CHAPTER: 1

Genetics of Life

JOE FREDY (HST)

The Nobel Prize in Chemistry for developing methodology of Gene Editing

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.

JOE FREDY (HST)

Structure of DNA

JOE FREDY (HST)

Structure of DNA

Rosalind Franklin
1920-1958

Maurice Wilkins
1916-2004

Francis Crick
1916-2004

James Watson
1928

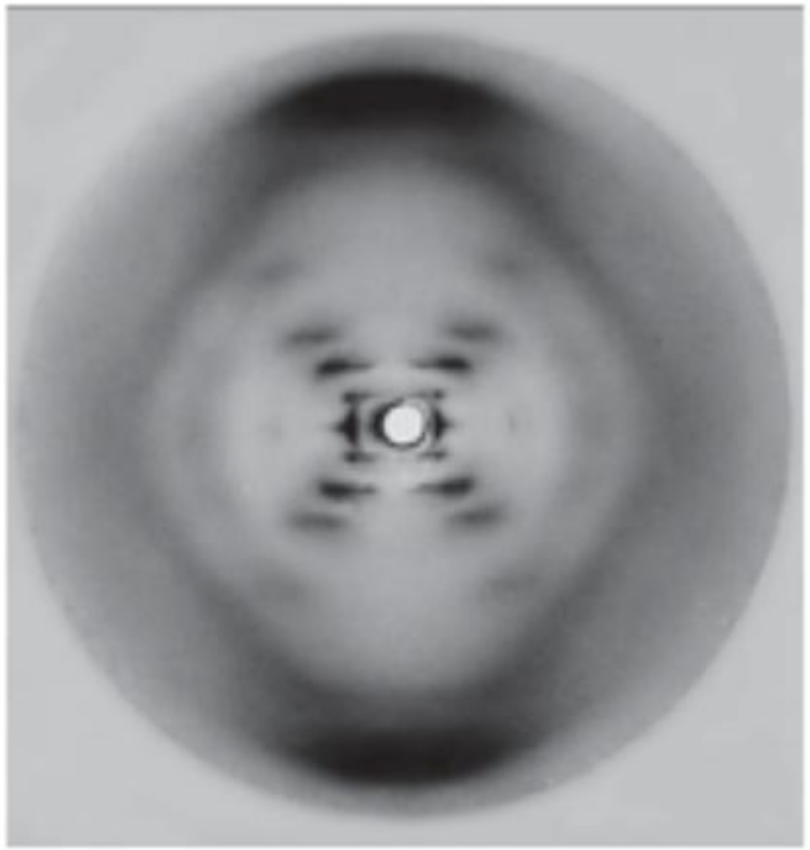
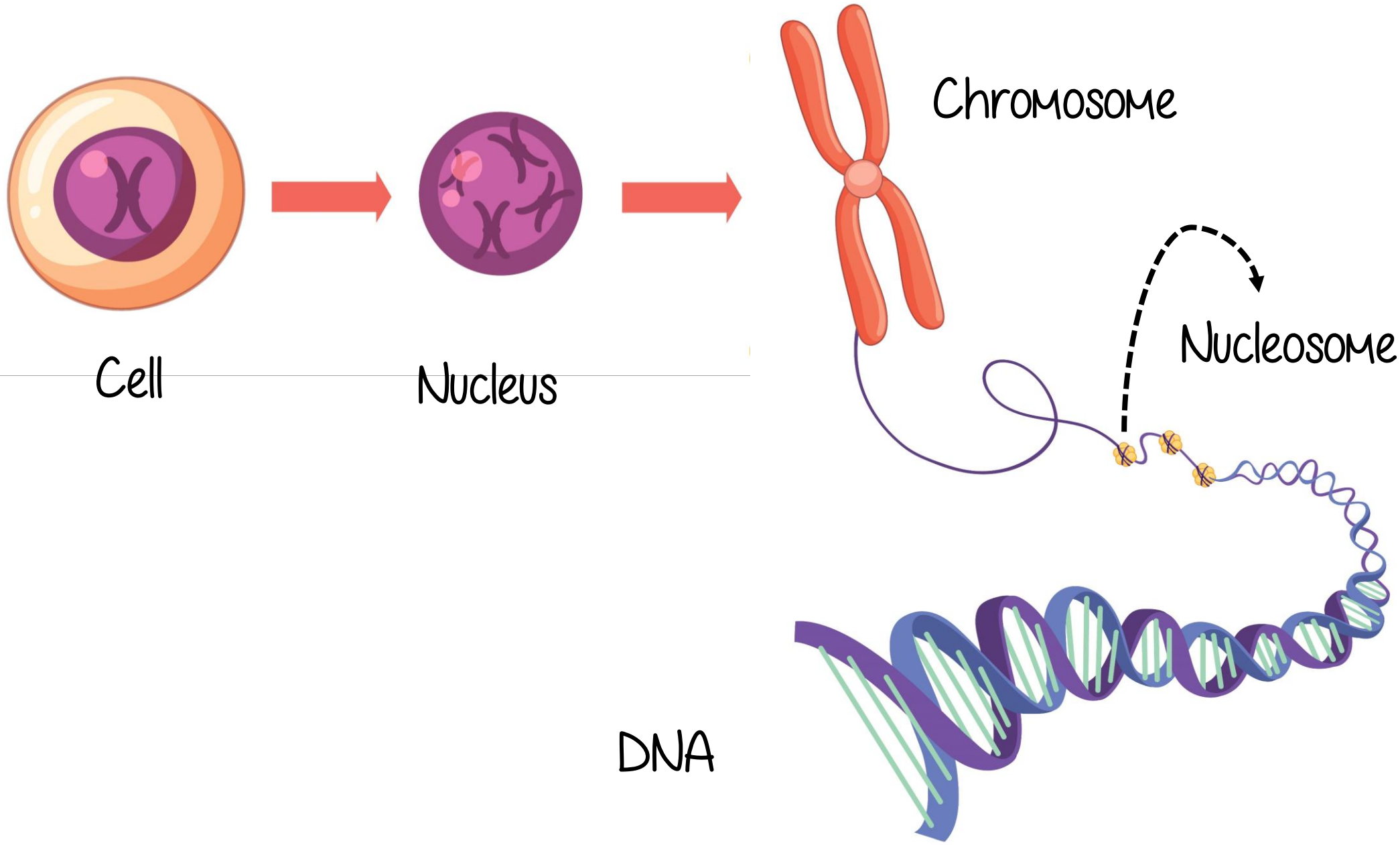


Photo 51

- 1953 James Watson along with Francis Crick presented the double helical model of DNA.
- The structure was proposed based on the X-ray diffraction studies conducted by Rosalind Franklin and Maurice Wilkins.
- The X-ray diffraction image obtained was called ‘Photo 51’.
- James Watson and Crick was Awarded the Nobel prize in Medicine 1962 for the discovery of double helical model.
- Roslind Franklin passed away at the age of 37 in 1958

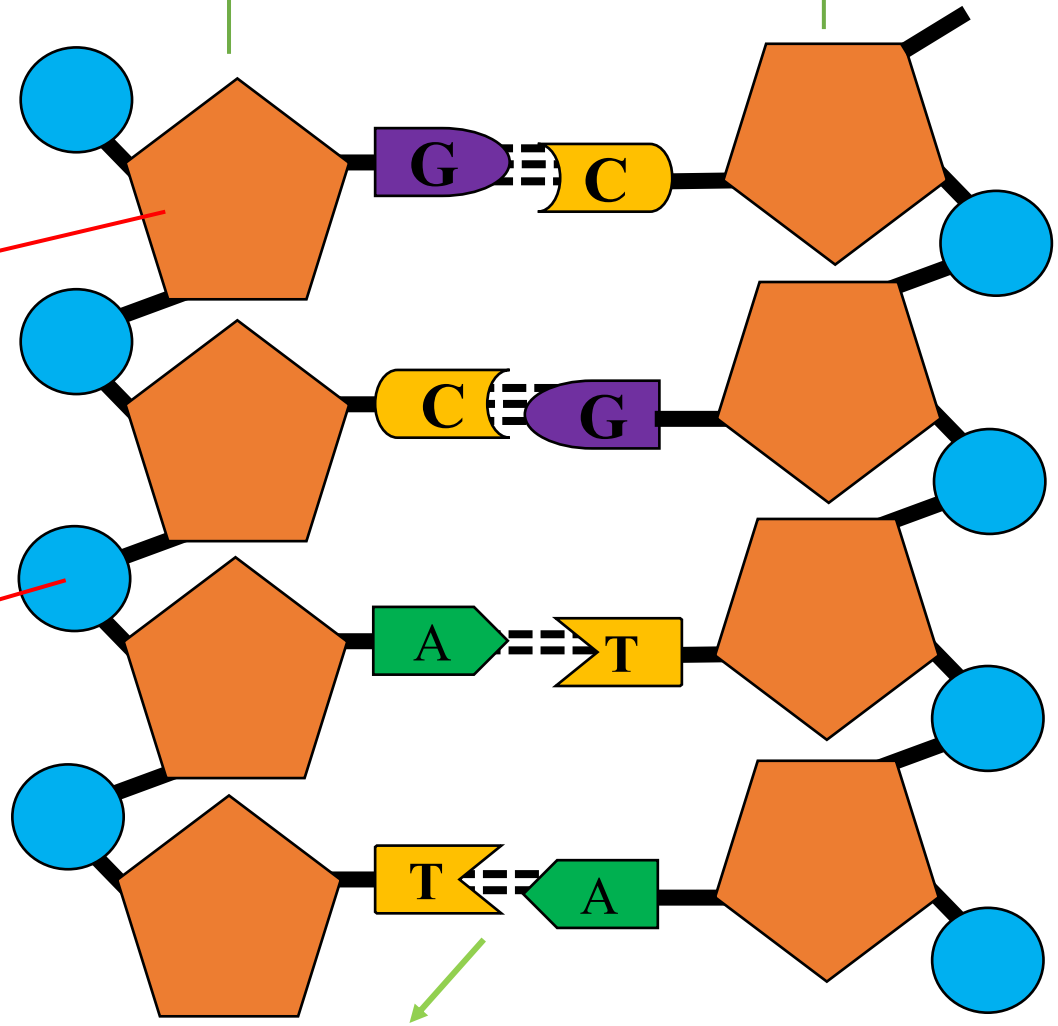


Double Helix Model of DNA

Strands: composed of sugar and phosphate

Sugar Molecule

Phosphate Molecule



Rungs: formed by the pairing of nitrogen bases

JOE FREDY (HST)

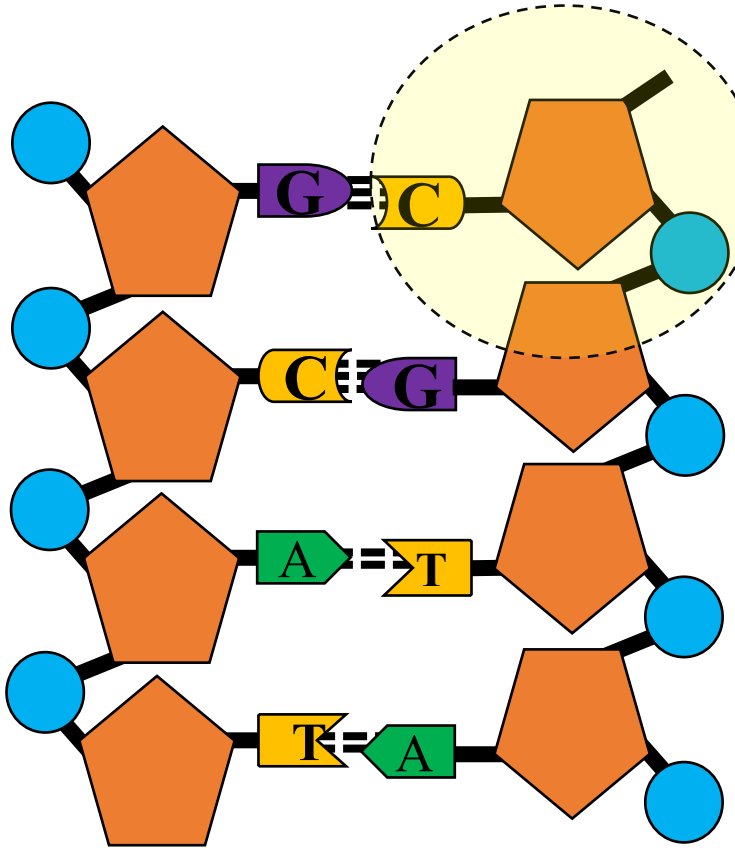
Phosphate: participates in the formation of bonds that link nucleotides together

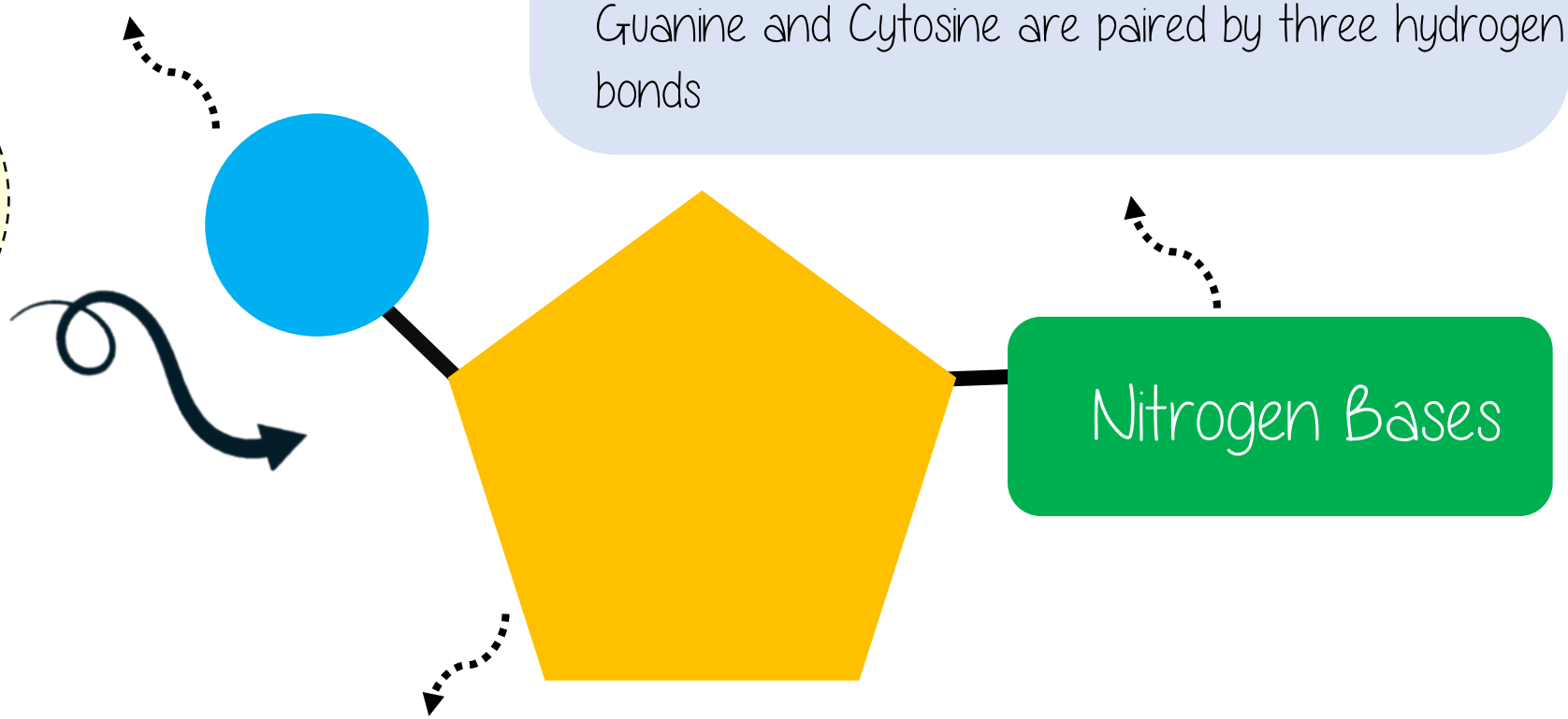
Adenine, Guanine, Thymine, Cytosine

Nitrogen containing alkaline compound. Always found in pairs

Adenine and Thymine are paired by two hydrogen bonds

Guanine and Cytosine are paired by three hydrogen bonds





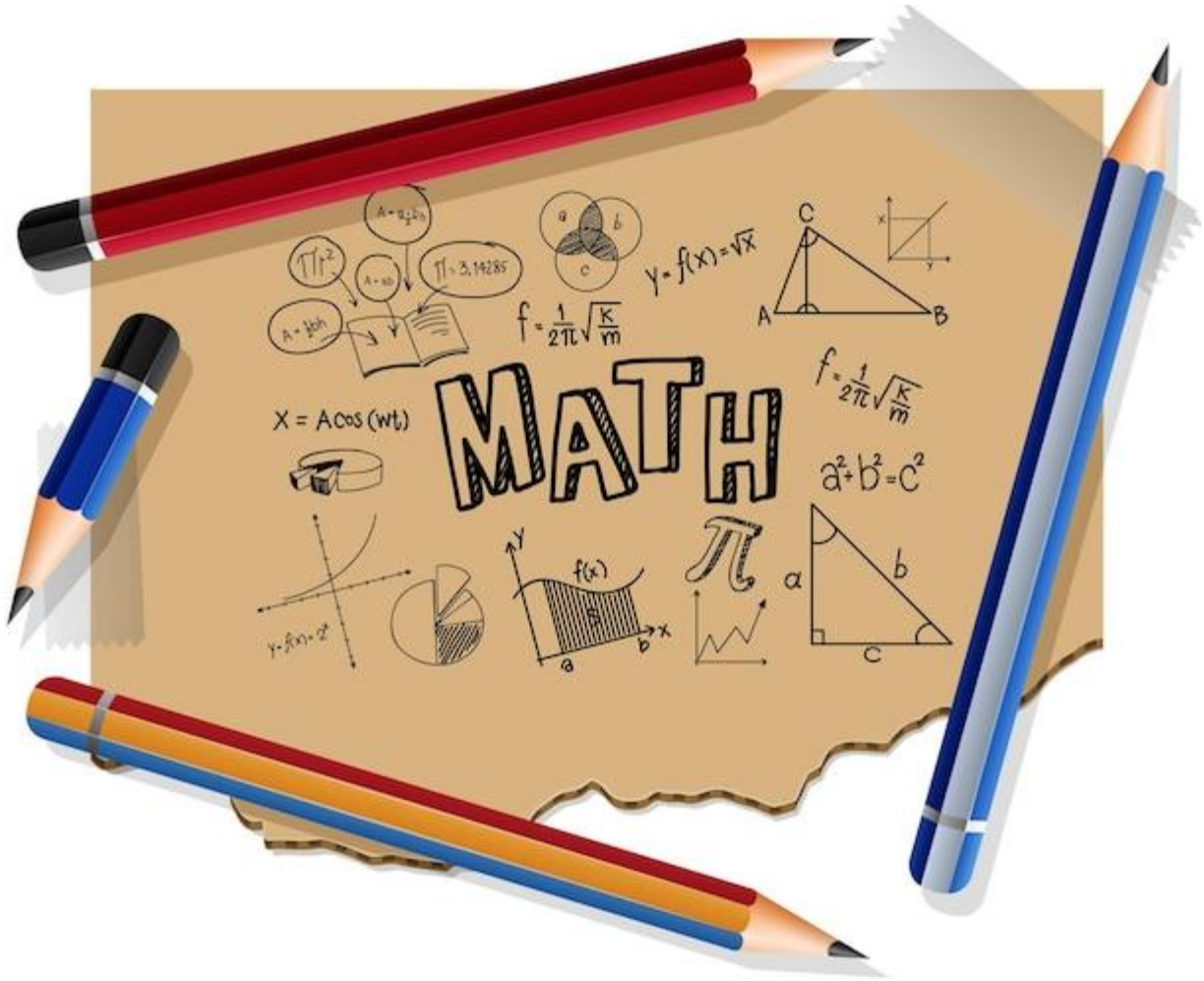
Deoxy Ribose sugar

5 carbon sugar

JOE FREDY (HST)

How does the normal sugar differ from a sugar molecule in DNA?

Normal Sugar	Ribose Sugar
• Is a six carbon compound • Metabolizes to produce energy	• pentose sugar (5 carbon compound) • Forms the backbone of DNA



Measuring Life: Biology & Numbers

JOE FREDY (HST)

Size of DNA

- DNA in each chromosome is about 2 inches (5cm).
- DNA from 46 chromosome of human cell if joined would be around 6ft (2cm).
- Human body is made up of trillions (one lakh crore) of cells.
- DNA's of all cell joins it would be 67 billion miles in length.
- Capable to wrap around the earth over two million times.

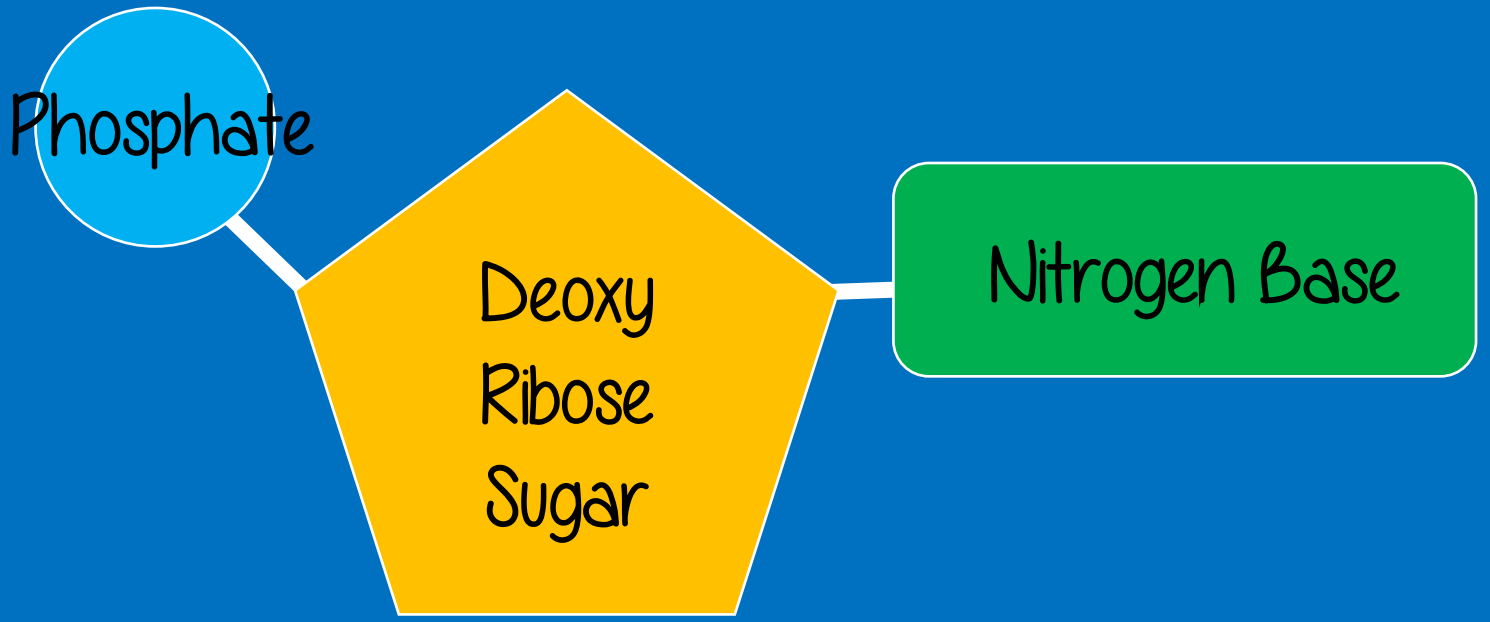
DNA

- DNA is found inside the nucleus.
- James Watson and Francis Crick proposed the double helical model.
- The ‘Photo 51’ obtained through X-ray diffraction serves as the basis for the double helical model of DNA
- The double helical model has mainly two parts.

1. Strands: Deoxy ribose sugar and phosphate
2. Rungs: Nitrogen bases joined by hydrogen bonds

- The base of DNA is nucleotide (Deoxy ribose sugar + Phosphate + Nitrogen Base)
- DNA condenses to form chromosome.
- Histones are protein that help in the condensation to form chromosome.

Nucleotide



Deoxy ribose sugar, phosphate nitrogen base together form the nucleotide

- Deoxy Ribose sugar: A five carbon sugar
- Phosphate: It helps in bonding adjacent nucleotide by forming bond with ribose sugar.

- Nitrogen Bases
  - Nitrogen containing alkaline compound
  - Adenine, Guanine, Thymine, Cytosine are the nitrogen bases.
  - Adenine and Thymine forms pairs by two hydrogen bonds
  - Guanine and Cytosine forms pairs by three hydrogen bonds

Complete the table

Number of strands in DNA	2
Molecules used to make strands	Deoxy ribose sugar and phosphate
Molecules used to make rungs	Nitrogen bases (Adenine, Guanine, Thymine, Cytosine)
Different type of nitrogen bases	Adenine, Guanine, Thymine, Cytosine
Formation of rungs	Pairs of Adenine and Thymine Pairs of Guanine and Cytosine
Mode of nitrogen base pairing	Adenine and Thymine bonded by two hydrogen bonds Guanine and Cytosine bonded by two hydrogen bonds
Molecules in a nucleotide	Deoxy ribose sugar + Nitrogen Base + Phosphate

Complete the table

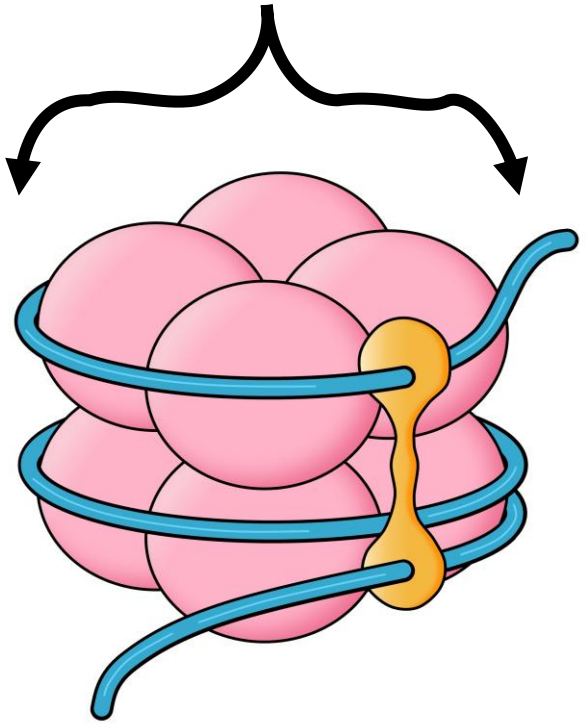
Number of strands in DNA	
Molecules used to make strands	
Molecules used to make rungs	
Different type of nitrogen bases	
Formation of rungs	
Mode of nitrogen base pairing	
Molecules in a nucleotide	

Histone Protein

- 8 histone protein forms an octamer
- DNA is wound around the histone protein
- They condense the DNA to form nucleosome / chromatid reticulum / chromosome.
- Human cell is 25 micro meter in diameter.
- Entire DNA inside the cell is about 2M
- Histone protein condenses the DNA in such a way that it fits into 25 micro meter cell
- Histone octamer and DNA forms nucleosome

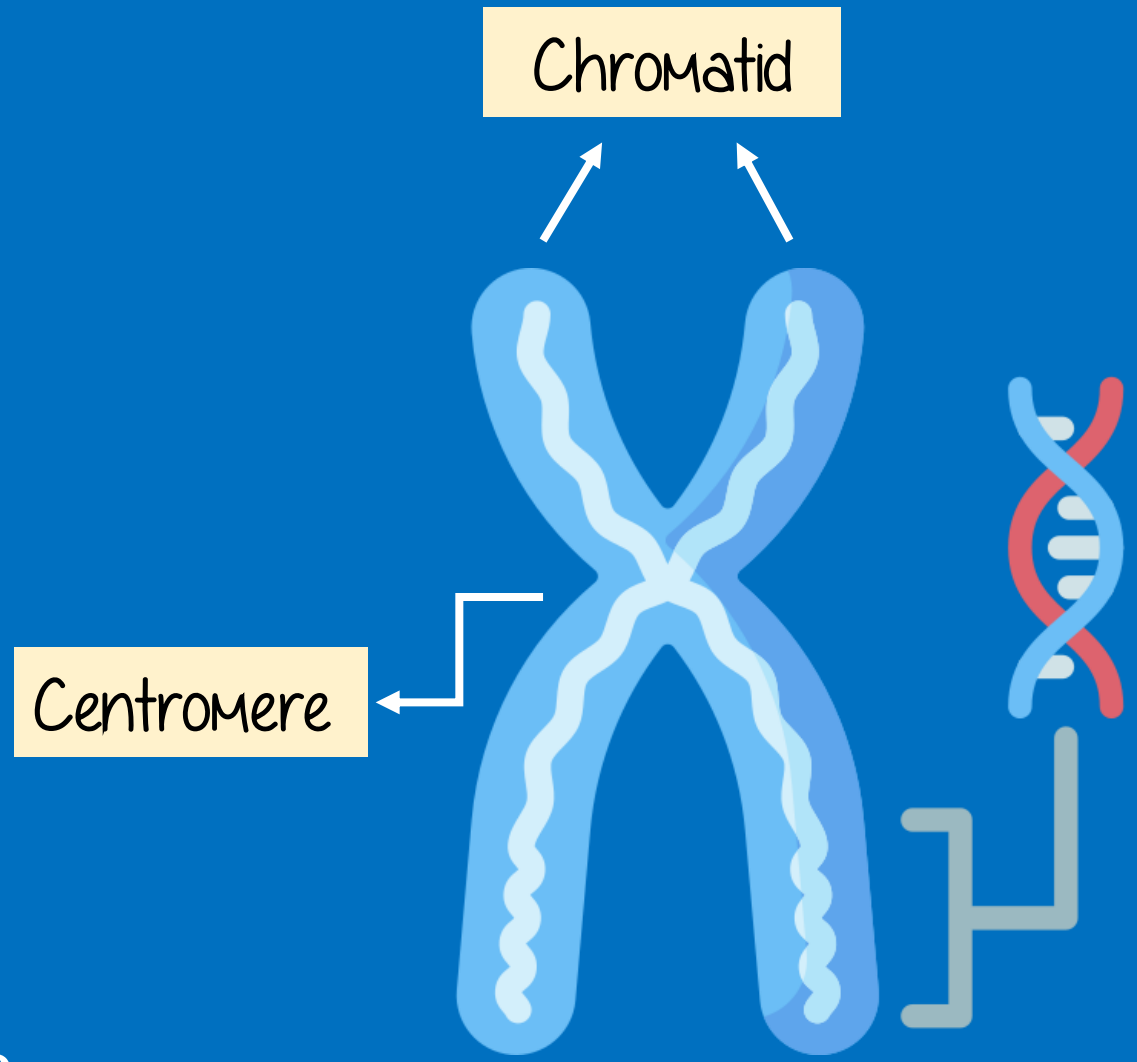
Nucleosome

(Histone protein + DNA)

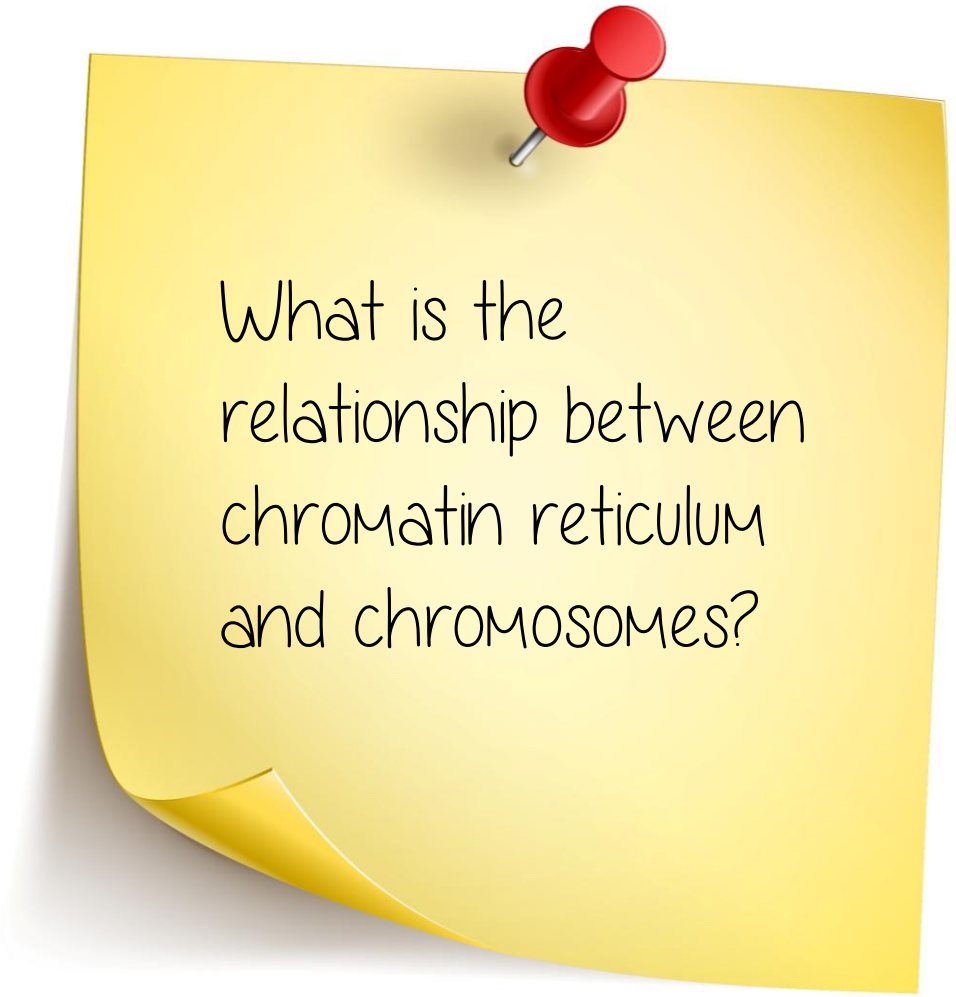


Chromosome

- DNA and Histone protein together form the basic building blocks of chromosome.
- Chromosome are formed by packing and recoiling the chains of nucleosome
- Histone helps the DNA to condense and form chromosome
- The centre of chromosome is called centromere.
- Chromatids are found attached to the centromere.

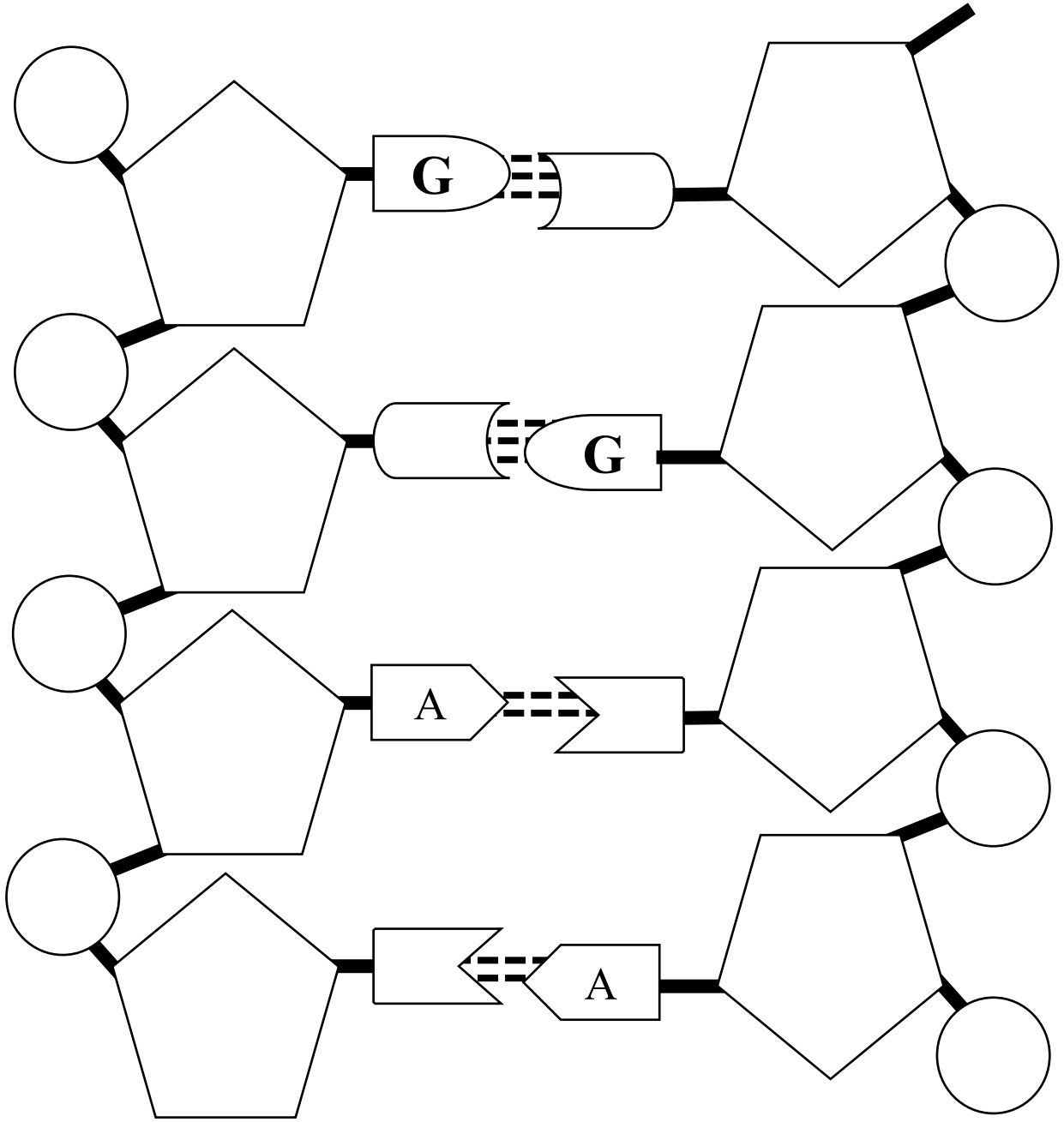


JOE FREDY (HST)



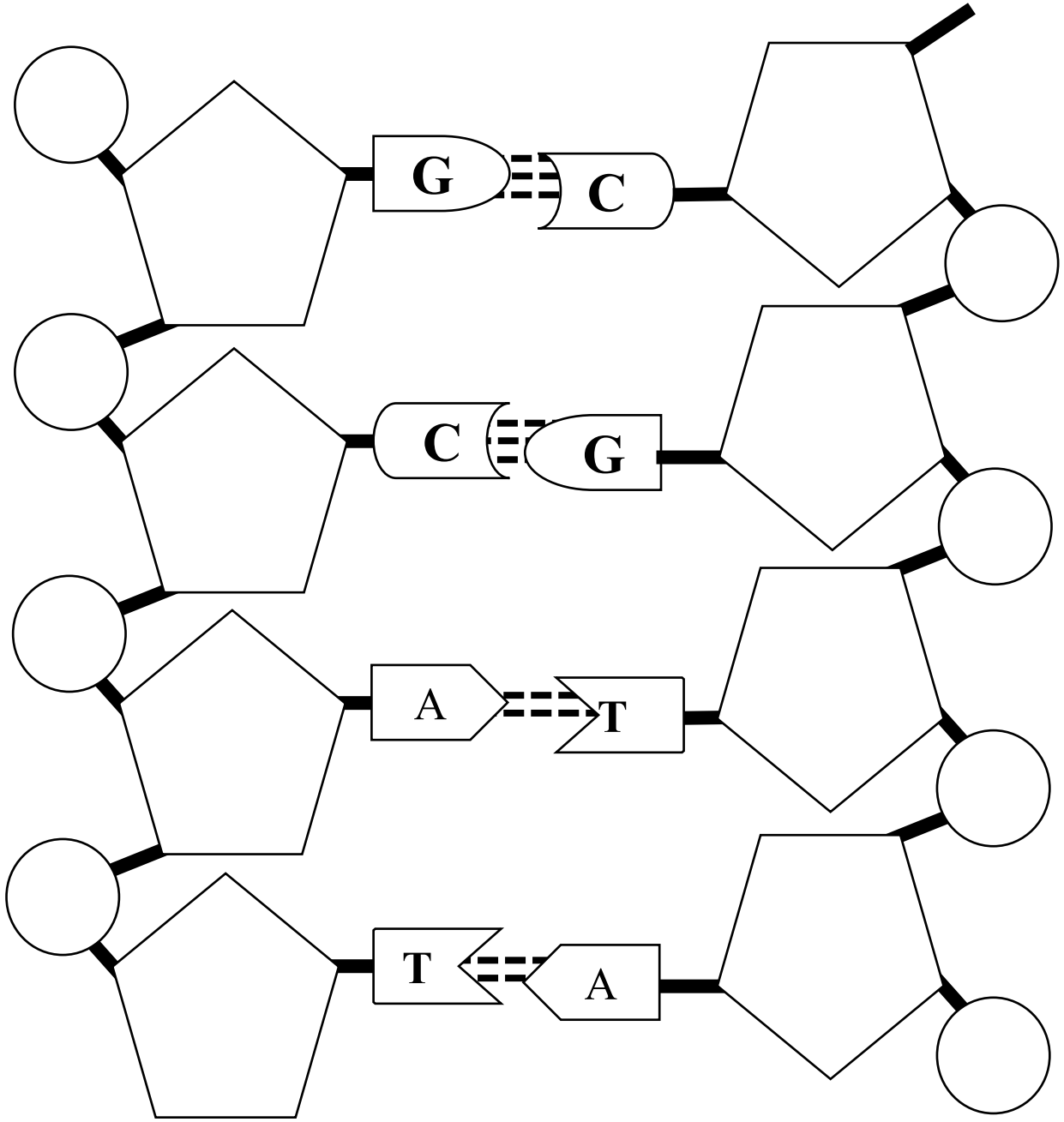
Thread like structure found before the cell enters cell division formed of DNA and histones. This chromatid reticulum undergoes further condensation during cell division to form chromosome

Complete the Figure



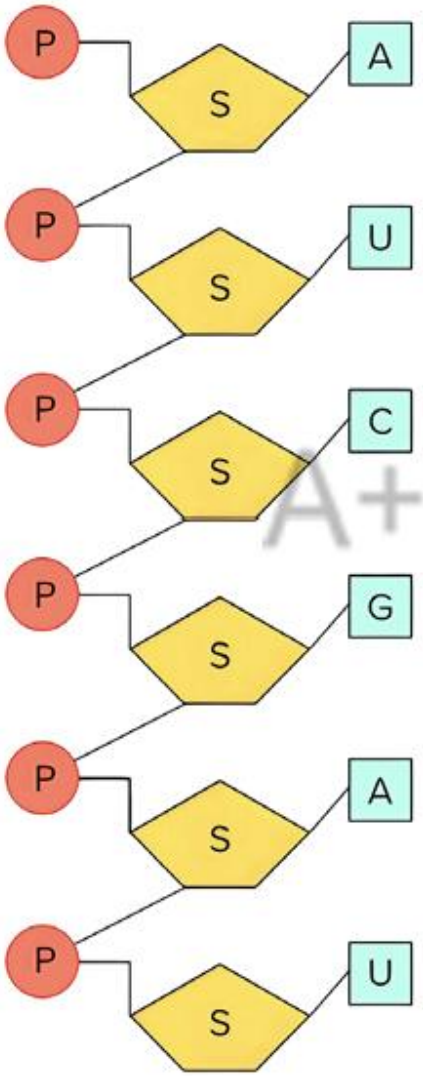
JOE FREDY (HST)

Complete the Figure



JOE FREDY (HST)

RNA



- 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.

Complete the Table

	Number of strands	Types of sugar molecules	Nitrogen bases
DNA			
RNA			

Complete the Table

	Number of strands	Types of sugar molecules	Nitrogen bases
DNA	2	Deoxy ribose	Adenine, Guanine, Thymine, Cytosine
RNA	1	Ribose sugar	Adenine, Guanine, Uracil, Cytosine

Human Chromosome

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 Ychromosome. The Ychromosome 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.

JOE FREDY (HST)

Complete the Table

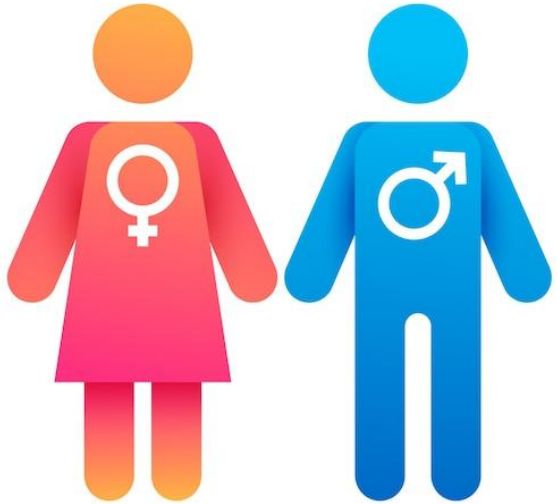
	Somatic Chromosomes	Sex Chromosomes
Number of Chromosome		
Function		
Peculiarity	• . • .	• • The SRY gene on the Y chromosome is responsible for the development of testis in the embryo.

Complete the Table

	Somatic Chromosomes	Sex Chromosomes
Number of Chromosome	22 pairs	2 pairs (X and Y)
Function	Controls physical characters	Involved in sex determination
Peculiarity	• A pair of identical chromosomes together form a homologous chromosome. • One of these is inherited from the mother and the other from the father.	• They are two types of sex chromosome . X 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.

Complete the Table

44+XX

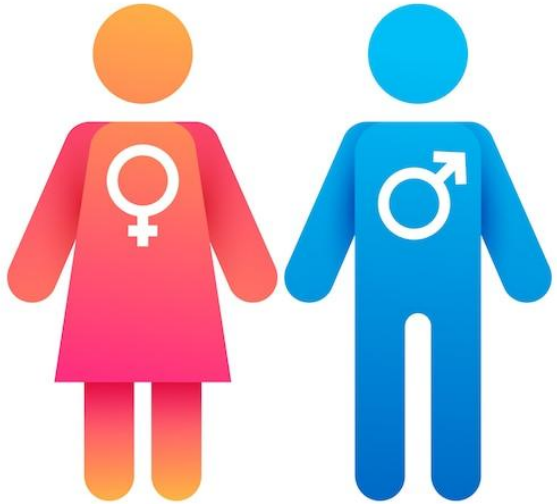


44+XY

	Genetic Constitution	Total number of chromosomes	Number of somatic chromosomes	Number and type of sex chromsome
Female	44+XX			
Male	44+XY			

Complete the Table

44+XX



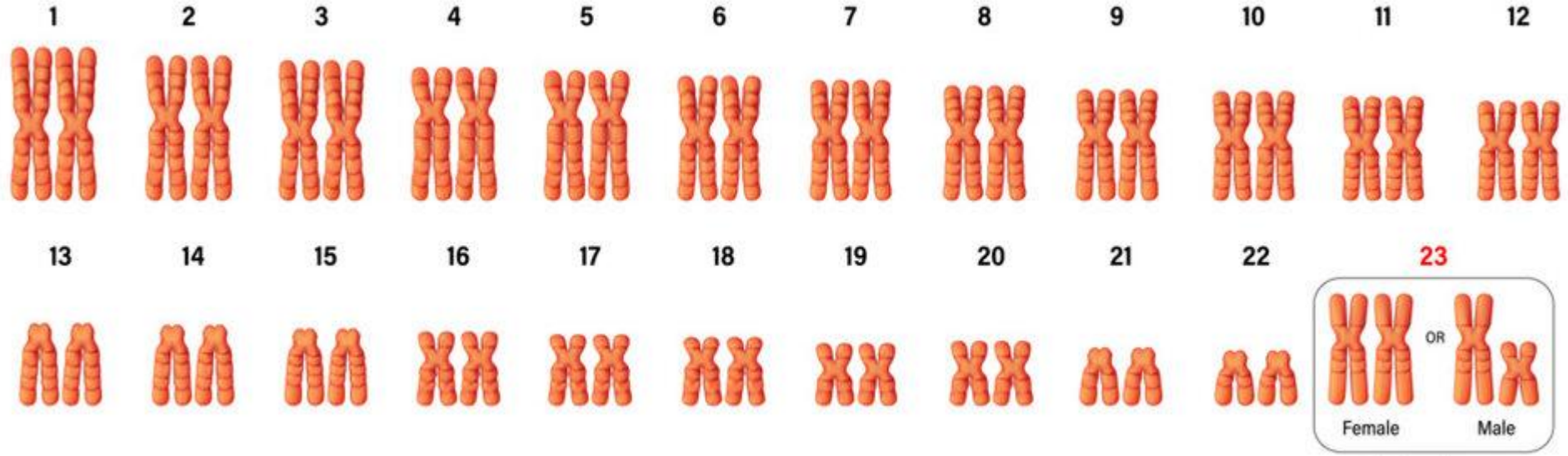
44+XY

	Genetic Constitution	Total number of chromosomes	Number of somatic chromosomes	Number and type of sex chromsome
Female	44+XX	46	44	2(X,X)
Male	44+XY	46	44	2(X,Y)

Humans beings are diploid organisms meaning they have two set of chromosome one from their father and other form their mother

Human Karyotype

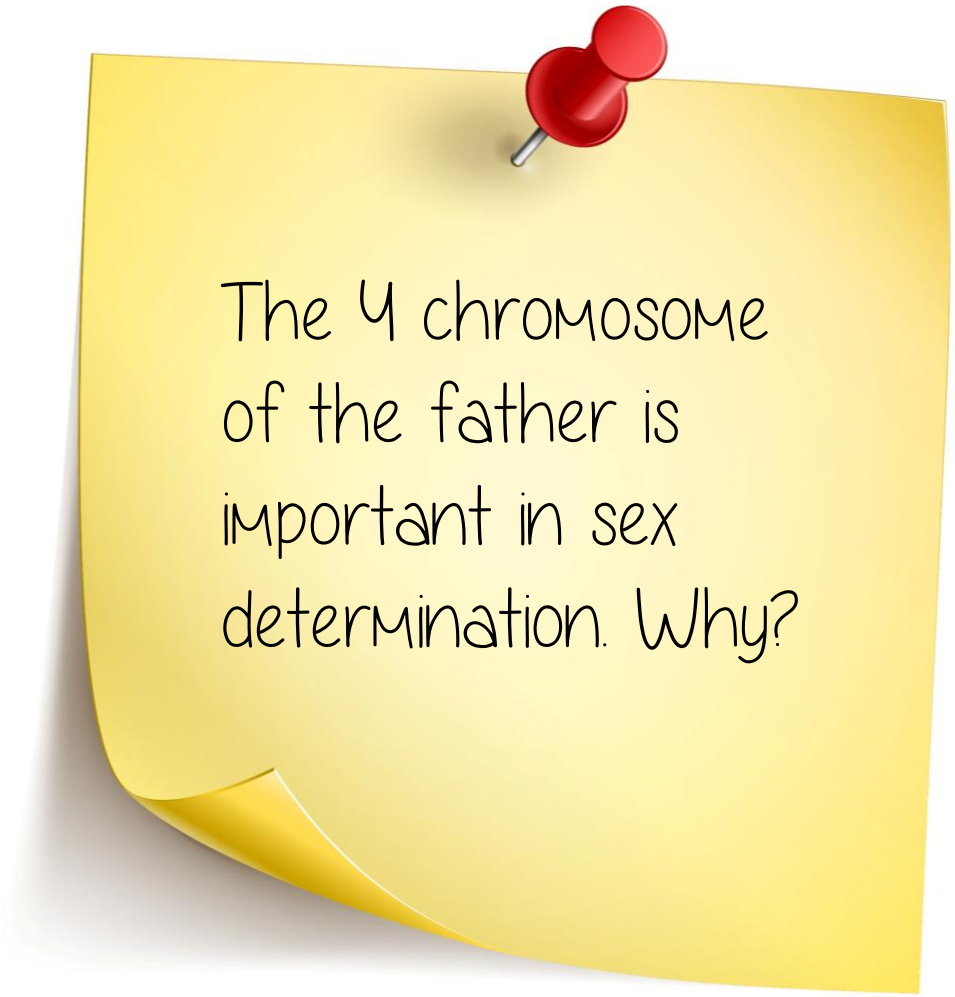
(A karyotype is the general appearance of the complete set of chromosomes in the cells of a species or in an individual organism, mainly including their sizes, numbers, and shapes)



Somatic Chromosome

Sex Chromosome

JOE FREDY (HST)



The Y chromosome are only found in male. So, he can only contribute the Y chromosome and SRY gene which determine the sex are only found in the Y chromosome..

JOE FREDY (HST)

Different Genetic Constitution

JOE FREDY (HST)

- 44+XX and 44+XY are considered as normal genetic constitution.
- There are variations in genetic constituents in humans.
- These variations influence the physical and mental development of individuals.
- Sex determination is complex and not only determined by genetic constitution, but also by other factors.

Turner Syndrome

44 + XO : Female with only one X chromosome

Triple X Syndrome

44 + XXX : Females with three X chromosomes

JOE FREDY (HST)

Klinefelter Syndrome

44 +XXY : Males with two X chromosomes

XYY Syndrome

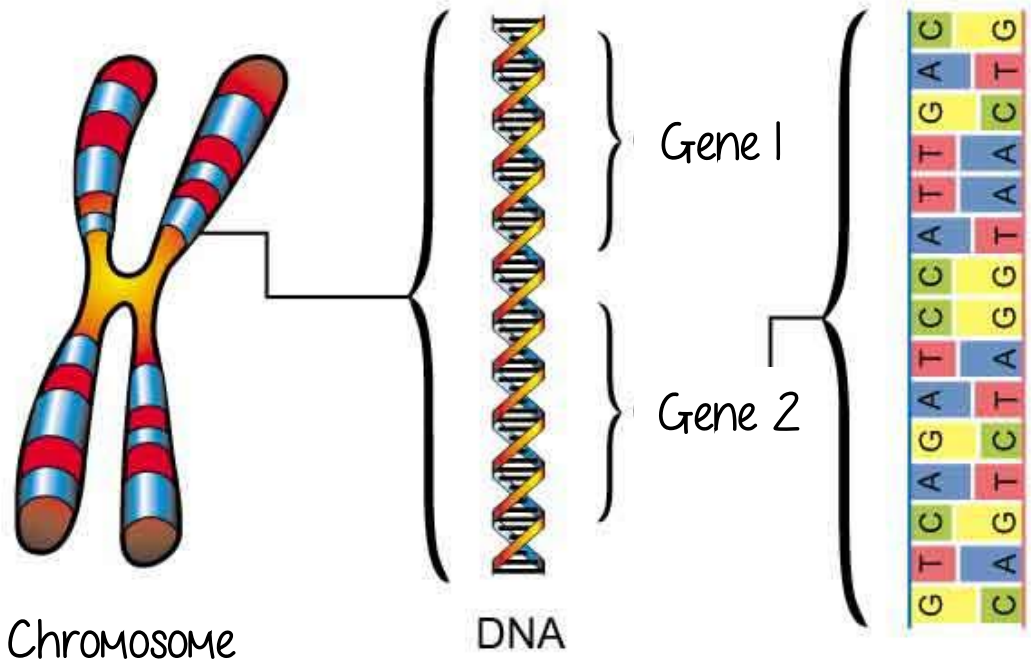
44 + XYY : males with one X chromosome and two Y chromosome

	Genetic constitution	Sex	Peculiarity
Turner Syndrome	44+XO		• • They have female sex organs but no female characters develop during adolescence.
Triple X Syndrome		Female	Females with three X chromosomes
Klinefelter Syndrome			• • Has male sex organs but may also exhibit female characteristics..
XYY Syndrome		Male	They are males and have one X chromosome and two Y chromosome.

	Genetic constitution	Sex	Peculiarity
Turner Syndrome	44+XO	Female	• Females with one X chromosome. • They have female sex organs but no female characters develop during adolescence.
Triple X Syndrome	44+XXX	Female	Females with three X chromosomes
Klinefelter Syndrome	44+XXY	Male	• Males with two X chromosome and one Y chromosome. • Has male sex organs but may also exhibit female characteristics..
XYY Syndrome	44+XYY	Male	They are males and have one X chromosome and two Y chromosome.

ജീൻ (Gene)

- Gene is specific sequence of nucleotide.
- Gene contains instructions for protein synthesis
- These proteins control metabolic activities and formation of characteristic features



JOE FREDY (HST)

Protein Synthesis

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.

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.

Protein Synthesis

- Through transcription with the help of enzymes mRNA is formed.
- mRNA moves into the cytoplasm
- Ribosome attaches to the mRNA. The ribosome contains rRNA
- tRNA brings amino acid based on the code (sequence of nucleotide).
- The ribosomal RNA combines with amino acids to make protein.

Complete the Table

Process	RNA	Function
Transcription		
Translation		

Complete the Table

Process	RNA	Function
Transcription	mRNA	Contains message for protein synthesis
Translation	tRNA	Carries specific amino acid based on the message in mRNA
	rRNA	amino acids to make protein.

Similarities and Difference in Characters

- 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.

JOE FREDY (HST)

GENETICS IN THE GARDEN

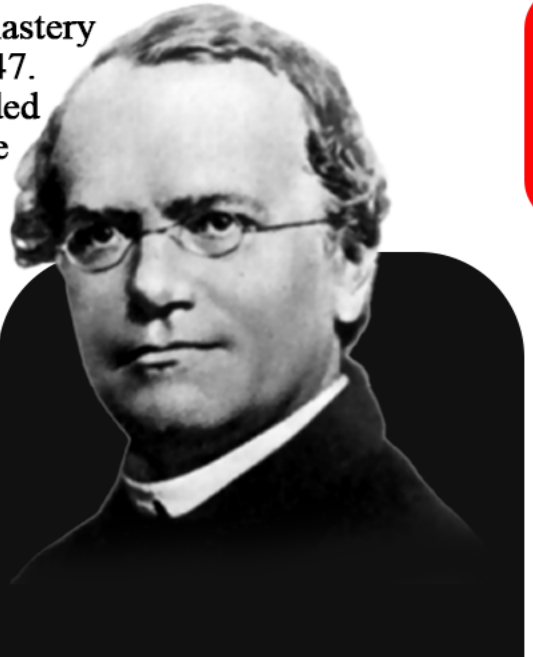
JOE FREDY (HST)

St. Johns Times

FATHER OF GENETICS

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.

Afterjoining 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.



1822 – 1884

Born on July, 1822 at Hyncice a small village of Northern Moravia which is now known as Czech Republic

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.

Mount Mendel in New Zealand's Paparoa Range was named after him in 1970 by the Department of Scientific and Industrial Research. In celebration of his 200th birthday, Mendel's body was exhumed and his DNA sequenced.

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.

Hugo De Vries

Carl Correns

Erich Von Teschermak

Genetics

- 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.

Mendel's Life

- He did most of his work in pea plant.
- 1865 he presented his findings in the Natural History Society at Brno.
- The following year, he published a thesis titled 'Experiments on Plant Hybridization.'
- 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 recognized the significance of Mendel's research.
- Mendel was born on 20 July 1822, Hyncice a small village in Czech Republic.
- He joined Augustinian monastery at Brno and became a priest in 1847.

Education

1851-1853 attended the University of Vienna and studies:-

- Physics
- Mathematics
- Natural Science
- Learned statical methods to analyse data scientifically

Mendel's Work

- 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.

Contributions

- The factors that Mendel found are now known to be genes.
- His findings contributed to the field of genetics
- Gregor Mendel’s conclusions are known as the Laws of Inheritance that is the fundamental genetic framework to understand heredity and variation.
- Published a thesis titled 'Experiments on Plant Hybridisation.'

MONOHYBRID CROSS

JOE FREDY (HST)

Mendel's Experiments

- Mendel initially conducted hybridization experiment by considering a pair of contrasting traits.
- This is known as monohybrid cross
- He considered the trait height.

He hypothesized 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.

JOE FREDY (HST)

JOE FREDY (HST)

Parents

X

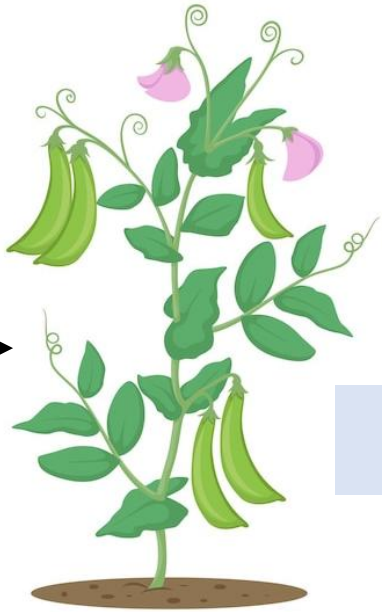
F_1 (Generation)

First Generation

Tall plant

F_2 (Generation)

Second Generation

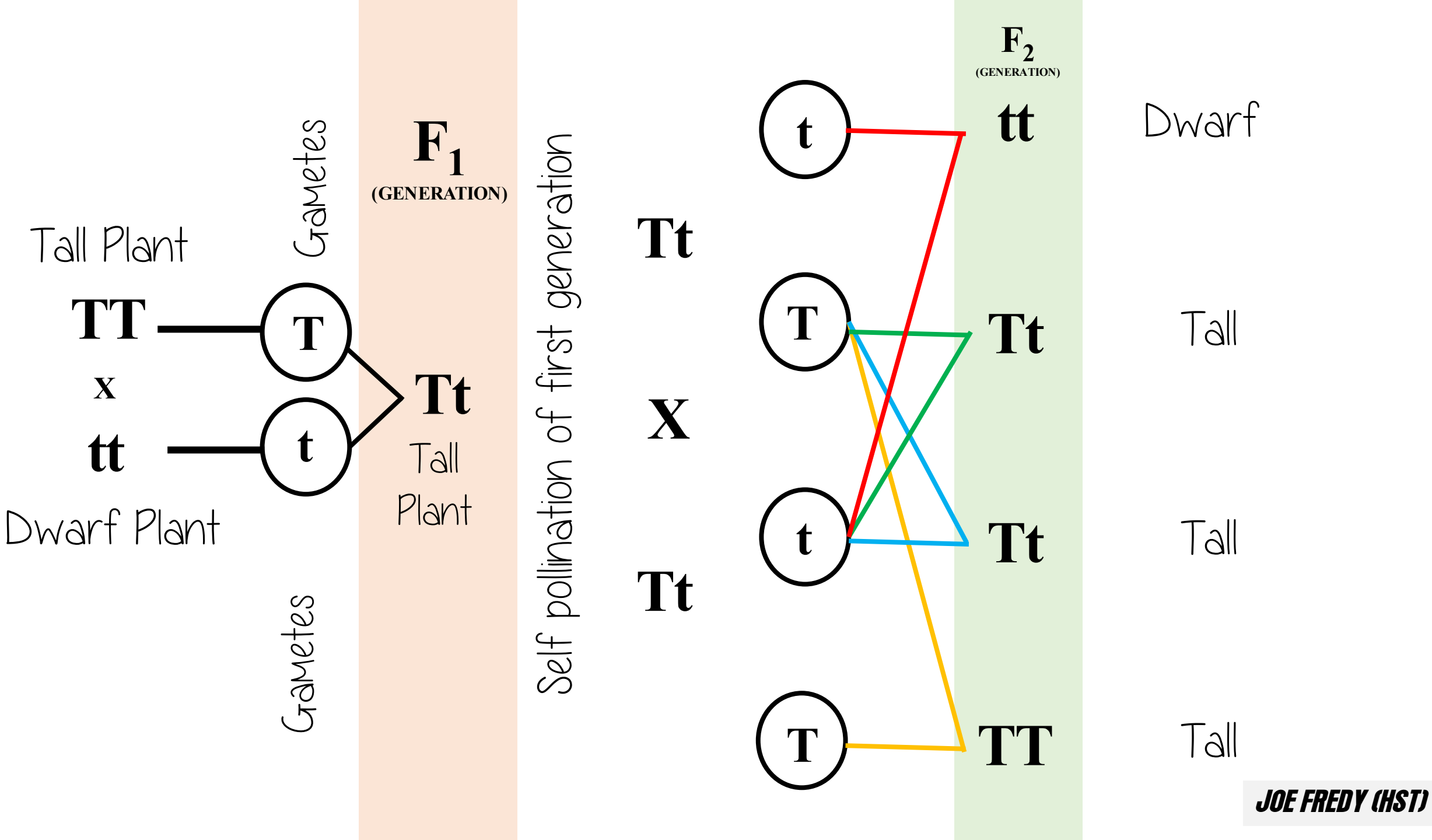


787 tall plants

1064 Plants

277 dwarf plants

The ratio between traits is approximately 3:1 (phenotypic ratio)


Tall Plant
TT
x
tt
Dwarf Plant
Gametes
T
t
F1
(GENERATION)
Tt
Tall
Plant
Gametes
Self pollination of first generation
Tt
X
Tt
F2
(GENERATION)
t
T
t
T
tt
Tt
Tt
TT
Dwarf
Tall
Tall
Tall
JOE FREDY (HST)

Tall Plant

TT

x

tt

Dwarf Plant

Gametes

T

t

Gametes

F_1
(GENERATION)

Tt

Tall Plant

Self pollination of first generation

Tt

X

Tt

F_2
(GENERATION)

tt

Tt

Tt

TT

. . . . . .

. . . . . .

. . . . . .

. . . . . .

Factors

- Mendel hypothesised that characters are passed to the offsprings through certain factors present in the chromosome.
- Later after Mendel's period these factors were discovered to be genes located in the DNA in the nucleus.
- There are different forms of gene called Alleles
- A gene has usually two alleles.
- Observable characters are called Phenotype. Eg:- Tall, Round, dwarf, Wrinkled … etc
- Genetic constitution responsible for characters are called Genotype. Eg:-TT, Tt, tt.. etc

JOE FREDY (HST)

Mendel's Postulates

- A trait is controlled by two factors.
- When pair of contrasting character are subjected to hybridization, only one is expressed in first generation while other is hidden.
- The trait that appears is first generation is called Dominant Trait.
- The trait that is hidden in first generation is called Recessive Trait.

- During gamete formation the factors that determine traits gets separated without mixing
- The phenotypic ration of dominant to recessive trait offspring of second generation is 3:1

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

Because the dominant trait tall hides the dwarf trait. So, only the tall character can be seen in the first generation or in combination of tall and dwarf alleles

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

It is because in Tt allele combination tall character is expressed and it is only in the second generation the tt combination appears. So, in this condition there is no influence of the dominant allele expressing the character in the first generation (i.e dwarf)

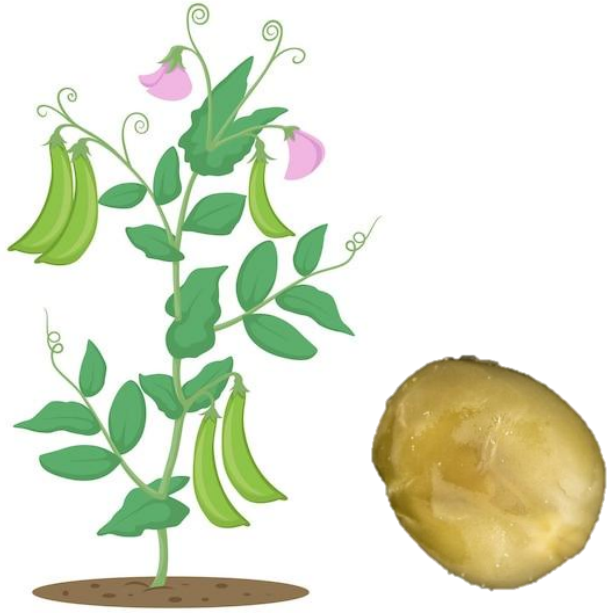
JOE FREDY (HST)

DIHYBRID CROSS

Gregor Mendel observed the inheritance of two pairs of contrasting traits of the same plant. This is known as dihybrid cross.

JOE FREDY (HST)

Tall and Round seed



TTRR → TR

Tall, round seed

ttrr → tr

Tall and Wrinkled seed

Self pollination of first generation

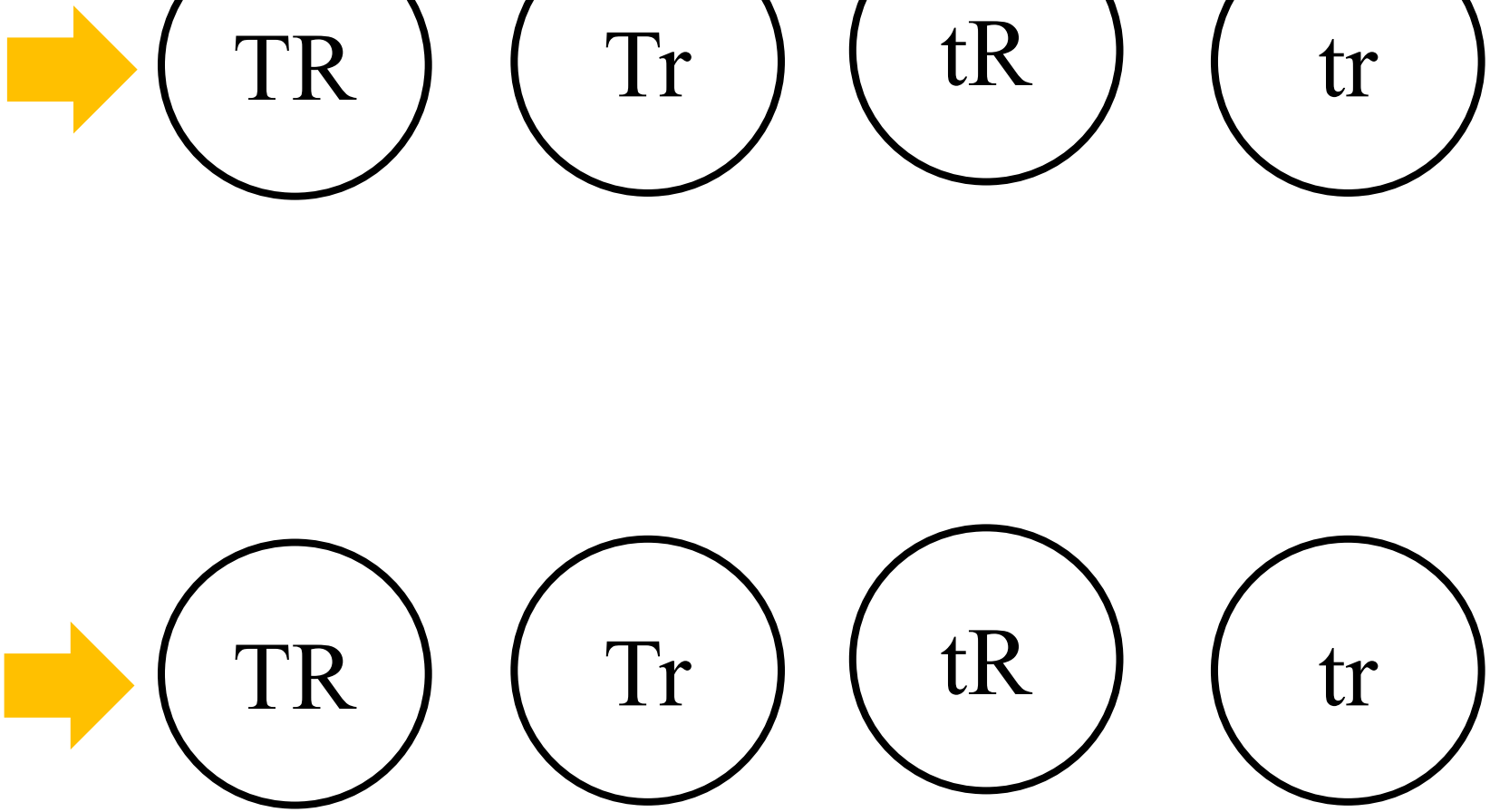
Tall, Round seed

TtRr

X

TtRr

Tall, Round seed



JOE FREDY (HST)

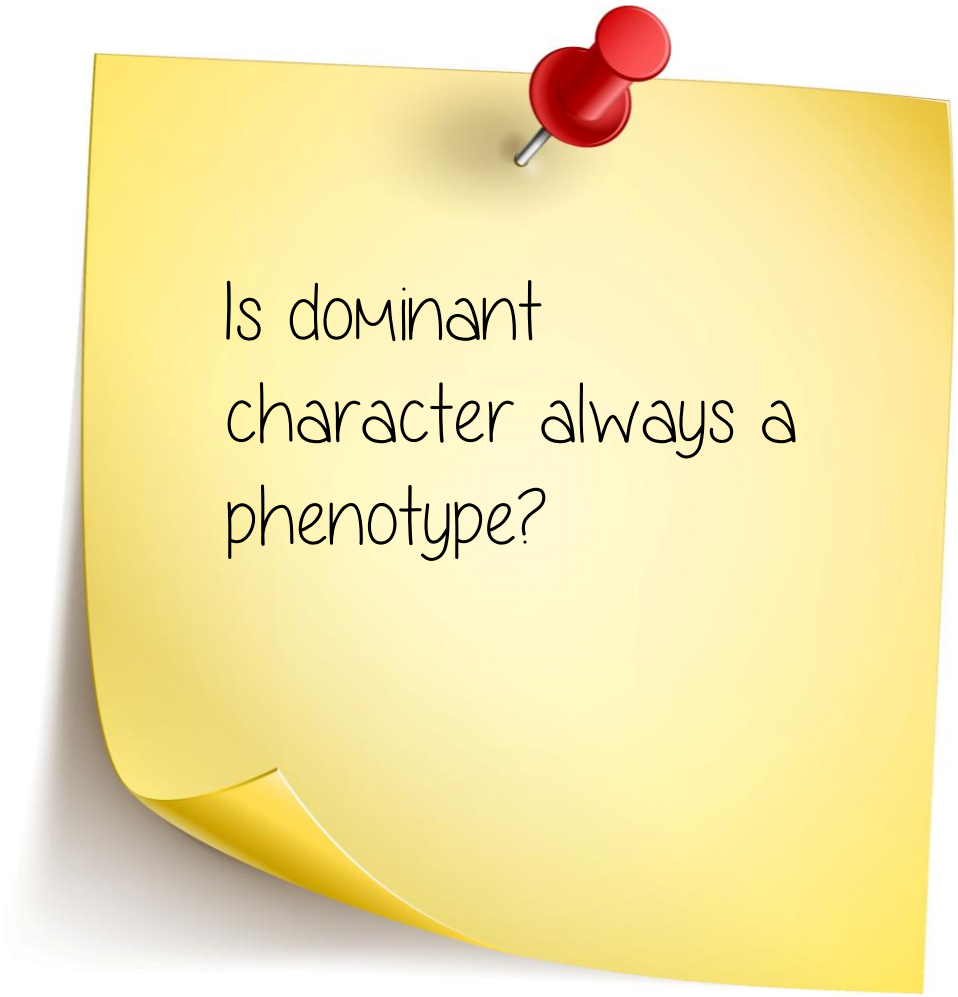
	TR	Tr	tR	tr
TR	**TTRR** Tall, Round			
Tr				
tR				
tr				

JOE FREDY (HST)

Gametes	TR	Tr	tR	tr
TR	**TTRR** Tall, Round	**TTRr** Tall, Round	**TtRR** Tall, Round	**TtRr** Tall, Round
Tr	**TTRr** Tall, Round	**TTrr** Tall, Wrinkled	**TtRr** Tall, Round	**Ttrr** Tall, Wrinkled
tR	**TtRR** Tall, Round	**TtRr** Tall, Round	**ttRR** Dwarf, Round	**ttRr** Dwarf, Round
tr	**TtRr** Tall, Round	**Ttrr** Tall, Wrinkled	**ttRr** Dwarf, Round	**ttrr** Dwarf, Wrinkled

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)



When dominant and recessive allele are together only dominant character is its phenotype. While only recessive alleles are present the recessive character is expressed.

Non-Mendelian Inheritance

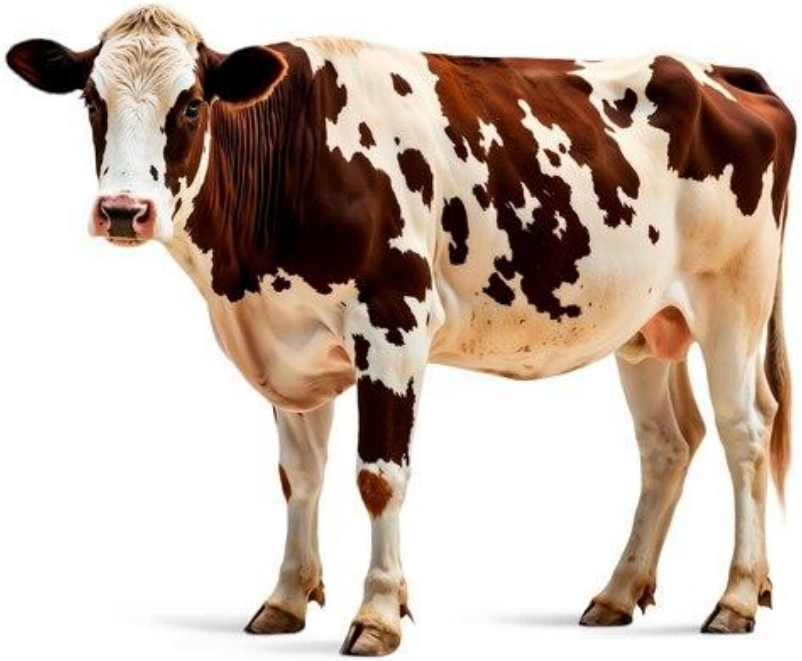
Mendel's laws were the foundation of genetics. However, it could not fully explain the diversity of traits observed in organisms. Later studies about the complex interaction among genes, environment and other factors revealed some of the limitations of Mendel's laws. This gave rise to the concept of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

Incomplete dominance

- 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.

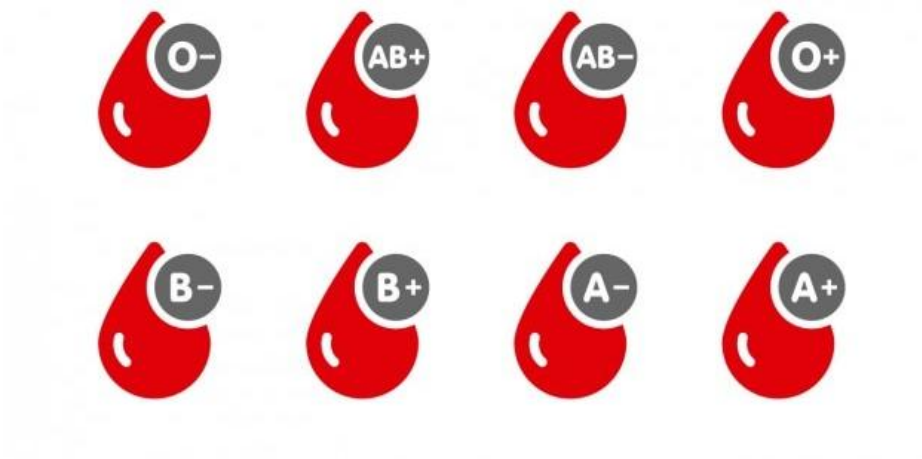
JOE FREDY (HST)

Co-dominance



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

Multiple Allelism



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

Polygenic inheritance

- 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.

JOE FREDY (HST)

Non-Medelian Inheritence	Reason	Peculiarity
Incomplete dominance		
Co-dominance		
Multiple Allelism	The gene that determines blood group in human beings has more than two alleles. Three alleles IA, IB and I determine the blood group.	
Polygenetic inheritance	More than one gene controls the colour of the skin. Theaction of these genes cause variation in the production of melanin that causes difference in skin colour.	

Non-Medelian Inheritence	Reason	Peculiarity
Incomplete dominance	A dominant allele cannot fully hide the allele of the recessive trait.	A red flowered four o'clock plant is hybridised with a white flowered plant, the resulting offspring will have pink flowers.
Co-dominance	Both alleles exhibit their traits at the same time.	Roan coat pattern, found on some cattle and horses
Multiple Allelism	The gene that determines blood group in human beings has more than two alleles. Three alleles IA, IB and I determine the blood group.	ABO blood grouping
Polygenetic inheritance	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.	Difference in skin colour

Behind the Colour Differences

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 differentgeographical regions, genetic variations suitable for the skin colour ofeach 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.

JOE FREDY (HST)

The pigment that gives colour to skin

.....

Factors that influence colour

- .....
- .....
- .....
- .....

Pigment that gives colour to skin

Melanin

Factors that influence skin colour

- The geographical region from where an individual's ancestors emerged
- Sunlight
- Diet
- Vitamin D

Genetic Processes Responsible for Variation

Crossing Over

Meiosis is the type of cell division that is responsible for the formation of gametes.

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

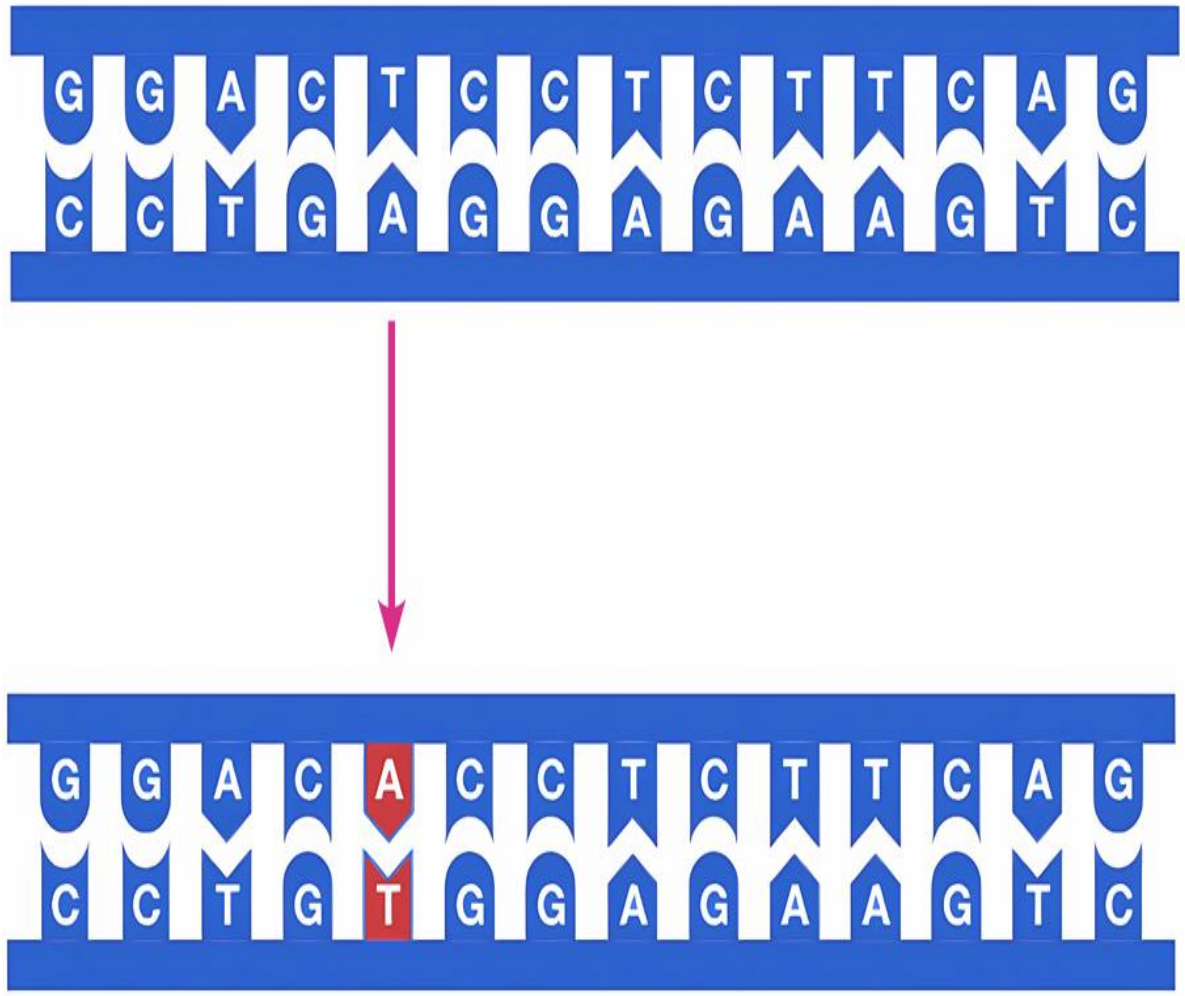
Chiasma

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.

- Crossing over takes place during a particular stage in meiosis
- Homologous chromosomes forms pairs
- The chromatids of form X shape called chiasma
- The parts that overlap are broken.
- The exchange of broken regions take place.
- The exchange causes recombination of alleles.
- This leads to the appearance of new traits in the offspring.

Mutation



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

JOE FREDY (HST)

What is mutataion?

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

Causes of mutation.

- Errors during DNA replication.
- Chemicals
- Radiation

Importance of mutation.

The mutated genes that are transferred through generation leads to variation in characters. Mutation plays a crucial role in process of evolution.

What is mutataion?

.....

.....

Causes of mutation.

- .....
- .....
- .....

Importance of mutation.

.....

.....


Genetics
Career
Opportunities
JOE FREDY (HST)

Branches of Genetics

- Molecular genetics
- Population genetics
- Medical genetics
- Cytogenetics
- Behavioural genetics
- Genomics

1. **Graduate programmes**: biotechnology, microbiology, bioinformatics etc. open doors to a wide range of career opportunities.
2. **Postgraduate level**: genetic counselling, genomics, medical genetics, and forensic science provide opportunities in healthcare, research, and education.
3. **Advanced degrees**: 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.

JOE FREDY (HST)