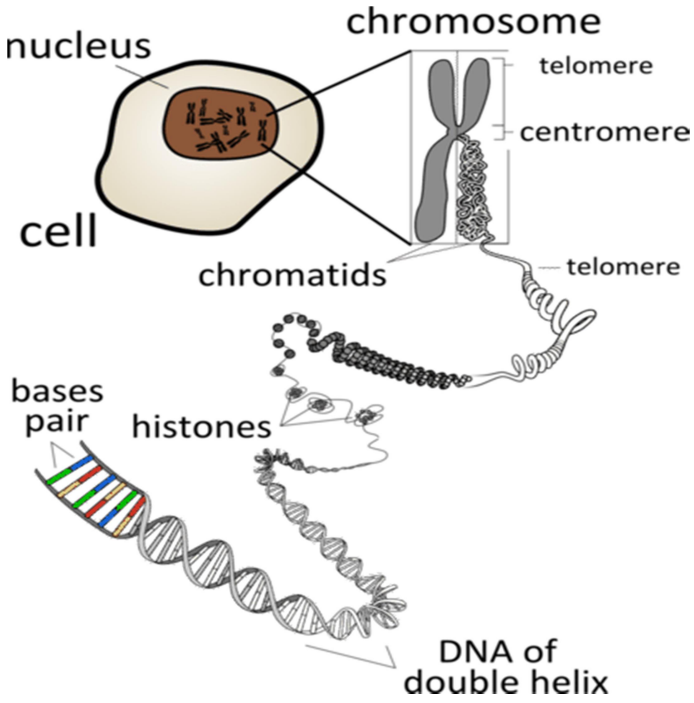
CHROMOSOME

In the Nucleus of each cell, the **DNA** molecule is packaged into thread-like structures called chromosomes.

- Each chromosome is made up of DNA tightly coiled many times around proteins that support its structure.
- The proteins that bind to the DNA to form eukaryotic chromosomes are traditionally divided into two classes: the histones and the non-histone chromosomal proteins.
- The complex of both classes of protein with the nuclear DNA of eukaryotic cells is known as chromatin.
- Chromatin are a highly compacted structure consisting of packaged DNA and necessary so as to fit DNA into the nucleus.
- The nucleosome is the smallest structural component of chromatin and is produced through interactions between DNA and histone proteins.

RELATION BETWEEN CHROMOSOME, DNA & GENE



STRUCTURE OF CHROMOSOME

Structurally, each chromosome is differentiated into three parts—

1. Pellicle
2. Matrix
3. Chromonemata

Pellicle

- It is the outer envelope around the substance of chromosome.
- It is very thin and is formed of achromatic substances.

Matrix

- It is the ground substance of chromosome which contains the chromonemata.
- It is also formed of non-genic materials.

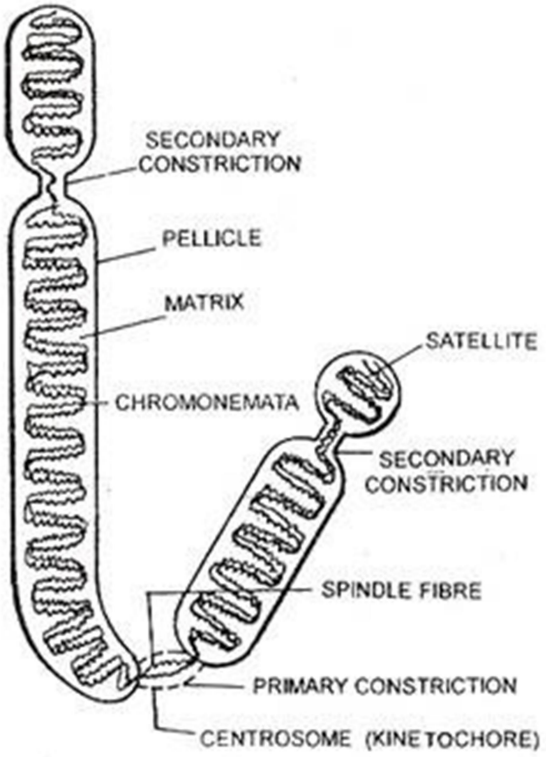
Chromonemata

- Embedded in the matrix of each chromosome are two identical, spirally coiled threads, the chromonemata.
- The two chromonemata are also tightly coiled together that they appear as single thread of about 800A thickness.
- Each chromonemata consists of about 8 microfibrils, each of which is formed of a double helix of DNA.

Main parts of chromosomes

- **Chromatid:** Each chromosome has two symmetrical structures called chromatids or sister chromatids which is visible in mitotic metaphase.
    - Each chromatid contains a single DNA molecule
    - At the anaphase of mitotic cell division, sister chromatids separate and migrate to opposite poles
- **Centromere and kinetochore:** Sister chromatids are joined by the centromere.
    - Spindle fibres during cell division are attached at the centromere
    - The number and position of the centromere differs in different chromosomes
    - The centromere is called **primary constriction**
    - Centromere divides the chromosome into two parts, the shorter arm is known as **'p' arm** and the longer arm is known as **'q' arm.**
    - The centromere contains a disc-shaped **kinetochore**, which has specific DNA sequence with special proteins bound to them
    - The kinetochore provides the centre for polymerisation of tubulin proteins and assembly of microtubules
- **Secondary constriction and nucleolar organisers:** Other than centromere, chromosomes possess secondary constrictions.
    - Secondary constrictions can be identified from centromere at anaphase because there is bending only at the centromere (primary constriction)

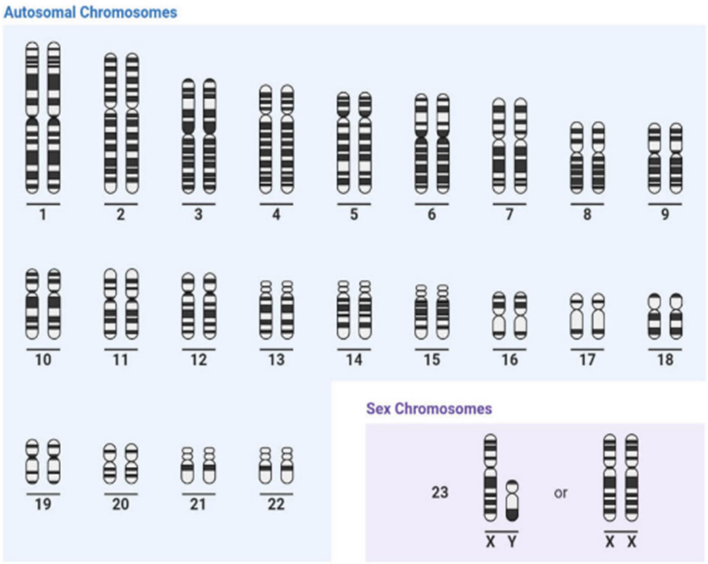
    - Secondary constrictions, which contain genes to form nucleoli are known as the **nucleolar organiser**
- **Telomere:** Terminal part of a chromosome is known as a telomere.
    - Telomeres are polar, which prevents the fusion of chromosomal segments
- **Satellite:** It is an elongated segment that is sometimes present on a chromosome at the secondary constriction.
    - The chromosomes with satellite are known as **sat-chromosome**
- **Chromatin:** Chromosome is made up of **chromatin.** Chromatin is made up of DNA, RNA and proteins. At interphase, chromosomes are visible as thin chromatin fibres present in the nucleoplasm. During cell division, the chromatin fibres condense and chromosomes are visible with distinct features.



TYPES OF CHROMOSOME

A. Autosomes and Sex Chromosomes

- Human chromosomes are of two types- autosomes and sex chromosomes.
- Genetic traits that are linked to the sex of the person are passed on through the sex chromosomes. The rest of the genetic information is present in the autosomes.
- Humans have 23 pairs of chromosomes in their cells, of which 22 pairs are autosomes and one pair of sex chromosomes, making a total of 46 chromosomes in each cell.



B. On the Basis of Number of Centromeres

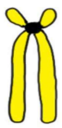
1. **Monocentric** with one centromere.
2. **Dicentric** with two centromeres.
3. **Polycentric** with more than two centromeres
4. **Acentric** without centromere. Such chromosomes represent freshly broken segments of chromosomes which do not survive for long.
5. **Diffused or non-located** with indistinct centromere diffused throughout the length of chromosome.

C. On the Basis of Location of Centromere

1. **Telocentric** are rod-shaped chromosomes with centromere occupying the terminal position, so that the chromosome has just one arm.


Telocentric

2. **Acrocentric** are also rod-shaped chromosomes with centromere occupying a sub-terminal position. One arm is very long and the other is very short.


Acrocentric

3. **Sub-metacentric** chromosomes are with centromere slightly away from the mid-point so that the two arms are unequal.


Submetacentric

4. **Metacentric** are V-shaped chromosomes in which centromere lies in the middle of chromosome so that the two arms are almost equal.

Metacentric

FUNCTIONS OF CHROMOSOME

1. Genetic Code Storage
2. Sex Determination
3. Control of Cell Division
4. Formation of Proteins and Storage

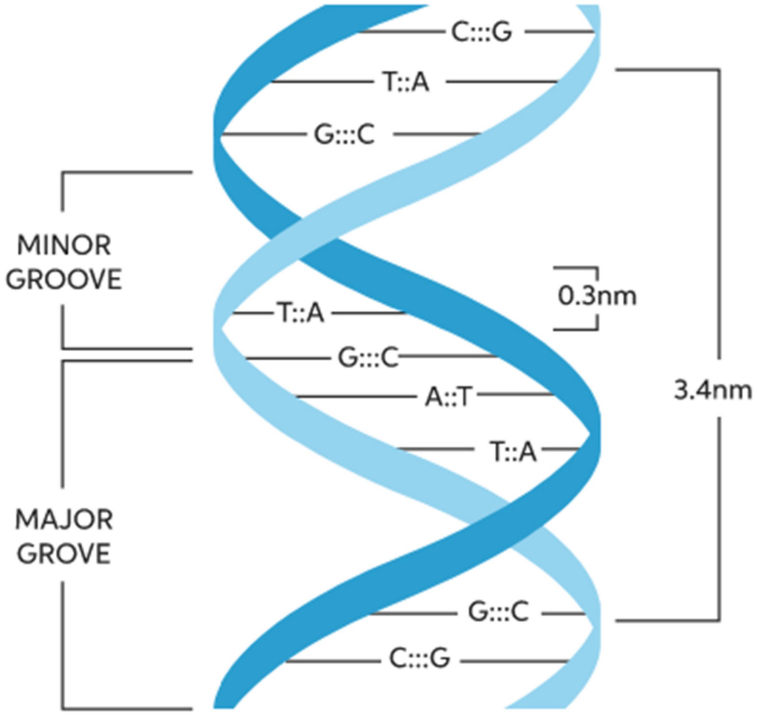
DNA (DE-OXY RIBONUCLEIC ACID)

Deoxyribonucleic acid, also abbreviated as DNA, is the principal informational macromolecule of the cell, which stores, translates and transfers the genetic information.

STRUCTURE

- The DNA molecule consists of two polynucleotide strands that are coiled around a common axis to form a right-handed double helix.
- The two strands run in opposite directions – one strand runs from 3' to 5' direction, while the other strand runs from 5' to 3' direction. They have opposite polarity and the polarity is due to phosphodiester linkage.
- The sugar–phosphate backbone remains on the outside, while the nitrogenous bases remain inside.
- The two strands of DNA are held together by hydrogen bonding between the nitrogenous bases on the opposite strands. The hydrogen bonds between the two strands are at a distance of 2.0nm.
- Two hydrogen bonds pairs Adenine and Thymine (A = T), whereas three hydrogen bonds pairs Guanine and Cytosine (G ≡ C).
- 11. The base pairs A = T and G ≡ C are known as complementary base pairs.

- The length of a complete turn of helix is 3.4 nm.There are 10 base pairs per turn of the helix.
- Each helix consists of a major groove, which is the site of bonding of specific protein and a minor groove.



TYPES OF DNA

	A	B	Z	H
1.	Shorter and wider	Normal reference strand–Watson and Crick model	Longer and thinner	It is a long stretch (part) of DNA with alternating T and C or polypurine/ polypyrimidine
2.	Right-handed double helix	Right-handed double helix	Left-handed double helix	Triple helix
3.	Distance between each turn is 2.3 nm	Distance between each turn is 3.6 nm	Distance between each turn is 3.8 nm	—
4.	11 base pairs per turn	10.5 base pairs per turn	12 base pairs per turn	—
5.	Stable in solutions devoid of water	Most stable under physiological conditions	Doubtful existence in physiological state	Helps in gene regulation

FUNCTION OF DNA

(a) DNA stores the complete genetic information required to specify (form) the structure of all the proteins and RNA's of each organism.

(b) DNA is the source of information for the synthesis of all cellular body proteins.

(c) It determines the activities of an organism throughout its life cycle, i.e., the period of gestation, birth, maturity, senescence and death.

(d) It determines the individuality and identity of a given organism.

(e) It duplicates (replicates to form two daughter DNA) itself and transfers one of the copy to the daughter cell during cell division, thus maintaining the genetic material from generation to generation.

GENE

a. **Gene**, unit of hereditary information that occupies a fixed position (locus) on a chromosome. Genes achieve their effects by directing the synthesis of proteins.

b. Genes are composed of deoxyribonucleic acid, except in some virus, which have genes consisting of a closely related compound called ribonucleic acid).

PROTEIN SYNTHESIS

Protein synthesis is the biological process by which cells produce proteins. Proteins are essential for growth, repair, enzyme activity, hormone production, immunity, and maintaining the structure and function of cells.

Protein synthesis occurs in **two main stages**:

1. **Transcription** (DNA → mRNA)
2. **Translation** (mRNA → Protein)

Flow of genetic information:

DNA → mRNA → Protein

Stages of Protein Synthesis

Stage 1: Transcription (DNA to mRNA)

Transcription is the process of copying genetic information from DNA into messenger RNA (mRNA). It occurs in the **nucleus** of eukaryotic cells.

Steps of Transcription

1. Initiation

- An enzyme called **RNA polymerase** binds to a specific DNA sequence called the **promoter**.
- The DNA double helix unwinds, exposing one DNA strand (the template strand).

2. Elongation

- RNA polymerase reads the DNA template strand in the **3′ → 5′** direction.
- It builds the mRNA strand in the **5′ → 3′** direction.
- RNA nucleotides pair with DNA bases:
    - DNA A → RNA U
    - DNA T → RNA A
    - DNA C → RNA G
    - DNA G → RNA C

3. Termination

- RNA polymerase reaches a **termination sequence**.
- The completed mRNA molecule is released.
- DNA rewinds into its double-helix structure.

mRNA Processing (Eukaryotes Only)

Before leaving the nucleus, the mRNA is modified:

- A **5′ cap** is added for protection and ribosome recognition.
- A **poly(A) tail** is added to increase stability.
- **Introns** (non-coding regions) are removed.
- **Exons** (coding regions) are joined together.

The mature mRNA then exits the nucleus through nuclear pores.

Stage 2: Translation (mRNA to Protein)

Translation is the process of converting the genetic code carried by mRNA into a chain of amino acids (a polypeptide). It occurs on **ribosomes** in the cytoplasm or on the rough endoplasmic reticulum.

Components Required

- **mRNA** – carries the genetic code.
- **Ribosome** – the site of protein synthesis.
- **tRNA (transfer RNA)** – brings amino acids to the ribosome.
- **Amino acids** – the building blocks of proteins.
- **Enzymes and energy (ATP/GTP)** – drive the process.

Structure of tRNA

Each tRNA has

- An **anticodon**, which pairs with an mRNA codon.
- An attachment site for a specific amino acid.

Steps of Translation

1. Initiation

- The ribosome binds to the mRNA.
- Translation usually begins at the **start codon (AUG)**.
- AUG codes for **methionine**, the first amino acid in most newly made proteins.

2. Elongation

- The ribosome reads mRNA **one codon at a time**.
- Each codon specifies one amino acid.
- Matching tRNAs deliver the correct amino acids.
- The ribosome forms **peptide bonds** between adjacent amino acids.
- It moves to the next codon and repeats the cycle.

3. Termination

- When a **stop codon** (UAA, UAG, or UGA) is reached, no tRNA binds.
- Release factors help free the completed polypeptide.
- The ribosome separates from the mRNA.

After Translation

The newly formed protein often undergoes **post-translational modifications**, such as:

- Folding into its functional three-dimensional shape.
- Addition of carbohydrate, phosphate, or lipid groups.
- Formation of disulfide bonds.
- Transport to its final destination within or outside the cell.

These modifications mainly occur in the **rough endoplasmic reticulum** and **Golgi apparatus**.

GENETIC PATTERNS OF INHERITANCE

Genetic patterns of inheritance describe the way genetic traits and disorders are passed from parents to their offspring through genes. Genes are the basic units of heredity and are located on chromosomes within the nucleus of every cell. Humans possess **23 pairs of chromosomes**, of which **22 pairs are autosomes** and **one pair consists of sex chromosomes (XX in females and XY in males)**. Every individual inherits one copy of each gene from the mother and one from the father. Variations or mutations in these genes determine whether a trait or genetic disorder is expressed. Understanding patterns of inheritance helps in predicting the likelihood of inherited diseases, providing genetic counseling, and making informed healthcare decisions.

1. Autosomal Dominant Inheritance

In an **autosomal dominant** pattern, a mutation in **only one copy** of a gene located on an autosome is sufficient to produce the trait or disease. An affected individual usually has one affected parent because the disorder tends to appear in every generation.

Characteristics

- Both males and females are equally affected.
- The trait is transmitted from one generation to the next (vertical transmission).
- Each child of an affected parent has a **50% chance** of inheriting the disorder.
- Unaffected individuals do not transmit the disease to their offspring.

Examples

- Huntington's disease
- Marfan syndrome
- Achondroplasia
- Familial hypercholesterolemia

2. Autosomal Recessive Inheritance

In **autosomal recessive** inheritance, a person must inherit **two mutated copies** of the gene (one from each parent) to develop the disease. Individuals with only one mutated gene are called **carriers** and usually do not show symptoms.

- Males and females are equally affected.
- The disorder often appears among siblings rather than in every generation.
- Parents of affected children are usually healthy carriers.
- If both parents are carriers:
    - 25% chance of an affected child
    - 50% chance of a carrier child
    - 25% chance of an unaffected child

Examples

- Cystic fibrosis
- Sickle cell anemia
- Phenylketonuria (PKU)
- Tay-Sachs disease

3. X-Linked Dominant Inheritance

In this pattern, the defective gene is located on the **X chromosome**, and **only one mutated copy** is needed for the disorder to occur.

Characteristics

- Females are more commonly affected than males because they have two X chromosomes.
- An affected father transmits the disorder to **all daughters** but **none of his sons**.
- An affected mother has a **50% chance** of passing the condition to each child, regardless of sex.
- The disorder usually appears in successive generations.

Examples

- Fragile X syndrome (some forms)
- Vitamin D-resistant rickets
- Rett syndrome

4. X-Linked Recessive Inheritance

In **X-linked recessive** inheritance, the defective gene is located on the X chromosome. Since males have only one X chromosome, they are more likely to express the disorder if they inherit the mutated gene. Females usually become carriers because they have a second normal X chromosome.

Characteristics

- Mostly affects males.

- Females are usually carriers and rarely affected.
- There is **no father-to-son transmission**.
- Carrier mothers have:
    - 50% chance of affected sons
    - 50% chance of carrier daughters

Examples

- Hemophilia A and B
- Duchenne muscular dystrophy
- Red-green color blindness
- G6PD deficiency

5. Y-Linked (Holandric) Inheritance

Y-linked inheritance involves genes located on the **Y chromosome**, which is present only in males.

Characteristics

- Only males are affected.
- An affected father passes the trait to **all of his sons**.
- Females are never affected or carriers.
- The trait is transmitted directly from father to son.

Examples

- Hypertrichosis of the ear
- Certain forms of male infertility

6. Mitochondrial (Maternal) Inheritance

Mitochondrial inheritance is caused by mutations in the DNA of **mitochondria**, the energy-producing organelles of cells. Since mitochondria are inherited only from the mother's egg, **only mothers transmit mitochondrial disorders**.

Characteristics

- Both males and females may be affected.
- Only affected mothers pass the mutation to their children.
- Affected fathers do not transmit the disorder.
- The severity of disease may vary because different cells contain different proportions of mutated mitochondria (heteroplasmy).

Examples

- Leber hereditary optic neuropathy (LHON)
- MELAS syndrome
- MERRF syndrome

7. Codominant Inheritance

In **codominant inheritance**, both alleles are fully expressed in the heterozygous individual, and neither allele masks the other.

Characteristics

- Both traits are expressed simultaneously.
- Neither allele is dominant or recessive.
- The offspring displays characteristics of both alleles.

Example

- **AB blood group**, where both A and B antigens are expressed equally.

8. Incomplete Dominance

In **incomplete dominance**, neither allele is completely dominant over the other, resulting in an intermediate phenotype.

Characteristics

- The heterozygous individual shows a blended characteristic.
- The phenotype is intermediate between the two homozygous forms.

Example

- Red-flowered plant × White-flowered plant → Pink-flowered offspring.

9. Multifactorial (Polygenic) Inheritance

Some traits and diseases are influenced by **multiple genes along with environmental factors** rather than a single gene mutation.

Characteristics

- Caused by the combined effects of many genes.
- Environmental factors such as diet, lifestyle, and exposure to toxins also influence expression.
- The inheritance pattern is complex and difficult to predict.

Examples

- Diabetes mellitus

- Hypertension
- Obesity
- Coronary artery disease
- Cleft lip and palate