

Biomedical
Science

Molecular
Genetics



BMS 23010B | Fall 2026-2027

Gene Expression

Overview of Genome Structure



Free Session

A+ is more than just a platform



[Academy Website](#)



[WhatsApp Community](#)



Four Words You Need First

Nucleus

The organelle that **contains DNA**.

DNA

The nucleic acid that carries the cell's **genetic information and hereditary characteristics** via its nucleotides.

Chromosome

A threadlike structure of nucleic acids and protein found in the nucleus of most living cells. Chromosomes are made up of **long lengths of DNA**.

Gene

A short length of DNA that carries the genetic code for a **particular characteristic or cell activity**. Different forms of the same gene are called **alleles** — dominant or recessive.

Nucleus



Chromosome



DNA



Gene



Nucleic Acids

DR. RAZA ONLY

- Nucleic acids are the **genetic materials** of life.
- They are **macromolecules essential to all known forms of life**.
- DNA serves as the genetic material in **all living organisms on earth**.

FOUR CRUCIAL CHARACTERISTICS

Replication

Storage of
information

Expression of
information

Variation by
mutation

DNA

Deoxyribonucleic acid

RNA

Ribonucleic acid

They carry information in cells and express that information **inside and outside the cell nucleus**.



The Nucleus & Chromatin

DR. KHAOULA ONLY

- A **double membrane-bound organelle**, located centrally and only in a eukaryotic cell.
- It **encloses the DNA** — the genetic material.
- The most important and defining feature of all higher organisms, plant and animal cells alike.
- Its main function: **control and coordinate the functioning of the entire cell**.

CHROMATIN — TWO STATES OF THE SAME MATERIAL

A **complex of genetic material** (DNA or RNA) and proteins found in a resting or non-dividing cell nucleus, classified into two types based on function.

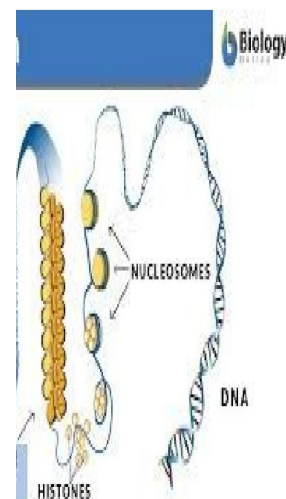
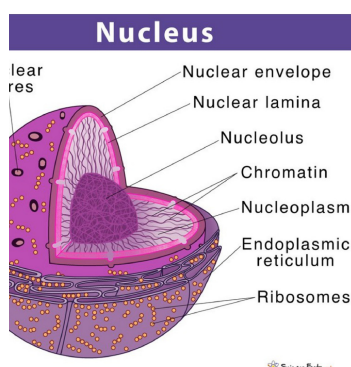
Heterochromatin

A functionally **inactive** form of chromatin, found near the nuclear envelope.

Euchromatin

A mild, **less condensed** form that is in a functionally **active** state.

Highly condensed and paired chromatin = a chromosome





How the Double Helix Was Found

Flagged in the lecture as **NOT IMPORTANT FOR EXAMS** — background story only.

1

Photo 51

Rosalind Franklin, an expert in X-ray crystallography, took the image known as Photo 51. She had done X-ray chromatography of DNA but was **unable to prove** the number of strands or the precise chemical organization of the molecule.

2

A rejected model

Two PhD students, **Watson and Crick**, working in the same lab, proposed a **three-stranded, inside-out model** — which Franklin rejected.

3

The photo changes hands

Her confidential lab data and Photo 51 were **shown to Watson and Crick** by another scientist. Watson recalled that on seeing it, "my mouth fell open and my pulse began to race." Photo 51 provided the **vital clue**.

4

The structure falls out

They found the relative distances of the repetitive elements and the dimensions of the monoclinic unit cell — which showed the molecule was in **two matching parts running in opposite directions**.

5

28 February 1953

Watson and Crick announced the **double-helix structure**: a spiral of two DNA strands, each a long chain of monomer nucleotides, wound around each other. The **Nobel Prize** followed in 1962. **Rosalind Franklin had died of ovarian cancer in 1958** and never learned how far they had relied on her data.



Three Forms of DNA

DR. KHAOULA ONLY

A-DNA

Right-handed — the spiral turns **clockwise** as it moves away from you.

One of the **less common** forms; typically found under **dehydrated conditions** or when DNA is bound to certain proteins.

B-DNA

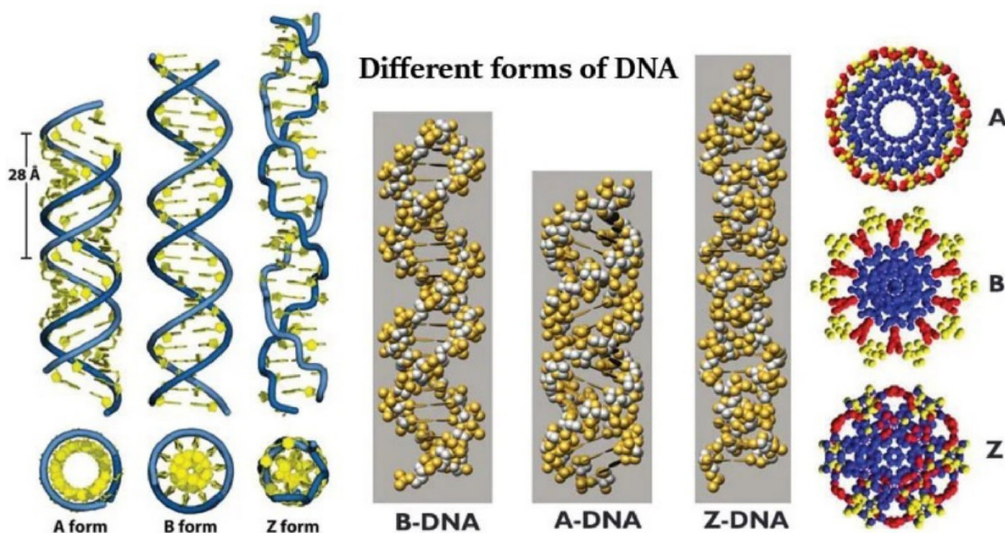
Right-handed, and the **most common and stable** form under physiological conditions.

The form most people picture when thinking about DNA structure.

Z-DNA

Left-handed — the spiral turns **counterclockwise**.

A **zigzag** pattern in its sugar-phosphate backbone gives it its name. Less common; often found in regions involved in **gene expression**.



The different forms of DNA

PART 2

Building Blocks



A nucleotide has three parts. Get those three right and the rest of the chemistry — the backbone, the directionality, the energy — follows from them.

[Base, sugar, phosphate](#)[Purines & pyrimidines](#)[Ribose vs deoxyribose](#)[Phosphates & ATP](#)[The phosphodiester bond](#)[5' to 3'](#)



From Base to Nucleotide

Nucleobase

+

Sugar

→

Nucleoside

Nucleoside

+

Phosphate

→

Nucleotide

1. Nucleobase

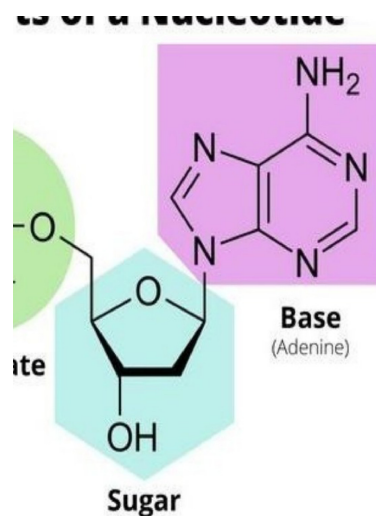
Either a **pyrimidine** or a **purine**. These are the **"letters"** of the genetic code that store information.

2. Nucleoside

The structural subunit that makes up nucleic acids. Consists of **two parts: a nucleobase and a sugar molecule**.

3. Phosphate

One phosphate gives a **nucleotide** (nucleoside monophosphate); two give **NDPs**; three give **NTPs**.



The three parts of a nucleotide



The Bases: Purines & Pyrimidines

Pyrimidines

Six-member single-ring.

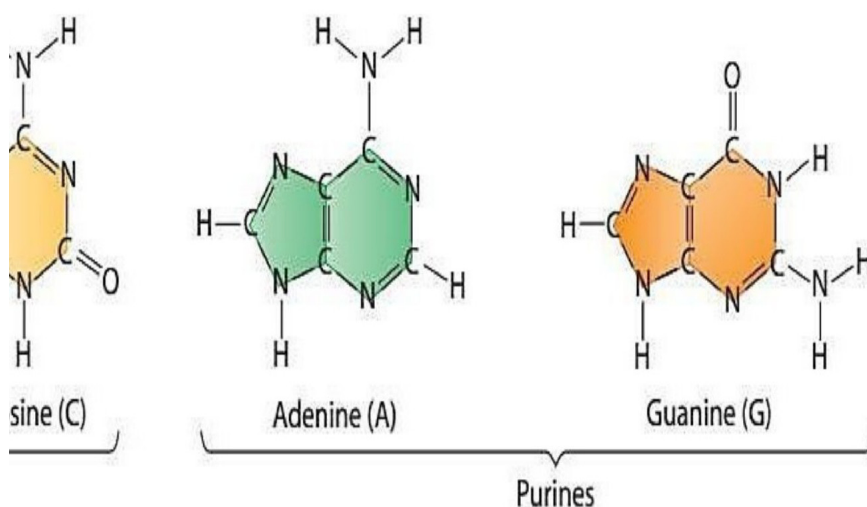
Cytosine (C), Thymine (T), or Uracil (U).

Purines

Nine-member double-ring.

Guanine (G) or Adenine (A).

In **DNA**, the purine bases are commonly **adenine and guanine**, whereas the pyrimidine bases are typically **thymine and cytosine**.



for the base-pairing rules in DNA, where
guanine pairs with cytosine (G-C), ensuring



The Sugars: Ribose vs Deoxyribose

The **pentose sugars** found in nucleic acids are what give them their names.

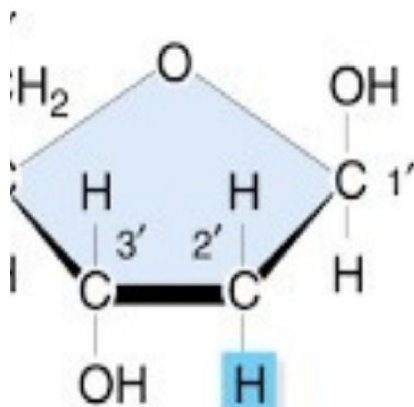
Ribose → RNA

Ribonucleic acids [RNA] contain **ribose**.

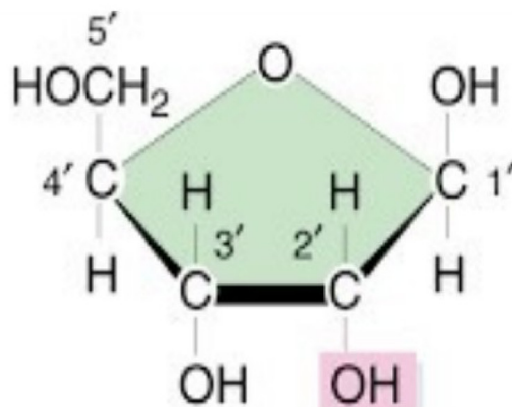
Deoxyribose → DNA

Deoxyribonucleic acids [DNA] contain **deoxyribose**.

The absence of a hydroxy group at the C2 position distinguishes DNA from RNA



Deoxyribose



Ribose



Phosphates & Energy

Monophosphate — nucleotide



Diphosphate — NDP



Triphosphate — NTP

DR. RAZA ONLY

Why triphosphate matters

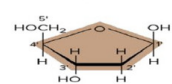
The triphosphate form is significant because it serves as the **precursor molecule** — a molecule that develops into a more mature or specialized form — during **nucleic acid synthesis** within the cell.

ATP and GTP

Adenosine triphosphate [ATP] and **guanosine triphosphate [GTP]** are important in **cell bioenergetics**, because of the large amount of energy involved in DNA replication.

(a) The separate entities

1. Deoxyribose sugar

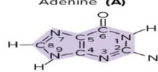
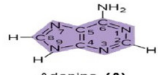


2. A phosphate group

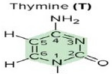
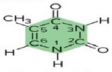


3. Four nitrogenous bases

Purines

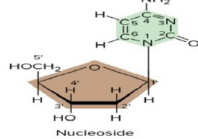


Pyrimidines

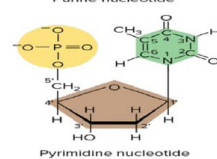
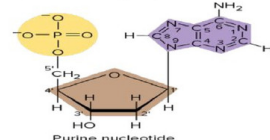


(b) Assembly into a nucleotide

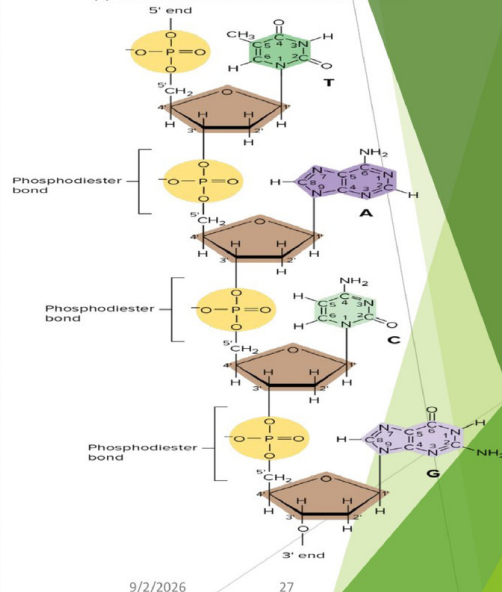
1. Attachment of base to sugar



2. Addition of phosphate



(c) Nucleotides linked in a directional chain



9/2/2026

27

Flagged IMPORTANT FOR EXAMS



The Phosphodiester Bond

1

It forms the backbone

Phosphodiester bonds link the nucleotides together to form a long chain, **creating the backbone** of DNA and RNA.

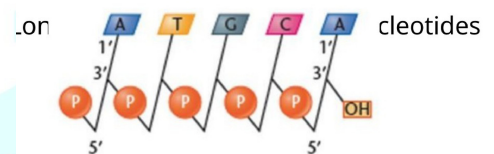
2

It provides stability

These are **strong covalent bonds**, making the backbone **resistant to cleavage** under normal cellular conditions — crucial for maintaining the integrity of genetic information during replication, transcription and translation.

Phosphodiester bond / linkage

Phosphodiester bond is the linkage between two mononucleotides consisting of a phosphate group linked to two sugars



DR. RAZA ONLY

HOW THE BOND ACTUALLY FORMS

- 1 The deoxyribose sugar has **five carbon atoms**, numbered **1' to 5'** — this numbering identifies where other molecules attach.
- 2 A nucleotide forms when a phosphate group attaches to the **5' carbon** of the deoxyribose sugar.
- 3 The bond forms between the **3' carbon of one nucleotide** and the **5' carbon of the next** — a continuous sugar-phosphate backbone with the bases sticking out like the **rungs of a ladder**.



Directionality, Replication & Repair

DR. KHAOULA ONLY

DNA and RNA run 5' → 3'

Why direction exists

The way phosphodiester bonds link nucleotides gives the strands **specific directionality**. Enzymes that work on DNA and RNA — DNA polymerase and RNA polymerase — **can only add nucleotides in the 5' to 3' direction**.

Why it matters

During replication and transcription, new phosphodiester bonds are catalyzed by those same enzymes. This is what allows the genetic code to be **accurately copied and expressed**.

The energy required to form phosphodiester bonds during these processes comes from **breaking high-energy bonds in nucleotide triphosphates** such as ATP and GTP.

WHEN THE BOND GOES WRONG

Mutations

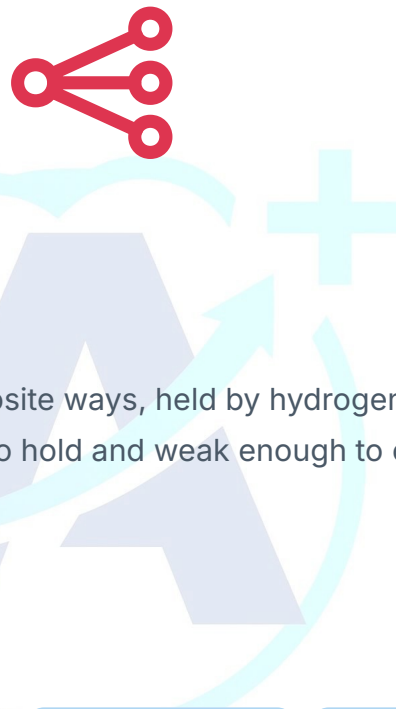
Any disruption or incorrect formation of these bonds can lead to **mutations**, which may result in genetic diseases or cancer.

Repair

DNA repair mechanisms often involve the **restoration of broken phosphodiester bonds** to maintain the stability of the genetic material.

PART 3

The Double Helix



Two strands, running opposite ways, held by hydrogen bonds that are strong enough to hold and weak enough to open.

[Helix anatomy](#)[Complementary pairing](#)[2 vs 3 hydrogen bonds](#)[In summary](#)[RNA & uracil](#)



Anatomy of the Double Helix

1

Two chains, one axis

Two DNA chains **spiral around an axis** with sugar-phosphate backbones on the outside and flat pairs of bases meeting in the middle.

2

Running opposite ways

One chain runs **5' to 3' upwards**, the other **5' to 3' downwards** — they are **antiparallel**.

3

Ten bases per turn

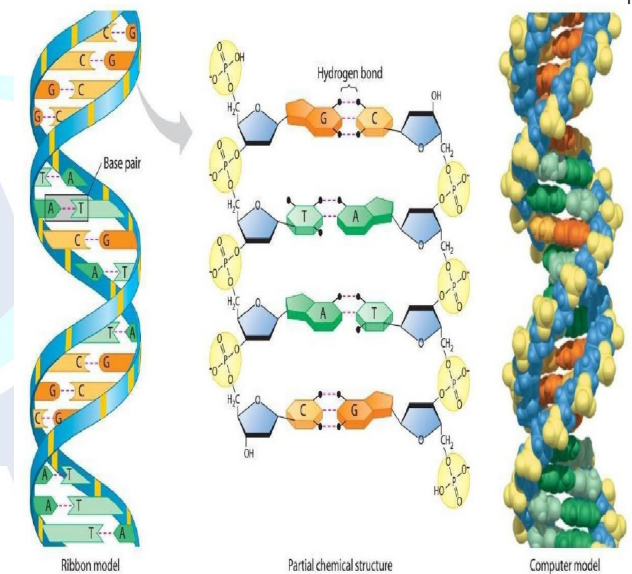
The two chains **wrap around each other every ten base pairs**.

4

Held by pairing

The chains are held together by complementary base pairing, **A with T** and **G with C** — purine with pyrimidine.

What is a double helix?



8/30/2023

29

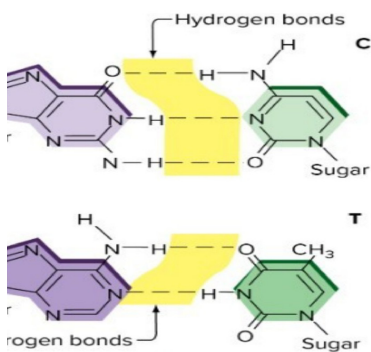


Complementary Base Pairing

A — T

G — C

- A **base pair** is a unit of double-stranded nucleic acids.
- It consists of **two nucleobases bound to each other by hydrogen bonds**.
- Base pairs form the building blocks of the double helix, and contribute to the folded structure of both DNA and RNA.
- The specific hydrogen bonding pattern lets the helix hold a **regular helical structure** that depends on its nucleotide sequence.

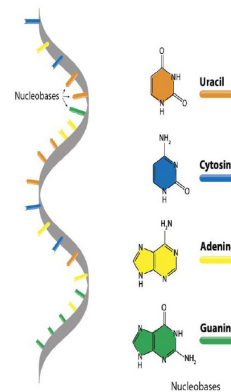


5.10 Complementary base pairing. An A on one strand can form two noncovalent hydrogen bonds with a T on the other strand. G on one strand can form three noncovalent hydrogen bonds with a C on the other strand. The size and shape of A:T and of G:C base pairs are such that they are filling the same amount of space between the two sugar-phosphate backbones of the double helix.

8/30/2023

26

Base Pairing



(Table IMPORTANT FOR EXAMS)

	DNA	RNA
Pentose sugar	Deoxyribose	Ribose
Base Composition	Adenine (A) Guanine (G) Cytosine (C) Thymine (T)	Adenine (A) Guanine (G) Cytosine (C) Uracil (U)
Number of strands	Double stranded (forms a double helix)	Single stranded

Flagged IMPORTANT FOR EXAMS



Why 2 vs 3 Hydrogen Bonds

A — T · two bonds

Adenine forms **two hydrogen bonds** with thymine. Stable enough to hold the strands together, but **separated relatively easily** during replication or transcription.

G — C · three bonds

Guanine forms **three hydrogen bonds** with cytosine — **stronger and more stable**. The extra bond gives G-C a **higher melting point**, so it resists being separated by heat.

WHY THE DIFFERENCE IS USEFUL

Stability

Having both pair types contributes to the overall stability of the molecule. **G-C rich regions** are more stable and less prone to denaturation than A-T rich regions.

Functionality

During strand separation, **A-T pairs are easier to break apart** — an advantage in regions of the genome where separation needs to happen more easily.

AND YET THE HELIX STAYS THE SAME WIDTH

Despite the difference in bond number, A-T and G-C pairs maintain a **uniform distance** between the strands. **Purine–pyrimidine pairing (A with T, G with C) ensures the width of the double helix remains constant.**



RNA & Why Uracil

DR. KHAOULA ONLY

- RNA molecules often **fold back on themselves** to form double-stranded regions of complementary base pairs.
- Just as in DNA, these folded strands are **anti-parallel** to one another.
- The result: double-stranded **"stems"** that end in single-stranded **"loops"**.
- Some animal viruses that use RNA as their genetic material contain double-stranded RNA.

SO WHY DOES RNA USE URACIL INSTEAD OF THYMINE?

Energy efficiency

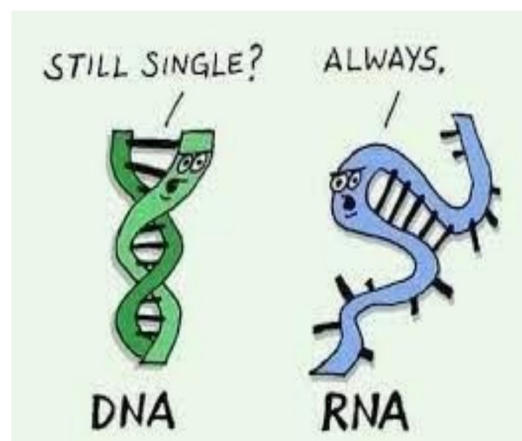
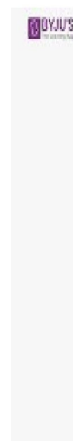
Uracil is **simpler and less expensive to produce** — chemically simpler than thymine because it **lacks the methyl group** ($-\text{CH}_3$). Since RNA is generally shorter-lived and more abundant, using uracil **saves energy**.

Error detection

Cytosine can spontaneously **deaminate to form uracil**. If uracil were normal in DNA, repair mechanisms could not tell damage from the real thing. By using thymine, cells **easily detect and repair** deaminated cytosine.

DNA forms a chain of base pairs, whereas RNA usually forms a chain of single bases.

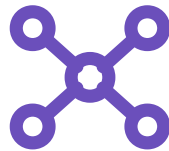
single
bases



8/30/2023 30

PART 4

Genomes



The same four letters, packaged three very different ways — loose in a bacterial cytoplasm, wound onto histones in a nucleus, or stripped to the minimum inside a virus.

[What is a genome](#)[Genomics vs genetics](#)[Prokaryote vs eukaryote](#)[Prokaryotic genome](#)[Eukaryotic genome](#)[Viral genomes](#)



What Is a Genome?

The genome is the **entire set of DNA instructions** found in a cell.

Where it lives

In humans, **23 pairs of chromosomes** in the cell's nucleus, plus a small chromosome in the mitochondria.

What it contains

Protein-coding genes and non-coding genes, regulatory sequences (non-coding DNA), and often a substantial fraction of **junk DNA** with no evident function.

A genome contains all the information needed for an individual to **develop and function**. It consists of nucleotide sequences of DNA — or RNA, in RNA viruses.

DR. RAZA ONLY

HOW BIG IS THE DNA MOLECULE IN A HUMAN?

6 feet

of DNA in every cell

3 billion

base pairs

30,000

genes

Stretched out, the DNA in one cell would be about **2 m long** — and all the DNA in all your cells put together would be about **twice the diameter of the Solar System**.



Genomics vs Genetics

DR. KHAOULA ONLY

Genomics

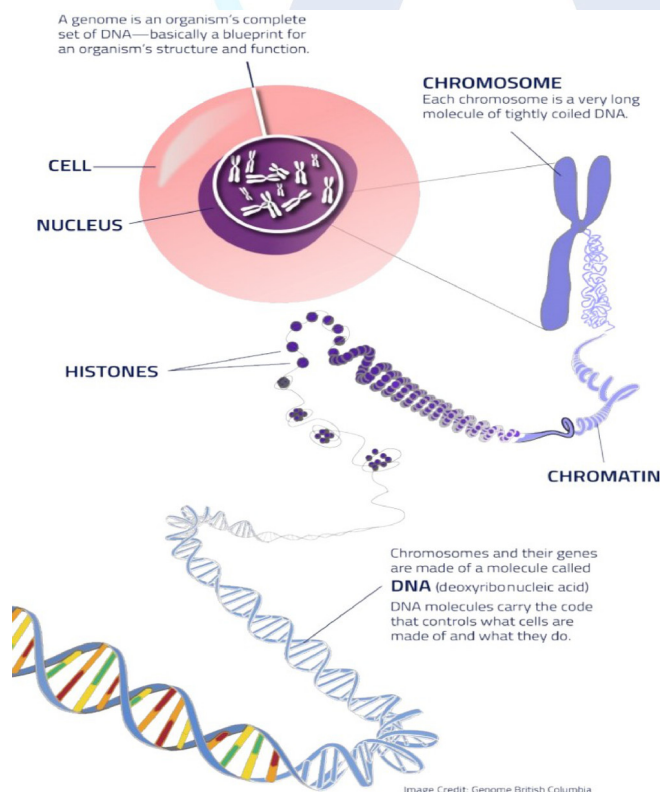
The study of an organism's **complete set of genetic information**. The genome includes both genes (coding) and non-coding DNA.

"Genome" = the complete genetic information of an organism.

Genetics

The study of **heredity** — the function and composition of **single genes**.

"Gene" = a specific sequence of DNA that codes for a functional molecule.



8/30/2023

33



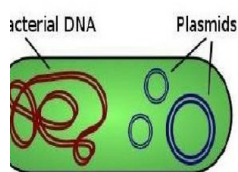
Prokaryotic & Eukaryotic Cells

Prokaryotes

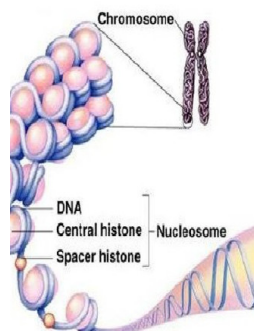
- Organisms that **do not have a nucleus**.
- Single-celled; no true cell organelles
- Genetic material **floats in the cytoplasm**
- **Haploid circular genome**
- Example: bacteria

Eukaryotes

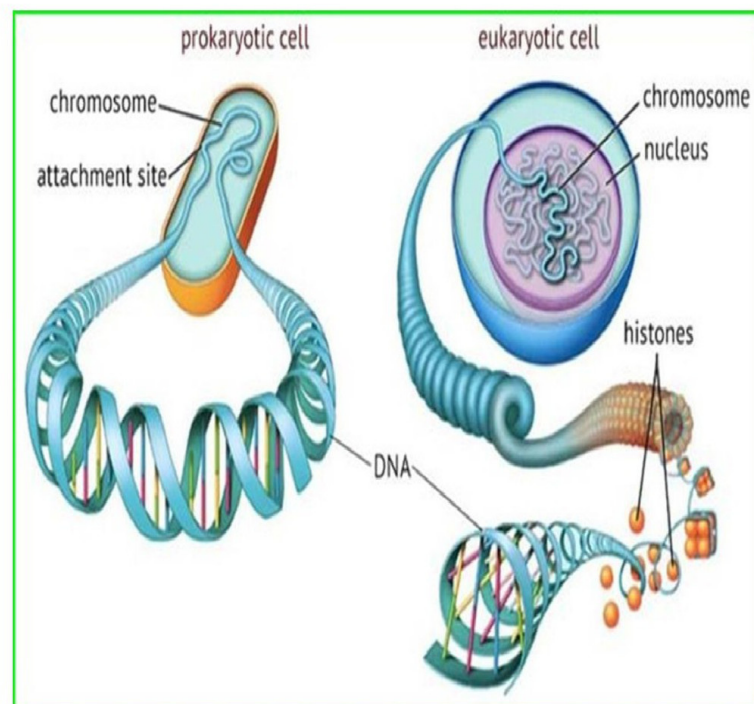
- Organisms that possess a **nucleus and true organelles**.
- Multi-cellular
- Genetic material inside the **membrane-bound nucleus**
- Genetic material is **linear, wrapped around histones**
- Example: humans, animals, fungi



Prokaryote



Eukaryote





Side by Side

DR. KHAOULA ONLY

FEATURE	PROKARYOTIC CELLS	EUKARYOTIC CELLS
Nucleus	No true nucleus (nucleoid region)	True nucleus with nuclear membrane
Size	Smaller (0.1–5 μm)	Larger (10–100 μm)
Organelles	No membrane-bound organelles	Membrane-bound organelles present
Genetic material	Single circular DNA, often with plasmids	Multiple linear chromosomes
Cell division	Binary fission	Mitosis and meiosis
Ribosomes	Smaller (70S)	Larger (80S) in cytoplasm, 70S in organelles
Cell wall	Peptidoglycan (bacteria), other polymers (archaea)	Cellulose (plants), chitin (fungi); absent in animals
Flagella	Simple, made of flagellin	Complex, made of microtubules (9+2 structure)
Metabolism	Diverse (aerobic, anaerobic, photosynthetic, chemolithotrophic)	Primarily aerobic; plants are photosynthetic
Examples	Bacteria, Archaea	Plants, Animals, Fungi, Protists



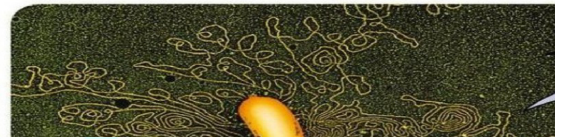
The Prokaryotic Genome

- Bacterial chromosomes are much **less complicated** than those in eukaryotes.
- They always consist of a **double-stranded (dsDNA)** molecule.
- That molecule is compacted into a **nucleoid**.

A **plasmid** is a small, circular, dsDNA molecule that is distinct from a cell's chromosomal DNA — **extrachromosomal DNA**.

Prokaryotic chromosomes are associated with **HU and H1 proteins**, structurally similar to molecules called **histones**.

(a)



E. coli with its DNA released by osmotic shock

DR. KHAOULA ONLY

Prokaryotic chromosomes are associated with **HU and H1 proteins**, structurally similar to molecules called **histones**.



Why Prokaryotic Genomes Are Simpler

DR. KHAOULA ONLY

1. Size and complexity

Smaller genome size — a typical bacterial genome might be a few million base pairs, whereas eukaryotic genomes can be billions.

Fewer genes — a few thousand, against tens of thousands in eukaryotes.

2. No nucleus

Lack of compartmentalization — genetic material is not separated from the rest of the cell. This simplicity reduces the need for the complex regulatory mechanisms associated with transport and modification.

3. No introns

Streamlined gene expression — the absence of introns simplifies transcription and translation. In eukaryotes, introns must be removed by **RNA splicing** before the mRNA can be translated.

4. Simpler regulation

Operon-based regulation allows simpler, more efficient control of gene expression, against the multiple layers eukaryotes use.

5. LESS NON-CODING DNA

Prokaryotic genomes contain **very little non-coding DNA**. Eukaryotes often have large amounts — repetitive elements, introns and regulatory regions — which contribute to their genome complexity.



The Eukaryotic Genome

1

Wound onto histones

In eukaryotes, DNA is **tightly wound around clusters of histone proteins** to form chromosomes.

2

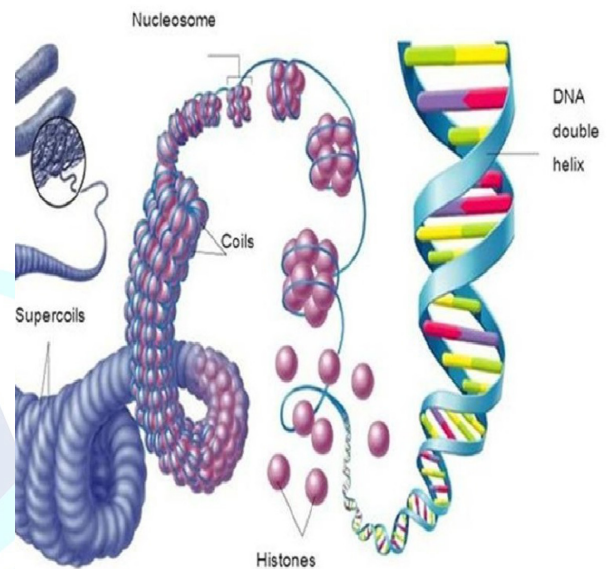
More material

In general, eukaryotic cells contain a **lot more genetic material** than prokaryotic cells.

3

Held in the nucleus

The nucleus is particularly important in eukaryotes, because it is the **location of a cell's DNA**.

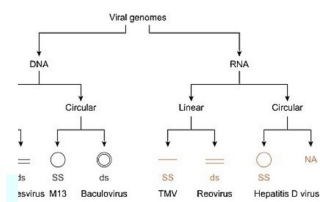
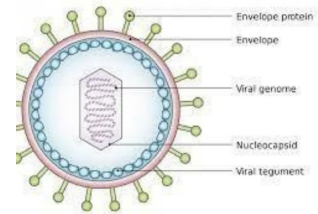




Viral Genomes — Diversity

DR. KHAOULA ONLY

- Viral chromosomes consist of nucleic acid — **either DNA or RNA**.
- That nucleic acid can be **either single or double stranded**.
- It may exist as a **circular structure**, or take the form of a **linear molecule**.



DNA viruses

Double-stranded (dsDNA) or single-stranded (ssDNA).

Herpesviruses (dsDNA), parvoviruses (ssDNA).

RNA viruses

dsRNA or ssRNA — positive-sense (+ssRNA) or negative-sense (–ssRNA).

Coronaviruses (+ssRNA), influenza (–ssRNA).

Retroviruses

ssRNA genome with **reverse transcription**.

HIV.

Genome size

Typically **small** — a few thousand to several hundred thousand base pairs or nucleotides. They often encode only **a few proteins**, relying heavily on the host cell's machinery.

Genome structure

Linear vs circular — adenoviruses vs papillomaviruses. **Segmented vs non-segmented** — influenza is segmented. Genes are **closely packed**, sometimes overlapping or using different reading frames.



How Viral Genomes Work

DR. KHAOULA ONLY

1

Attachment and entry

The virus attaches to the host cell and enters by various mechanisms.

2

Replication

DNA viruses replicate in the **nucleus**; RNA viruses in the **cytoplasm**.

3

Transcription and translation

The viral genome is transcribed into **mRNA** and translated into proteins.

4

Assembly and release

New viral particles are assembled and released to **infect new cells**.

DR. KHAOULA ONLY

WHAT MAKES VIRAL GENOMES SPECIAL

High mutation rates — lack of proofreading

Genetic flexibility — recombination and reassortment

Dependence on host machinery

Minimalism — only essential proteins

Unique replication — reverse transcription



Over to You

DR. KHAOULA ONLY

RNA viruses often have **higher mutation rates** than DNA viruses, due to the lack of proofreading mechanisms during replication.

How might this **high mutation rate influence the development of antiviral drugs**, and **what approaches could be taken to design treatments** that remain effective against rapidly mutating RNA viruses?

— Find and get back to me with your answers.

YOUR NOTES





To Summarise...

BUILD 3**Parts of a nucleotide**

- Nitrogenous base — purine or pyrimidine
- 5-carbon sugar — ribose or deoxyribose
- One, two or three phosphate groups

BASES ATGC**Pairing rules**

- Purines: adenine, guanine — double ring
- Pyrimidines: cytosine, thymine, uracil
- A pairs with T; G pairs with C

BONDS 2 & 3**Hydrogen bonds**

- A-T has two — easier to separate
- G-C has three — higher melting point
- Purine-pyrimidine keeps the width constant

FORMS 3**Forms of DNA**

- A-DNA right-handed, dehydrated conditions
- B-DNA right-handed, most common and stable
- Z-DNA left-handed, zigzag backbone

GENOME 23**Human chromosome pairs**

- Plus a small mitochondrial chromosome
- 6 feet of DNA, ~3 billion base pairs per cell
- Coding genes, regulatory and junk DNA

CELLS 3**Ways to package a genome**

- Prokaryote: circular, in the cytoplasm
- Eukaryote: linear, on histones, in a nucleus
- Viral: DNA or RNA, single or double stranded

A REMINDER ON THE MARKING

Sections carrying a **blue rule** come only from Dr. Khaoula Errafii's lecture; a **green rule** only from Dr. Afsheen Raza's lecture. Everything else is taught in both.