

- The 2020 Nobel Prize in Chemistry

Recipients: Shared by Emmanuelle Charpentier and Jennifer A. Doudna.

Field of Study: Methodology of Gene Editing.

Core Discovery: A technology known as CRISPR-Cas 9.

- What is CRISPR-Cas 9?

It is a specific process used for gene editing.

It functions by bringing about desirable changes within the genes of DNA.

The development of this technology was made possible by a deeper scientific understanding of DNA

- Potential Impact and Applications

The discovery is considered revolutionary due to its potential in several sectors:

Medicine: Advancing therapies for genetic diseases and providing new avenues for the treatment of cancer.

Agriculture: Developing crops that are naturally resistant to pests and various diseases.

## **Location and Structure of DNA**

### 1. The Location of DNA

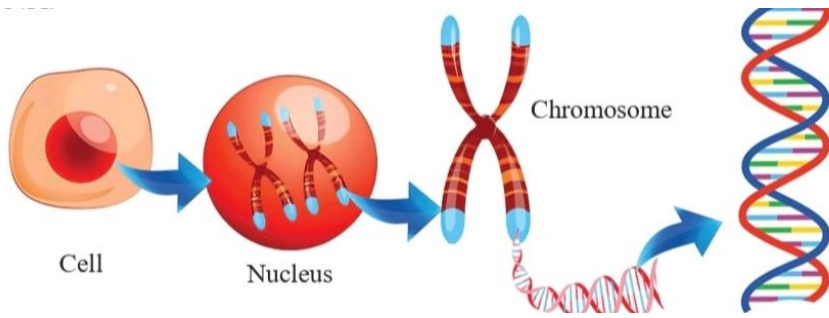
Understanding the location of DNA involves a hierarchical look at the cell structure:

Cell: The basic unit of life.

Nucleus: The specialized organelle within the cell that houses genetic material.

Chromosome: Within the nucleus, DNA is organized into thread-like structures called chromosomes.

DNA (Deoxyribonucleic Acid): The molecule that carries genetic instructions, found tightly coiled within the chromosomes.



## The Discovery of DNA Structure

The identification of the **double helical model** of DNA in **1953** is considered one of the greatest leaps in biological studies.

- **Key Scientists:**
  - **James Watson & Francis Crick:** Proposed the double helix model.
  - **Rosalind Franklin & Maurice Wilkins:** Conducted the X-ray diffraction studies that provided the foundational data for the model.
- **The Crucial Evidence:**
  - **Photo 51:** A famous X-ray diffraction image of DNA taken by **Rosalind Franklin**. This image provided the essential structural information needed to determine the double helix shape.
- **The Nobel Prize:**
  - In **1962**, James Watson, Francis Crick, and Maurice Wilkins were awarded the **Nobel Prize in Medicine** for their contributions to the discovery of the double helix model.
  - **Historical Note:** Rosalind Franklin passed away in 1958 at the age of 37, before the Nobel Prize was awarded.

## Structure of DNA

### The Double Helix Model

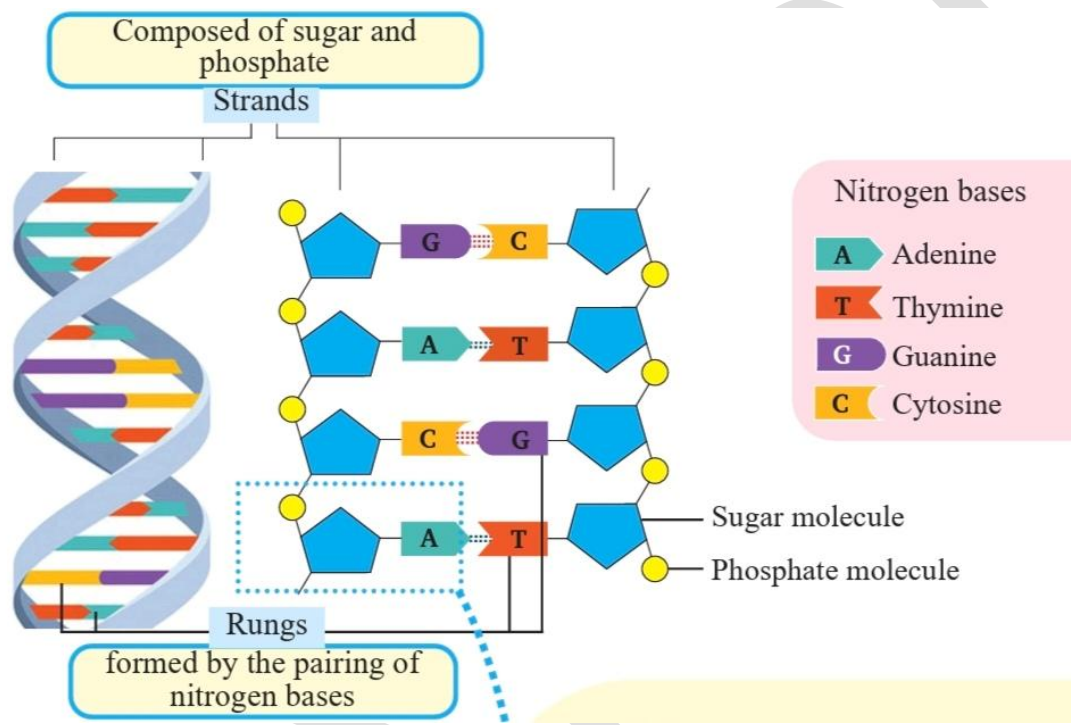
DNA is structured as a **double helix**, which resembles a twisted ladder.

- **Strands (The Backbone):** The two outer sides of the ladder are composed of alternating **sugar** and **phosphate** molecules.
- **Rungs (The Steps):** The horizontal steps of the ladder are formed by the **pairing of nitrogen bases**.

# The Nucleotide: The Basic Building Block

A nucleotide is the fundamental unit that makes up DNA. Each nucleotide is composed of three specific components:

1. **Deoxyribose Sugar:** A 5-carbon sugar that forms the central part of the nucleotide.
2. **Phosphate Group:** Participates in the formation of chemical bonds that link individual nucleotides together to form a long chain.
3. **Nitrogen Base:** A nitrogen-containing alkaline compound attached to the sugar



## Size of DNA

**Individual Chromosome:** The DNA in each chromosome is about 2 inches (5cm) long.

**Single Cell Length:** If DNA from 46 chromosomes of a human cell joins together, it would be around 6 feet in length (2m).

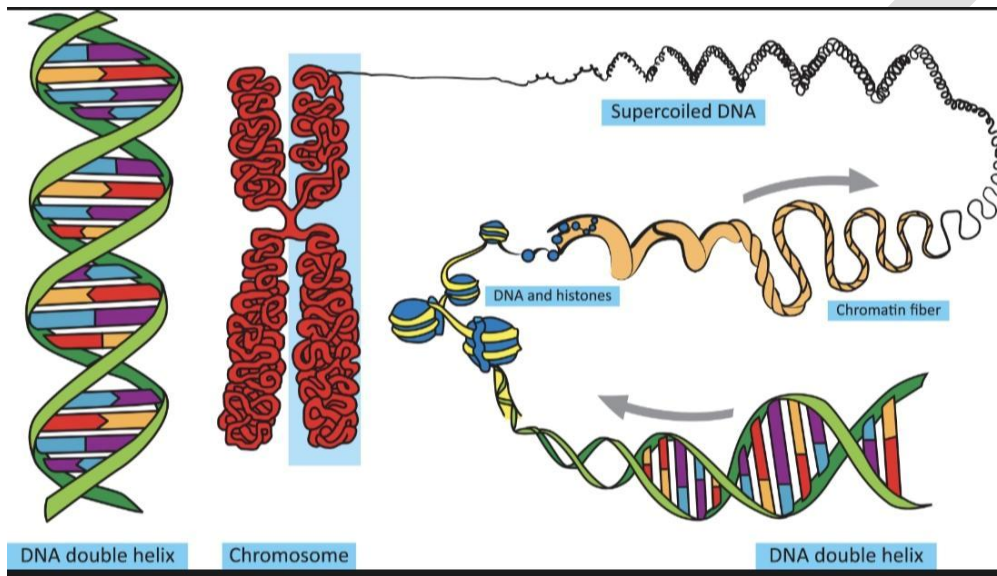
**Body-wide Scale:** The human body is made up of trillions (one lakh crore) of cells.

**Total Human DNA:** If the DNAs of all the cells join together, it would be about 67 billion miles in length.

**Comparison:** This total length is capable enough to wrap around the Earth over two million times.

Q:1. How does such a large DNA fit into the chromosomes of tiny cells?

Ans: DNA uses a process called supercoiling. It wraps tightly around special proteins called histones (like thread around a spool). These spools pack together closely to form chromatin, which eventually condenses into the compact chromosomes visible during cell division. This allows 2 meters of DNA to fit into a space smaller than a speck of dust.



Q:2. How does normal sugar differ from a sugar molecule in DNA?

Normal Sugar (Sucrose): This is a complex sugar (disaccharide) used for energy.

DNA Sugar (Deoxyribose): This is a simple 5-carbon sugar (pentose). It is called "deoxy-ribose" because it is missing one oxygen atom at the second carbon position compared to ribose sugar. Its primary job is structural (forming the "backbone" of DNA) rather than providing energy.

### **Components and Formation**

- Primary Components: DNA and Histone proteins.
- Histone Octamer: Eight histone proteins join together to form this structure.
- Nucleosome: The structure formed when DNA strands wind around a histone octamer.
- Chromosome Formation: Formed by packing, coiling, and recoiling numerous nucleosomes.

### **Parts of a Chromosome**

- Chromatids: The parts of a chromosome connected by a centromere.

- Centromere: The connection point between chromatids.

Q: 1. How does such a larger DNA fit into the chromosomes of tiny cells?

Ans: Through the formation of nucleosomes and extreme coiling/recoiling (supercoiling) around histone proteins.

Q: 2. How does normal sugar differ from a sugar molecule in DNA?

Ans: Normal sugar (sucrose) is for energy; DNA sugar is Deoxyribose, a 5-carbon pentose sugar lacking one oxygen atom, used for structure.

Q: 3. What is the relationship between chromatin reticulum and chromosomes?

Ans: Chromatin reticulum is the uncoiled thread-like state. During cell division, it condenses and coils to form distinct chromosomes.

## **HUMAN CHROMOSOME**

- Each species possesses a specific number of chromosomes.
- Humans have 46 chromosomes (23 pairs) in each somatic cell.

### **Somatic Chromosomes**

- Definition: Chromosomes that control physical characteristics.
- Quantity: There are twenty-two pairs (44 chromosomes) of somatic chromosomes.
- Homologous Chromosomes: A pair of identical chromosomes together form a homologous chromosome.
- Inheritance: In each pair, one chromosome is inherited from the mother and the other from the father.

### **Sex Chromosomes**

- Definition: Chromosomes involved in sex determination.
- Types: There are two types—X chromosome and Y chromosome.
- X Chromosome: Comparatively larger.
- Y Chromosome: Comparatively smaller than the X chromosome.
- SRY Gene: Located on the Y chromosome; responsible for the development of testis in the embryo.

Q1: How many chromosomes do humans have?

Ans: Humans have 46 chromosomes, which exist as 23 pairs.

Q2: Why are chromosomes seen in pairs?

Ans: Chromosomes are seen in pairs because sexually reproducing organisms receive one set of chromosomes from each parent. Each chromosome from the paternal set has a corresponding (homologous) chromosome in the maternal set that carries genes for the same traits.

Q3: What determines the male sex in humans according to the text?

Ans: The presence of the Y chromosome, specifically the SRY gene located on it, is responsible for the development of testes, which determines the male sex.

### **Genetic Constitution**

The chromosomal makeup of a human is represented as the sum of somatic chromosomes and sex chromosomes.

Female Genetic Constitution: 44 + XX

Group	Genetic Constitution	Total Number of Chromosomes	Number of Somatic Chromosomes	Number and Type of Sex Chromosomes
Female	44 + XX	46	44	2 (XX)
Male	44 + XY	46	44	2 (XY)

Male Genetic Constitution: 44 + XY

Q: 1 Are there multiple organisms with the same number of chromosomes?

Ans: Yes, different species can have the same number of chromosomes. For example, both Potatoes and Chimpanzees have 48 chromosomes. However, the genetic information and arrangement on these chromosomes differ significantly between species.

Q:2 What are the Chromosomal Similarities and Differences in human?

- Similarity: Both males and females have the same number of somatic chromosomes (44).

- Difference: Females possess two identical X chromosomes (XX), while males possess one X and one Y chromosome (XY).

### **Different Genetic Constitutions**

While 44+XX (Female) and 44+XY (Male) are normal, variant genetic constitutions influence physical and mental development.

- 44+X0: Females with only one X chromosome. Condition: Turner syndrome.
- 44+XXX: Females with three X chromosomes. Condition: Triple-X syndrome.
- 44+XXY: Males with two X chromosomes and one Y chromosome. Condition: Klinefelter syndrome.
- 44+XYY: Males with one X chromosome and two Y chromosomes. Condition: XYY syndrome.

### Complexity in Gender Development

- Turner Syndrome: Individuals have female sex organs but may not develop female sexual characteristics at the beginning of adolescence.
- Klinefelter Syndrome: Individuals have male sex organs but may also exhibit female characteristics.

Q: The Y chromosome of the father is important in sex determination. Why?

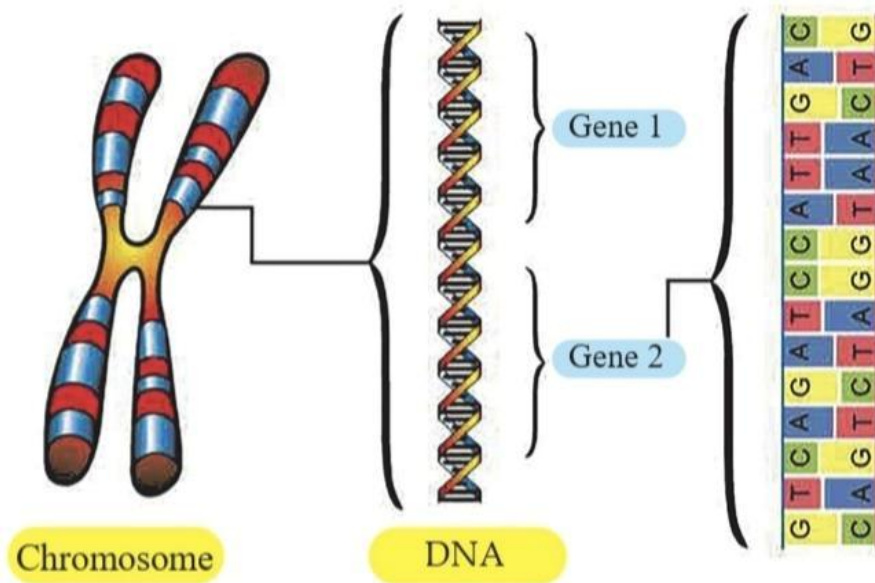
Ans: The Y chromosome carries the SRY (Sex-determining Region Y) gene. If a sperm carrying a Y chromosome fertilizes the egg, the SRY gene triggers the development of male sex organs. Since only fathers possess a Y chromosome (XY), they are the determining factor for the sex of the offspring.

### **Gene**

- Definition: A gene is a specific sequence of nucleotides in DNA.
- Function: Genes provide instructions for the synthesis of proteins.
- Role of Proteins: Proteins synthesized based on genetic instructions are responsible for the formation of characteristic features and controlling metabolic activities.

Q: Where are genes found?

Ans: Genes are found on the DNA strands which are tightly packed within the chromosomes inside



the cell nucleus.

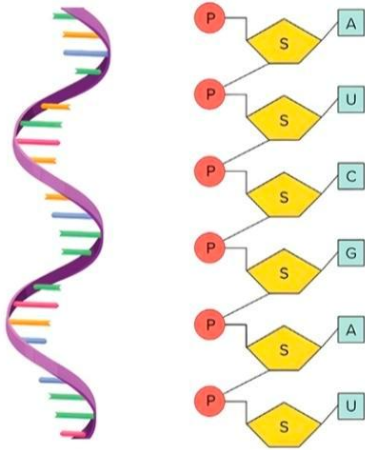
### RNA (Ribonucleic acid)

- Role: RNA plays a crucial role in the synthesis of proteins.
- Composition: Like DNA, RNA is also made up of units called nucleotides.
- Nucleotide Components: Each RNA nucleotide contains three parts:
  1. A ribose sugar
  2. A phosphate group
  3. A nitrogenous base
- Number of Strands: Most RNAs consist of a single strand.
- Nitrogenous Bases in RNA

RNA contains the following four nitrogenous bases:

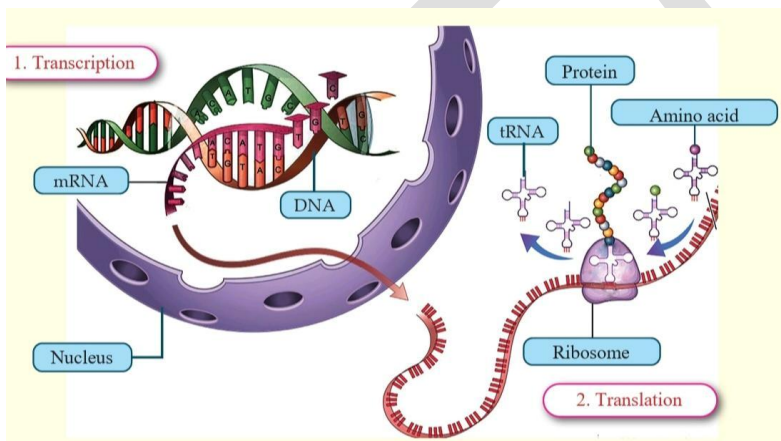
1. Adenine (A)
2. Guanine (G)
3. Uracil (U) – Specific to RNA
4. Cytosine (C)

	Number of strands	Type of sugar molecule	Nitrogen bases
DNA			
RNA			



## Protein Synthesis

### Stage 1: Transcription



- Location: Occurs inside the Nucleus.
- Process: mRNA (messenger RNA) is formed from a specific nucleotide sequence (gene) in DNA.
- Role of Enzymes: Various enzymes help in the formation of mRNA from the DNA template.
- Function of mRNA: The mRNA carries the messages for protein synthesis from the DNA in the nucleus to the ribosomes in the cytoplasm.

### Stage 2: Translation

- Location: Occurs at the Ribosome in the cytoplasm.

- Role of tRNA: tRNAs (transfer RNA) carry specific amino acids to the ribosome based on the message carried by the mRNA.
- Role of rRNA: rRNAs (ribosomal RNA), which are parts of ribosomes, combine these amino acids to make a protein chain.
- Final Product: A specific protein is formed by the linking of amino acids in a particular sequence.

Q1: How are proteins synthesized in the body?

Ans: Proteins are synthesized as a result of the action of genes through two main processes: transcription (formation of mRNA from DNA) and translation (assembly of amino acids into proteins at the ribosome).

Q2: What is the role of mRNA in protein synthesis?

Ans: mRNA (messenger RNA) acts as a carrier of genetic information. It is formed in the nucleus from DNA and carries the specific "message" or instructions to the ribosome in the cytoplasm for building a protein.

Q3: Distinguish between the roles of tRNA and rRNA.

Ans: tRNA (transfer RNA) is responsible for carrying specific amino acids to the ribosome according to the mRNA code.

rRNA (ribosomal RNA) is a structural component of the ribosome that helps in linking these amino acids together to form a protein chain.

Q4: Where do transcription and translation occur in the cell?

Ans: Transcription occurs inside the nucleus, while translation occurs at the ribosomes located in the cytoplasm.

Q5: What is Transcription?

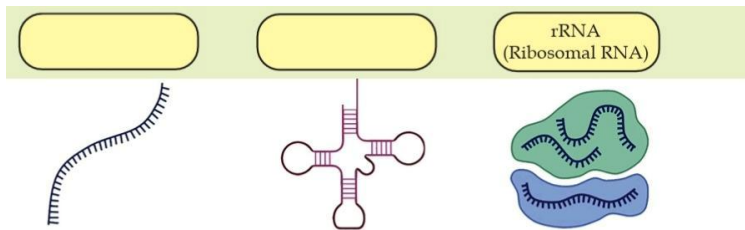
Transcription is the first stage of protein synthesis where a specific gene on the DNA is copied into a single-stranded mRNA molecule. This occurs inside the nucleus.

Q6: What is Translation?

Translation is the second stage where the ribosome reads the mRNA "message." tRNA molecules bring specific amino acids to the ribosome, where rRNA helps link them together to create a functional protein. This occurs in the cytoplasm.

### Different Types of RNAs

There are three primary types of RNA involved in protein synthesis, each with a unique structure and



function:

mRNA (Messenger RNA)	tRNA (Transfer RNA)	rRNA (Ribosomal RNA)
Carries the genetic information (message) from DNA in the nucleus to the ribosome in the cytoplasm.	Carries specific amino acids to the ribosome for protein assembly based on the mRNA message.	The primary component of the ribosome; helps in the formation of peptide bonds between amino acids.

Q1: Proteins are synthesized as a result of the action of genes. The process occurs in two main stages: Transcription and Translation.

What are the various RNAs involved in protein synthesis?

mRNA, tRNA, and rRNA. Each plays a distinct role: mRNA provides the instructions, tRNA provides the building blocks (amino acids), and rRNA provides the machinery.

Q2: How do proteins influence features?

Genes carry the code for proteins. Once a protein is made, it might become a structural part of the body (like hair or skin) or an enzyme that controls chemical reactions. By controlling what proteins are made, genes control the physical and mental traits of the organism.

Q3: Why do offspring resemble their parents?

Because offspring inherit the same genes (and thus the same protein-building instructions) from their parents. Differences occur because the specific combination of genes is unique to each individual.

## **Heredity and Variation**

Heredity: The transmission of characteristics from parents to their offspring.

Variation: The characters expressed in offspring that differ from their parents.

Genetics: The branch of science that deals with genes, heredity, and variation.

### **Gregor Johann Mendel (1822–1884)**

- Known as the 'Father of Genetics'.
- Conducted experiments on pea plants (*Pisum sativum*) in the garden of his monastery. •Studied seven pairs of contrasting traits
- Published his findings in 1866 titled 'Experiments on Plant Hybridisation'. His work was rediscovered in 1900 by Hugo de Vries, Carl Correns, and Erich Von Tschermak.

## **Mendel's Experiments**

Gregor Mendel conducted hybridization experiments in pea plants (*Pisum sativum*) to understand how traits are inherited.

He initially focused on a monohybrid cross, which involves the study of a single pair of contrasting traits at a time.

### **The Monohybrid Cross (Height Trait)**

- Parental Generation (P): Mendel crossed a pure-bred Tall plant with a pure-bred Dwarf plant.
- First Generation (F1): All the offspring obtained in this generation were Tall. The dwarf trait seemed to disappear.
- Second Generation (F2): When the F1 tall plants were self-pollinated, both tall and dwarf plants appeared in the next generation
- Ratio: Approximately 3:1 (3 Tall : 1 Dwarf).

Characters Considered: The primary character was plant height, with two contrasting traits: Tallness and Dwarfness.

Dominant and Recessive Traits: In the F1 generation, only the tall trait was expressed, making it the Dominant trait. The dwarf trait, which remained hidden in F1 but reappeared in F2, is the Recessive trait.

Q1. Why did Mendel choose pea plants (*Pisum sativum*) for his experiments?

Ans: Mendel chose pea plants because they have distinct contrasting traits, a short life cycle, and are easy to grow and self-pollinate or cross-pollinate.

Q2. Distinguish between Phenotype and Genotype.

Ans: Phenotype refers to the physical appearance or observable traits of an organism (like being tall). Genotype refers to the actual genetic makeup or the combination of alleles (like TT or Tt) that results in that appearance.

Q3. In Mendel's experiment, the dwarf trait disappeared in the F1 generation but reappeared in the F2 generation. Why?

Ans: In the F1 generation, the dwarf trait is recessive and is masked by the dominant tall trait (Tt). However, when F1 plants are self-pollinated, the alleles segregate, and some offspring in the F2 generation receive two recessive alleles (tt), allowing the dwarf trait to be expressed again.

Q4. What is the phenotypic ratio of a monohybrid cross in the F2 generation?

Ans: The phenotypic ratio is approximately 3:1 (3 Tall : 1 Dwarf).

### From Factors to Genes

Gregor Mendel hypothesized that characters are passed from parents to offspring through certain factors transmitted through gametes.

These factors were later discovered to be genes, which are located in the chromosomes within the cell nucleus.

## Alleles

A gene that determines a specific character can have different forms. These different forms of genes are called Alleles.

Most genes have two alleles.

For plant height:

Allele T represents Tallness.

Allele t represents Dwarfness.

## **Mendel's Postulates from Monohybrid Cross**

- A trait is controlled by two factors.
- When a pair of contrasting traits is subjected to hybridisation, only one of the contrasting traits is expressed in the offspring of the first generation (F1) and the other remains hidden.
- The trait that appears in the first generation is called the dominant trait, and the hidden trait is called the recessive trait.
- The trait hidden in the first generation reappears in the second generation (F2).
- When gametes are formed, the factors that determine traits get separated without mixing.
- The ratio of dominant to recessive traits in the offspring of the second generation is 3:1.

## **Dihybrid Cross**

A dihybrid cross is a cross between two organisms that are identical hybrids for two traits. Mendel used plant height and seed shape for this experiment.

### Parental Generation (P)

- Parent 1: Tall and Round seeded plant (TTRR)
- Parent 2: Dwarf and Wrinkled seeded plant (ttrr)
- Gametes: TR and tr

### First Generation (F1)

- Result of crossing: TtRr
- Phenotype: All plants are Tall and Round seeded.

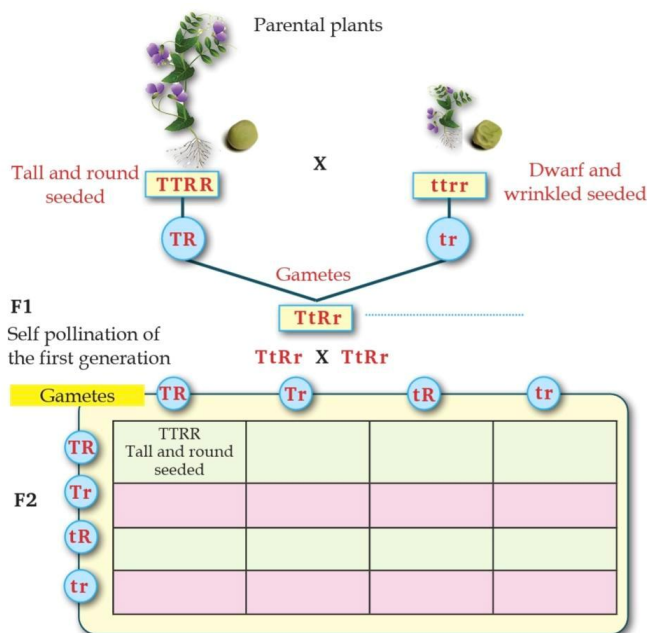
### Second Generation (F2)

Obtained by self-pollination of the F1 generation (TtRr x TtRr).

- Gametes produced by F1: TR, Tr, tR, tr
- Phenotypic Ratio: 9:3:3:1

### Mendel's Postulates

"When two or more different traits are combined, each trait is inherited independently to the next generation without mixing each other. A pair of alleles in an organism does not influence the



separation of another pair of alleles."

### Non-Mendelian Inheritance

Mendel's hypothesis does not apply to all traits. Non-Mendelian inheritance describes patterns where traits do not follow the simple dominant-recessive relationship.

#### **1. Incomplete Dominance**

- Peculiarity: If a red flowered four o'clock plant is hybridised with a white flowered plant, the resulting offspring will have pink flowers.
- Reason: A dominant allele cannot fully hide the allele of the recessive trait.

## **2. Co-dominance**

- Peculiarity: Roan coat pattern, found on some cattle and horses.
- Reason: Both alleles exhibit their traits at the same time.

## **3•Multiple Allelism**

- Peculiarity: ABO blood group in humans.
- Reason: The gene that determines blood group in human beings has more than two alleles. Three alleles IA, IB and I determine the blood group.

## **4.Polygenic Inheritance**

- Peculiarity: Difference in skin colour.
- Reason: More than one gene controls the colour of the skin. The action of these genes cause variation in the production of melanin that causes difference in skin colour.

## **Behind the Colour Difference**

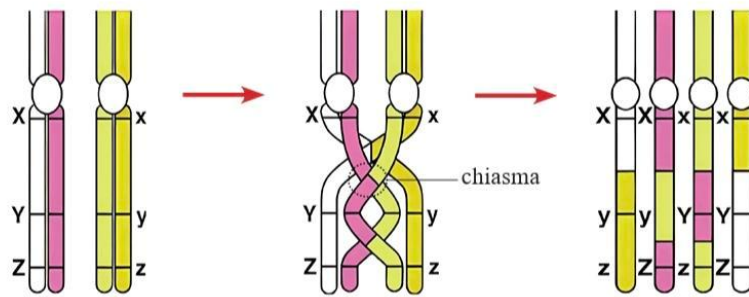
Melanin is the primary pigment that gives colour to skin. The amount of melanin production is controlled by multiple genes, leading to the wide variety of skin tones observed in humans.

### Factors Influencing Skin Colour:

- Geographical Region: The area where an individual's ancestors emerged is a major factor.
- Sunlight Intensity: Genetic variations suitable for the skin colour of each region have occurred based on sunlight intensity.
- Environmental Factors: Sunlight, diet, and vitamin D also influence skin colour.
- Genetic Diversity: The human race is genetically diverse, and skin colour is just one aspect of this diversity.

## **Genetic Processes Responsible for Variations**

### **Crossing Over**



### **The Process of Crossing Over:**

Meiosis is the type of cell division responsible for the formation of gametes. Crossing over occurs during the first phase of meiosis.

1. Pairing: Homologous chromosomes (identical chromosomes inherited from the parents of an organism) pair up.
2. Chiasma Formation: The point of contact between the paired chromosomes is called the chiasma. The chromatids break at this region.
3. Exchange and Recombination: The broken segments are exchanged with each other. This exchange causes a recombination of alleles.

### **Role of Crossing Over:**

Crossing over leads to the appearance of new traits in the offspring. It is one of the chief processes responsible for variations among individuals.

### **Mutation**

Mutation is the sudden heritable change in the genetic constitution of an organism. It involves changes in the structure or sequence of genes (DNA).

#### **Reasons for Mutation**

- Errors during DNA replication: Mistakes occurring when the cell copies its genetic material.
- Exposure to chemicals: Certain environmental chemicals (mutagens) can alter DNA structure.
- Radiations: Exposure to harmful radiations (like UV rays, X-rays) can cause genetic changes.