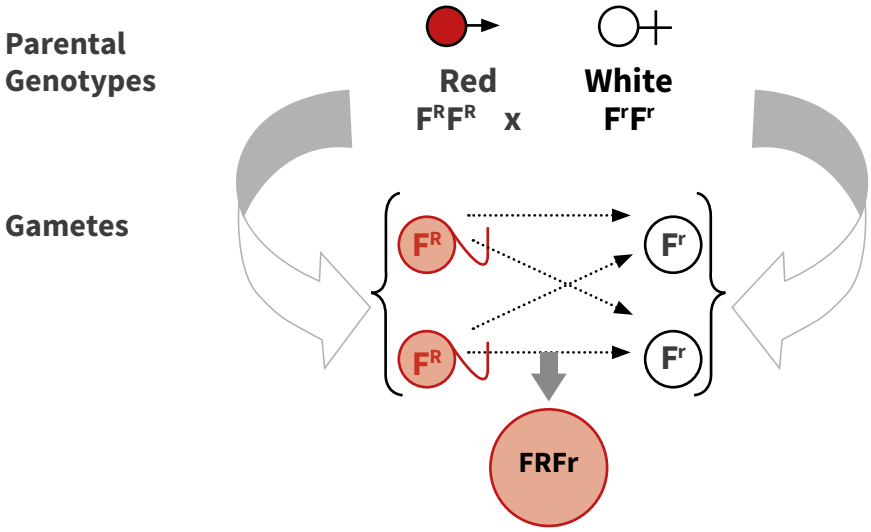
It is also known as intermediate inheritance and is characterized because the heterozygote presents an intermediate phenotype to that produced by the homozygous individuals. That is, the heterozygote does not manifest the same phenotypic relationship as the dominant homozygote, as is the case with Mendelian inheritance. When there is incomplete dominance between two alleles, the phenotypic ratios in the F2 are 1:2:1, and the phenotype describes the genotype, different from the classic Mendelian ratio of 3:1.

In this case, and for the analysis of the problems related to this principle, a symbology is used that consists of employing a capital base letter, the same for the genes, and a superscript that can be a letter, number, or symbol indicating the variability of the gene for that same trait. For example:

Red phenotype : F^RF^R White phenotype
: F^rF^r Pink phenotype
: F^RF^r

Example:

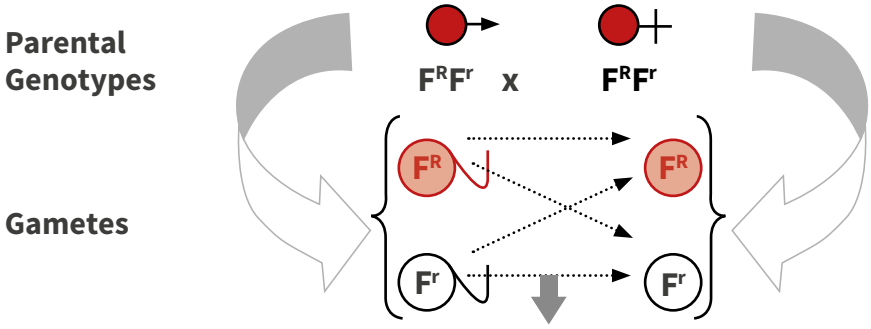
The coat color in Shorthorn cattle presents three types of color: red, white, and roan, where roan is produced when a red animal is crossed with a white one.



Zygotes F1: All type $F^R F^r$

All roan

If we self-cross the F1, we get the following in the F2:



Zygotes F2:	FR FR	¼	Red
	FR Fr	2/4	Roan
	Fr Fr	1/4	White phenotypic ratio 1:2:1

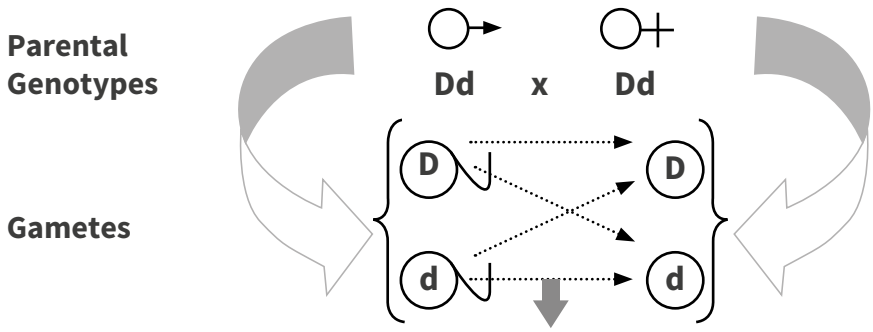
7.2 LETHAL GENES

Lethal genes cause an individual to die during the prenatal period or between birth and the onset of sexual maturity. This means that individuals carrying lethal genes in their genome will never produce offspring.

When lethal genes are present, the phenotypic ratios in the F2 are 1:2, and the phenotype reflects the genotype, different from the classic Mendelian 3:1 ratio.

Example:

In the Dexter dairy cattle breed, there is a gene that, in its homozygous (recessive) state, causes the death of the calf shortly after birth. If carrier individuals are crossed, what is the expected phenotypic and genotypic ratio in the adult F1 offspring?



Live-born individuals: 1/4 normal, 1/2 carrier, and 1/4 affected.

Adult F1 individuals: Since all live-born individuals do not reach adulthood, the 4/4 ratio for live-born individuals becomes 3/3, where: 1/3 are normal DD, and 2/3 are carriers Dd.

7.3 MULTIPLE ALLELES

Until now, in all the cases studied, a gene represented only two different forms of expression. However, the same gene may have multiple forms of expression or manifestation. In this case, we say that we have a series of multiple alleles. In this type of alleles, a hierarchy of dominance is typically established, indicating which type of alleles are more dominant, which are in an intermediate, and which are more recessive.

Example:

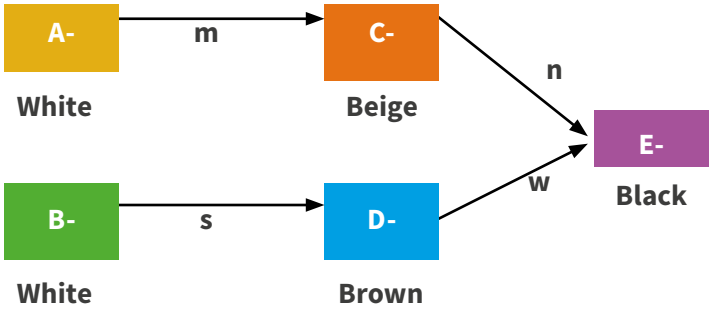
In Drosophila, eye color is governed by a series of multiple alleles that cause the color to vary from red or wild type (w+) through coral (wco), blood (wbl), eosin (we), cherry (wch), peach (wa), honey (wh), buff (wbf), nuanced (wt), pearl (wp), ivory (wi), to white (w). Each allele in the system, except for w, produces pigment, but the alleles successively produce less pigment as the dominance hierarchy decreases: w+>wco>wbl>we>wch>wa>wh>wbf>wt>wp>wi>w. The wild-type allele (w+) is completely dominant, and w is completely recessive compared to the other alleles that form the allele series.

In rabbits, there is a type of multiple alleles that express different colors in the fur N: allows the total production of the typical gray color; n^{ch}, when in homozygous condition, removes the yellow pigment from the fur, producing a silver-gray color called chinchilla; n^{ch}, when heterozygous with other alleles lower in the dominance hierarchy, produces a light gray fur; n^{h} produces a white rabbit with black extremities, called Himalaya; n does not produce pigment, resulting in an albino rabbit. The dominance hierarchy is: $N>n^{ch}>n^{h}>$. Based on this, the following phenotypes can be observed:

POSSIBLE GENOTYPES	PHENOTYPES
NN, Nn^{ch}, Nn^{h}, Nn	Fully colored
$n^{ch}\,n^{ch}$	Chinchilla
$N^{ch}\,n^{h}$, $n^{ch}\,n$	Light gray
$N^{h}\,n^{h}$, $n^{h}\,n$	Himalaya
nn	Albino

7.4 GENE INTERACTIONS

Gene interactions refer to the action that two or more genes can exert to express the same trait or phenotype. For example, consider the following metabolic pathway:



The above metabolic pathway can be understood as follows:

- To express the white color, only the presence of gene A- or B- is required.
- To express the beige color, the presence of gene A- and C- and the enzyme m is required.
- To express the brown color, the presence of genes B- and D- and the enzyme s is required.
- To express the black color, genes A-, C- and E- and enzymes m and n, or genes B-, D- and E- and enzymes s and w, must necessarily be present.

Many traits in living organisms are due to the reciprocal action between two or more pairs of genes.

Example:

Bateson and Punnett discovered that the type of comb in hens is due to this phenomenon. Wyandotte chicken have a rose comb, produced by the interaction of the R and p genes. The Brahma breed has a pea comb, produced by the interaction of the r and P genes. Andalusian hens have a simple comb type caused by the r and p genes in a homozygous recessive state. When rose-combed birds (RRpp) are crossed with simple-combed birds (rrpp), the F1 chicks develop rose combs, and in F2, the phenotypic ratios of three rose combs for one simple comb are obtained. The same occurs when pea-combed birds (rrPP) are crossed with simple-combed birds (rrpp): all the F1 chicks have pea combs, and in F2, for each one with a simple comb, three pea combs are produced.

When birds with rose combs (RRpp) are crossed with pea-combed birds (rrPP), the R and P genes act reciprocally to produce a comb with a walnut appearance (RrPp) in F1. In F2, the following segregation is presented: 9/16 with a walnut comb, 3/16 with a rose comb, 3/16 with a pea comb, and 1/16 with a simple comb.

- 9/16 rose comb: RRpp, Rrpp
- 3/16 pea comb: rrPP, rrPp
- 3/16 simple comb: rrpp
- 1/16 walnut comb: RrPp

The R and P genes act as complementary genes to produce the walnut comb, and the r and p genes complement each other to produce the comb in simple form.

7.5 APPLICATION PROBLEMS

1. Demonstrate through these problems, using the appropriate symbology, how the epistatic ratios contemplated in Table 1.3.4 change when conducting the test cross to one of the parents.

2. The fur color in guinea pigs is conditioned by a homozygous genotype, the cream color by a heterozygous genotype, and the white color by a homozygous genotype. When yellow guinea pigs are crossed with each other, all the offspring are yellow; when white guinea pigs are crossed with each other, all the offspring are white; when yellow and white guinea pigs are crossed, all their offspring are cream and mating between cream individuals with each other, they produce yellow, cream, and white offspring in the 1:2:1 ratio. Use the corresponding symbology to represent the respective crosses.

3. The shape of radishes can be long (RL RL), round (RA RA), or oval (RLRA). If long radishes are crossed with round ones, what phenotypic and genotypic ratios will be found in the F1 and F2?

4. In Shorthorn cattle, the (R–) allele for red fur color is not dominant over white color (R´). Roan color is produced by the heterozygous condition (RR´). A farmer has red, white, and roan animals in his herd and wishes to have animals of a single color. What procedure would you advise this farmer to follow?

5. Colon polyps (an abnormality of the large intestine) and Huntington's chorea nervous disorder are diseases of genetic origin in men, both governed by dominant genes located on non-homologous chromosomes. A man who is homozygous recessive for this abnormality but carries the dominant gene for chorea marries a woman who carries the dominant gene for the same and who is homozygous recessive for chorea. Diagram the mating between these two individuals and indicate the ratios in which their children are expected to have one of the two abnormalities, both or neither.

6. It is known that a pair of codominant alleles in soybeans determine the color of the leaves. The F O FO genotype produces the dark color; the pale green color is produced by the FP FP genotype, and the yellow color is produced by the FO FP genotype, which has few chloroplasts. Due to this phenomenon, the seeds do not reach maturity. If dark green plants are pollinated with pale green plants and the F1 and F2 generations are obtained, what would be the phenotypic and genotypic ratios of F2?

7. In hens, there is a lethal (l) recessive gene linked to the X chromosome. If a heterozygous male for this lethal gene is crossed with a normal hen, what will be the ratio of males and females obtained among their offspring?

8. Tay-Sachs is a recessive hereditary disease and is so severe that it causes death in the early years of life in individuals who suffer it when found in homozygous state. It is thought that abnormally short fingers (brachyphalangy) are due to the heterozygous genotype for a lethal gene, with BB individuals being normal. What phenotypes are expected among children and adolescents born to parents with brachyphalangy and heterozygous for Tay-Sachs disease?

9. In some plants of a rare species, the flowers can be red, white, or pink. It is known that this trait is determined by two genes, red (Cr) and white (Cb), with intermediate inheritance. What will the offspring be like from a cross between pink-flowered plants and red-flowered plants?

10. A series of multiple alleles governs mice's coat color intensity. B = full color, b = diluted color, and bl = lethal in homozygous state. The dominance order is: B > b > bl. A fully-colored mouse carrying the lethal gene is mated with a diluted-colored color mouse also carrying the lethal gene. What will the viable offspring look like?

CHAPTER 8
NON-MENDELIAN CROSSES II

Before delving into the contents corresponding to Non-Mendelian Crosses II, it is important to introduce and deepen on two statistics commonly used in genetics, which were not covered in depth in Chapter 3. For their explanation, the student must first handle the concepts of Mendelian genetics, crosses, and Mendelian variations I.

8.1 MULTIBINOMIAL EXPANSION OR TEST OF TRINOMIAL PROPORTIONS

The binomial distribution can be generalized to accommodate any number of variables. If events e1, e2, e3…. ek occur k1,k2,k3…. kn times, it is respectively:

$$\frac{N!}{k2!k3\ldots.kn!} \quad p1^{k1}p2^{k2}p3^{k3}\ldots.pn^{kn}\ k1!$$

Where:

N!	= equals the total number of individuals in the population
$k1!\ k2!k3\ldots.kn!$	= equals the number of individuals for each phenotypic class
$p1^{k1}p2^{k2}p3^{k3}\ldots.pn^{kn}$	= equals the independent probability for each phenotype

Example 1:

The blood types of humans are under the genetic control of a pair of codominant alleles. In families with six offspring where both parents are MN type, what is the probability of finding three children with blood type M, two with type MN, and one with type N?

Parents: $L^{M}L^{N}$ X $L^{M}L^{N}$

F1: ¼ $L^{M}L^{M}$ = type M
½$L^{M}L^{N}$ = type MN
¼ $L^{N}L^{N}$ = type N

Let p1 be the probability that a child is type M = ¼, p2 the probability that a child is type MN = ½, and p3 the probability that a child is type N = ¼.

Let K1 = required number of children with type M = 3, K2 = required number of children with type MN = 2, and K3 = required number of children with type M = 1.

N = 6

By replacing the formula, we have:

$$(p1 + p2 + p3) = \frac{6!}{3!2!1!} (1/4)^3 (1/2)^2 (1/4) = 0.059 \text{ o } 5.9\ \%,$$

This would be the probability that in families of six offspring, three are of blood type M, two of blood type Mn, and one of blood type N.

Example 2:

In Shorthorn cattle, the genotype (**RR**) produces a red phenotype; the genotype (**R'R'**) produces the white phenotype; and roan individuals are of genotype (**RR'**). A farmer has eight roan cows that were mated with roan bulls in his herd. What is the probability that two of the eight expected calves will be red, five roan, and one white?

The problem could be solved using the following formula: $(p + q + r)^8$, but it more practical to use the trinomial proportions formula:

Parents: **RR'** X **RR'**

F1: ¼ **RR** = red color
½ **RR'** = roan color
¼ **R'R'** = white color

Let p1 be the probability that the offspring has red color = ¼, p2 the probability that the offspring has roan color = ½, and p3 the probability that the offspring has white color = ¼.

Let K1 = required number of offspring of red coat = 2, K2 = required number of offspring of roan coat = 5, and K3 = required number of offspring of white coat = 1.

$$N = 8$$

By replacing the formula, we have:

$$(p1 + p2 + p3) = \frac{8!}{2!5!1!}(1/4)^2 (1/2)^5 (1/4)^*100 = 8{,}20\ \%.$$

The probability that out of eight expected offspring by the farmer, two will be red, five will be roan, and one will be white from the cross between **RR'** X **RR'** parents is 8.20%.

8.2 THE CHI-SQUARE TEST OF PHENOTYPIC RATIOS (X^2)

The chi-square is a non-parametric measure of dispersion applied to binomial populations. A binomial population is one in which a qualitative trait is measured and distributed according to the binomial expression.

The chi-square is a statistical tool that estimates the probability of discrepancies between observed phenotypic ratios and those expected for a given inheritance pattern and determines whether these discrepancies are significant or small enough to be attributed to chance.

Example:

From a cross between two pure strains of Drosophila melanogaster flies, a total of 524 individuals were obtained in the F2, distributed into the following phenotypic classes: 290 flies with red eyes and black bodies, 90 flies with red eyes and yellow bodies, 100 flies with white eyes and black bodies, and 44 flies with white eyes and yellow bodies.

a. Propose a hypothesis to explain these results.
b. Outline the cross and compare the observed results with the expected ones.

a. Ho : O = E ; there is concordance

Selection of the probability level against which the null hypothesis will be tested (5%).

Ha = O ≠ E; there is no concordance.
Ho is rejected if:

X^2	≥	$X^2\infty$ (gl)
calculated		table value

Ho is accepted if:

X^2	<	$X^2\infty$ (gl)
Calculated		table value

a. Calculation of expected F2 ratios

To calculate the expected phenotypic ratios in F2, we are based on the values obtained and phenotypes obtained experimentally. It is concluded that the dominant traits are red eye (**AA**) and black body (BB), while white eye (aa) and yellow body (**bb**) are recessive. Thus, the cross is as follows:

AABB X aa**bb**

F1: All individuals will be (**A**aB**b**); i.e., red eyes and black body. By cross them each other, we have:

AaB**b** **X** **A**aB**b**

9/16 will be **A**_**B**_ individuals, i.e., with red eyes and black bodies. 3/16 will be **A**_**bb** individuals, i.e., with red eyes and yellow bodies. 3/16 will be **aaB**_ individuals, i.e., with white eyes and black bodies. 1/16 will be **aabb** individuals, i.e., with white eyes and yellow bodies.

Thus, we determine the number of individuals that would be expected for each phenotypic class as follows:

$\frac{9}{16}$ x 524 = 295 flies with red eyes and black body

$\frac{3}{16}$ x 524 = 98 flies with red eyes and yellow body

$\frac{3}{16}$ x 524 = 98 flies with white eyes and black body

$\frac{1}{16}$ x 524 = 33 flies with white eyes and yellow body

When organizing the data in a chi-square table, we have:

Phenotypes	Observed Values (O)	Expected VAlues (E)	(O-E)	$(O-E)^2$	$(O-E)^2/E$
Red eyes – black body	290	295	-5	25	0.08
Red eyes – yellow body	90	98	-8	64	0.65
White eyes - black body	100	98	2	4	0.04
White eyes - white body	44	33	11	121	3.66
Totals	**524**	**524**	**/**	**/**	**4.43**

The chi-square value corresponds to 4.43

The degrees of freedom for this case are three; by placing the chi-square value obtained in our exercise for three degrees of freedom in a chi-square distribution table, we can see that this value is between 20% and 30% probability of occurrence. In other words, we can say that the null hypothesis is accepted.

8.3 PRECAUTIONS IN THE USE OF CHI-SQUARE IN GENETICS

- It should not be used with percentages derived from frequencies to calculate expected or observed ratios.
- It is important for studying numerical frequencies in qualitative inheritance.
- It should not be used with samples where the total number of observed individuals (N) is less than the total number of individuals required to obtain the correct phenotypic ratios.

8.4 EPISTASIS

When a gene suppresses or inhibits the expression of another gene that is not an allele, this is called epistasis. The gene that is expressed is called epistatic, while the non-allelic gene that is inhibited or repressed is called hypostatic.

The genes that cause the epistasis phenomenon can be located on the same chromosome or different chromosomes.

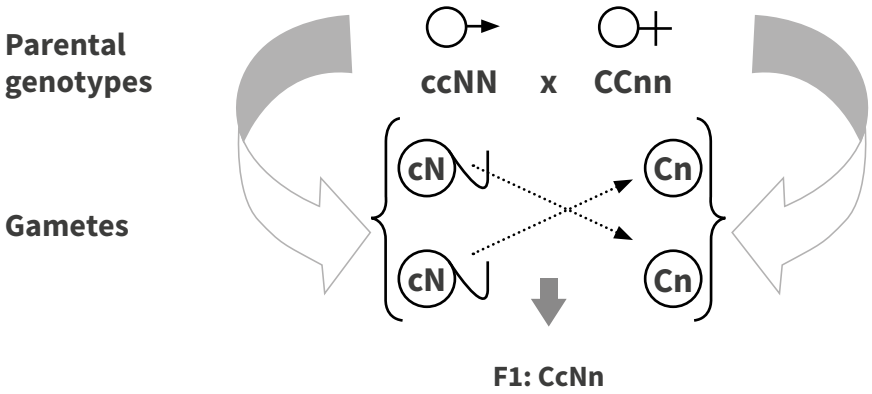
There are six different types of gene interactions involving epistasis, three of which manifest with three phenotypes, while the other three have only two phenotypes, each with a different designation.

Simple Recessive Epistasis

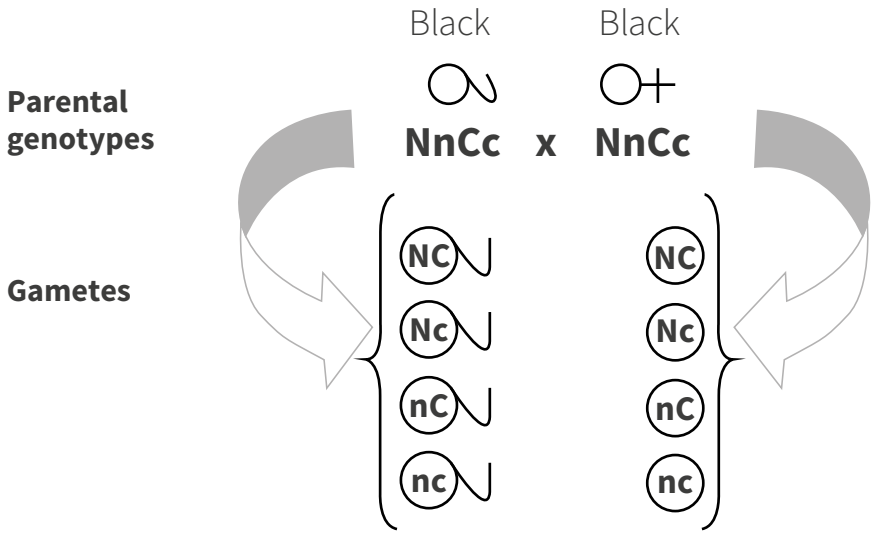
When the recessive genotype of a locus (for example, yy) masks the expression of the alleles of the Z locus, it is said that the Y locus exhibits recessive epistasis over the Z locus. Only if the dominant allele is present at the Y locus can the alleles of the hypostatic Z locus be expressed. Therefore, the yy gene in its homozygous state is epistatic to the Z and z genes, causing the 9:3:3:1 ratio to convert into 9:3:4, resulting in three phenotypes.

Example:

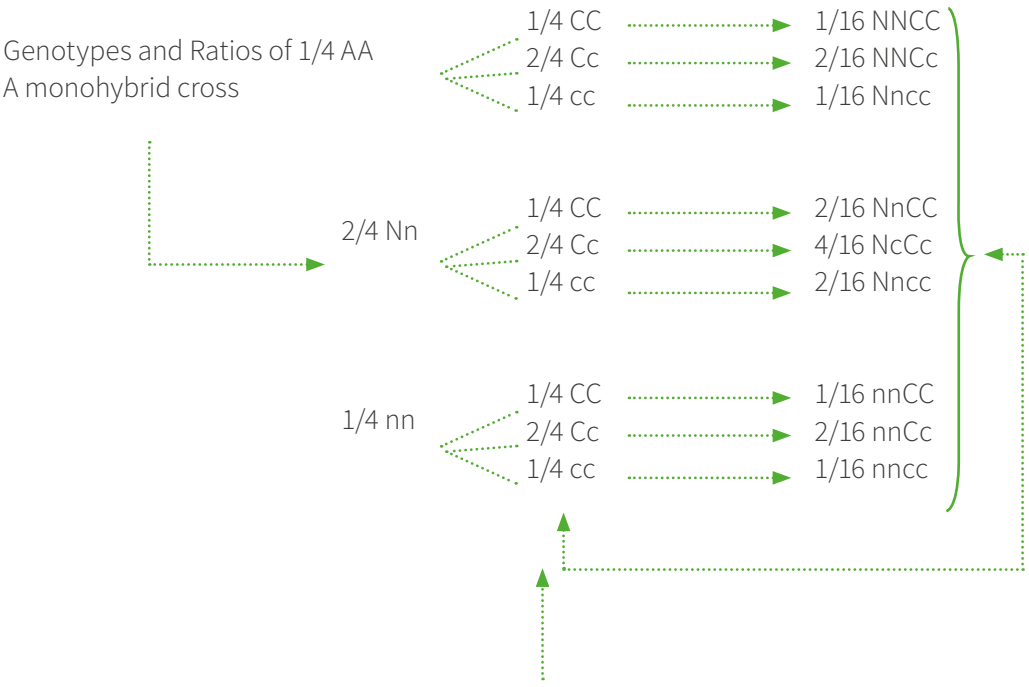
The mice have three types of colors: black, brown, and albino. Black (N–) is dominant to brown (nn); pigment (C–) is dominant to albino (cc), but cc is epistatic to both N– and nn. If we cross a homozygous albino mouse (ccNN) with a homozygous brown female (CCnn), what would be the expected ratios in the F2 offspring?



They are all black. When crossing all the F1 individuals with each other



Empleando el árbol gamético tenemos:



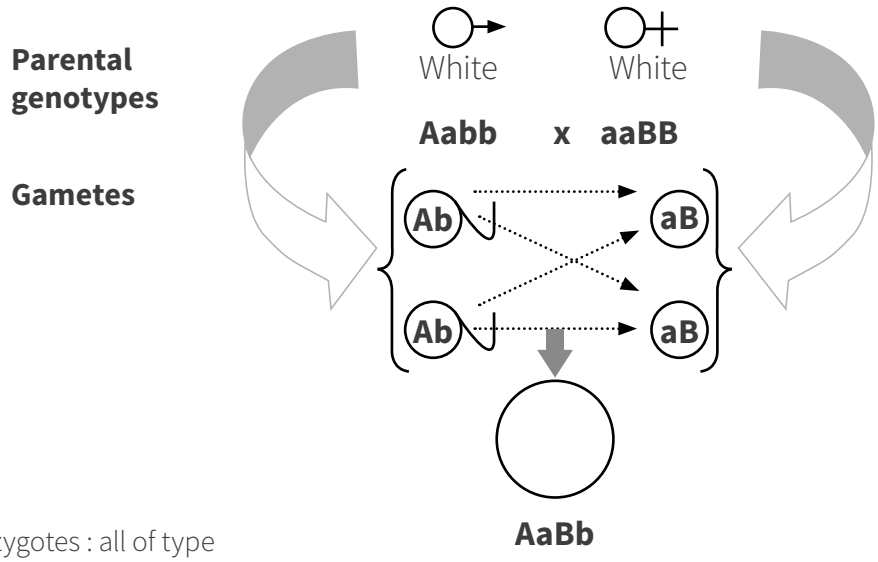
Considering that the cc gene is epistatic to N- and nn, then: 9/16 are black mice, 3/16 are brown mice, and 4/16 are albino mice. This results in a phenotypic ratio of 9:3:4.

Double Recessive Epistasis

This occurs if recessive alleles at one or both loci produce the same phenotype; however, when dominant alleles are present together, they complement each other and result in a different phenotype. The result of this gene interaction is two phenotypes in a 9:7 ratio.

Example:

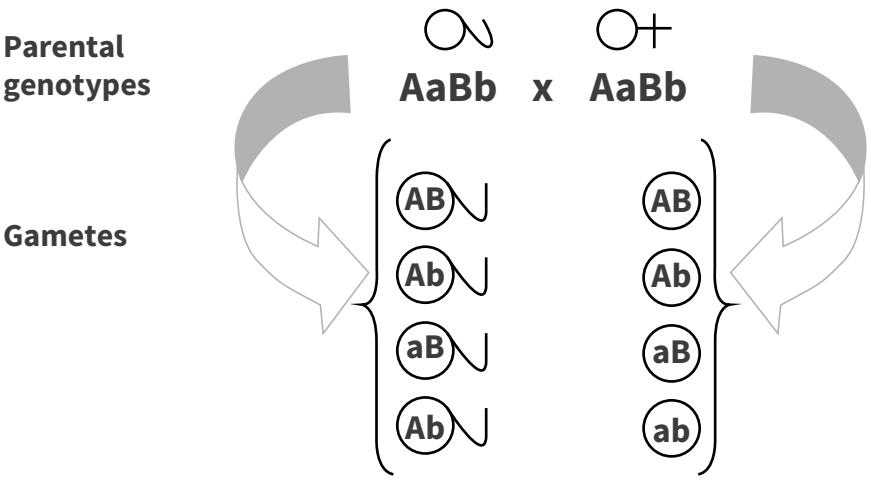
In a type of plant, the genes aa and bb are epistatic to their respective alleles (i.e., A– and B–) and produce white flowers; when plants have the genes A– and B–, their flowers are violet. Determine the phenotypic ratios of F1 and F2 from a cross between homozygous, white-flowered plants with AAbb and aaBB genotypes.



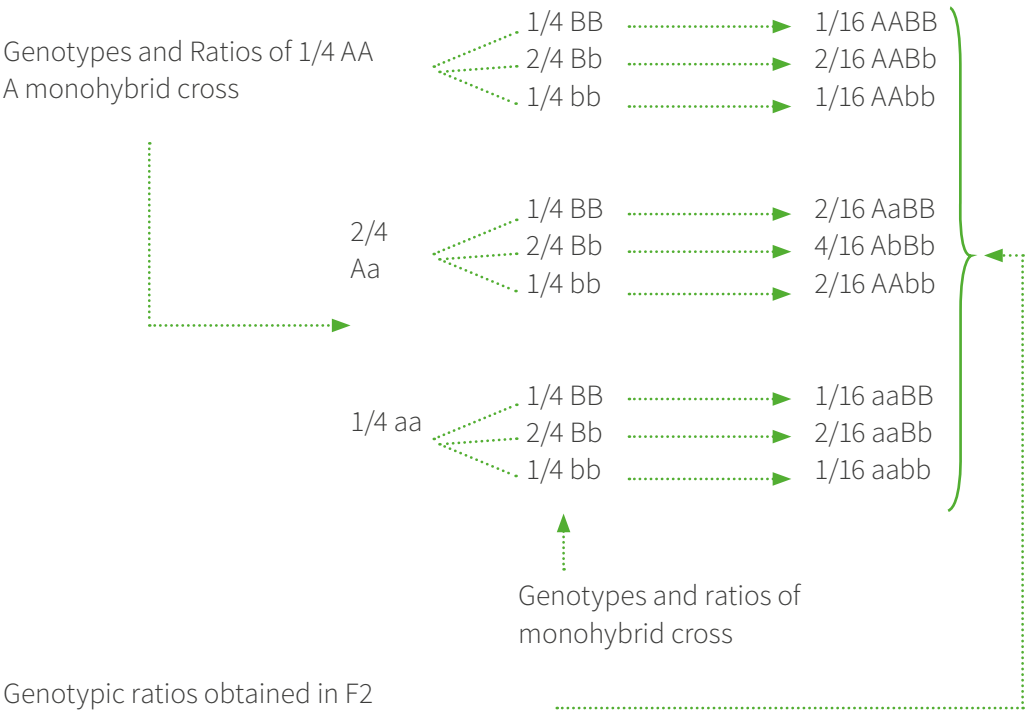
F1 zygotes : all of type

Plants with purple flowers

When crossing all the F1 individuals with each other, we have:



To determine the zygotes generated in the F2, we can use the gametic tree as follows:



Considering that the aa and bb genes are epistatic to A- and B-, the results are: 9/16 are plants with purple flowers, and 7/16 are plants with white flowers. This results in a phenotypic ratio of 9:7.

Simple Dominant Epistasis

If the dominant allele at one locus, for example, the allele M, produces a certain phenotype regardless of the allelic condition at the other locus (N), it is said that locus M is epistatic to locus N. The allele M can express itself in the presence of either N or n; only when the individual's genotype is homozygous recessive at the epistatic locus (mm) the alleles at the hypostatic locus (N or n) are expressed. In this regard, the M-N- and M-nn genotypes produce the same phenotype, while mm N- and mmnn result in two additional phenotypes; therefore, the classic ratio of 9:3:3:1 converts into 12:3:1, thus yielding three phenotypes.

Example:

In dogs, the T- gene governs the white color and is epistatic to the N- genes for black color and nn for brown color. If we cross a white dog with a TTNN genotype with a brown female with a ttnn genotype, what ratios would be obtained in the F2?

Using the same procedure as the previous problems, the following genotype ratios are obtained in F2: 9/16 are of the T-N- genotype, 3/16 are of the T-nn genotype, 3/16 are of the ttN- genotype, and 1/16 is of the ttnn genotype.

Considering that the T- gene is epistatic of N- and nn, the results are: 12/16 will be white dogs, 3/16 will be black dogs, and 1/16 will be brown dogs. Thus, the phenotypic ratio is 12:3:1.

Note. The student will carry out the explanation using the gametic tree diagram.

Double Dominant Epistasis

In this type of genetic interaction, the same phenotype can occur if two dominant genes or one dominant gene and one recessive gene are involved; recessive genes manifest a different phenotype. Essentially, individuals will not show the condition. In this genetic interaction, the phenotypic ratio is 15:1, and two phenotypes are obtained.

Example:

In poultry, certain breeds have feathers on their legs, while most breeds lack them. Apparently, the presence of feathers on the legs is due to the interaction of dominant genes, which are epistatic to recessive genes. What phenotypic ratio will be obtained in the F2 generation resulting from the cross between poultry with feathers on the legs (of genotype FFPP) and poultry without feathers on the legs (of genotype ffpp)?

Following the same procedure used in the previous problems, the following genotypic ratios are obtained in F2: 9/16 are of the F-P- genotype, 3/16 are of the F-pp genotype, 3/16 are of the ffP- genotype, and 1/16 is of the ffpp genotype.

Considering that the F- and P- and F-pp and ffP- genes produce poultry with feathers on the legs and ffpp genotype produces poultry without feathers on the legs, 15/16 poultry with feathers on the legs and 1/16 poultry without feathers on the legs are obtained. Thus, we obtain a phenotypic ratio of 15:1.

Note. The student will carry out the explanation using the gametic tree diagram.

Epistasis with Cumulative Effect

The classic 9:3:3:1 ratio becomes 9:6:1 when the dominant condition (either homozygous or heterozygous) at either locus (but not both) produces the same phenotype. This type of interaction produces three different phenotypes.

Example:

The red color of Duroc pigs is produced by the interaction between the H- and C- genes; the yellow color by the interaction of the hh and C- or H- and cc genes, and the white color, which is quite rare, is produced when the animal is homozygous recessive for the hhcc genes. From the cross between a yellow HHcc boar and a yellow hhCC sow, what will be the phenotypic expressions for the F2 generation?

Following the same procedure used in the previous problems, the following genotypic proportions are obtained in F2: 9/16 are of the H-C- genotype, 3/16 are of the H-cc genotype, 3/16 are of the hhC- genotype, and 1/16 is of the hhcc genotype.

Considering that the H- and C- genes produce red individuals, that the hhC- and H-cc genes produce yellow individuals, and that the hhcc genotype produces white

individuals, 9/16 red pigs, 6/16 yellow pigs, and 1/16 white pigs are obtained. Thus, we obtain a phenotypic ratio of 9:6:1.

Note. The student will carry out the explanation using the gametic tree diagram.

TABLE 5. *Summary of Epistatic Ratios Involving Two Gene Pairs*

Genotypes	A – B -	A - bb	aaB -	aabb
Normal ratio	9	3	3	1
Simple dominant epistasis	12		3	1
Simple recessive epistasis	9	3	4	
Duplicated genes with cumulative effect	9	6		1
Double dominant epistasis	15			1
Double recessive epistasis	9	7		
Interaction of dominants and recessives	13		3	

Source: Stansfield (2002)

8.5 ALLOSOMAL AND AUTOSOMAL INHERITANCE

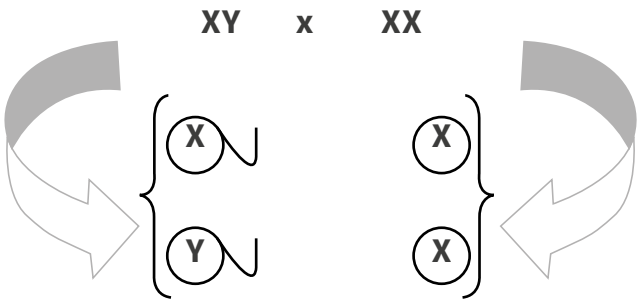
Sex is a biological character genetically determined in almost all sexed species. In most eukaryotic species, individuals belong to one of two sexes. In turn, in those most known species, such as animals and plants, the existence of the sexes is related to reproduction. Males and females produce different gametes through meiotic cell division that unite during fecundation process, resulting in offspring. In protists and fungi, reproduction is almost always asexual by mitosis, regardless of whether the sexes exist. Moreover, when they do exist, they are not usually morphologically differentiated; they are instead distinguished because cells of different sexes react with each other when they meet, producing sexual spores through meiosis (Stansfield, 2002).

8.6 MECHANISMS OF SEX DETERMINATION

It is known that mammals, Drosophila and humans, have a clearly identifiable sexual dimorphism in a pair of chromosomes: the allosomes or sex chromosomes (XX for females and XY for males) (Stansfield, 2002).

Therefore, if we cross a female and a male, the female will only produce gametes of X type while the male will produce gametes of X and Y types. In species with this mechanism of sexual determination, the female is known as the homogametic sex (since it produces gametes of only one type), and the male is called the heterogametic sex (since it produces two different types of gametes). Let us see in a scheme how this process occurs:

Sex of the offspring XX or female and XY or male



When we speak of sex-linked characteristics or characters, we refer to those present on the X chromosome, specifically in the non-homologous end.

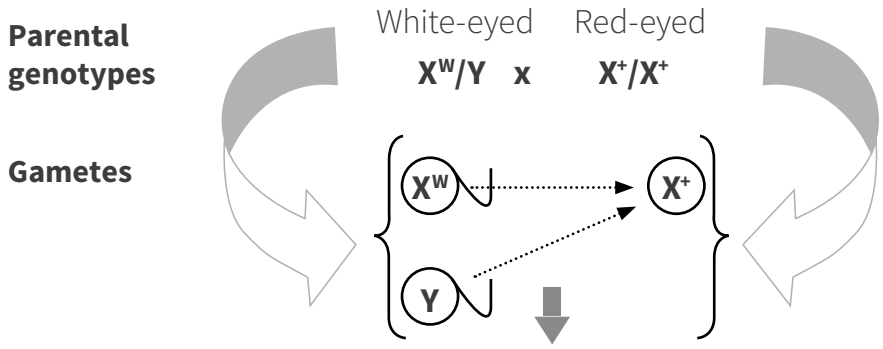
In the case of birds, the male is the homogametic sex (ZZ) since it has two equal chromosomes, and the female is the heterogametic sex (ZW). Therefore, the expression of sex-linked genes follows a scheme of alternating inheritance in the sexes, from mothers to granddaughters, through their carrier offspring.

If, for example, a Plymouth Rock rooster (with barred plumage) is crossed with a black Minorca hen, all the offspring will be barred. If, on the contrary, the crossbreeding is made between a black Minorca rooster and a barred Plymouth Rock hen, this female transmits the barred character only to her male offspring, while the females will be uniformly black. The distinction between the sexes can be seen from hatching; chicks have a lighter spot on their heads than the rest of their black feathers, while females are black.

Likewise, another type of sex determination can be found: the XO or ZO mechanism, which indicates the deficiency of one of the sex chromosomes (Stansfield, 2002).

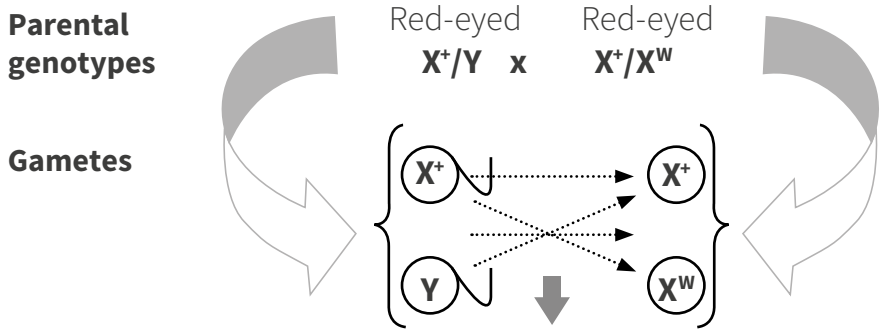
8.7 SEX-LINKED INHERITANCE IN DROSOPHILA

Thomas H. Morgan, circa 1920, gave one of the earliest pieces of evidence for sex-linked inheritance during his studies on eye color in Drosophila. Morgan established that the inheritance pattern of white eye color was a sex-linked characteristic, considering that red is both the standard eye color and dominant over white for flies. His experiment consisted of crossing a pure white-eyed (w) male with a pure red-eyed (++) female; in this way, he obtained in his F1 that all females and all males were red-eyed, indicating that the white-eyed trait was recessive (Stansfield, 2002).



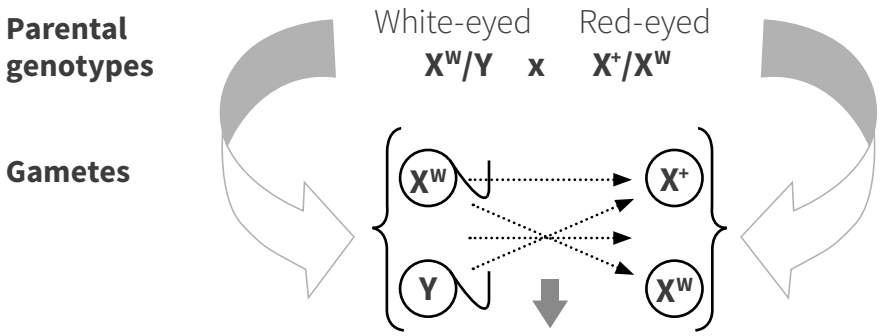
F1: X^+X^W red-eyed females : X +Y red-eyed males

The cross of F1 males and females with each other resulted in a phenotypic ratio of 3:1 in F2, which consists of 3/4 red-eyed individuals (2/4 females and 1/4 males) and 1/4 white-eyed individuals (males only).



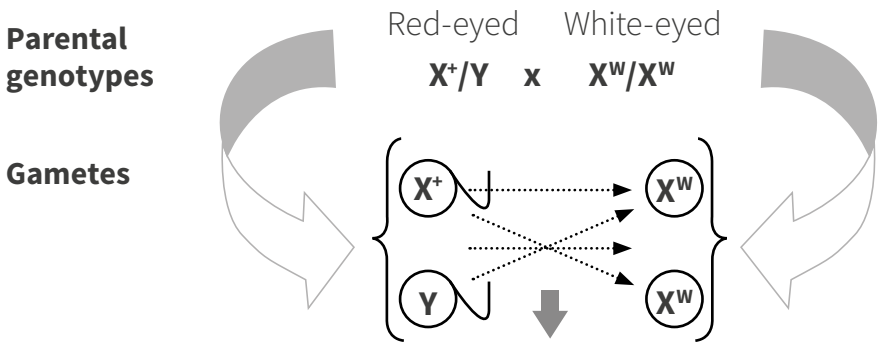
F2 :$X^{+}X^{W}$ 1/4 red-eyed females, $X^{+}X^{+}$ 1/4 red-eyed females, $X^{+}Y$ 1/4 red-eyed males, and $X^{W}Y$ 1/4 white-eyed males. Total: 2/4 red-eyed females, 1/4 red-eyed males, and 1/4 white-eyed males.

Subsequently, white-eyed males were crossed with red-eyed females (carriers), who were descendants of the previous cross, and red-eyed males, red-eyed females, white-eyed males, and white-eyed females were obtained (ratio 1: 1: 1: 1).



Generation: $X^{W}X^{+}$ 1/4 red-eyed females, $X^{W}X^{W}$ 1/4 white-eyed females, X+Y 1/4 red-eyed males, and $X^{W}Y$ 1/4 white-eyed males.

Finally, in a cross between pure white-eyed females and red-eyed males, all females were red-eyed and all males were white-eyed (ratio 1: 1).



Generation: $X^{+}X^{W}$ 1/2 red-eyed females $X^{w}Y$ and 1/2 white-eyed males.

This criss-cross inheritance is typical of sex-linked genes and particularly results because the Y chromosome does not carry alleles homologous to the white-eye locus of the X chromosome. Indeed, in most organisms, the Y chromosome has virtually no linked genes. Thus, males carry only one allele for sex-linked characters.

In typical diploid organisms with sex-determining mechanisms (XX - XY), a characteristic governed by a sex-linked recessive gene commonly manifests as follows:

- Males have this characteristic more frequently than females of the species.
- Females do not have it unless it also appears in the paternal parent.
- It rarely appears in the parent and child. It only occurs if the female parent is a carrier.
- The character can be transmitted through a series of carrier females; if so, the kinship relationship between affected males is established through females.

On the contrary, a characteristic governed by a sex-linked dominant gene commonly manifests as the following (Stansfield, 2002):

- Females have it more frequently than males of the species.
- If a male shows the characteristic, all his female offspring will have it.
- It is not transmitted to any offspring if the mother does not exhibit the characteristic.

8.8 SEX-LINKED INHERITANCE IN MAMMALS

Despite the extensive of research conducted in animals to detect sex-linked inheritance, only a few characters have been detected. For example, about 20 sex-linked genes have been found in mice. In domestic cats, it has been found that males can be black or yellow, while females can be black, tortoiseshell patterned (heterozygous), or yellow; these colors are governed by sex-linked inheritance (Stansfield, 2002).

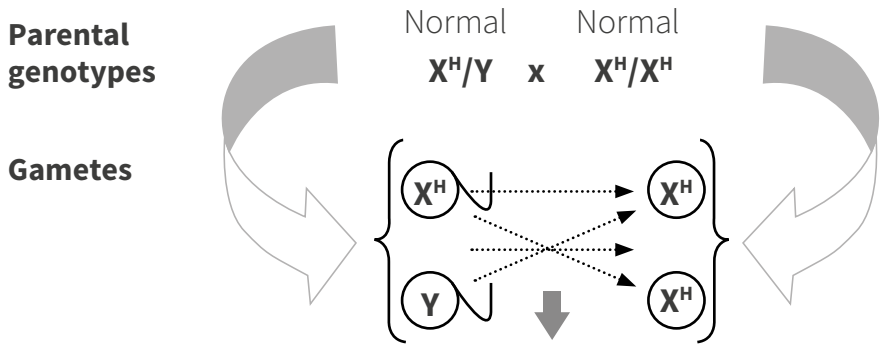
8.9 SEX-LINKED INHERITANCE IN HUMANS

Humans have 22 pairs of chromosomes (autosomes) and a sexual pair: the XX for female sex and the XY for male sex. For this reason, the male that determines the sex of a new being.

After analyzing the genes found in the non-homologous segment of humans' X chromosome, we can highlight, among others, genes causing color blindness, hemophilia, congenital cataracts, muscular dystrophy, testicular feminization

syndrome, absence of central incisors, distichiasis, and white patch of hair in the occipital region, etc. These characters are governed by both recessive and dominant genes (Stansfield, 2002).

To understand how they are inherited in humans, let us take the example of hemophilia, where, in terms of genotype, a normal female can be X^HX^H or X^HX^h A female hemophiliac would have the recessive gene for hemophilia XhXh on both chromosomes. In the cross between a normal man and a normal woman all children would be normal regardless of their sex.



Descendants: 1/2 normal X^HX^H females and 1/2 normal XH^Y males. All individuals resulting from the cross are normal.

Table 6 lists all the cross types, gametes, and ratio to be expected in the progeny of a sex-linked X^H YX^h and allele pair.

TABLE 6. *Types of Crosses, Gametes, and Expected Ratios in Offspring for a Sex-Linked Allele Pair XH and Xh*

Types of cross	Gametes		Progeny	
	Female	Male	Genotypes	Phenotypes
X^HX^H x X^HY	X^H, X^H	X^H, Y	X^HX^H X^HY	All normal
X^HX^h x X^HY	X^H , X^h	X^H , Y	X^HX^H X^HX^h X^HY X^hY	1/2 normal 1/2 transmitters 1/2 normal 1/2 affected

Types of cross	Gametes		Progeny	
	Female	Male	Genotypes	Phenotypes
$X^hX^h \times X^HY$	X^h , X^h	X^H , Y	X^HX^h X^hY	All transmitters All affected
$X^HX^H \times X^hY$	X^H , X^H	X^h , Y	X^HX^h X^HY	All transmitters All normal
$X^HX^h \times X^hY$	X^H , X^h	X^h , Y	X^HX^h X^hX^h X^HY	1/2 transmitters 1/2 affected 1/2 normal
			X^hY	1/2 affected
X^hX^h x X^hY	X^h , X^h	X^h , Y	X^hX^h X^hY	All affected

8.10 HOLANDRIC INHERITANCE IN HUMANS

Genes located on the non-homologous part of the Y chromosome determine the holandric inheritance. This inheritance is transmitted through the paternal line since any father who possesses the gene will transmit it to his sons and none of his daughters will have such characteristic; therefore, they will not transmit it to their progeny.

The classic example of this type of inheritance in humans is the character of ear hair (hypertrichosis) in men (Stansfield, 2002).

8.11 AUTOSOMAL INHERITANCE

This type of inheritance is associated with autosomes, i.e. the whole set of chromosomes that are part of the individual, excluding the sex chromosomes. In this kind of inheritance, two types are distinguished. One is autosomal dominant inheritance, which manifests in all generations and is transmitted only by affected individuals. Unaffected individuals or individuals that do not carry the character will not manifest it to their offspring, and the appearance and transmission of the character are not influenced by sex, i.e., males and females have the same probabilities of possessing or transmitting it. The other type of autosomal inheritance is recessive inheritance, which can manifest itself in an individual only if his/her parents are carriers of the character, so it is homozygous for this gene. This type of inheritance does not manifest itself in all generations.

Some diseases resulting from autosomal dominant and recessive inheritance are neurofibromatosis, achondroplasia, Marfan syndrome, myotonic dystrophy, sickle cell disease, Gaucher disease, cystic fibrosis, phenylketonuria, among others. Mendel's principle of inheritance can explain this type of inheritance (Stansfiend, 2002).

8.12 SEX-LINKED INHERITANCE

The genes for sex-influenced inheritance are carried on the autosomes, or homologous portions of the sex chromosomes, and their manifestation depends on the individual's sex. In the heterozygote, the genes manifest as dominant in the male and recessive in the female. An example of this type of inheritance is the presence or absence of horns in sheep.

TABLE 7. *Inheritance of Horns in Sheep; Sex-Influenced Character*

Genotype	Male phenotype	Female phenotype
CC	with horns	with horns
Cc	with horns	Hornless
cc	Hornless	Hornless

Another type of inheritance influenced by sex occurs in the European dairy cattle breed (Ayrshire) in which two types of hair colors are present: mahogany and white and red and white. If the male is heterozygous, its colors are mahogany and white, whereas if the female is heterozygous, she has red and white colors (Stansfield, 2002).

TABLE 8. *Inheritance of Coat Color in European Ayrshire Dairy Cattle*

Genotype	Male phenotype	Female phenotype
RR	Mahogany and white	Mahogany and white
Rr	Mahogany and white	Red and white
rr	Red and white	Red and white

Another type of inheritance influenced by sex is related to rooster's and hen's plumage. There is a known dominant gene (P) related to producing hen plumage in the hen. Females always have hen plumage regardless of their genotype (PP), (Pp), or (pp) because they do not have male hormones to produce rooster plumage. In roosters, the P gene prevents them from developing rooster plumage, so PP and Pp genotypes have hen plumage. Moreover, roosters of the pp genotype have rooster plumage. This character is limited to the rooster and is caused by the interaction of genes with hormones produced by the male.

TABLE 9. *Inheritance of Plumage in Roosters and Hens*

Genotype	Rooster phenotype	Hen phenotype
PP	Hen plumage	Hen plumage
Pp	Hen plumage	Hen plumage
pp	Rooster plumage	Hen plumage

In humans, two types of inheritance influenced by sex are premature baldness and shortness of the index finger, both dominant in men and recessive in women (Stansfield, 2002).

TABLE 10. *Sex-Influenced Inheritance in Humans: Premature Baldness and Shortness of the Index Finger*

Genotype	Men phenotype	Female phenotype	Genotype	Men phenotype	Female phenotype
AA	Bald	Bald	DD	Short finger	Short finger
Aa	Bald	Not bald	Dd	Short finger	Long finger
Aa	Not bald	Not bald	dd	Long finger	Long finger

The explanation of the reason for the type of inheritance influenced by sex, in the case of animals, is caused by the production of hormones in their endocrine glands, which are poured into the blood and influence the cellular activity of the tissues in joint action with the individual's own genes.

8.13 APPLICATION PROBLEMS

1. When homozygous yellow rats are crossed with homozygous black rats, the entire F1 is gray. After crossing F1 individuals, 20 yellow, 56 gray, 4 cream, and 16 black individuals were produced in F2.

- **a.** Indicate the appropriate symbols for each of the color's genotypes.
- **b.** How many of the 96 rats in F2 would be expected to be gray?
- **c.** How is the inheritance of these colors?

2. The white color in pumpkin fruit is caused by a dominant gene (A), and its recessive allele (a) gives rise to a colored fruit. The yellow color of the fruit is governed by a hypostatic gene of independent distribution (P), while the green fruit is by its recessive allele (p). When dihybrid individuals mate, the offspring have a ratio of 12 whites: 3 yellow: 1 green. What ratio of fruit color is expected from mating:

- **a.** AApp x AaPp
- **b.** AaPp x aapp
- **c.** AaPp x AaPp

3. Two types of genes located on chromosomes three and nine are in maize, which are dominant and produce a color aleurone: B1 and B2 genes, respectively. All other combinations result in a colorless aleurone. Two pure, uncolored strains are mated in F1 all colored.

- **a.** What are the genotypes of the F1 parents?
- **b.** What phenotypic ratios can be expected in F2?
- **c.** What is the genotypic ratio of color in F2?

4. In onion the color of the bulb is given by two pairs of alleles. Moreover, all offspring are red when a pure red strain is crossed with a pure white strain. In F2, they result from several F1 crossovers: 94 white, 76 yellow, and 218 red onions.

- **a.** To what epistatic ratio do these data approximate? Use X.
- **b.** What is the name of this type of gene interaction?

5. The R and S genes are necessary for hearing and speaking normally in humans. Any combination of one of the dominants with its recessive non-allele in its homozygous state and the two recessives in the homozygous state causes an individual to be deaf-mute.

a. What type of gene action is this?
b. From the mating of individuals RrSs x RrSs, RrSs x RRSs, RrSs x rrss, find the ratio of normal and deaf-mute individuals in each cross.

6. The red color of Duroc Jersey pigs is produced by the interaction of the R and A genes, the yellow color by the interaction of the r and A or R and a genes, and the white color, which is quite rare, is produced when the animal is recessive for the r and a genes.

a. Determine the phenotypic ratios to be obtained from the following crosses: Rraa x rrAA, Rraa x rrAa, RrAa x RrAa, and RrAa x RRAA.
b. If a producer has red and yellow animals in his/her herd and wishes to have yellow animals, what procedures would you suggest from a genetic point of view?

7. In Drosophila, the gene for yellow body color is recessive and sex-linked. Its y+ dominant allele produces wild-type body color. What phenotypic ratios are expected from crosses between:

a. Yellow male x yellow female
b. Yellow female x wild male
c. Wild (homozygous) female x yellow male
d. Wild-type female carrier x yellow male.

8. Sex determination in the plant genus Melandrium is similar to that in humans. A sex-linked gene (l) is lethal in homozygous females. When found in hemizygous condition in males (lY), it produces a yellowish-green stain. The homozygous or heterozygous condition of the wild-type allele (Ll or LL) in females or the hemizygous condition in males (LY) produces a standard dark green color. Predict the phenotypic ratio expected from a cross between heterozygous females and yellowish-green males.

9. An autosomal recessive gene (tra), transforms a female (X/X) Drosophila into a phenotypic male in a homozygous condition. These “transformed” males are sterile. The gene has no effect in males (XY). A cross is made between a female heterozygous at the tra locus and a male homozygous recessive for the same gene. What sex ratio is expected in F1 and F2?

10. There is a dominant sex-linked B gene that produces white bars in the black plumage of adult chicks, as seen in the barred Plymouth Rock breed. Newly hatched chicks that will later be barred show a white spot on the top of the head.

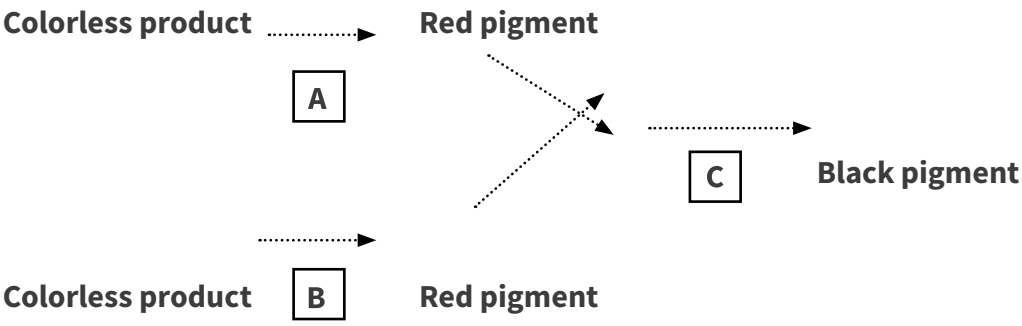
a. Diagram to F2 the cross between a homozygous barred male and a non-barred female.
b. Diagram the reciprocal cross to F2 between a homozygous non-barred male and a barred female.
c. Are both crosses useful for sexing F1 chicks at hatching?

11. Black, sepia, and albino are phenotypes of laboratory guinea pig coats. Individual animals (not necessarily pure lines) exhibiting these colors were crossed with each other; the results are shown in the table below, where the abbreviation B is used for black, S for sepia, C for cream, and A for albino. The number of individuals obtained for each phenotypic class were:

Crosses	Parental phenotypes	Offspring phenotypes			
		N	**S**	**C**	**A**
1	NXN	22	0	0	7
2	NXA	10	9	0	0
3	CXC	0	0	34	11
4	SXC	0	24	11	12
5	NXA	13	0	12	0
6 7 8 9 10	NXC NXS NXS SXS CXA	19 18 14 0 0	20 20 8 26 0	0 0 6 9 15	0 0 0 0 17

a. Deduce the mode of inheritance of these coat colors by choosing your own genetic symbols. Indicate all genotypes of parents and offspring.
b. If the black offspring of crosses 7 and 8 are crossed with each other, what genotypic and phenotypic proportions would be obtained?

12. A, B, and C are independently secreted genes that control the production of a black pigment in animals. These genes are involved in the following metabolic pathway.



a, b, and c are the alleles of the respective genes. Crossing a pure black individual for the three genes with a recessive individual for the same three genes results in F1 black individuals. These individuals self-cross to produce F2.

a. What ratio of the F2 will be colorless?
b. What ratio of F2 will be red?
c. What ratio will be black?

If the F1 black individuals are test-crossed, what is the probability that they will all be black?

BIBLIOGRAPHY

Griffiths, A. J. F., Gelbart, W. M., Miller, J. H., & Lewontin, R. C. (2002). Genética moderna (Modern Genetic Analysis). McGraw-Hill (Interamericana de España), Madrid.

Forero, G. y Bernal, L. (2013a). Módulo de genética básica. Universidad Nacional Abierta y a Distancia. Colombia.https://xdoc.mx/documents/universidad-nacional-abierta-ya-distancia-5dd44d8032680

Forero, G. y Bernal, L. (2013b). Aula virtual inteligente para el curso de genética básica. Universidad Nacional Abierta y a Distancia. Colombia. https://xdoc.mx/documents/universidad-nacional-abierta-ya-distancia-5dd44d8032680

Karp, G. (2019). Biología celular y molecular. Ed. McGraw-Hill.

Magnaplus. (s.f.). ADN. Acido desoxirribonucleico. Plataforma educativa – Magnaplus. https://www.magnaplus.org/articulo/-/articulo/AD3155/adn-acido-desoxirribonucleico-

Sadava, D. y Purves, W. (2009). Vida / Life: La ciencia de la biología / The Science of Biology. Ed. Médica Panamericana.

Raisman, J., & González M. (2008a). Hipertextos de Biología. http://www.biologia.edu.ar/cel_euca/celula1.htm

Raisman, J., & González M. (2008b). Hipertextos de Biología. http://www.biologia.edu.ar/plantas/cell_vegetal.htm

Mosso Elizabeth., (2010a). Biología. Editorial Maipue. https://elibro-net.bibliotecavirtual.unad.edu.co/es/ereader/unad/78974?page=78

Mosso Elizabeth., (2010b). Biología. Editorial Maipue. https://elibro-net.bibliotecavirtual.unad.edu.co/es/ereader/unad/78974?page=85

Burriel Martí, F., & Lucena Conde, F. (2017). Química analítica cualitativa. In Química analítica cualitativa.

Coll, V. B. (2007). Estructura y propiedades de los Ácidos Nucleicos. Química Aplicada a la Ingeniería Biomédica.

Sello Editorial
Universidad Nacional
Abierta y a Distancia

**UNIVERSIDAD NACIONAL ABIERTA
Y A DISTANCIA (UNAD)**

Sede Nacional José Celestino Mutis
Calle 14 Sur 14-23
PBX: 344 37 00 - 344 41 20
Bogotá, D.C., Colombia

www.unad.edu.co

9 780201 379624