

Biomedical
Science

Molecular
Genetics



BMS23010C | Fall 2026-2027

Molecular Genetics & Processes

Molecular Structure of Chromosomes



Free Session

A+ is more than just a platform



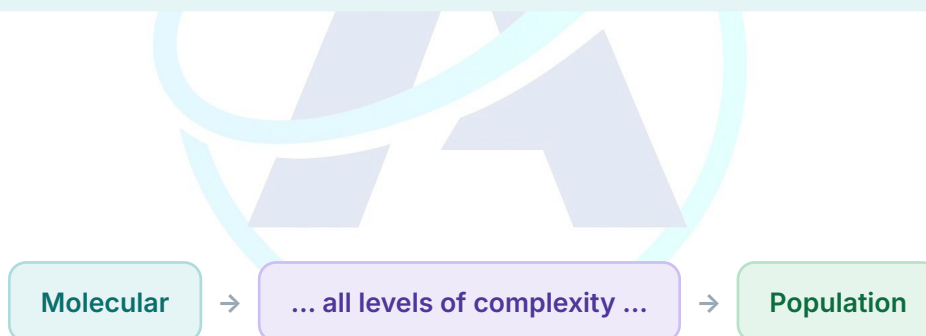


What is Genetics?

Genetics is the branch of biology that deals with **heredity and variation**.

A UNIFYING DISCIPLINE

Unifying discipline in biology allowing us to understand how life can exist at **all levels of complexity**, ranging from the molecular to the population level.



Genetics is centered on the **study of genes**:

A gene is classically defined



unit of heredity

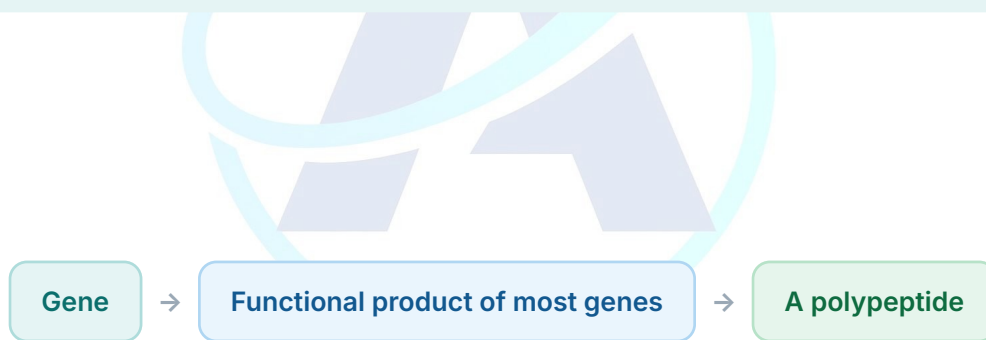


What is a Gene?

What is a **gene** at a molecular level?

DEFINITION

A **gene** is a segment of **DNA** (DeoxyriboNucleic Acid) that contains the information to produce a **functional product**.



GENES ARE COMMONLY DESCRIBED ACCORDING TO THE WAY THEY AFFECT TRAITS

Eye color

Hair texture

Height...

Living Cells Are Composed of Biochemicals

- 1 DNA is formed from the linkage of **nucleotides**, producing a very long macromolecule.
- 2 The DNA associates with **proteins** to form a **chromosome**.
- 3 The chromosomes are located within a membrane-bound organelle: the **nucleus**.

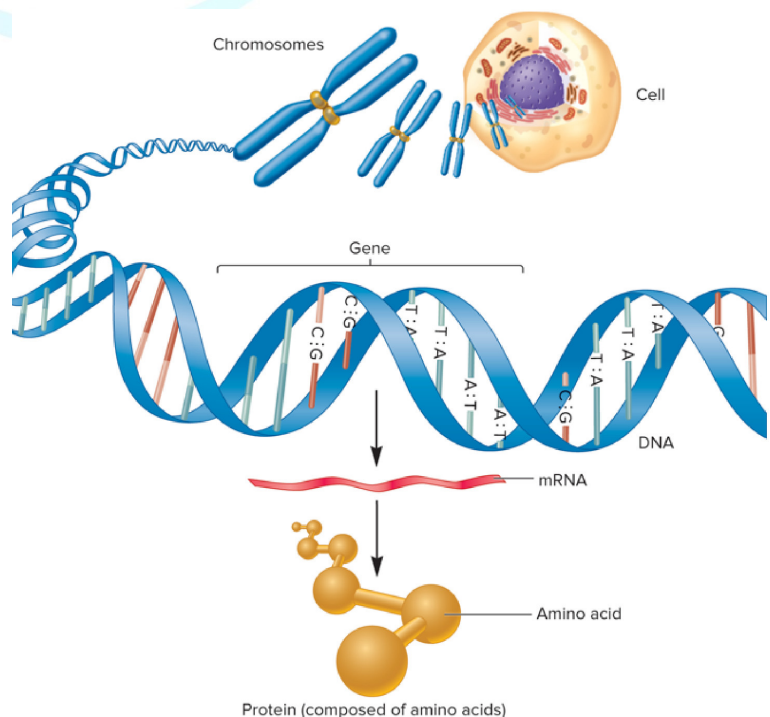
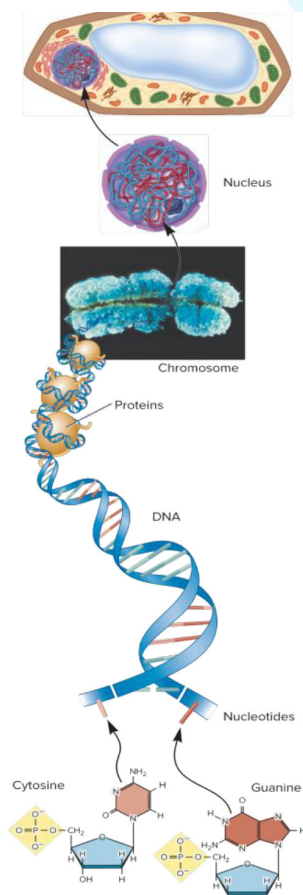


DNA, the Molecule of Life

The adult human body is composed ? of cells.

MOST HUMAN CELLS CONTAIN THE FOLLOWING

- ? human chromosomes, found in ? pairs
- ? of DNA
- Approximately ? genes coding for proteins that perform most life functions
- Approximately **3 billion** DNA base pairs per set of chromosomes, containing the bases **A, T, G, and C**



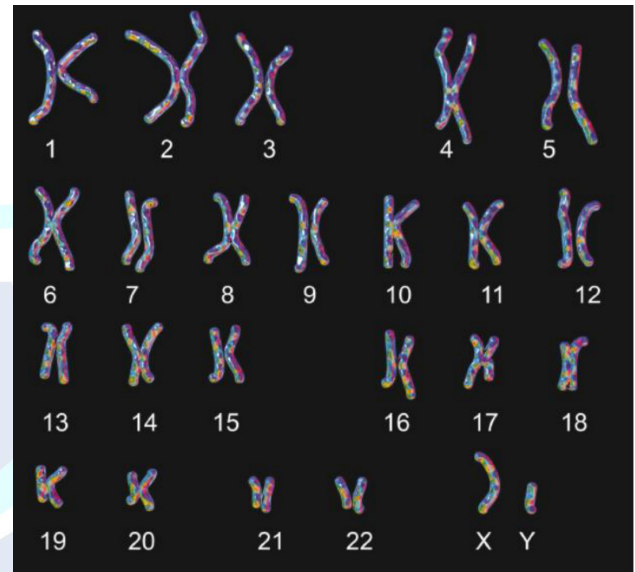


Chromosomes & the Karyotype

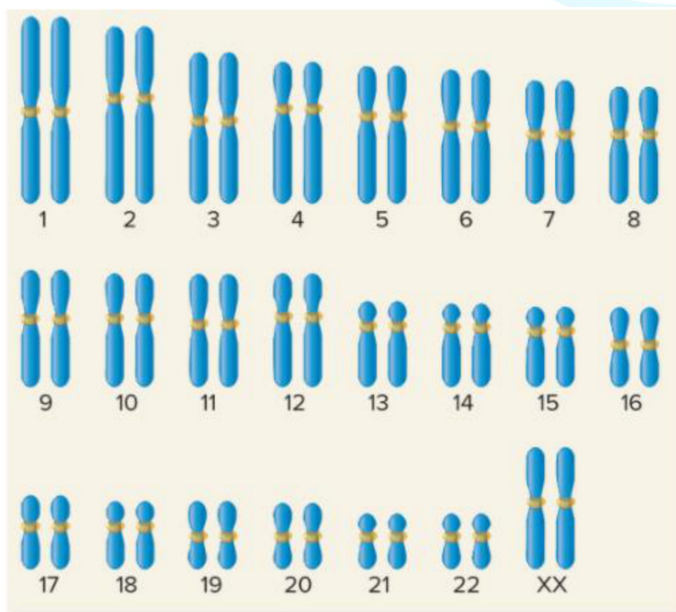
In living cells, the DNA is found within large structures known as **chromosomes**.

Karyotype

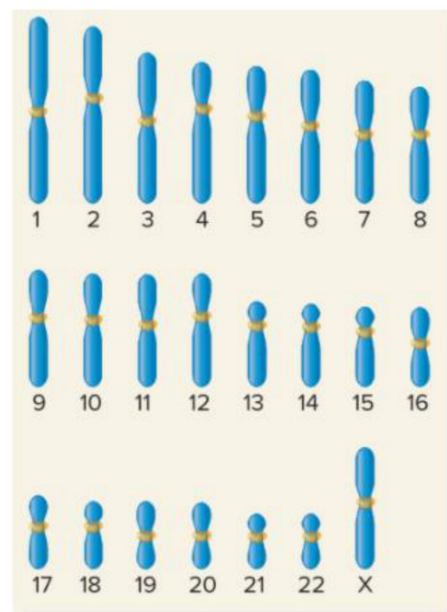
Photographic representation of all the chromosomes. It reveals **how many chromosomes** are found within an actively dividing **somatic cell**.



A micrograph of the 46 chromosomes found in a cell from a human...



(a) Chromosomal composition found in human somatic cells of females (chromosomes)



(b) Chromosomal composition found in a human gamete (chromosomes)



Proteins Determine Cell Structure & Function

Each cell contains many different **proteins** that determine cell structure and function.

TUBULIN

Assembles into large structures known as **microtubules**, which provide the cell with **internal structure and organization.**

ENZYMES

Which **accelerate chemical reactions**, important category of proteins.

Catabolic enzymes

Use of energy

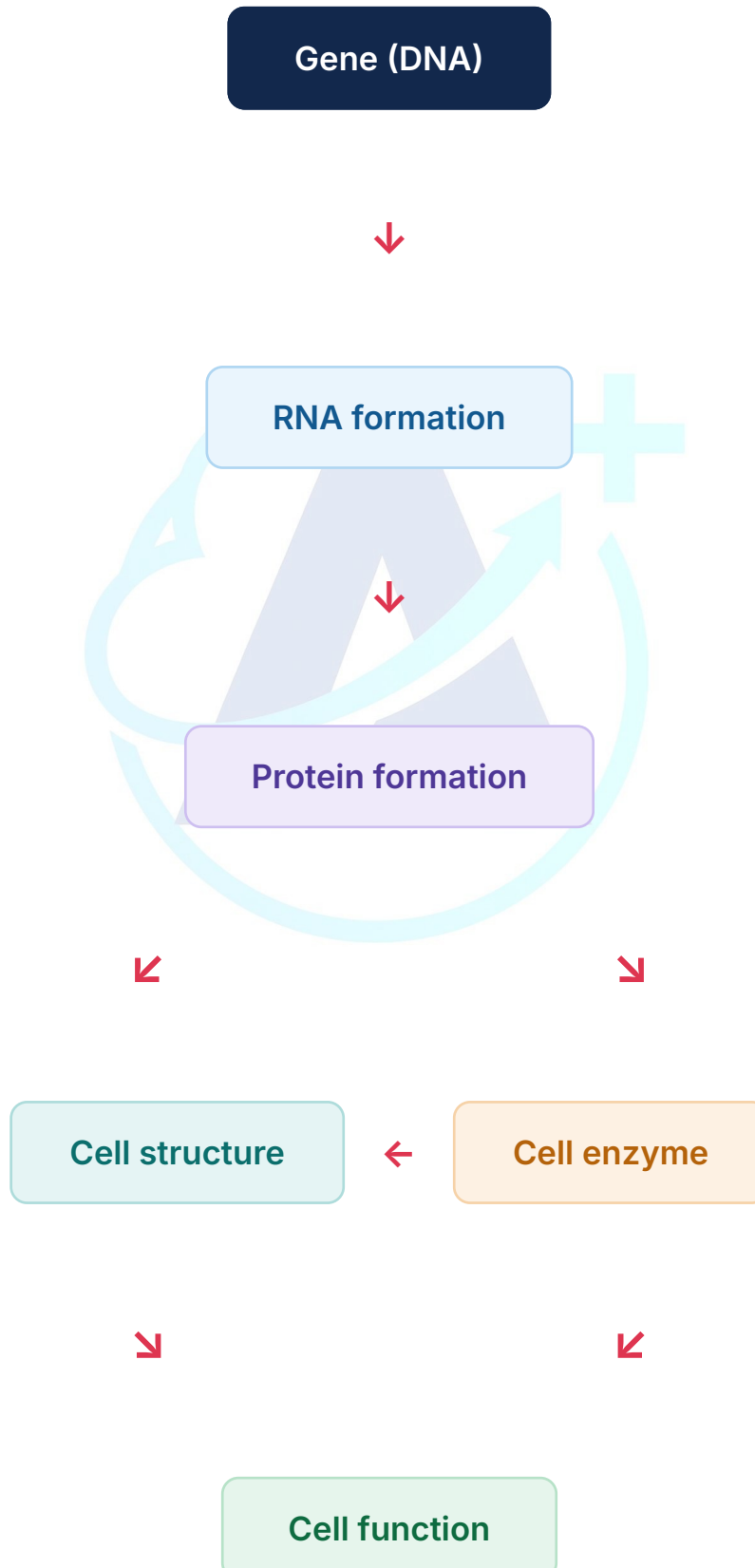
Anabolic enzymes

Synthesis of molecules...

The construction of a cell greatly depends on its proteins that are involved in **anabolism.**



From Gene to Cell Function





DNA Stores the Information for Protein Synthesis

DNA is composed of a linear sequence of **nucleotides** — adenine (A), thymine (T), guanine (G), or cytosine (C).

A — adenine

T — thymine

G — guanine

C — cytosine

DNA sequences within most genes contain the information to direct the order of amino acids within **polypeptides** according to the **genetic code**.

DNA SEQUENCE

AMINO ACID SEQUENCE

ATG GGC CTT AGC

Methionine Glycine Leucine Serine

TTT AAG CTT GCC

Phenylalanine Lysine Leucine Alanine



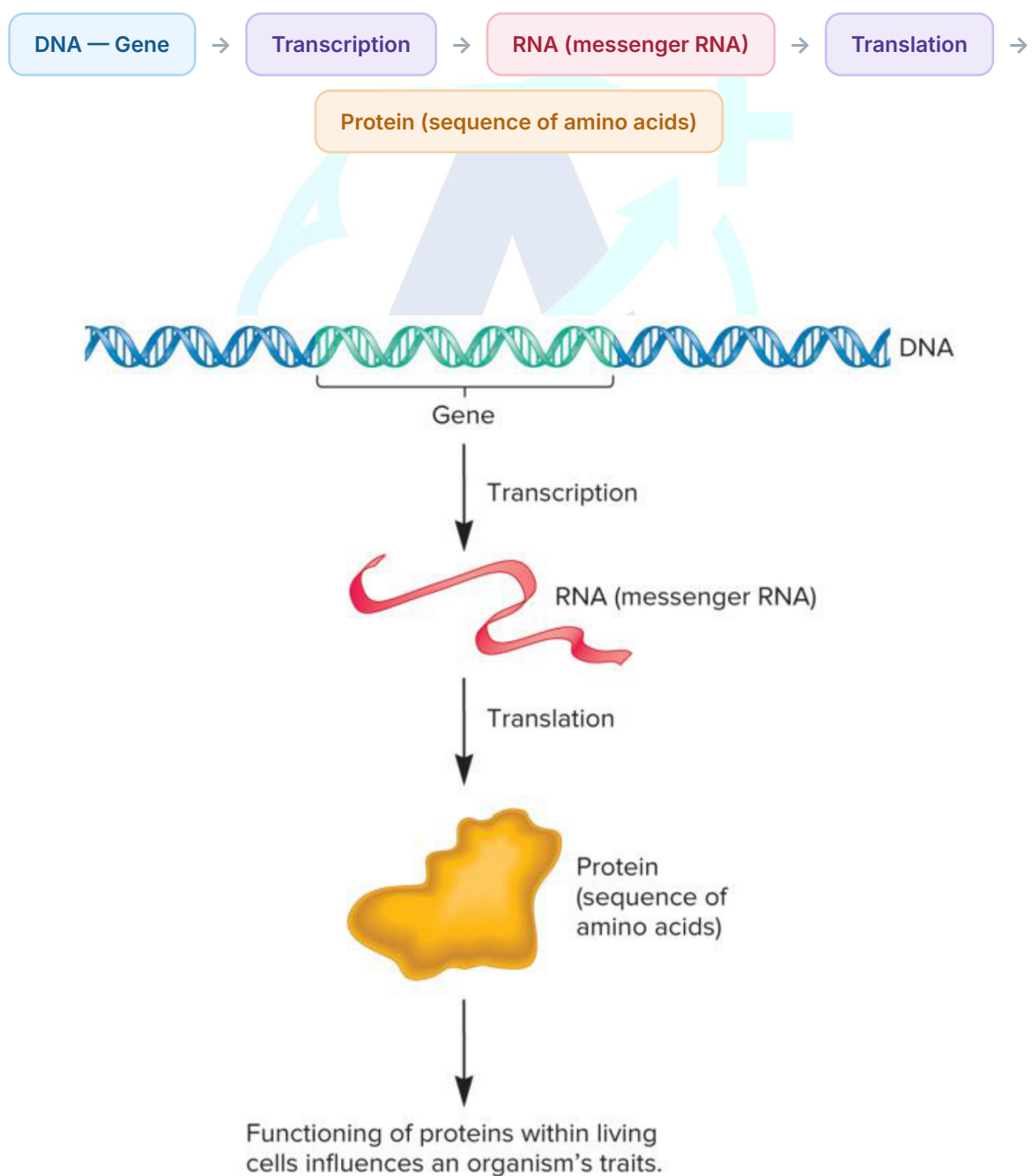
The Genetic Code

		Second Letter					
		U	C	A	G		
1st letter	U	UUU Phe UUC UUA Leu UUG	UCU UCC Ser UCA UCG	UAU Tyr UAC UAA Stop UAG Stop	UGU Cys UGC UGA Stop UGG Trp	U C A G	3rd letter
	C	CUU CUC Leu CUA CUG	CCU CCC Pro CCA CCG	CAU His CAC CAA Gln CAG	CGU CGC Arg CGA CGG	U C A G	
	A	AUU AUC Ile AUA AUG Met	ACU ACC Thr ACA ACG	AAU Asn AAC AAA Lys AAG	AGU Ser AGC AGA Arg AGG	U C A G	
	G	GUU GUC Val GUA GUG	GCU GCC Ala GCA GCG	GAU Asp GAC GAA Glu GAG	GGU GGC Gly GGA GGG	U C A G	



Gene Expression

The information in DNA is accessed during the process of **gene expression**.

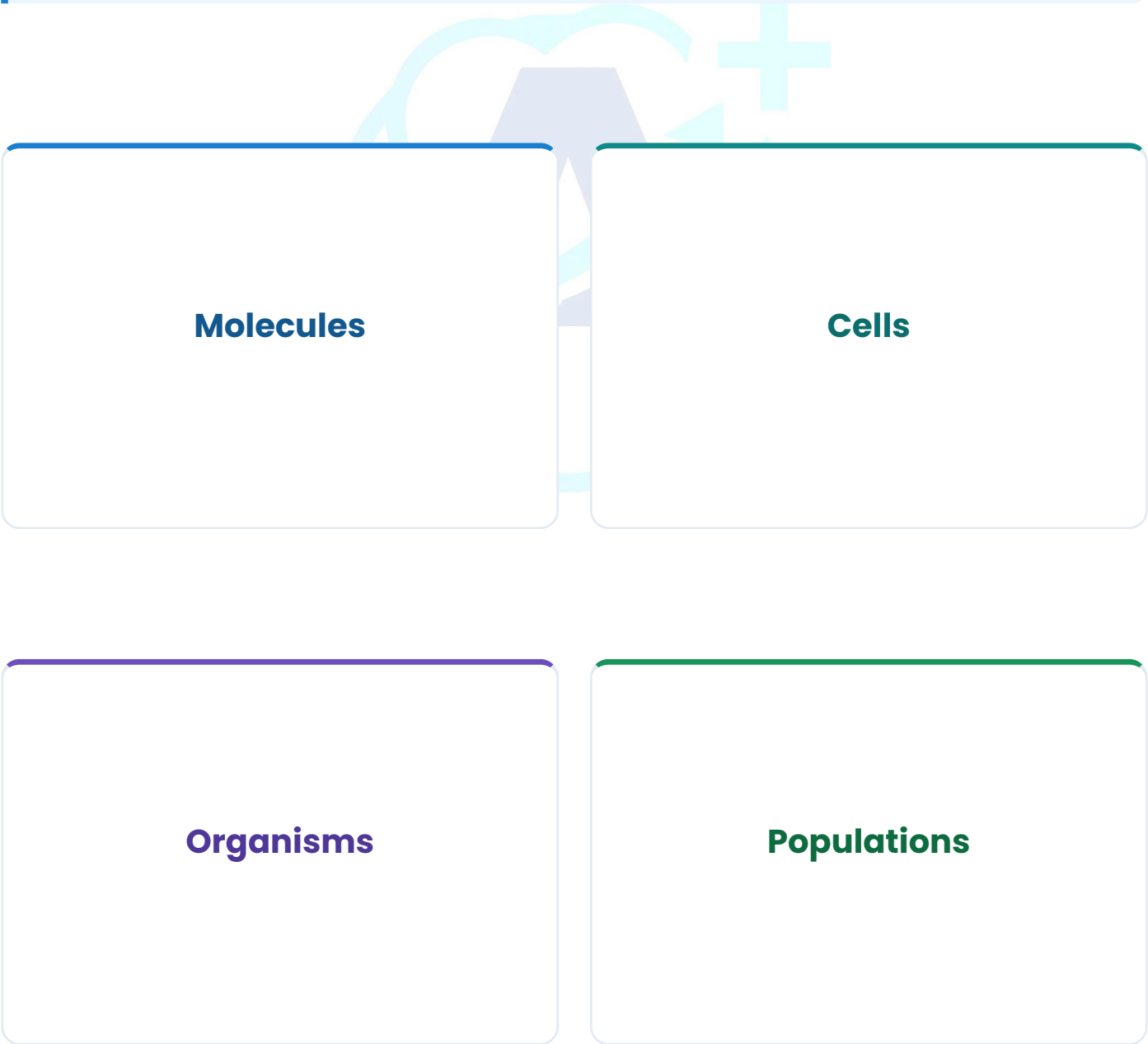




The Relationship Between Genes and Traits

The molecular expression of genes leads to an **organism's traits**.

Observations and theories of Genetics span **four levels** of biological organization:



Molecules

Cells

Organisms

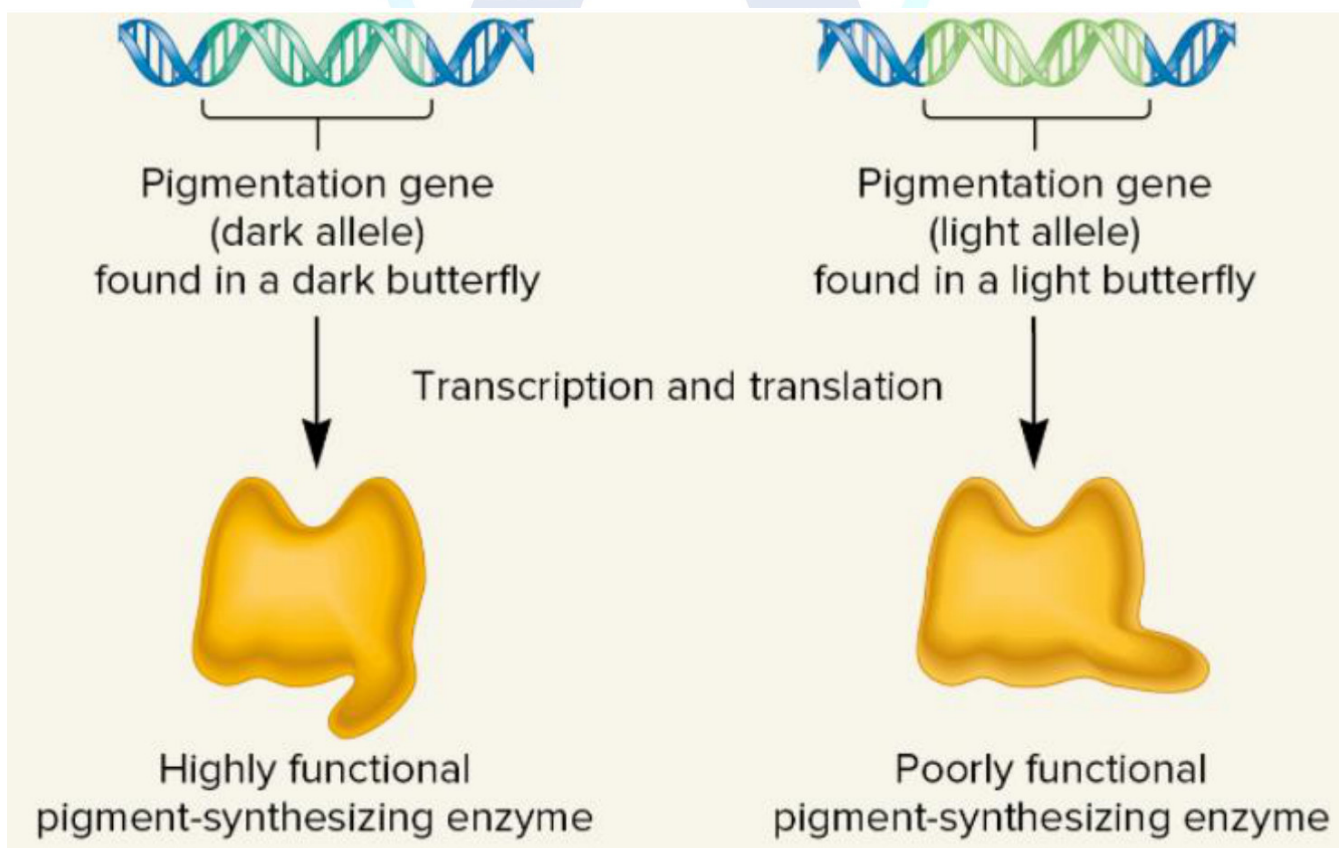
Populations



Molecular Level

Genes are expressed at the **molecular level**.

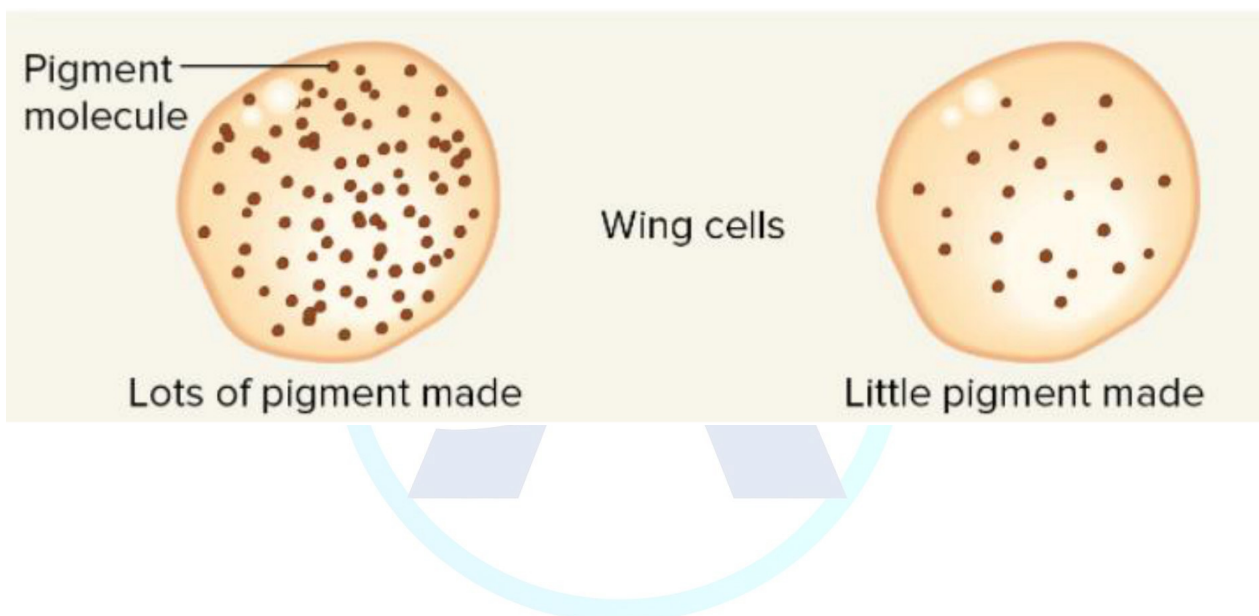
Gene **transcription** and **translation** lead to the production of a particular **protein**.





Cellular Level

Proteins often function at the **cellular level**. The function of a protein within a cell affects the **structure and working** of that cell.



Organism Level

The trait of dark butterfly color is an observation at the **organism level**. Yet the trait is rooted in the **molecular characteristics** of the organism's cells.





Population Level

The occurrence of a trait within a species is an observation at the **population level**.

Trait predominates within a population



Promotes the reproductive success of the members of the population



Evolution of beneficial traits



Dark butterflies usually live in forested areas.



Light butterflies usually live in unforested areas.



Inherited Differences Are Due to Genetic Variation

Variation in traits among members of the same species is **very common**.

People have **brown hair** / **blond hair**

Roses have **red flowers** / **white flowers**

GENETIC VARIATION

Differences in **inherited traits** among individuals within a population.



Why do these two frogs look so different?





Changes in the Nucleotide Sequence of DNA

Changes in the nucleotide sequence of DNA underlie the **genetic variation** that we see among individuals.

At the molecular level, genetic variation can be attributed to **different types of modifications**:

- 1 Small or large differences can occur within gene sequences. When such changes initially occur, they are called **gene mutations**.
- 2 Major alterations can also occur in the **structure of a chromosome**. A large segment of a chromosome can be lost, rearranged, or reattached to another chromosome.
- 3 The **total number of chromosomes**. In some cases, an organism may inherit one too many or one too few chromosomes. In other cases, it may inherit an extra set of chromosomes.

EXAMPLE

A person with **Down syndrome** has **47 chromosomes** rather than the common number of 46, because of an extra copy of chromosome 21.



Check Your Understanding

1 At which of the following levels can gene expression be observed?

1. Molecular and cellular levels

2. Organism level

3. Population level

4. All of the above

2 Variation in the traits of organisms may be attributable to

1. gene mutations.

2. alterations in chromosome structure.

3. variation in chromosome number.

4. all of the above.

3 A human skin cell has 46 chromosomes. A human sperm cell has

1. 23.

2. 46.

3. 92.

4. None of the above is the number of chromosomes in a sperm cell

4 Evolutionary change caused by natural selection results in species with

1. greater complexity.

2. less complexity.

3. greater reproductive success in their environment.

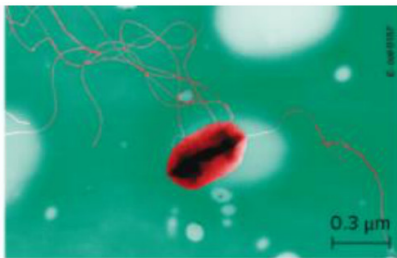
4. the ability to survive longer.



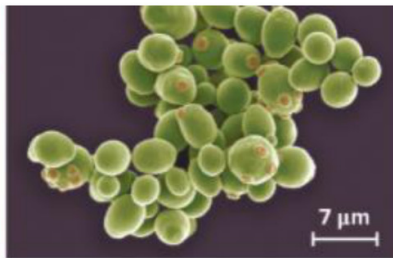
Fields of Genetics

Genetics is a **broad discipline** encompassing molecular, cellular, organism, and population biology.

Experimentally, geneticists often focus their efforts on **model organisms**.



(a) *Escherichia coli*



(b) *Saccharomyces cerevisiae*



(c) *Drosophila melanogaster*



(d) *Caenorhabditis elegans*



(e) *Mus musculus*



(f) *Arabidopsis thaliana*

Can you think of another example of a **model organism**?

THREE MAJOR FIELDS OF GENETICS

Transmission

Molecular

Population genetics

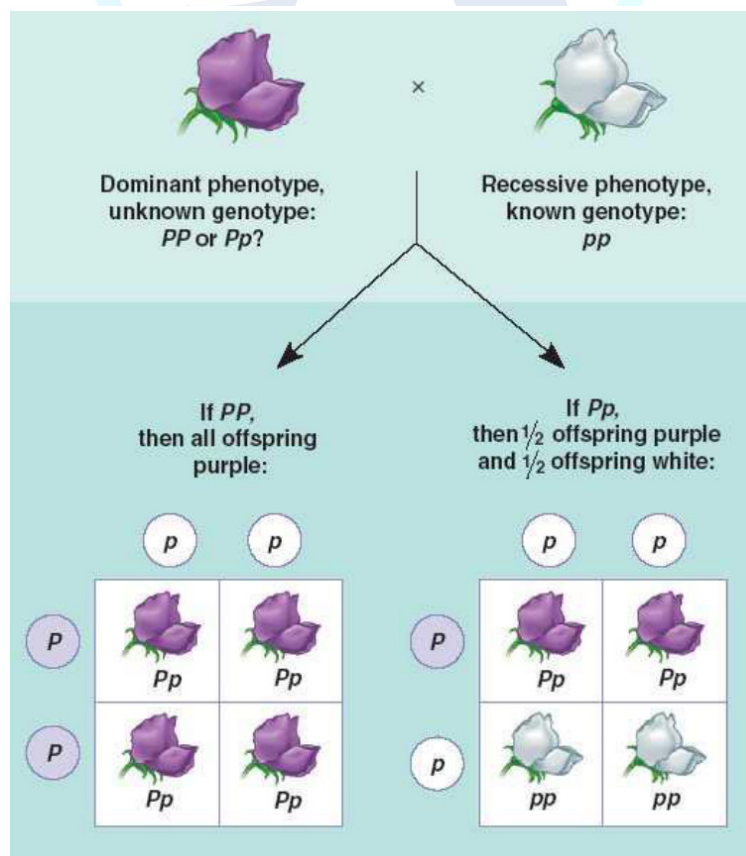


Transmission Genetics

Explores the **inheritance patterns of traits** as they are passed from parents to offspring.

Fundamental technique used by a transmission geneticist is the **genetic cross**:

The breeding of **two selected individuals** and then analyzing their offspring in an attempt to understand how traits are passed from parents to offspring.





Molecular Genetics

Focuses on a **biochemical understanding** of the hereditary material.

- 1 Understand how the genetic material works at the **molecular level**.
- 2 Understand the molecular features of DNA and how these features underlie the **expression of genes**.
- 3 Study **mutant genes** that have abnormal function.

Analyzes the effects of a gene mutation that eliminates the function of a gene: **loss-of-function mutation** — which reveals the role of the functional, nonmutant gene.



Population Genetics

Is concerned with **genetic variation and its role in evolution**.

- Population geneticists have developed **mathematical theories** to explain the prevalence of certain alleles within populations of individuals.
- Population geneticists are particularly interested in genetic variation and how that variation is related to an **organism's environment**.



To Summarise...

SCOPE

4

Levels of organization

- Molecules, cells, organisms, populations
- Genetics is a unifying discipline in biology
- Spans the molecular to the population level

GENE

DNA

A segment of DNA

- Contains information for a functional product
- That product is usually a polypeptide
- Described by the traits they affect

CELLS

46

Chromosomes and karyotype

- DNA + proteins form a chromosome in the nucleus
- Karyotype shows all chromosomes of a somatic cell
- A skin cell has 46; a sperm cell has 23

CODE

ATGC

The genetic code

- DNA is a linear sequence of nucleotides
- Sequences direct the order of amino acids
- Transcription then translation give a protein

VARIATION

3

Types of modification

- Gene mutations within gene sequences
- Alterations in chromosome structure
- Variation in total chromosome number

FIELDS

3

Branches of genetics

- Transmission — inheritance patterns
- Molecular — the hereditary material
- Population — variation and evolution

MODEL ORGANISMS STUDIED BY GENETICISTS

Escherichia coli

Saccharomyces cerevisiae

Drosophila melanogaster

Caenorhabditis elegans

Mus musculus

Arabidopsis thaliana