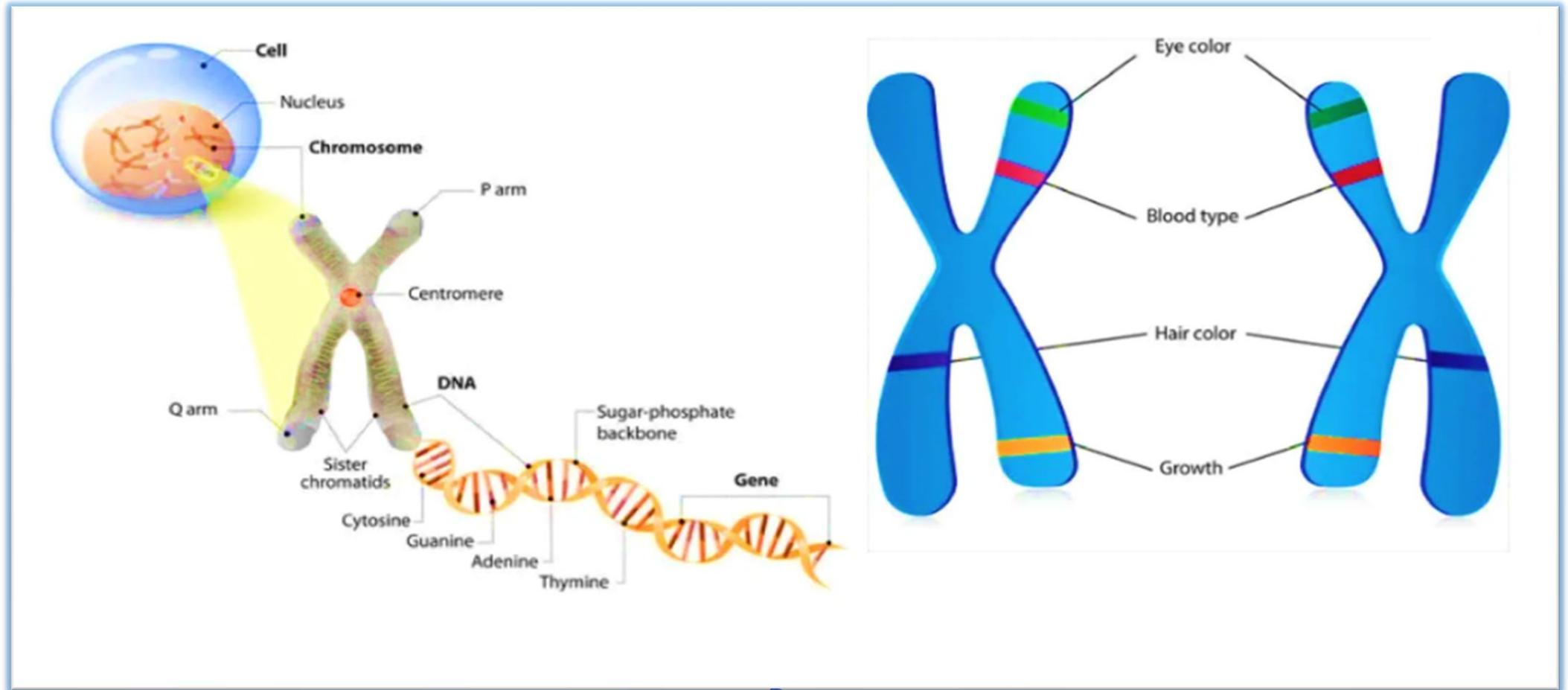


# Basics of Genetics



By

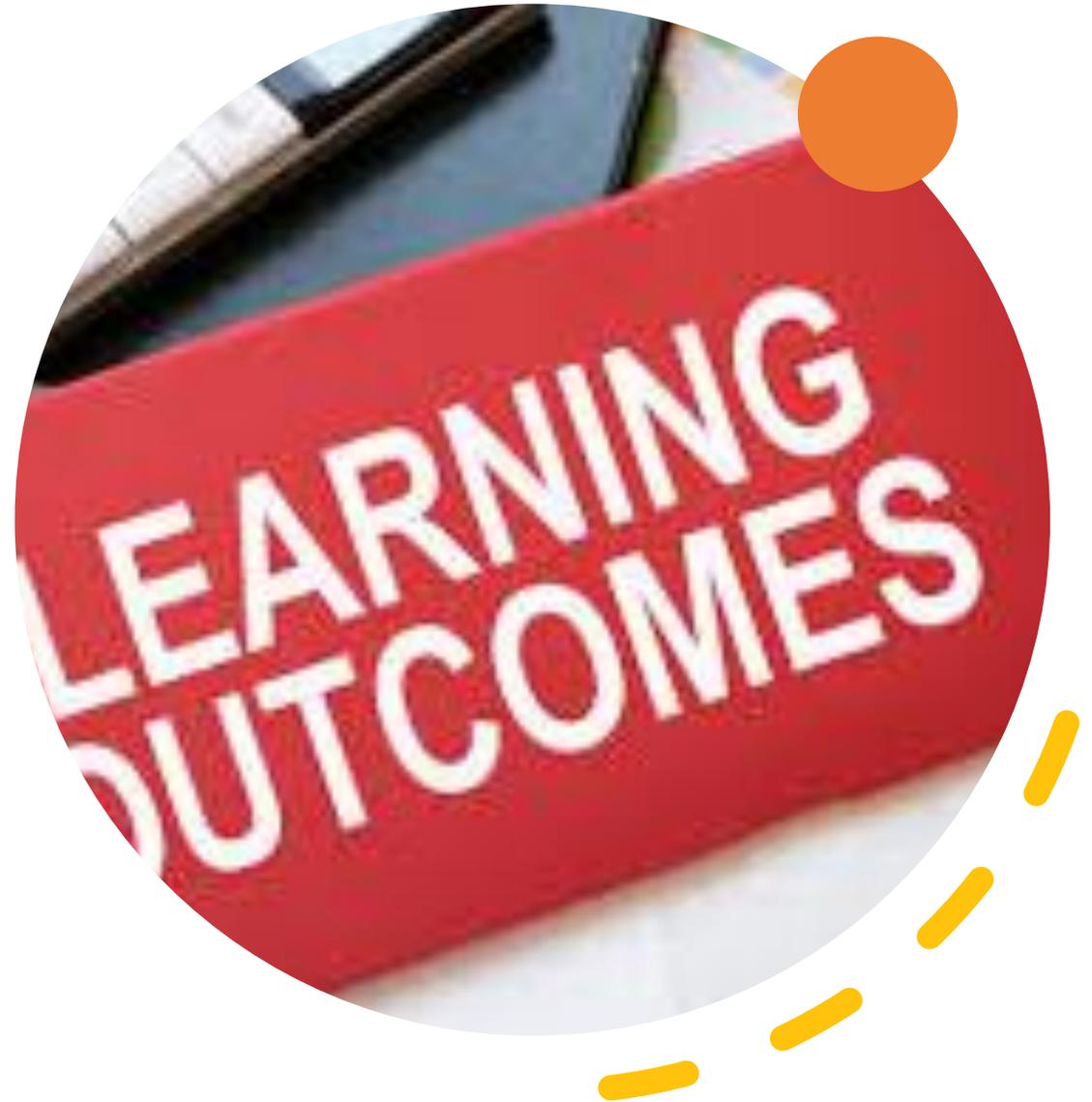
**Dr: Aya Ahmed El-Hanafy**

Associate Professor of Biochemistry & Molecular Biology

# Learning outcomes

*By the end of lecture, students should be able to:*

- Understand the concept of genetics in brief.
- Define chromosomes, DNA, genome and genes.
- Identify different Terminology related to Genetics.

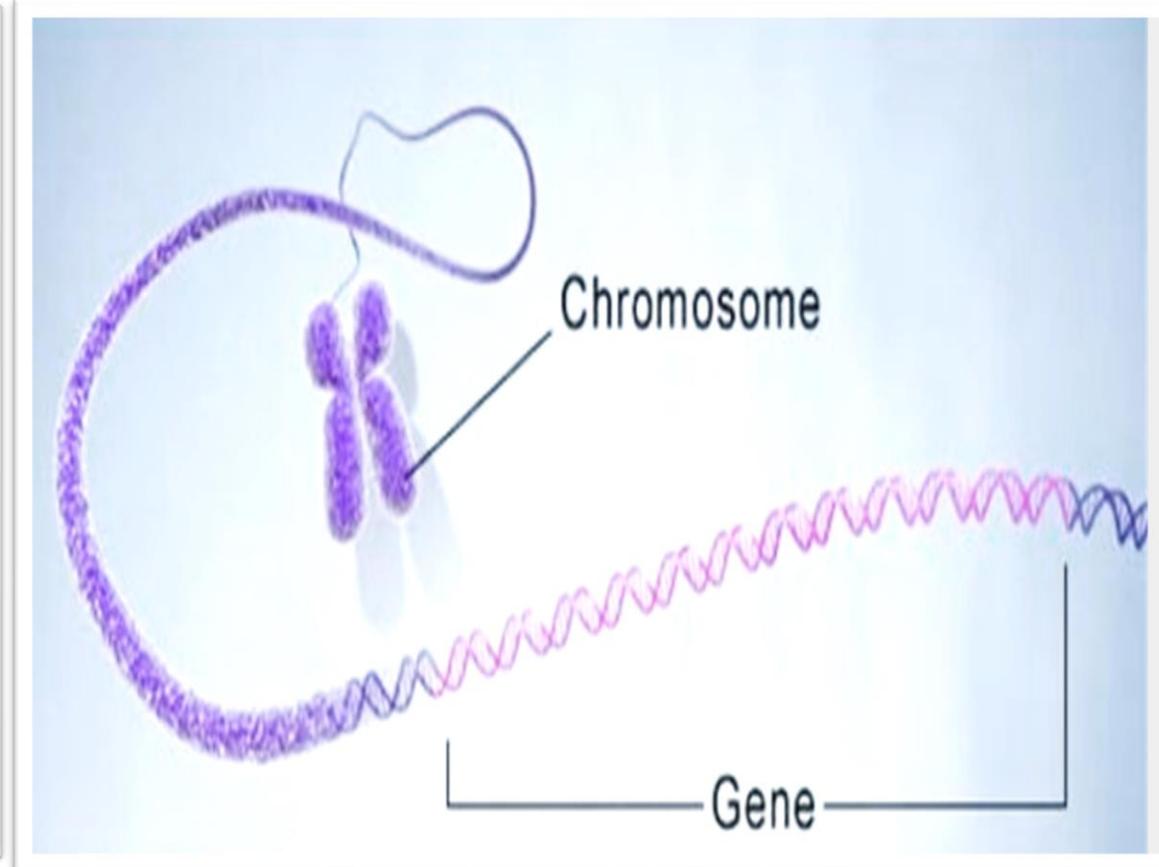
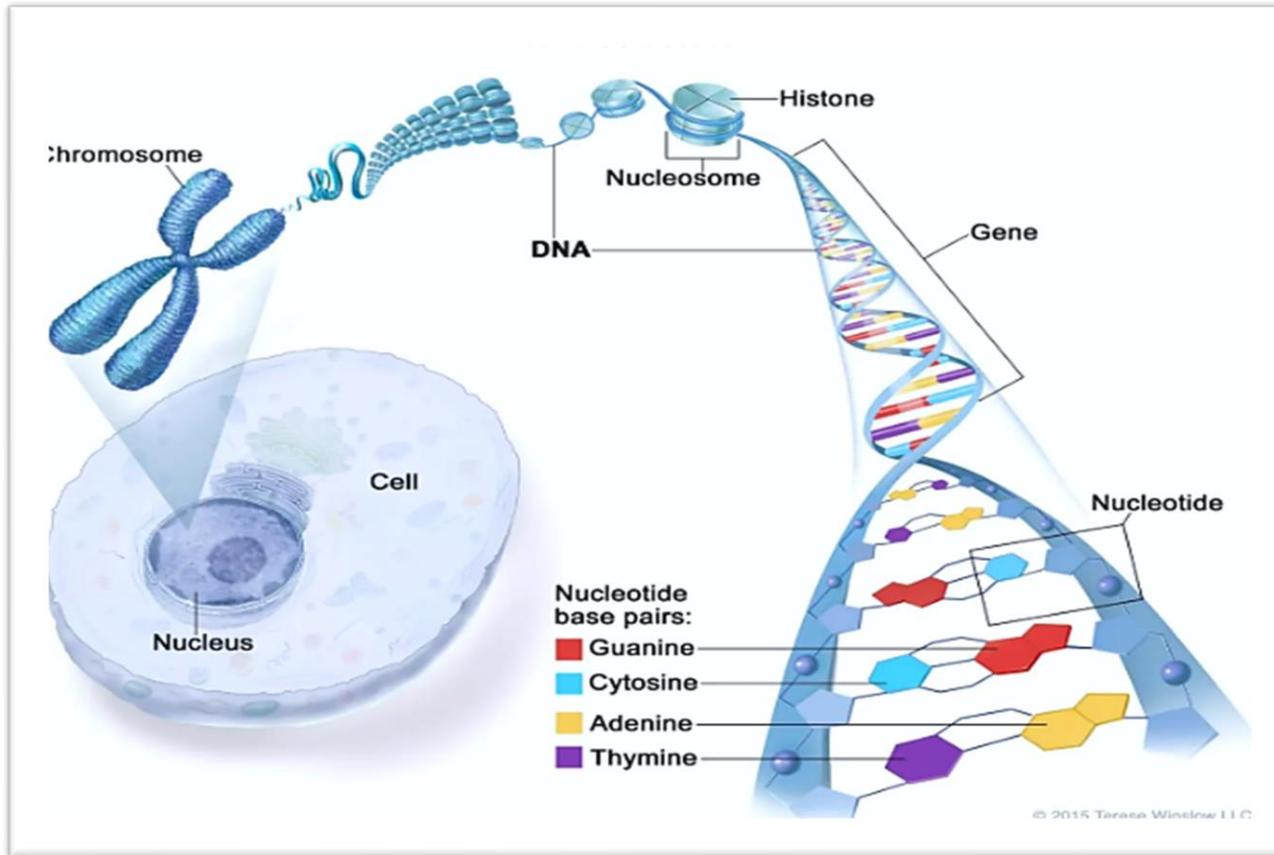


# Introduction : WHAT IS GENETICS?

- Genetics refers to the study of genes, heredity & genetic variation in the living organism.
- It is the field of science that examines how traits are passed from one generation to the next.

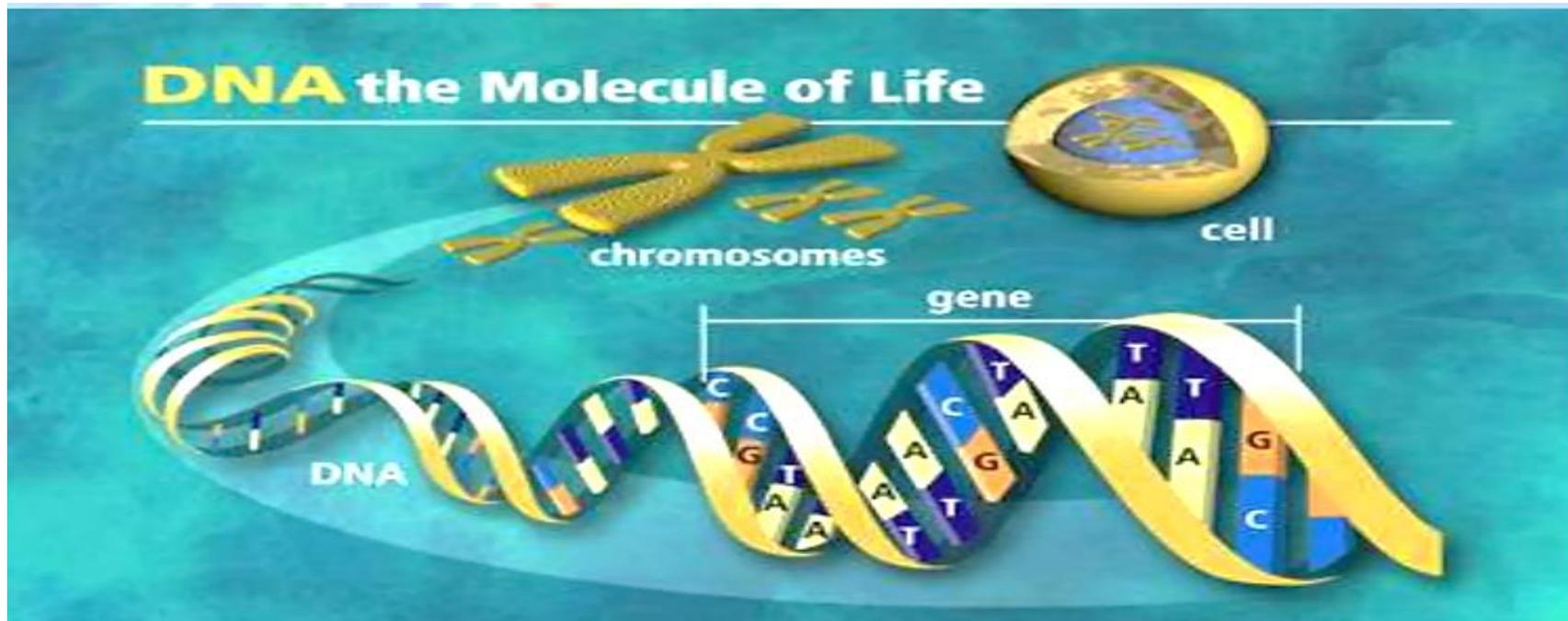


# Genetic Information (DNA, Genes & Chromosomes)



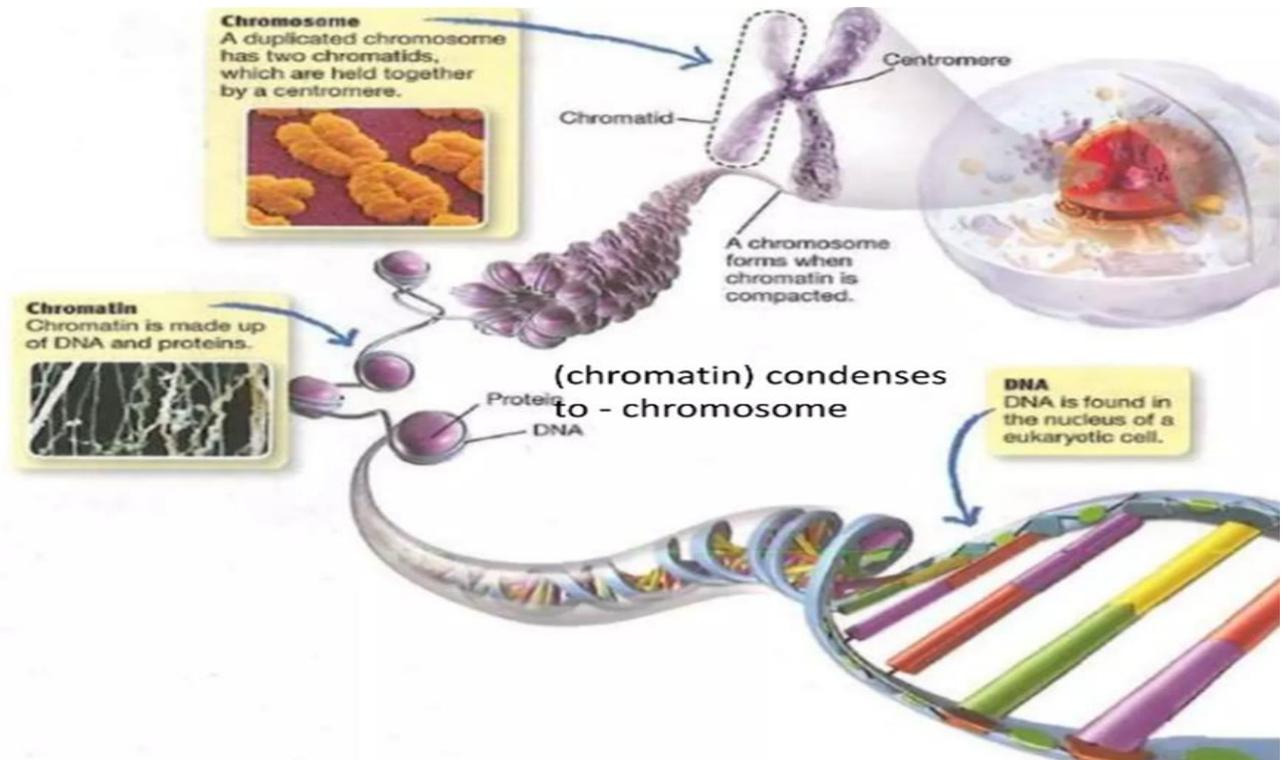
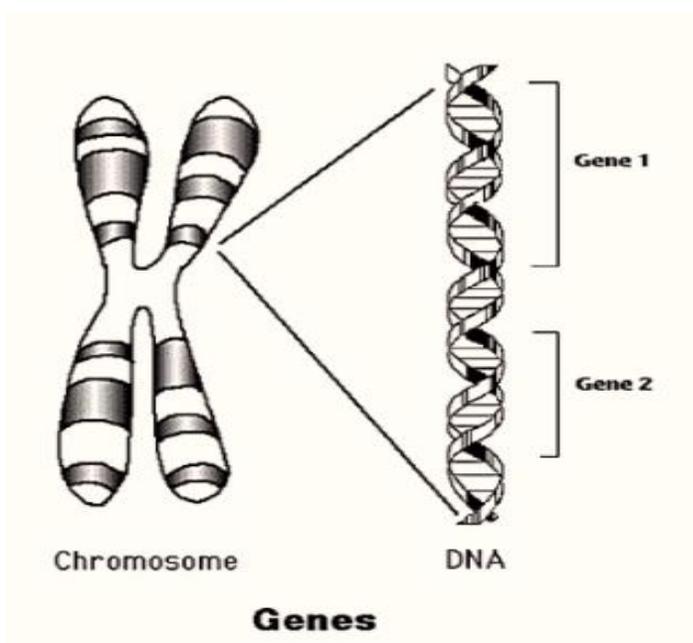
# DNA, Genes & Chromosomes

- At the core of genetics is the DNA, a molecule that contains instructions for building and maintaining living organisms.



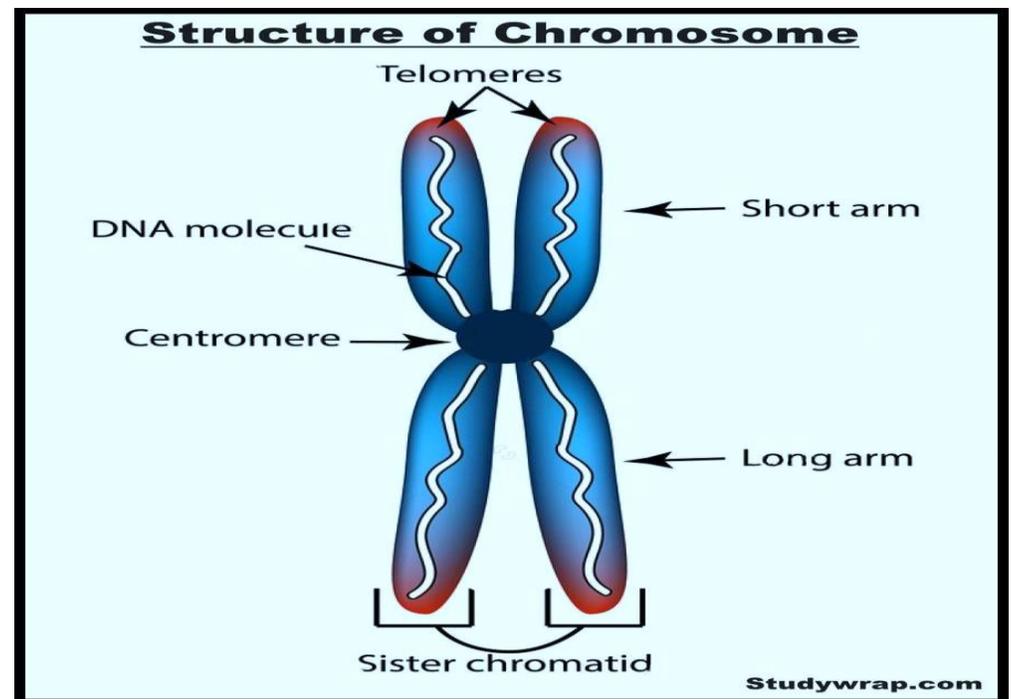
# Chromosome: Structure

- ❑ Chromosome is a thread-like structure made of protein & one molecule of DNA with collection of genes.
- ❑ It acts as a carrier unit of inheritance (the storage unit for genes inside the nucleus).



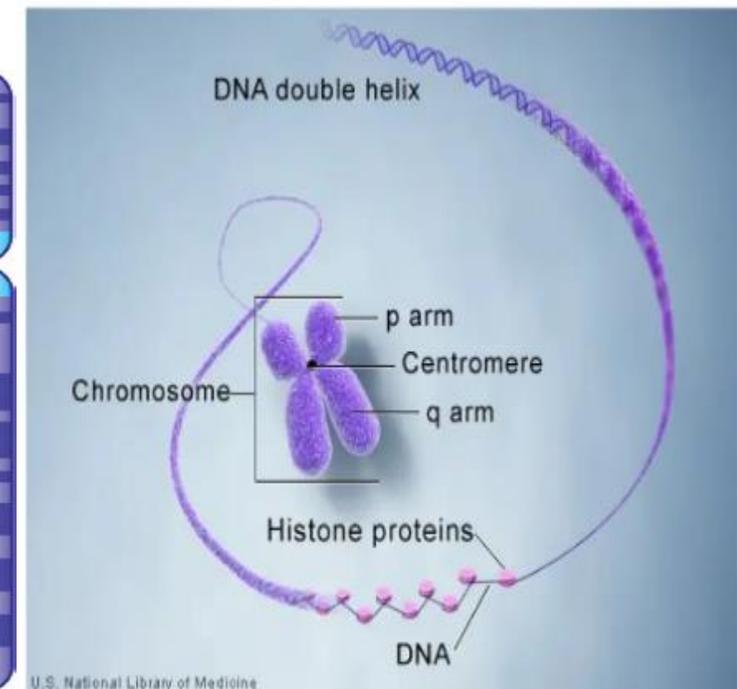
# Chromosome: Structure

- Each chromosome formed of **2 sister chromatid** linked by a **centromere**
- Each has a **short arm** called 'p' & a **long arm** called 'q'.
- These arms are divided into **region**, **bands & sub-bands**, numerically, e.g., 7q21.2 means long arm of chromosome 7, region 2, band 1 & sub-band 2.



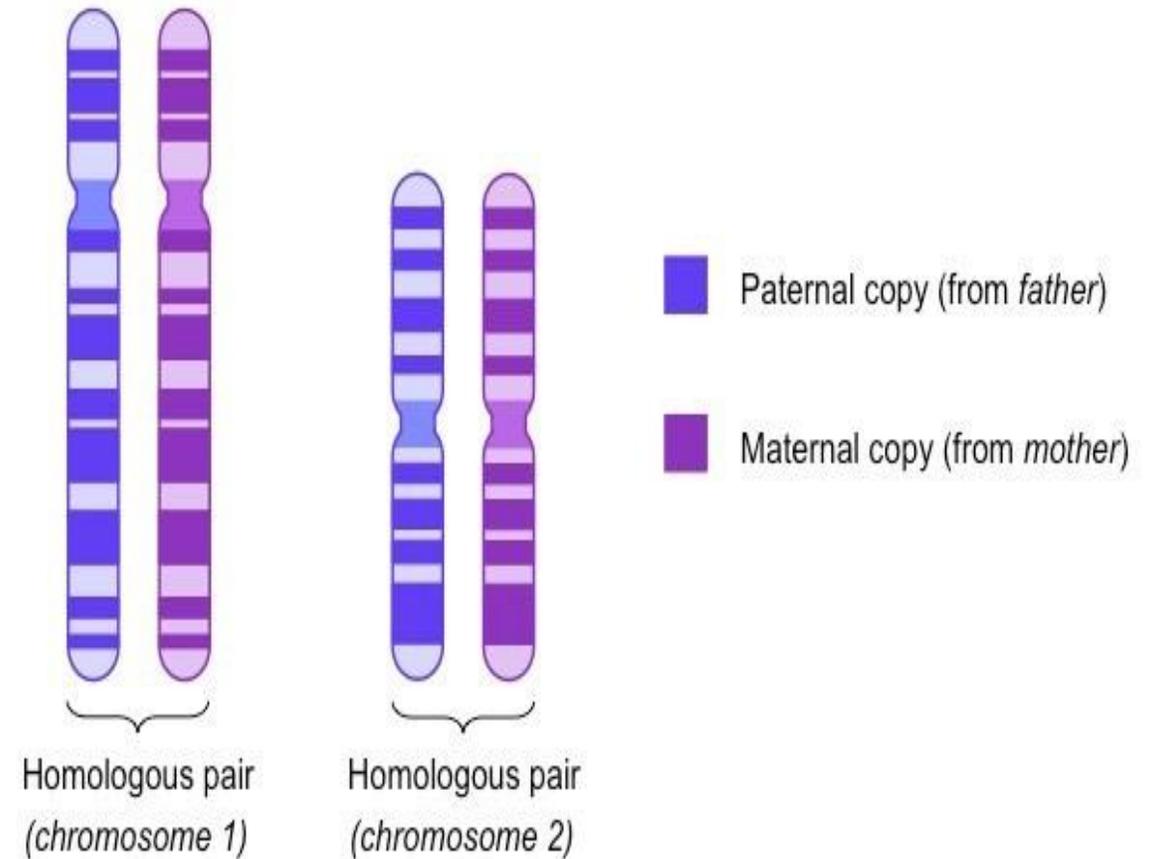
X - Chromosome

Arm	Region	Band	Sub-band
p	2	2	1, 2, 3
		1	1, 2, 3, 4
	1	1	1, 2, 3, 4
		1	1
q	1	1	1, 2
		2	1, 2
		3	1, 2, 3
		1	1, 2, 3, 4, 5
	2	2	1, 2, 3, 4, 5, 6, 7, 8
		3	1, 2, 3, 4, 5, 6, 7, 8
		4	1, 2, 3, 4, 5, 6, 7, 8
		5	1, 2, 3, 4, 5, 6, 7, 8



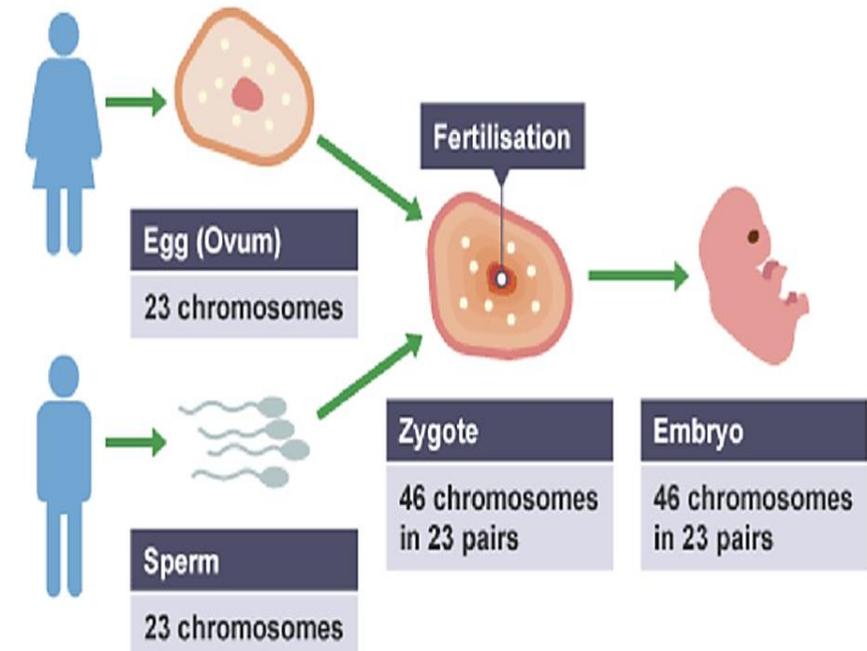
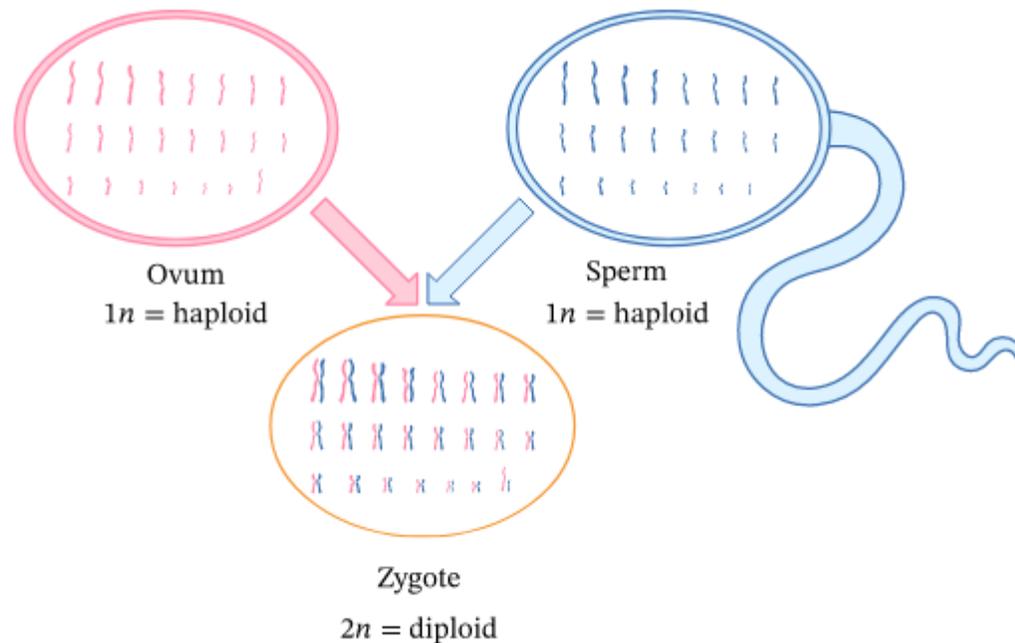
# Number of chromosomes

- In **Somatic cells**, the total number is 46 arranged in 23 pairs named as **'diploid' (2n)**.
- One member of each chromosome is derived from **the mother** & the other is from **the father**



# Number of chromosomes

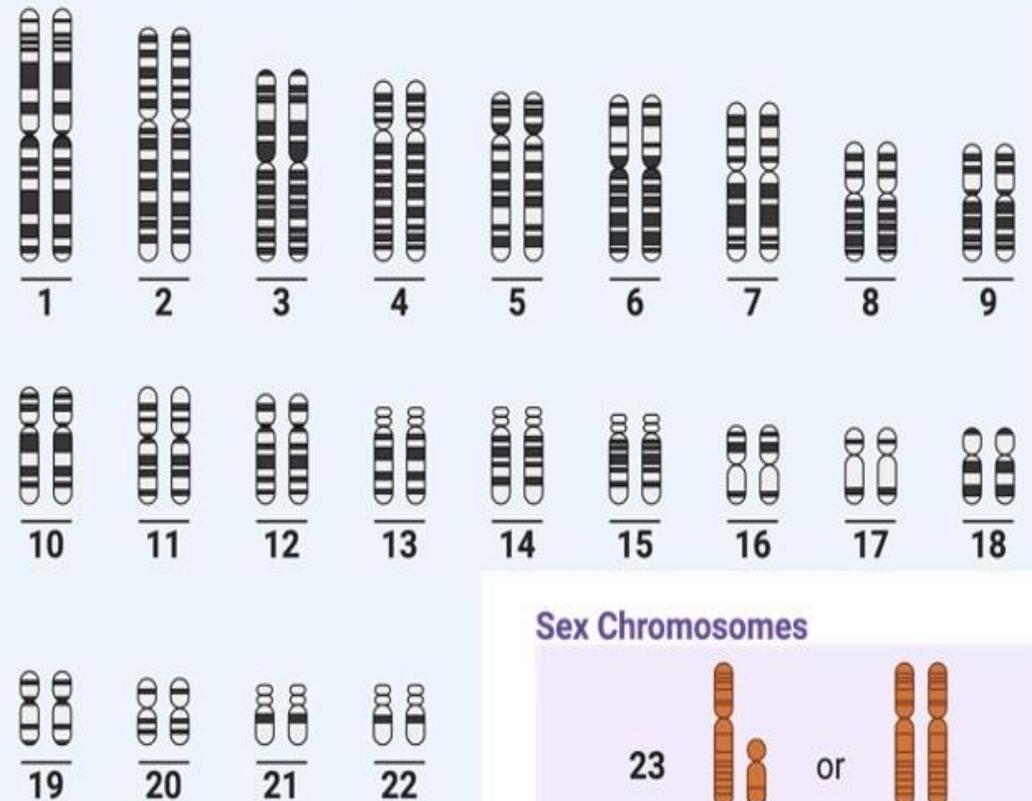
- While **in gametes (male sperm or female egg)** the number is half of the somatic cell “**23**” (Single, not paired) and known as a ‘**haploid**’ (**n**) set of chromosomes, which is the result of meiosis during reproduction.



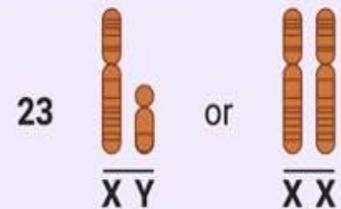
# Autosomes & Sex chromosomes

- 22 pair chromosomes are identical and Known as “Autosomes”
- While the 23rd pair is Known as “Sex chromosome”.
- Women have two ‘X’ chromosomes (XX) for their 23rd pair, and men have one ‘X’ chromosome and one ‘Y’ chromosome (XY).

Autosomal Chromosomes



Sex Chromosomes



# Types of Chromosomes

According to **position of centromere** chromosome divided into:

**Metacentric chromosome:**

centromere located in the center.

**Sub-metacentric chromosome**

centromere is in intermediate position.

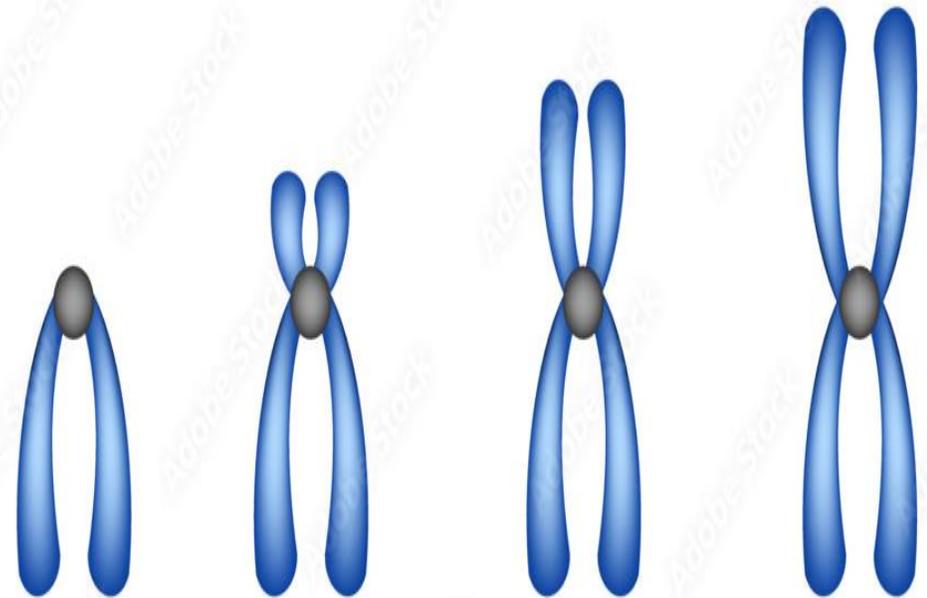
**Acrocentric chromosome**

centromere is close to the end.

**Telocentric chromosome**

centromere is located at the terminal end.

## TYPES OF CHROMOSOMES



Telocentric

Acrocentric

Submetacentric

Metacentric

# Homologous Chromosomes

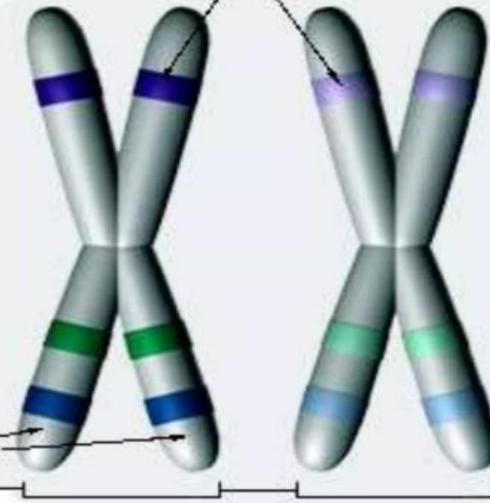
- Homologous chromosomes are **chromosome pairs (one from each parent)** that are similar in length, centromere location, and gene position.

Homologous chromosomes contain DNA that codes for the same genes. In this example, both chromosomes have all the same genes in the same locations (represented with colored strips), but different 'versions' of those genes (represented by the different shades of each color).

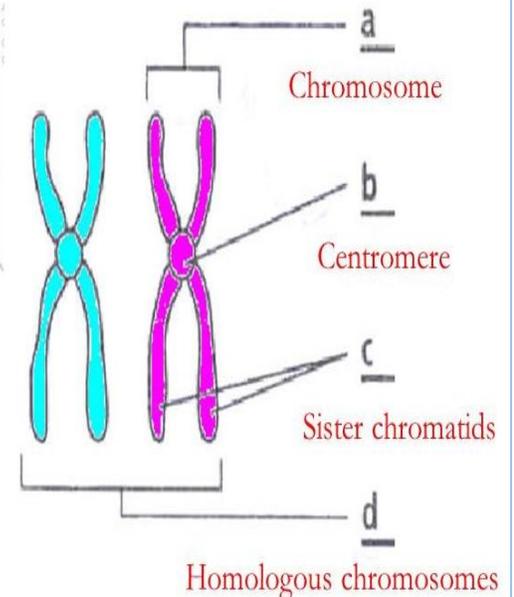
Homologous regions code for the same gene.



Sister chromatids are exact replicas... but homologous chromosomes are not.

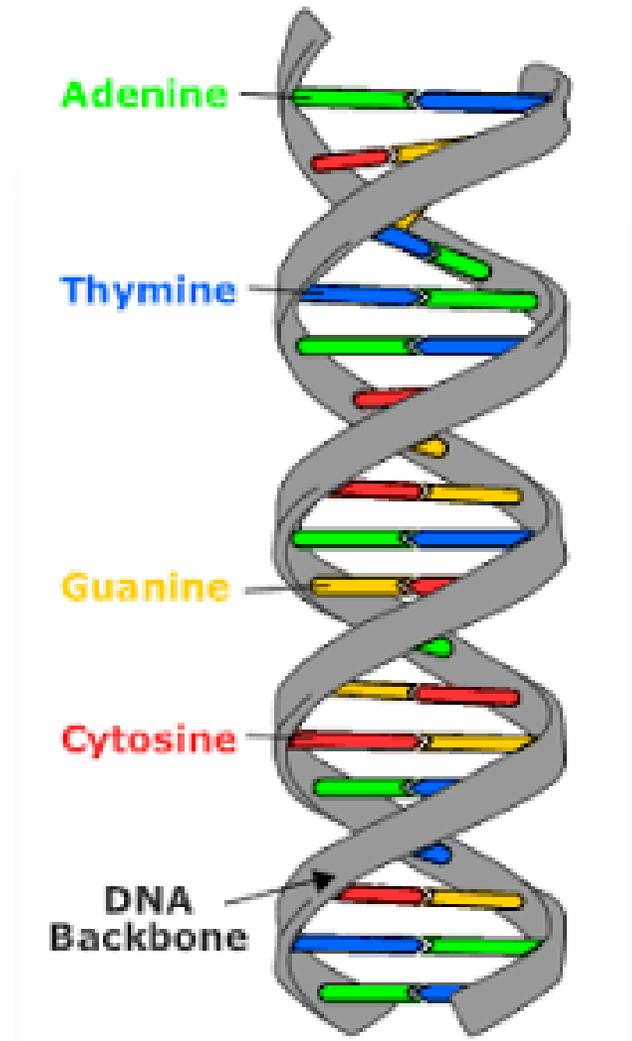


- Homologous chromosomes
- Sister chromatids
- Centromere
- Chromosome



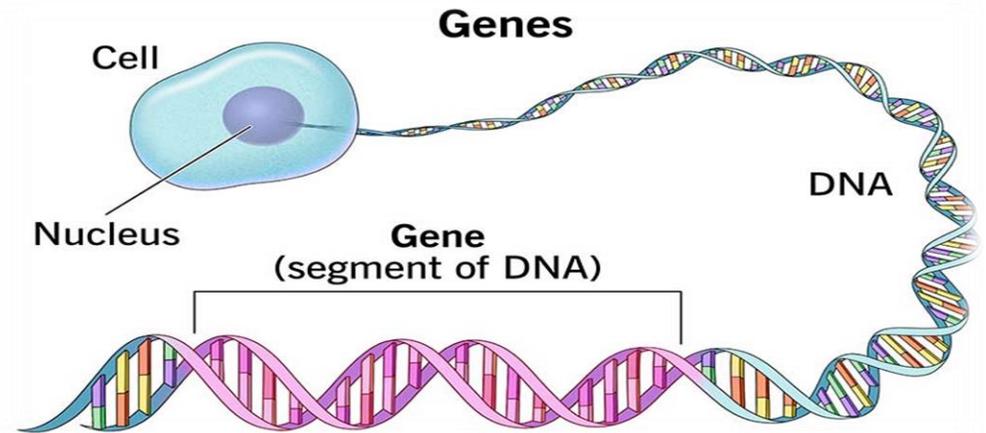
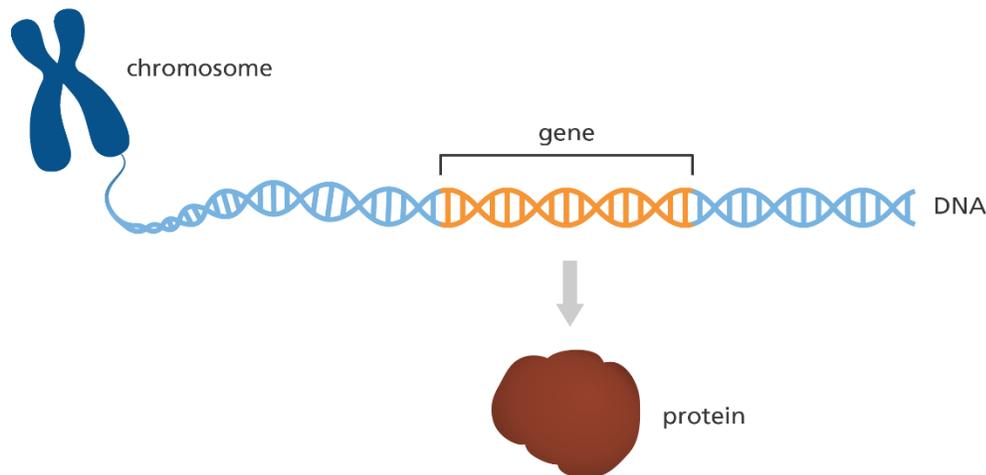
# DNA

- Deoxyribonucleic acid, or DNA, is the basic material of genetics.
- A DNA molecule consists of **two strands twisted together**, each made up of sequence of **nucleotides**-adenine, guanine, cytosine, and thymine.
- It **stores** an organism **genetic information** essential for the synthesis of functional **proteins**.



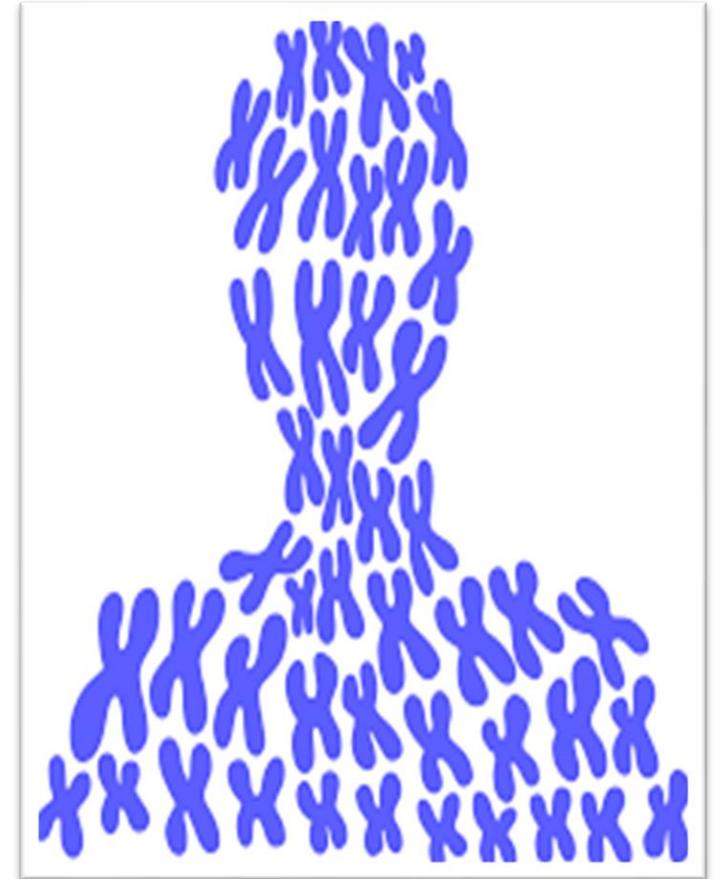
# Gene

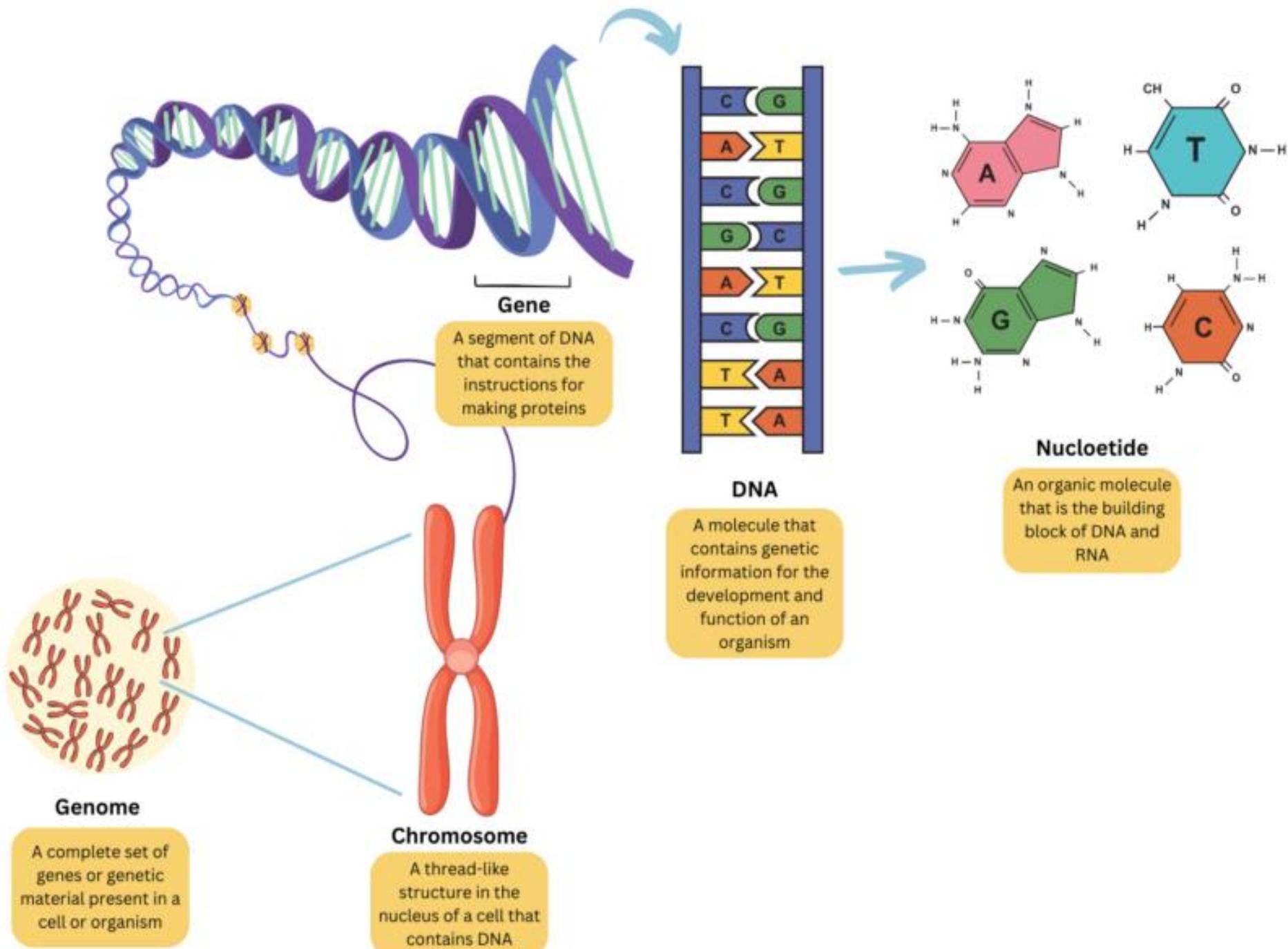
- Gene – basic unit of genetic information “Unit of hereditary” which provide instructions for making protein and determine the inherited characters
- Gene is a **segment of DNA** along the chromosome which carry specific sequence of nucleotide codes for a single specific protein.
- Human beings have an estimated **20,000 to 25,000 genes**.



# Genome

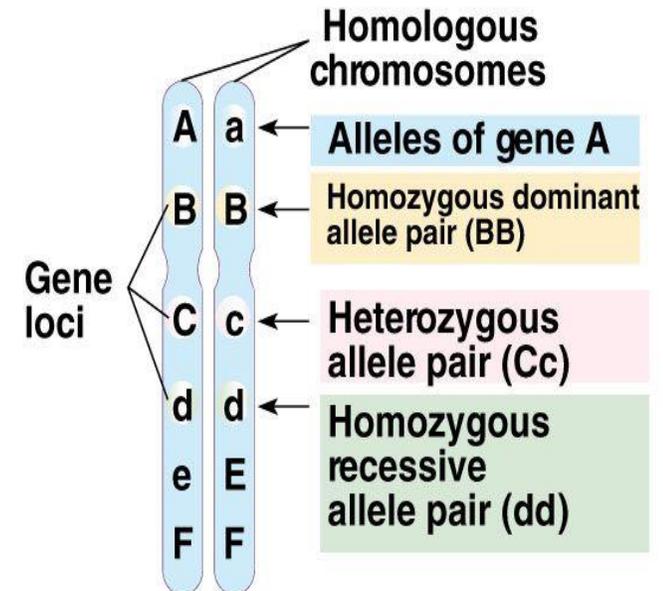
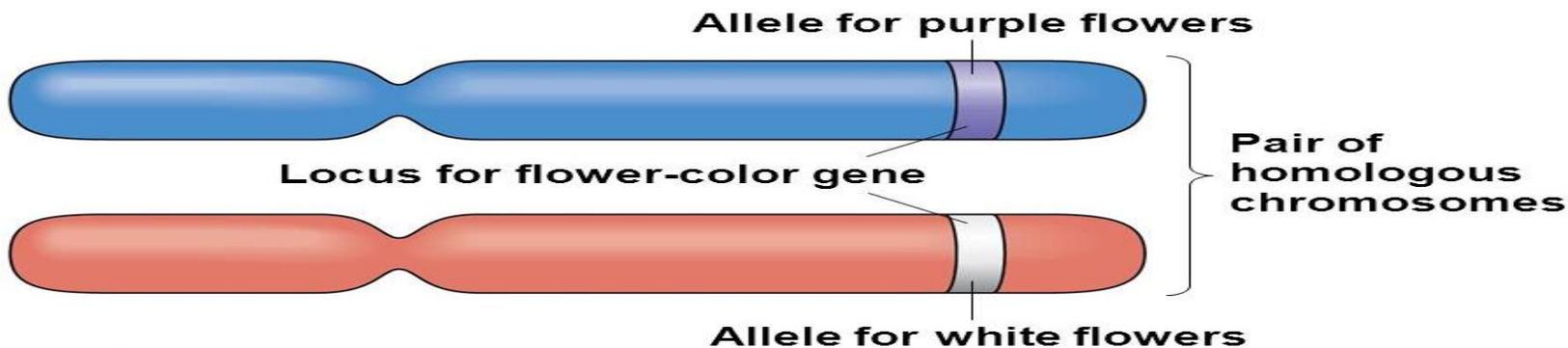
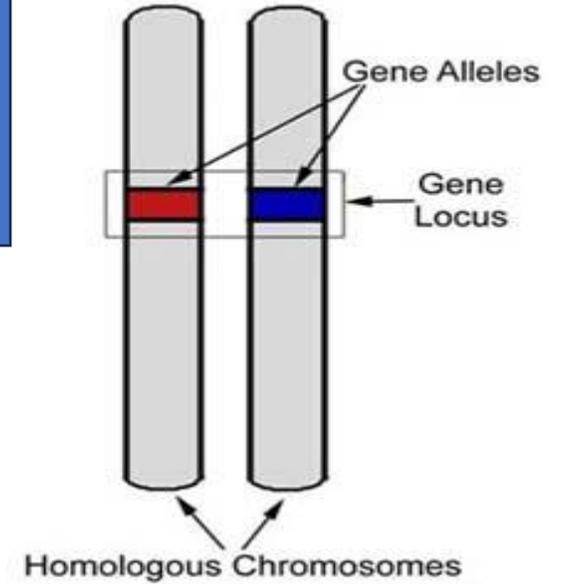
- Genome is the complete set of genes in an individual. It represents the collection of all genetic material in a particular cell or organism.
- Its size is generally given as its total number of base pairs
- The human genome composed approximately of **3 billion base pair** of DNA.
- **Only 1.5 %** of the human genome is made up of protein-coding genes





# Locus & Allele

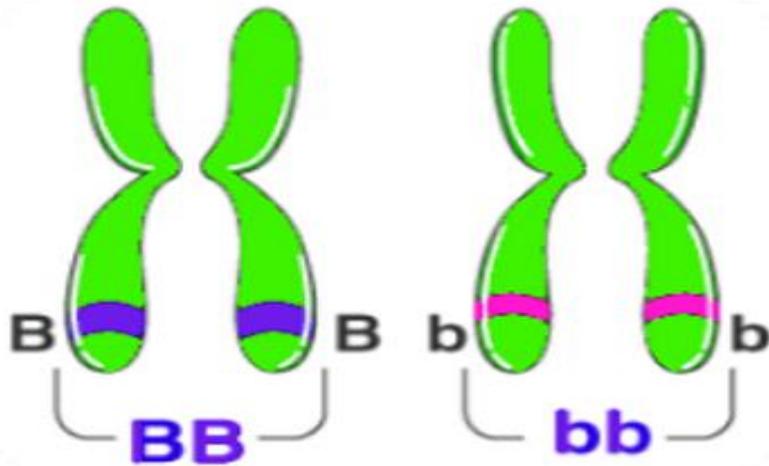
- **Locus:** Specific site (location) (fixed positions) of certain gene on a certain chromosome.
- **Allele:** Alternative form ( Variant ) of a gene that occupy a specific locus on chromosome
- A single allele for each locus is inherited separately from each parent. One allele is inherited from father and the other from mother



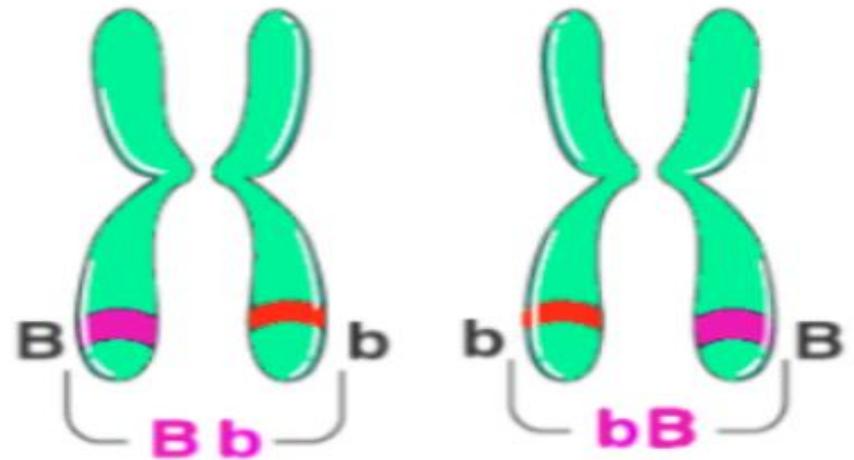
# Homozygous & Heterozygous

- **Homozygous:** Both alleles of a pair are the same
- **Heterozygous:** Both alleles of a pair are not the same (different)

## Homozygous vs Heterozygous in genetics



Homozygous = Same Alleles



Heterozygous = Different Alleles

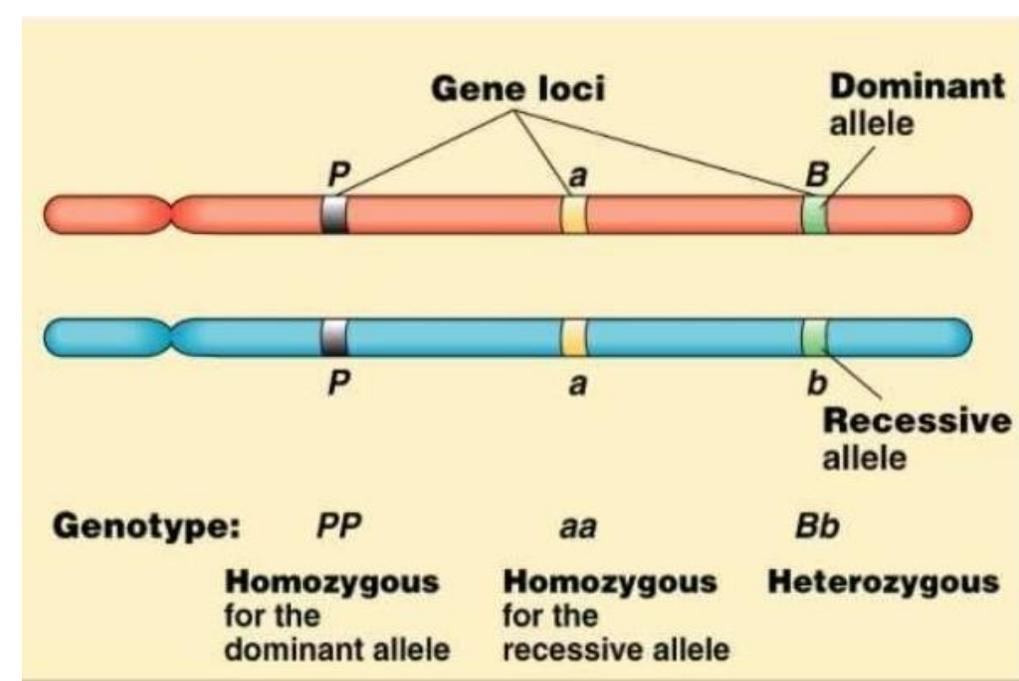
# Dominant & Recessive

## □ Dominant allele (Capital letter):

- The **allele** of a gene that **always express& masks or suppresses the expression** of an alternate allele
- The trait appears in the homozygous dominant & heterozygous condition

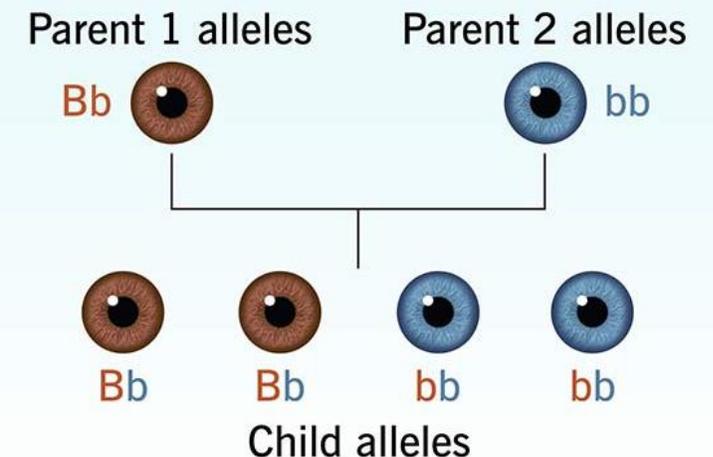
## □ Recessive allele (small letter):

- an **allele that is masked by a dominant allele** (Express only in absence of dominant allele)
- The trait appears only in homozygous recessive.



Alleles - different versions of genes (B/b)

Ex. Child's possible eye color depends on the allele received from each parent



# Genotype versus Phenotype

- **Genotypes:** the 2 alleles of a gene at each locus, constitute the individual's genotype at the locus. eg. BB, Bb, bb.
- **Phenotypes:** The expression of a genotype (the physical appearance of an individual) is termed a phenotype. For example, hair color, weight, or the presence or absence of a disease.

**Genotype**



**Genetic Code**

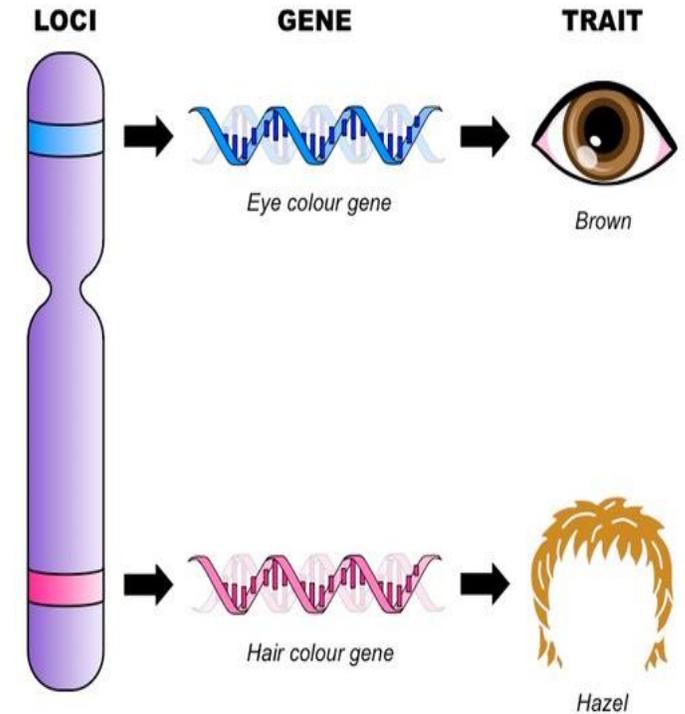
**Phenotype**



**Physical Expression**

# Trait

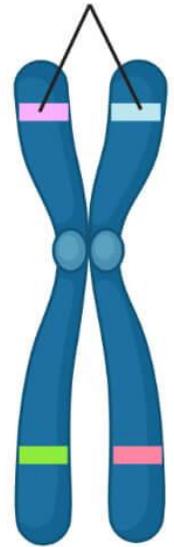
- Trait is any **genetically determined characteristics of the individual**
- **Dominant traits**- traits that are expressed.
- **Recessive traits**- traits that are covered up.



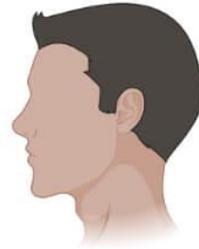
# Trait

Locus

## Genes and Loci



Hair Color Gene



Trait (Black Hair)



Eye Color Gene

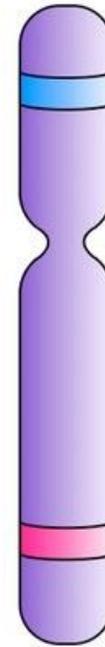


Trait (Brown Eye)

LOCI

GENE

TRAIT



Eye colour gene



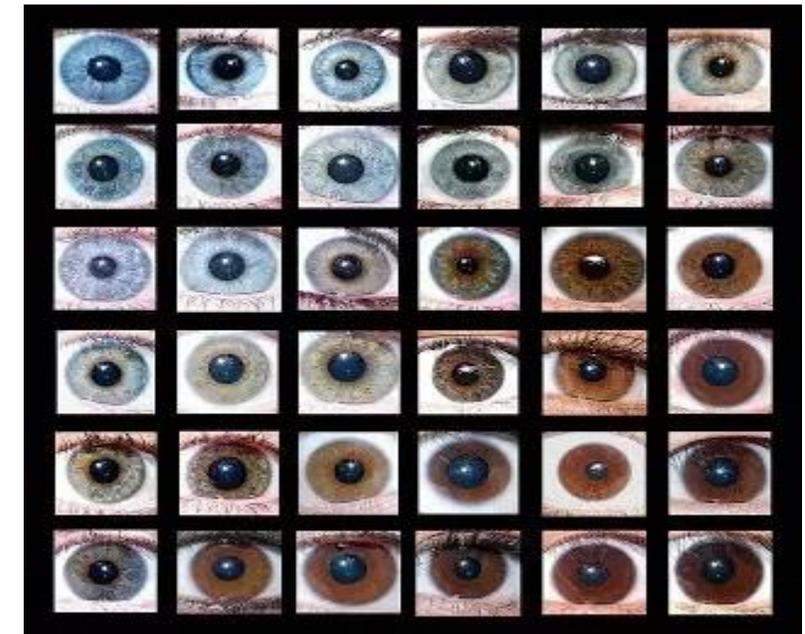
Brown



Hair colour gene



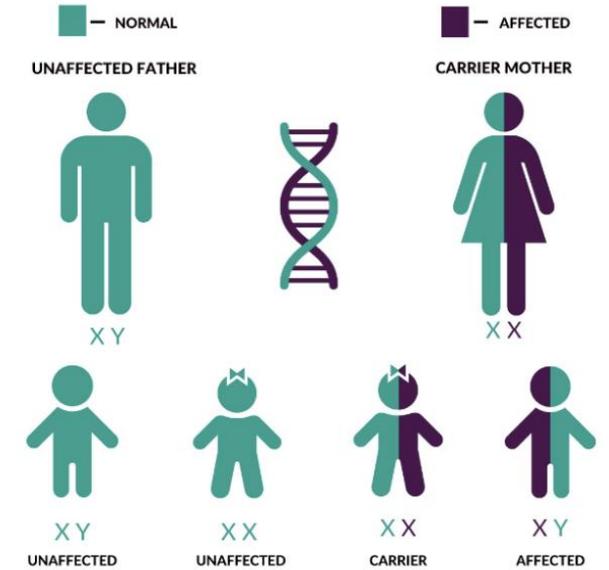
Hazel



An allele is a version of a gene. **Consider eye color.** Everyone has a gene for eye color, but there are many, many ways that the gene(s) could be slightly different. Each variation is an allele, and each allele will produce a slightly different effect.

# Inheritance

- Inheritance is **how traits, or characteristics, are passed on from generation to generation.**
- Gregor Mendel is considered "The Father of Genetics"
- Mendel demonstrated that **traits are passed from parents to offspring through discrete factors, genes, located on chromosomes.** His work showed that some traits are dominant over others and that segregation and independent assortment of alleles allows for prediction of phenotypic ratios in offspring.





**THANK YOU**