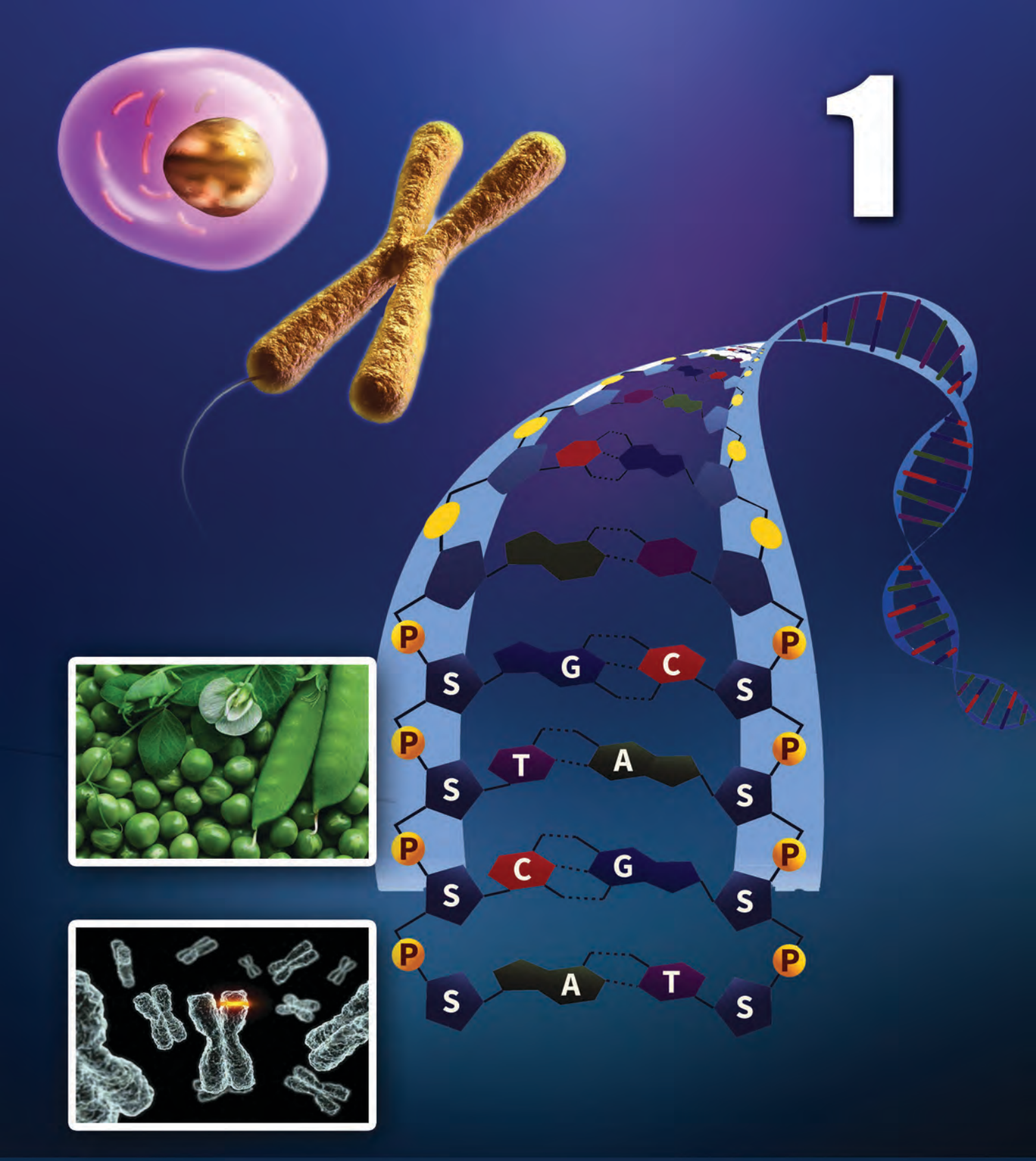


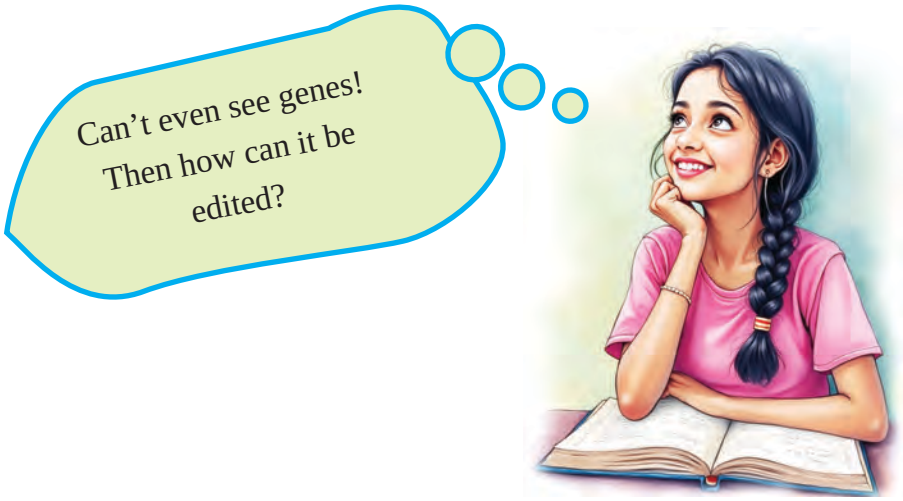

# Genetics of Life

## The Nobel Prize in Chemistry for developing methodology of Gene Editing

Jennifer A Doudna  Emmanuelle Charpentier

The Nobel Prize of 2020 in Chemistry was shared by Emmanuelle Charpentier and Jennifer A Doudna for their contributions in the field of gene editing. The award is for the discovery of a technology called CRISPR-Cas 9, a gene editing process which can bring desirable changes in the genes in DNA. This discovery is expected to make revolutionary advances in genetic disease therapy and treatment of cancer. It can also be used to develop crops that are resistant to pests and diseases.

You have read the extract related to the technology which can bring revolutionary changes in the treatment of diseases and other fields.




Didn't you too have this doubt?

A deeper understanding of DNA (Deoxyribonucleic acid) and genes paved the way for gene editing.

Let us understand the location and ultra structure of DNA and gene.

Observe illustration 1.1 and make notes on the location of DNA.

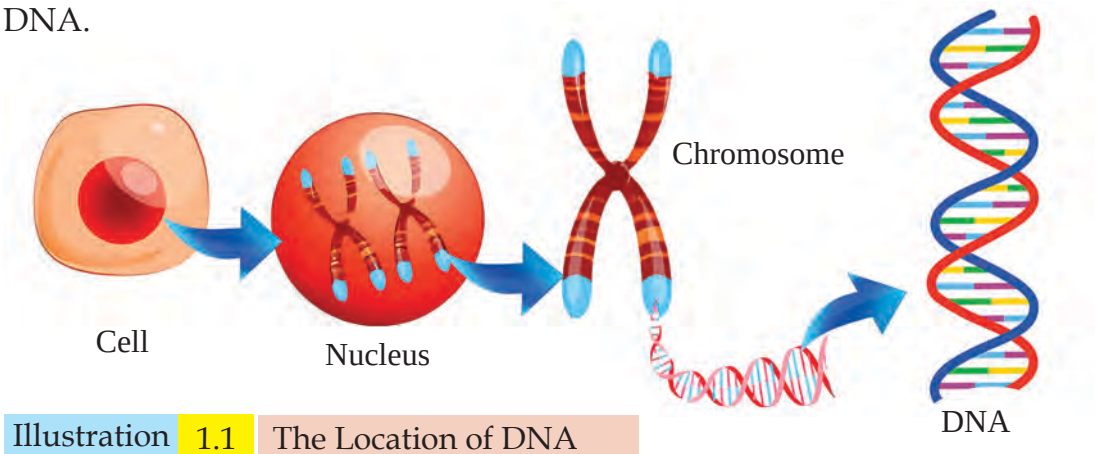



Illustration 1.1 The Location of DNA

You have understood the location of DNA. Discovery of the structure of the nucleic acid DNA was one of the greatest leaps in the field of biological studies.

## Structure of DNA

In 1953, James Watson along with Francis Crick had presented the double helical model of DNA. They proposed the structure of DNA based on the X-ray diffraction studies conducted by Rosalind Franklin and Maurice Wilkins. The crucial information that led to this discovery was obtained from the famous 'Photo 51', an X-ray diffraction image of DNA taken by Rosalind Franklin. Rosalind Franklin passed away at the age of 37 in 1958. James Watson, Francis Crick and Maurice Wilkins were awarded the Nobel Prize in Medicine in 1962 for their contributions on the discovery of the double helix model of DNA.

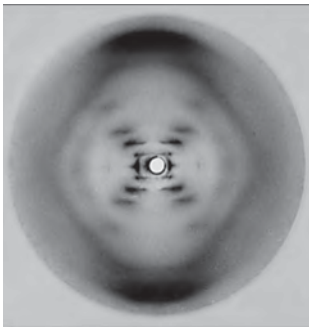
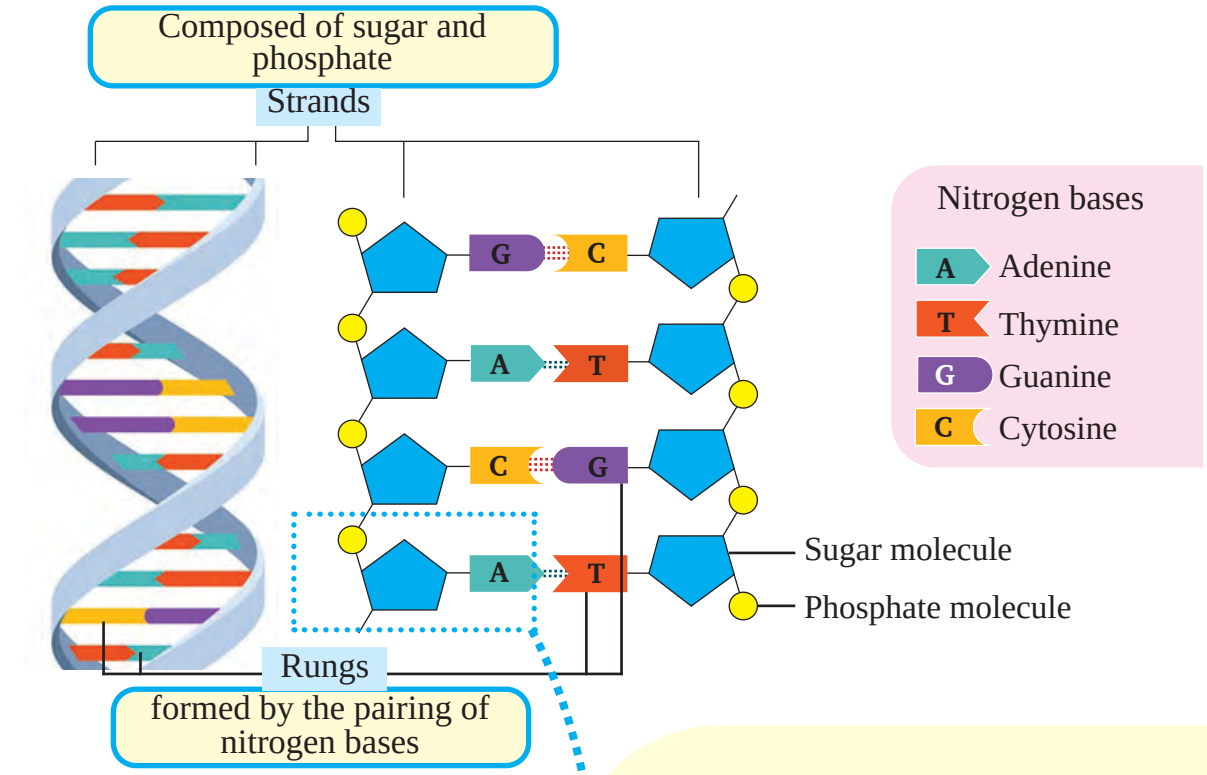


Photo 51

Rosalind Franklin 1920-1958 | Maurice Wilkins 1916-2004 | Francis Crick 1916-2004 | James Watson 1928

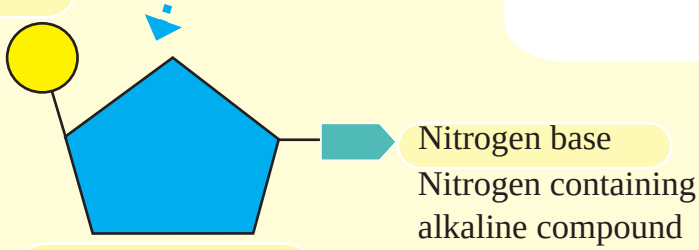
Analyse the illustration 1.2 to gain an understanding on the structure of DNA and complete the work sheet 1.1.




Double helix model of DNA

**Nucleotide**

The basic building block of DNA Each nucleotide is composed of a deoxyribose sugar, a phosphate group, and a nitrogen base.




Participates in the formation of bonds that link nucleotides together

Illustration 1.2 Structure of DNA

## Worksheet

| | |
|---|---|
| 1. Number of strands in DNA | |
| 2. Molecules used to make strands | |
| 3. Molecules used to make rungs | |
| 4. Different types of nitrogen bases | |
| 5. Formation of rungs | |
| 6. Mode of nitrogen base pairing | |
| 7. Molecules in a nucleotide | |

### Worksheet 1.1 The structure of DNA

Prepare the double helix model of DNA by using locally available waste materials and display it in class.

How does normal sugar differ from a sugar molecule in DNA? Find out.

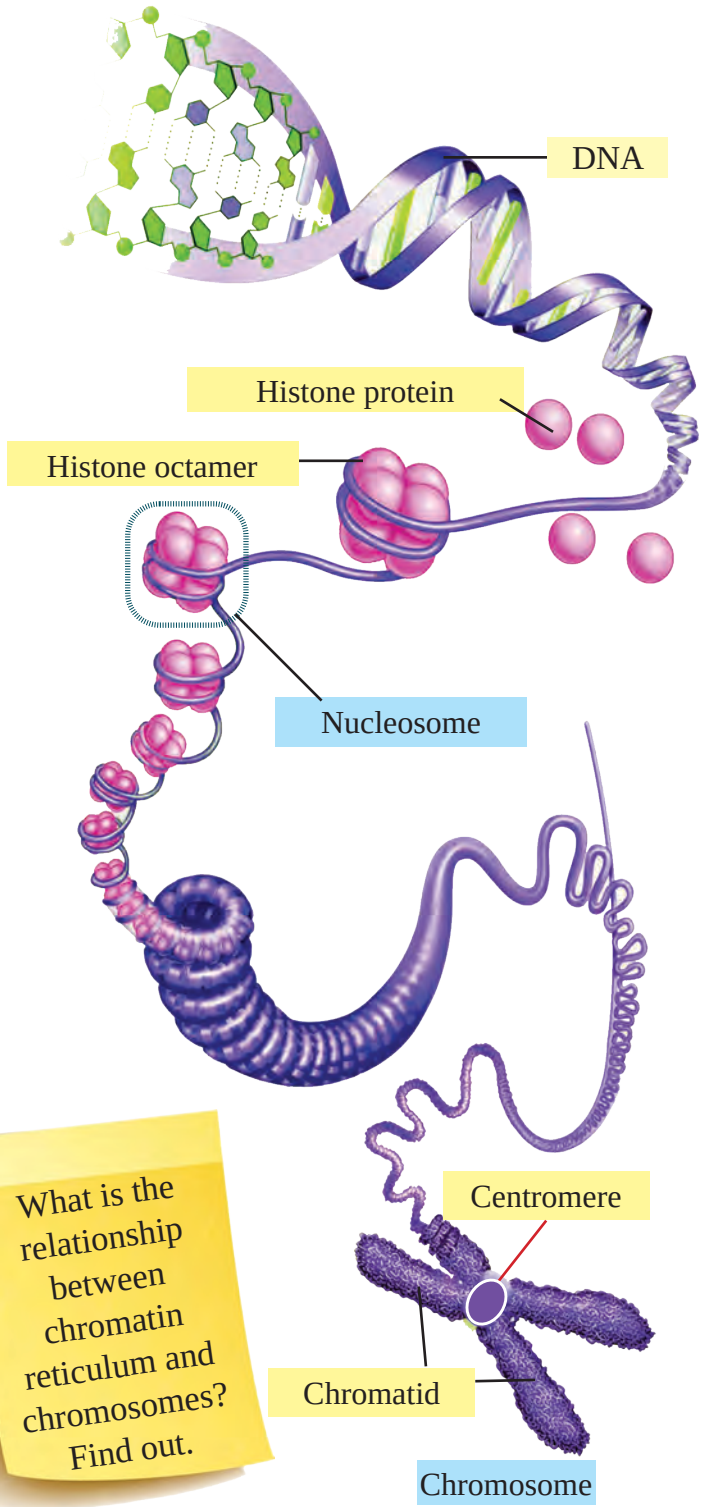
### Size of DNA

The DNA in each chromosome is about 2 inches (5cm) long. If DNA from 46 chromosomes of a human cell joins together, it would be around 6 feet in length (2m). The human body is made up of trillions (one lakh crore) of cells. If the DNAs of all the cells joins together, it would be about 67 billion (one billion = 100 crore) miles in length. It is capable enough to wrap around the Earth over two million times!

How does such a larger DNA fit into the chromosomes of tiny cells?

Haven't you listened to the child's doubt?

Analyse the illustration 1.3 and description based on the indicators and find the answer to the child's doubt.




What is the relationship between chromatin reticulum and chromosomes? Find out.

## Chromosome

DNA and histone proteins are the primary components of a chromosome. Eight histone proteins join together to form a histone octamer. DNA strands wind around this **octamer** to form a structure called **nucleosome**. The chromosomes are formed by packing and coiling numerous nucleosomes and recoiling the chains of nucleosomes. **Chromatids** are the parts of a chromosome which are connected by means of centromere.

## Indicators

- Building blocks of chromosomes
- Histone and nucleosome
- Formation of chromosome
- Chromatid, centromere

Illustration 1.3 The structure of chromosome

Each species possess a specific number of chromosomes. How many chromosomes do humans have? How do they appear? Analyse the illustration 1.4 based on the indicators, understand human chromosomes and prepare notes.

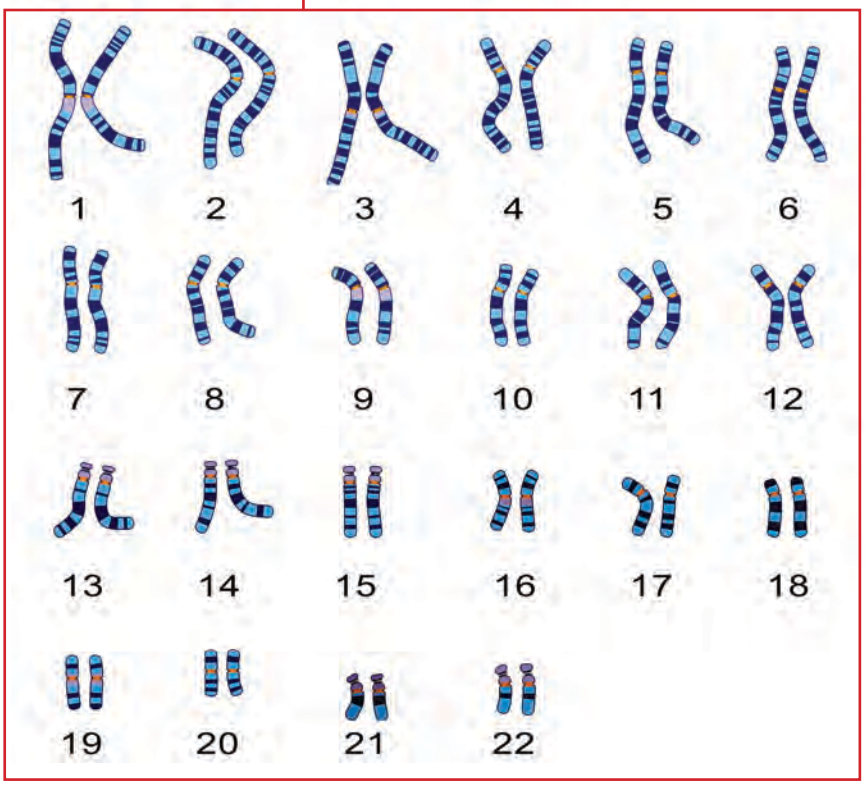
## Human Chromosomes

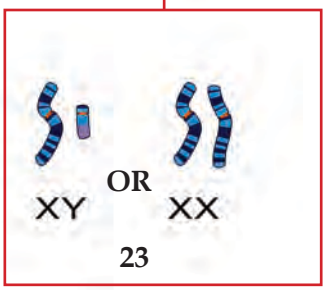
### Somatic chromosomes

These are chromosomes that control physical characteristics. There are twenty two pairs of somatic chromosomes. A pair of identical chromosomes together form a homologous chromosome. One of these is inherited from the mother and the other from the father.

### Sex Chromosomes

These are the chromosomes which are involved in sex determination. They are of two types. X chromosome and Y chromosome. The Y chromosome is comparatively smaller than that of the X chromosome. The SRY gene on the Y chromosome is responsible for the development of testis in the embryo.




Why are chromosomes seen in pairs? Find out.

Illustration 1.4 Human chromosomes

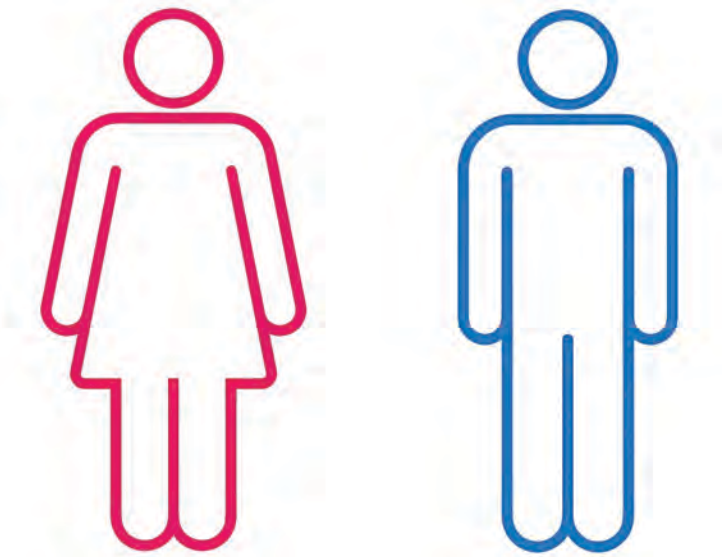
Are there multiple organisms with the same number of chromosomes? Find out.

## Indicators

- Different types of chromosomes
- Somatic chromosomes and their functions
- Homologous chromosomes
- Sex chromosomes and their function

Prepare a table including the chromosome number of various organisms that are seen in your surroundings and display it in class.

Pay attention to the given genetic constitution of a female and a male and complete table 1.1. Analyse the information in the table and draw inferences of chromosomal similarities and differences between a female and a male.



44 + XX

44 + XY

| | Genetic constitution | Total Number of chromosomes | Number of somatic chromosomes | Number and type of sex chromosomes |
|---|---|---|---|---|
| Female | | | | |
| Male | | | | |

Table 1.1 Human Chromosomes

### Different genetic constitutions

Though, 44+XX and 44+XY are considered as normal genetic constitutions, many different kinds of genetic constitutions are seen in humans. These variant genetic constitutions influence the physical and mental development of the individuals. Gender determination is a complex process which not only depends upon genetic constitution, but also on other factors. Some examples for variant genetic constitutions:

- 44+X0: Females with only one X chromosome. They have the condition called Turner syndrome.
- 44+XXX: Females with three X chromosomes. They have triple-X syndrome.
- 44+XXY: Males with two X chromosomes and one Y chromosome. They have Klinefelter syndrome.
- 44+XYY: Males with one X chromosome and two Y chromosomes. They have XYY syndrome.

Gender determination may become complex in individuals with these genetic constitutions mentioned above. For example, females with Turner syndrome may have female sex organs. However, they may not develop female sexual characteristics at the beginning of their adolescence. Males with Klinefelter syndrome may have male sex organs, but they may also exhibit female characteristics.

The Y chromosome of the father is important in sex determination. Why? Find out.

You have understood that genes provide instructions as to how our body should function. Where are these genes found? How do they perform? Analyse illustration 1.5, their description and prepare notes out of it.

## Gene

Gene is a specific sequence of nucleotides in DNA. Proteins, which are synthesised according to the instructions of genes, are responsible for the formation of characteristic features and for controlling metabolic activities.

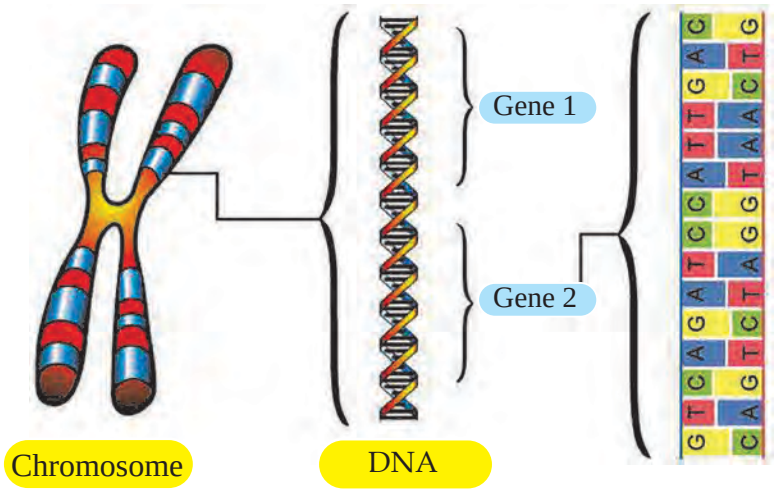



Illustration 1.5 Genes

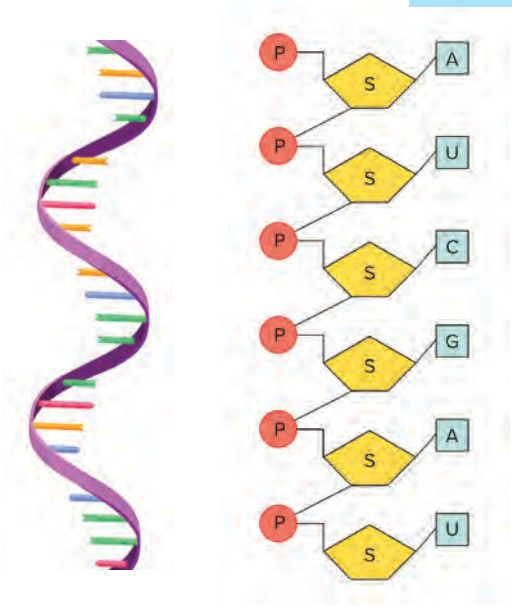



Illustration 1.6 Structure of RNA

The nucleic acid RNA also plays a crucial role in the synthesis of proteins.

Analyse illustration 1.6 and the description to understand the structure and characteristic features of RNA.

## RNA (Ribonucleic acid)

RNA is another type of nucleic acid, similar to DNA. They are also made up of nucleotides. Each of the nucleotide contains a ribose sugar, a phosphate group, and a nitrogenous base. The nitrogen bases in RNA are Adenine, Guanine, Uracil, and Cytosine. Most of the RNAs have a single strand.

Didn't you understand the structure of RNA? Compare this with the structure of DNA and complete the table 1.2.

| | Number of strands | Type of sugar molecule | Nitrogen bases |
|---|---|---|---|
| DNA | | | |
| RNA | | | |

Table 1.2 DNA, RNA Comparison

## Protein Synthesis

Have you understood that the proteins are synthesised as a result of the action of genes? Prepare a note on various stages of protein synthesis by analysing the illustration 1.7 and the description based on the indicators.

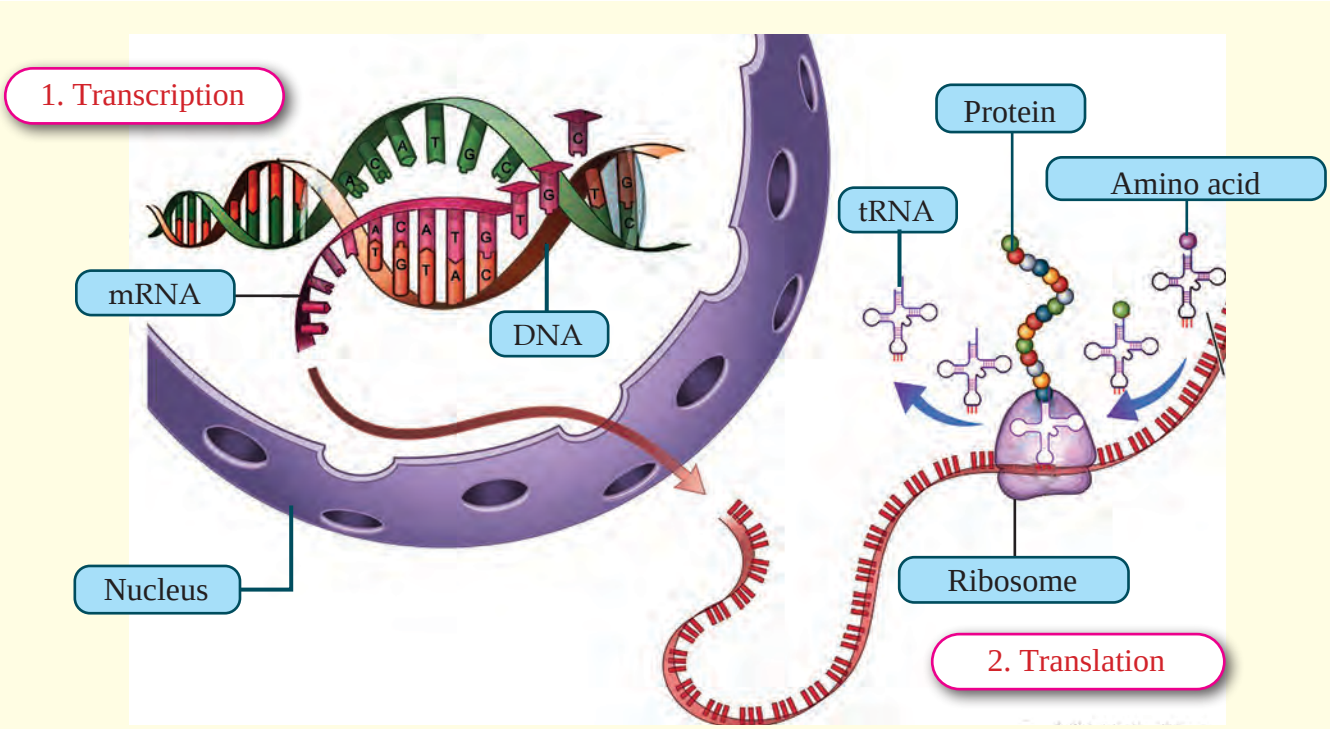



Illustration 1.7 Protein Synthesis

**1. Transcription**

mRNA is formed from a specific nucleotide sequence (gene) in DNA with the help of various enzymes. The mRNA contains messages for protein synthesis.

**2. Translation**

tRNAs (transfer RNA) carry specific amino acids to the ribosome based on message in the mRNA that has reached the ribosome from the nucleus. The rRNAs (ribosomal RNA), which are part of ribosomes combine amino acids to make protein.

## Indicators

- Stages of Protein Synthesis
- Processes take place in the nucleus
- Processes take place in the cytoplasm

Prepare a flow chart that shows the various stages of protein synthesis and display it in the classroom.

Various RNAs involved in protein synthesis are given in the illustration 1.8. Complete it by including their name and function.

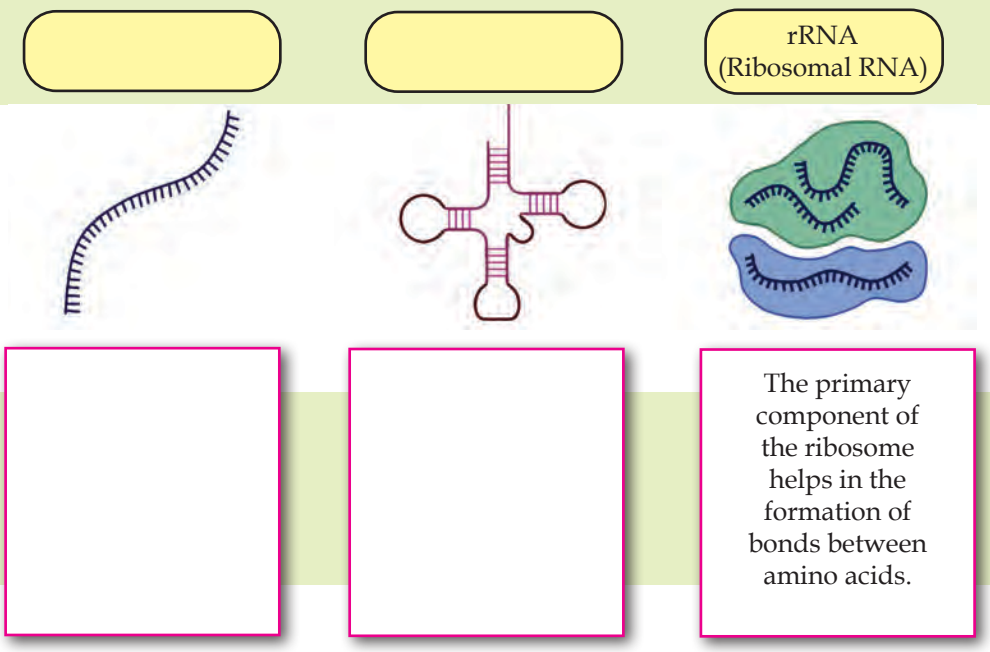



Illustration 1.8 Different types of RNAs

To understand how proteins, which are synthesised according to the direction of genes, influence the characteristic features of organisms, observe the pictures given and find out the similarities and differences between parents and offsprings.

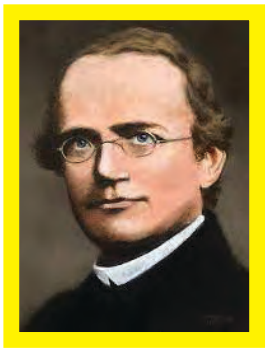
## Similarities and Differences in Characters

Some characteristics of parents are also found in their children. Isn't it also common to see certain characters in children which differ from their parents?

Heredity refers to the transmission of characteristics from parents to their offspring. Variations are characters expressed in offspring, that differ from their parents. Genes inherited from parents are responsible for both heredity and variations.

The historical path that led science to the discovery of genes is pivotal. Analyse the given description to get a deeper understanding.

Mendel Museum of Masaryk University in Brno



Gregor Johann Mendel
1822-1884

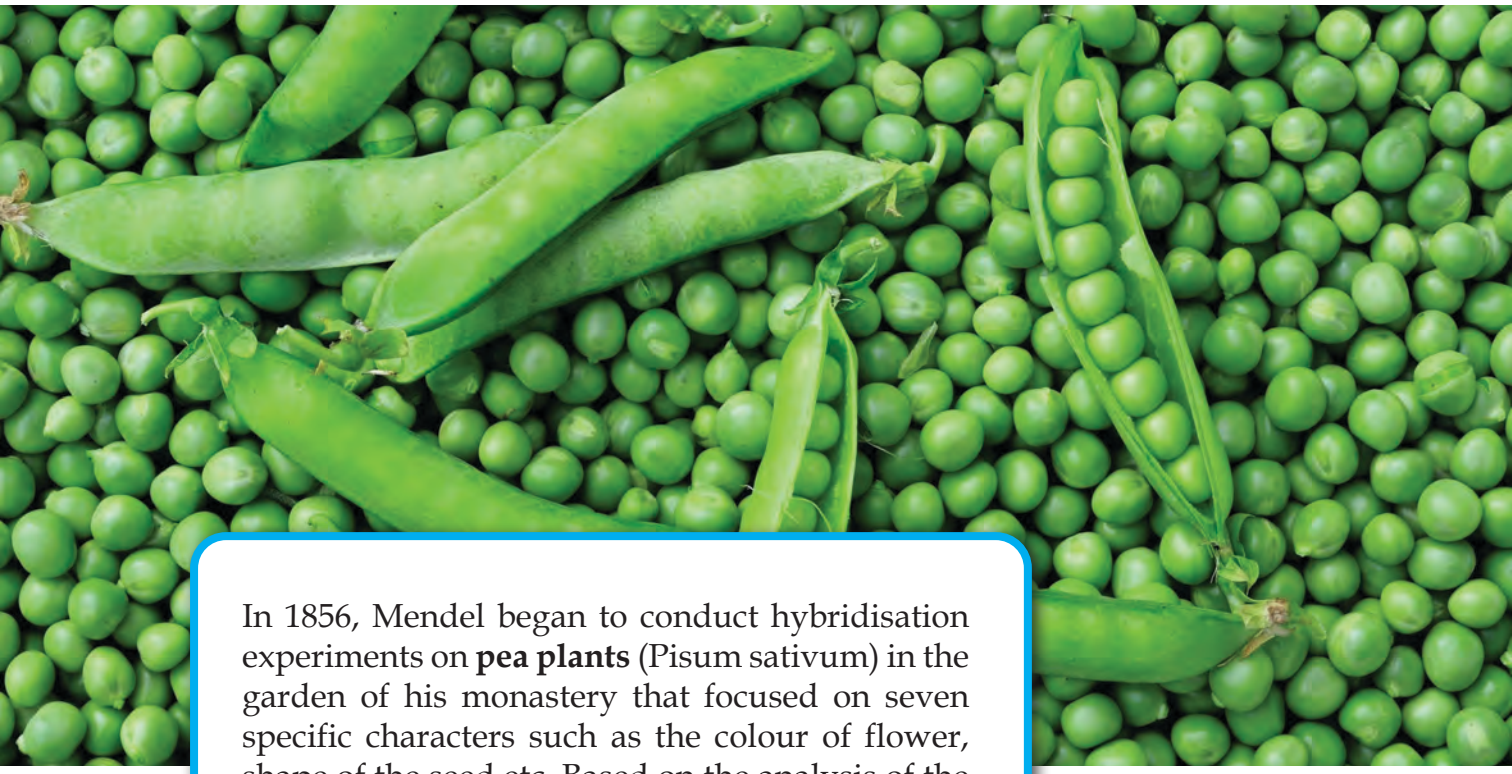
## Genetics in the Garden

Genetics is the branch of science that deals with genes, heredity, and variation. Gregor Johann Mendel's experiments on pea plants (Pisum sativum) and the conclusions he drew out of hybridisation experiments laid the foundation for the field of genetics. Therefore, he is considered as the father of genetics.

### Know the scientists

**Gregor Johann Mendel**

Gregor Johann Mendel was born on 20 July, 1822, at Hyncice a small village of Northern Moravia, which is now known as Czech Republic. After joining the Augustinian monastery at Brno, he became a priest in 1847. Between 1851 and 1853, he attended the University of Vienna where he studied Physics, Mathematics, and Natural sciences, and learned statistical methods to analyse data scientifically.



In 1856, Mendel began to conduct hybridisation experiments on **pea plants** (Pisum sativum) in the garden of his monastery that focused on seven specific characters such as the colour of flower, shape of the seed etc. Based on the analysis of the experimental result, he explained that a pair of factors controls each character and represented those factors using symbols. These factors are now known to be genes. Gregor Mendel's conclusions are known as the **Laws of Inheritance**. These laws provide the fundamental genetic framework to understand heredity and variation.

In 1865, he presented his findings in the Natural History Society at Brno. The following year, he published a thesis titled 'Experiments on Plant Hybridisation.' However, the scientific community of that time largely ignored Mendel's discoveries. Gregor Mendel passed away in 1884.

In 1900, sixteen years after his death, botanists Hugo de Vries, Carl Correns, and Erich von Tschermak recognised the significance of Mendel's research. With this, Mendel's findings were accepted as the foundation of the science of genetics. Genetics has grown into the most extensive branch of science through numerous contributions of various scientists.

Hugo de Vries

Carl Correns

Erich Von Tschermak

## Mendel's Experiments

Mendel initially conducted hybridisation experiments by considering a single pair of contrasting traits. This is known as a monohybrid cross. The hybridisation experiment conducted considering the trait of the height is depicted in the illustration 1.9. Analyse it based on the indicators, and form inferences.

Parental plants

Tall

X

Dwarf

Tall

First generation ($F_1$)

He hypothesised that there must be certain factors within the seed that control traits. To find out what might have happened to the factor responsible for dwarfness, he self-pollinated the plants obtained in the first generation to produce a second generation of plants.

Second generation ($F_2$)

1064 plants

787 Tall

277 Dwarf

The ratio between the traits is approximately 3:1

Illustration 1.9 Monohybrid Cross

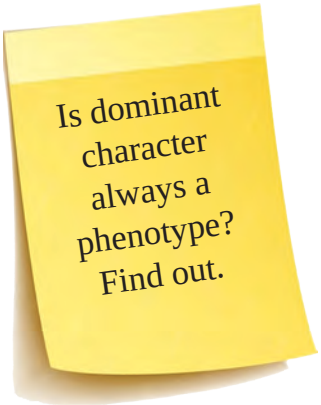
## Indicators

- The characters considered and their traits
- Dominant and recessive traits in the first generation
- Importance of self-pollination
- Traits in the second generation

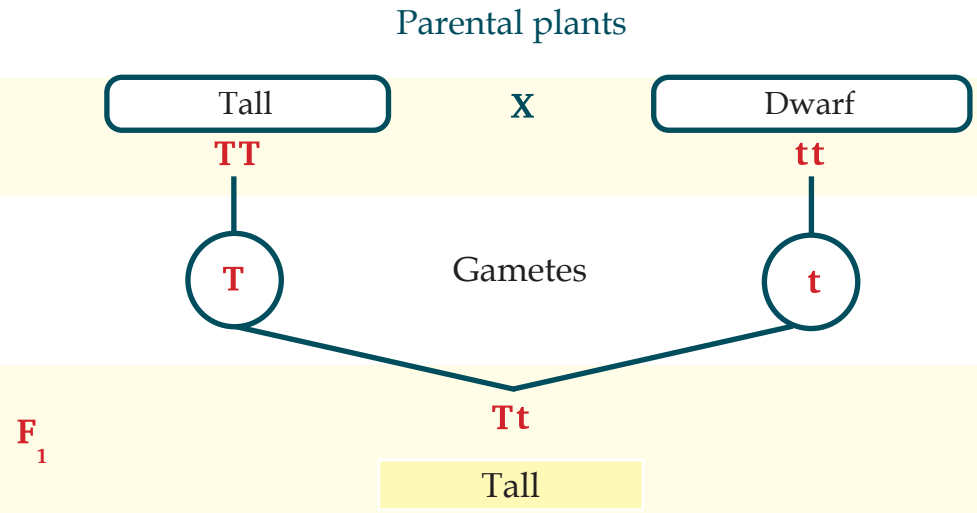
While conducting hybridisation experiments based on the contrasting traits of six other characteristics in pea plants, Mendel obtained results similar to the first experiment.

### Factors

Gregor Mendel hypothesised that characters from parents are passed on to offsprings through certain factors that are transmitted through gametes. It was only after Mendel's period that, these factors were discovered to be genes that are located in chromosomes in the nucleus. A gene that determines a character can have different forms. These different forms of genes are called alleles. A gene usually has two alleles. The hybridisation experiment shown in illustration 1.9, the different alleles that determine the character of height are represented by ***T*** and ***t***. The allele ***T*** represents tall and the allele ***t*** represents dwarf. The observable characteristics of an organism are called phenotype, and the genetic constitution responsible for these characteristics are called genotype.




Observe the illustration 1.10 of the hybridisation experiment where the factors controlling the traits are represented using symbols. Complete the illustration, discuss and prepare notes based on the indicators.




Self pollination of the first generation

Tt X Tt

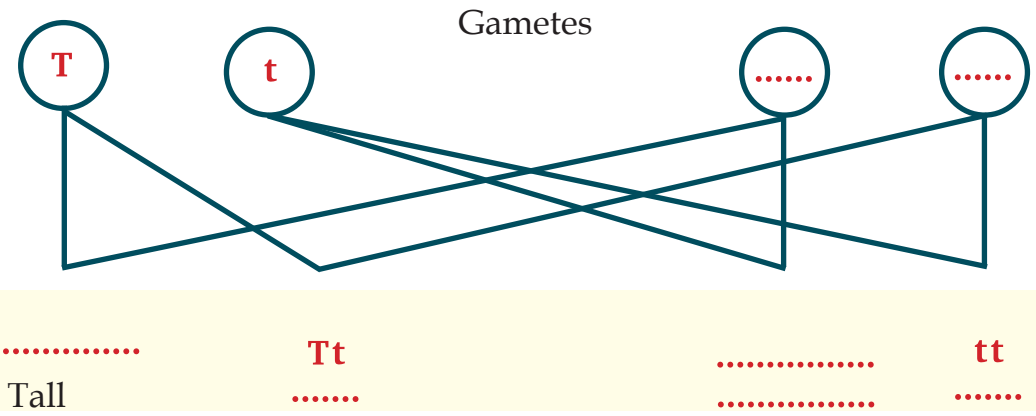



Illustration 1.10 Monohybrid cross

## Indicators

- Phenotype and genotype of the parental plants
- Phenotype and genotype of first generation plants
- Genotype of the tall parental plant and the first generation plant
- Ratio of the traits in the second generation

The following are the inferences drawn by Gregor Mendel from monohybrid cross. Analyse them and answer the questions given below.

### Mendel's Postulates

- 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 and the other remains hidden. The trait that appears in the first generation is called dominant trait and the hidden trait is called recessive trait. The trait hidden in the first generation reappears in the second generation.
- When gametes are formed, the factors that determine trait gets separated without mixing.
- The ratio of dominant to recessive traits in the offspring of the second generation is 3:1.

? Why was a plant with intermediate height not formed by the combination of tall and dwarf?

? Hasn't the character that is not expressed in the first generation appeared in the second generation? How would that be?

In the next stage, he observed the inheritance of two pairs of contrasting traits of the same plant. This is known as dihybrid cross. Analyse the illustration 1.11 of hybridisation experiment conducted by considering height of the plant and shape of the seed based on the given indicators, and note down your inferences.

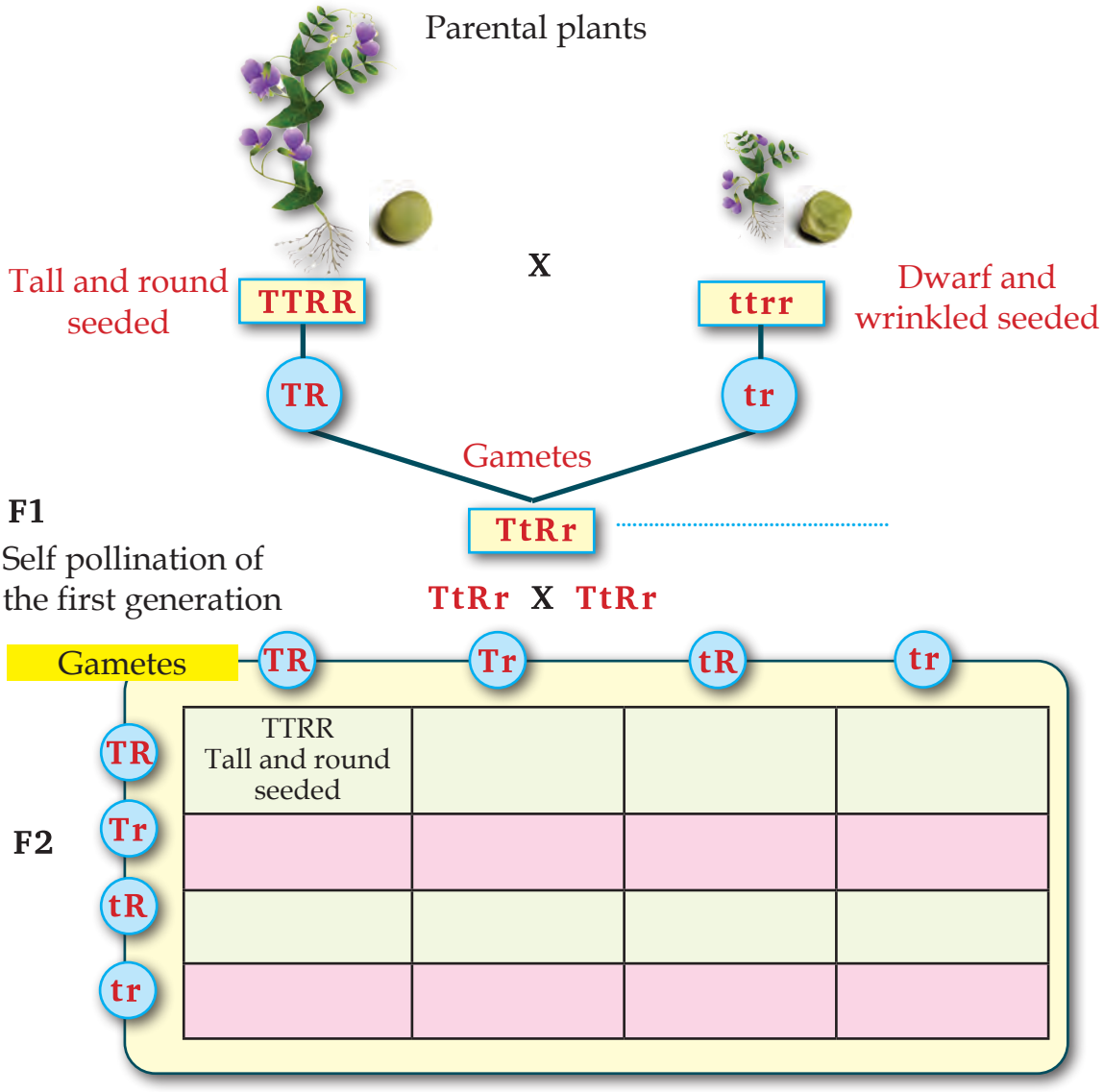



Illustration 1.11 Dihybrid cross

## Indicators

- Characters considered and their contrasting traits
- Phenotype and genotype of the parental plants
- Dominant and recessive traits of the first generation
- Alleles of the gametes produced by the first generation
- Phenotype of the plants in the second generation
- Phenotypes observed in the second generation that differed in contrast to the parental plants and their genotypes
- Phenotypic ratio of the second generation

The inference that Mendel drew from this experiment is given below. Analyse it and find out the answer to the given question.

**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.)

? Characters that are not found in the parent plants are found in the second generation. Why?


Don't you notice the child's doubt when she observed the flowers? What is your opinion?

Mendel's laws were the foundation of genetics. However, it could not fully explain the diversity of traits observed in organisms. Later studies about the complex interaction among genes, environment and other factors revealed some of the limitations of Mendel's laws. This gave rise to the concept of Non-Mendelian Inheritance.

Analyse various situations given below and find out how they differ from Mendel's hypothesis and then answer the child's doubt.

## Non Mendelian Inheritance

If a red flowered four o'clock plant is hybridised with a white flowered plant, the resulting offspring will have pink flowers.

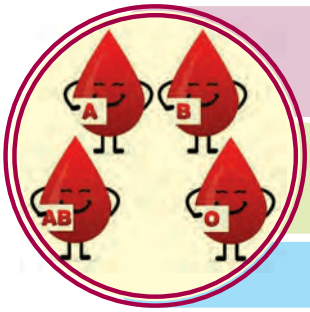
A dominant allele cannot fully hide the allele of the recessive trait.

Incomplete Dominance

Roan coat pattern, found on some cattle and horses

Both alleles exhibit their traits at the same time.

Co-dominance




ABO blood group in humans

The gene that determines blood group in human beings has more than two alleles. Three alleles IA, IB and i determine the blood group.

Multiple allelism

Difference in skin colour

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.

Polygenic inheritance

*Peculiarity* *Reason* *Name of the inheritance*

Table 1.3 Non Mendelian Inheritance

Explore more situations and examples of Non Mendelian Inheritances and present in the class.

### Behind the colour difference

Melanin is the primary pigment that gives colour to skin. The amount and type of melanin determine the colour of the skin. The geographical region from where an individual's ancestors emerged is a major factor that influences skin colour. As the intensity of sunlight varies in different geographical regions, genetic variations suitable for the skin colour of each region have occurred. Environmental factors such as sunlight, diet, and vitamin D also influence skin colour. The human race is genetically diverse, and skin colour is only one aspect of this diversity.

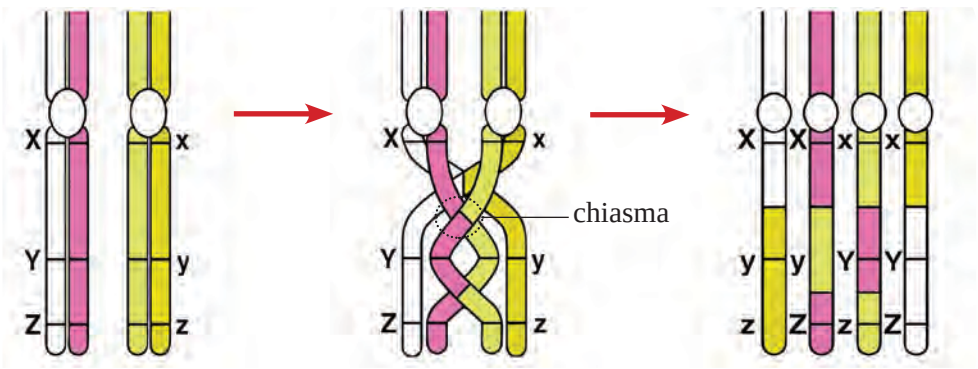

Haven't you noticed the child's doubt?

The characteristics the offspring receives from parents may not always be the same. The chief genetic processes which are responsible for this diversity among individuals are given below. Analyse them and find out the answer to the child's doubt.

## Genetic processes responsible for variations

### Crossing over

Meiosis is the type of cell division that is responsible for the formation of gametes. Analyse the illustration 1.12 to gain an understanding about the process of crossing over, which occurs during the first phase of meiosis.




Pairing of homologous chromosomes (Identical chromosomes inherited from the parents of an organism)

The point of contact of the paired chromosomes is called **chiasma**. The chromatids break at this region and the broken segments are exchanged with each other.

This exchange causes a recombination of alleles. This leads to the appearance of new traits in the offspring.

Illustration 1.12 Crossing over

### Indicators

- Phase in which crossing over occurs
- Process of crossing over
- Role of crossing over in the formation of variations

## Mutation

**Mutation** is the sudden heritable change in the genetic constitution of an organism.

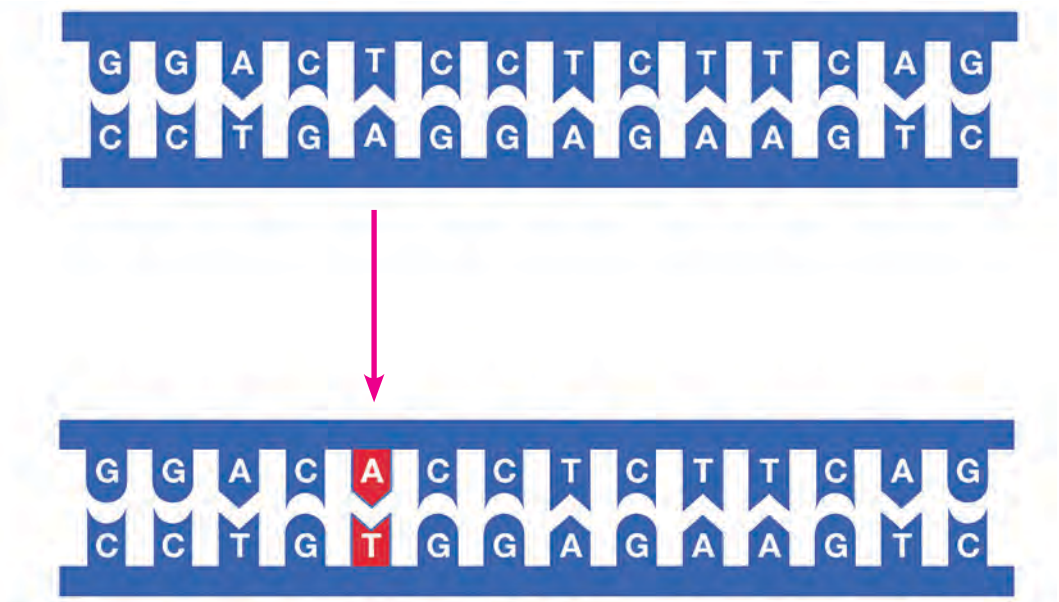



Illustration 1.13 Mutation

Mutations can be caused by errors during DNA replication, exposure to certain chemicals, radiations, etc. Mutation causes changes in genes. These genes are transferred through generations which leads to variations in characters. Mutations play a crucial role in the process of evolution.

### Indicators

- Mutation
- Reasons for mutation
- Importance of mutation

Haven't you understood that recombination of alleles during fertilisation, crossing over, mutation, etc. causes variations in organisms?

Collect more information on the processes that cause variations and prepare a slide presentation.

## Genetics- Career Opportunities

Genetics is a broad field with various branches, that includes molecular genetics, population genetics, medical genetics, cytogenetics, behavioural genetics, and genomics.

Graduate programmes in genetics and related fields such as biotechnology, microbiology, bioinformatics etc. open doors to a wide range of career opportunities. At the postgraduate level specialised studies in genetic counselling, genomics, medical genetics, and forensic science provide opportunities in healthcare, research, and education. Advanced degrees such as a Ph.D in Genetics equip students for careers in cutting-edge research, pharmaceuticals, agriculture, and other industries. The world of genetics is so vast that it offers endless opportunities for new discoveries and research. For those who are curious about the mysteries of life, genetics is a scientific field that offers more possibilities to explore more domains.

Genetics is the branch of science that unravels the mysteries of life. Genetics provides an understanding about how genes function and how they get transmitted from one generation to the next. Advances in this field have led to significant developments in genetic engineering. We have covered certain steps that lead to the answer to the child's doubt regarding gene editing. We will be able to get a complete answer as we go through the upcoming chapters.

Discoveries in genetics help us to understand and explain how evolution takes place. The combination of various genetic processes determines the characteristics of an organism. These processes are fundamental to the diversity and evolution of life. We will understand how it is, in the next chapter.

## Let us Assess

1. Are basic building blocks of DNA and RNA the same? Explain.
2. Analyse the statements and choose the appropriate one.
   - i. $F_1$ has similarity with both the parents.
   - ii $F_1$ has no similarity with any of the parents' character intermediate to them.
   - iii $F_1$ has similarity with one of the parents
     - a) i - Dominance, ii - Incomplete dominance, iii - Co-dominance
     - b) i - Incomplete dominance, ii - Dominance, iii - Co-dominance
     - c) i - Co-dominance, ii - Incomplete dominance, iii - Dominance
     - d) i - Dominance, ii - Co-dominance, iii - Incomplete dominance
3. Which of the following is contributed by organisms that reproduce sexually, to their offspring?
   - a) All genes
   - b) Half of their genes
   - c) One fourth of their genes
   - d) Double the number of genes
4. A tall pea plant with purple flowers (dominant character) is crossed with a dwarf plant with white flowers.
   - a) Illustrate the dihybrid cross of these and write the $F_2$ ratio.
   - b) Did characters that differ from the parents appear in the $F_2$ generation? Why?
   - c) If both the genes are not assorting independently, how does it affect the $F_2$ ratio?
5. How does dominance, co-dominance and incomplete dominance differ from one another?
6. Different phenotypic ratios are obtained in monohybrid and dihybrid cross. Why? What does it indicate about the inheritance of characters?
7. Even though a gene responsible for certain characters has more than two alleles, why does that particular gene have only two alleles in an individual ?

8. Although the DNA possesses all genetic information for protein synthesis, RNA is also required for protein synthesis. Why?
9. How do co-dominance and multiple allelism function in the determination of blood group in the ABO blood grouping in human beings? Explain.
10. All ova formed in females contain one type of sex determining chromosome. Why?

## Extended activities

1. Present the process of transcription and translation in the class, using coloured beads or paper strips to indicate nucleotides.
2. Prepare and present a time-line animation in class that depicts the steps in the development of genetics.
3. Conduct Mendel's hybridisation experiment on pea plants using dices or beads to represent alleles.
4. Collect data and draw conclusion on how sex determination in various species takes place and the influence of environmental factors on it.
5. Discuss the scientific, social and cultural dimensions of skin colour variation.