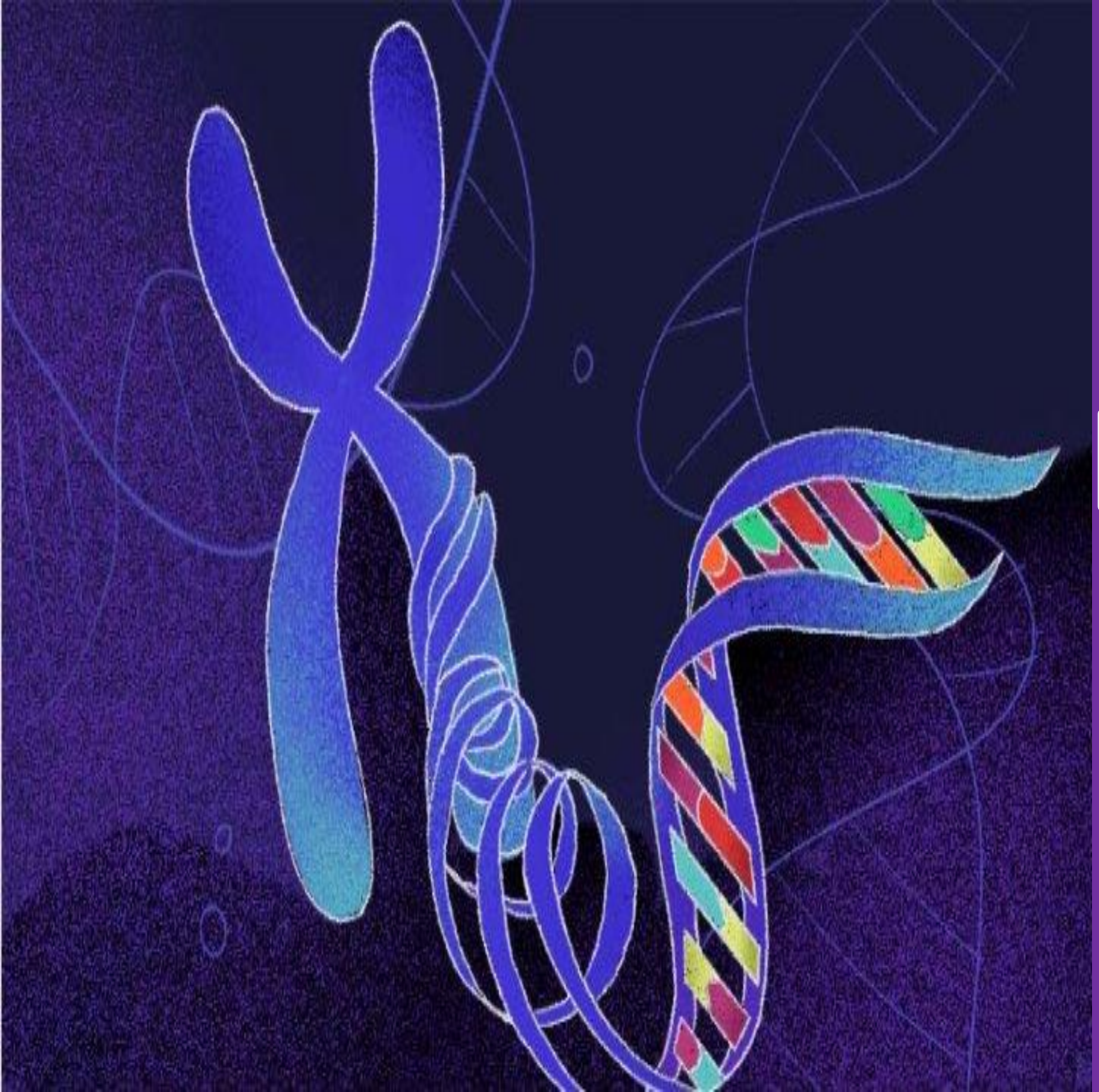


Genetics and Epigenetics

Introduction to Biology

Chapter: 10

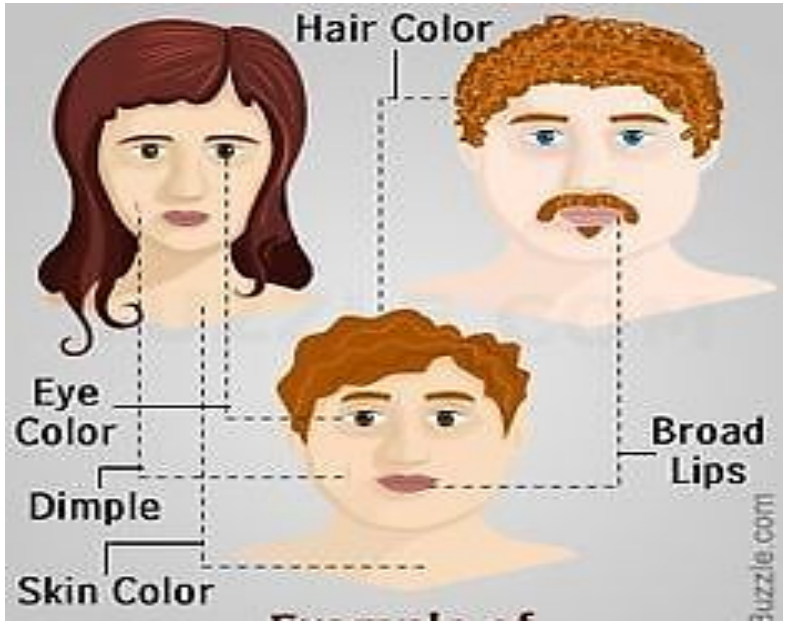
BRAC UNIVERSITY

Inspiring Excellence

Have you ever wondered why children have similarities to both their parents?

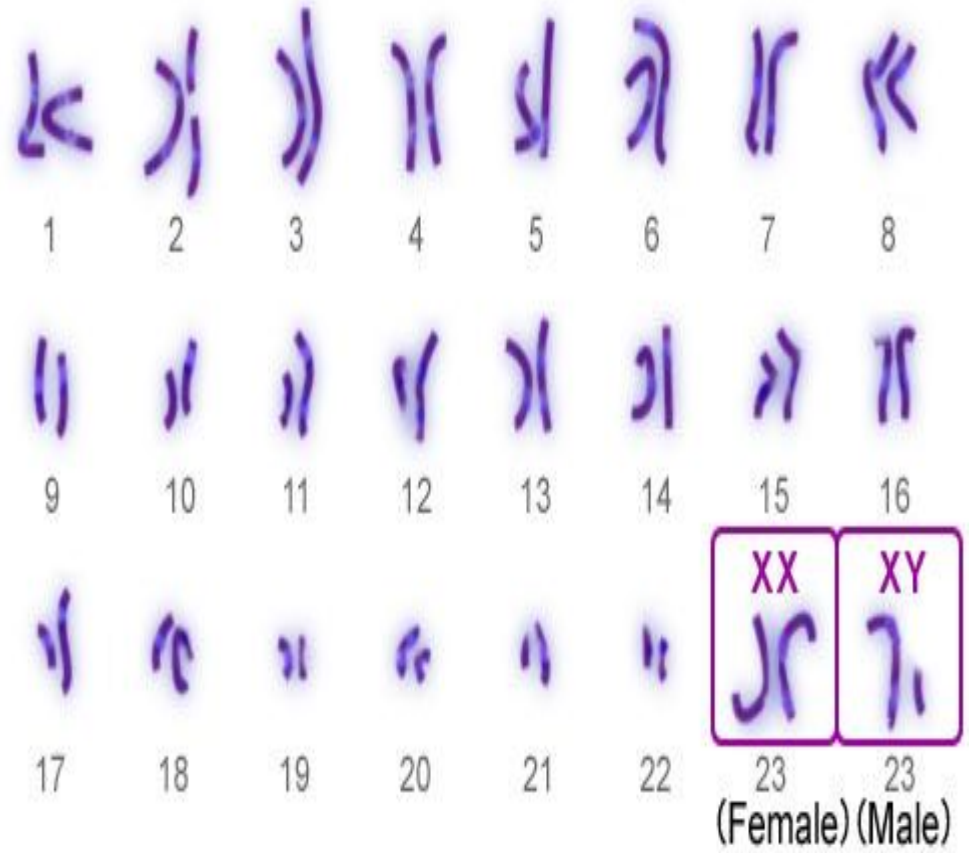
- We acquire these similar characteristics (traits) through a process called **inheritance** (heredity).

- Inheritance occurs through transfer of genetic material from parents to their offspring.



Chromosomes

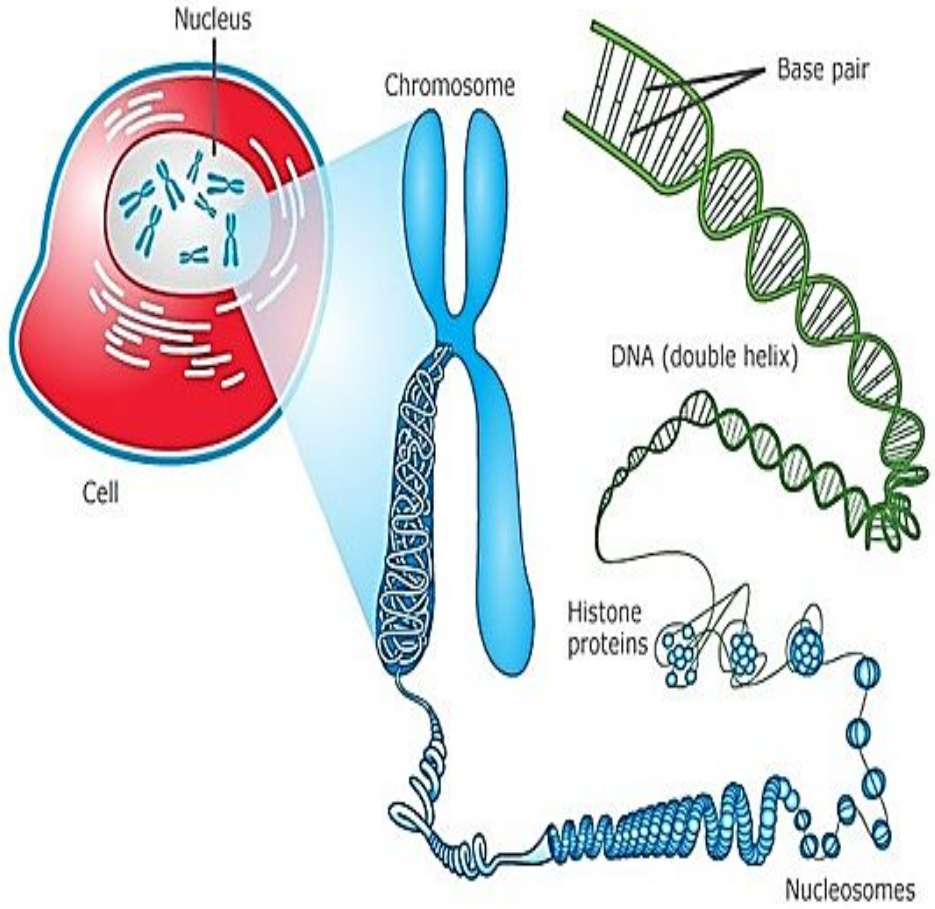
- They are present in the nucleus of the cell (eukaryotes) or present in the cytoplasm (prokaryotes).
- Different organisms have different numbers of chromosomes.
- Humans have 23 pairs of chromosomes (46 in total): 1 from each pair comes from father and one from mother.



Karyotyping

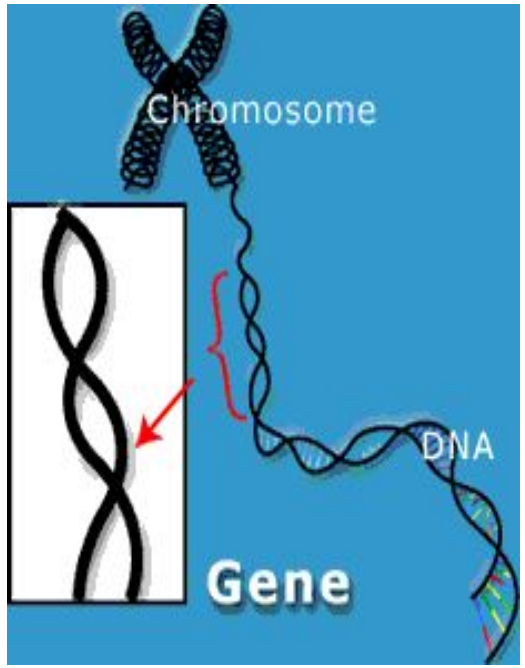
Chromosomes

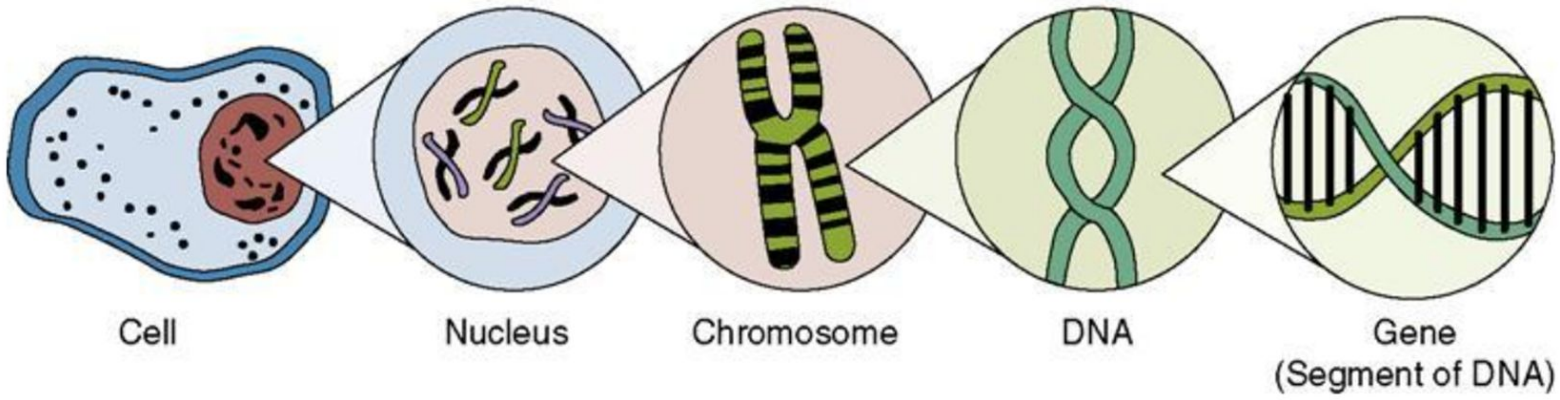
- Thread like structures that contain an organism's **DNA**.
- The DNA are coiled in proteins called **histones (nucleosomes).**



Genes

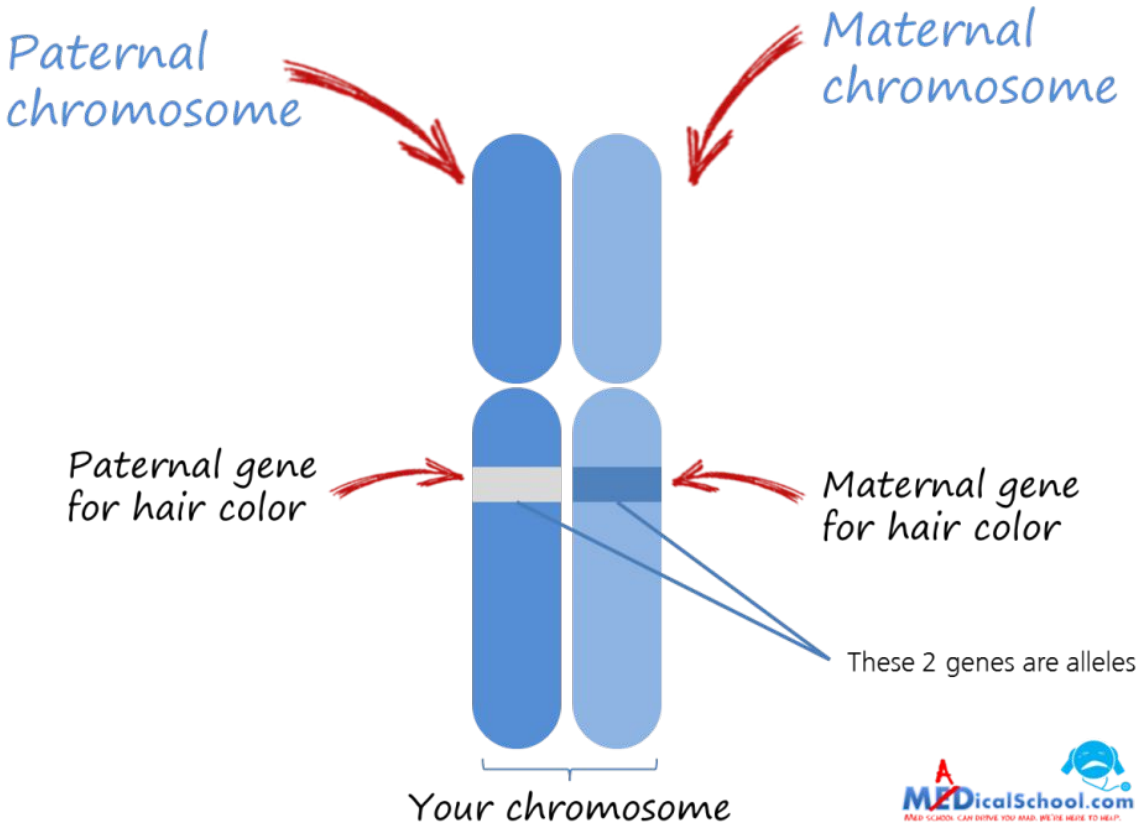
- Units of information about specific traits
- A sequence of DNA
- Passed from parents to offspring
- The complete set of DNA of an organism is called its **genome**.
- The study of genes, genetic variation and heredity is called **Genetics**.





Alleles

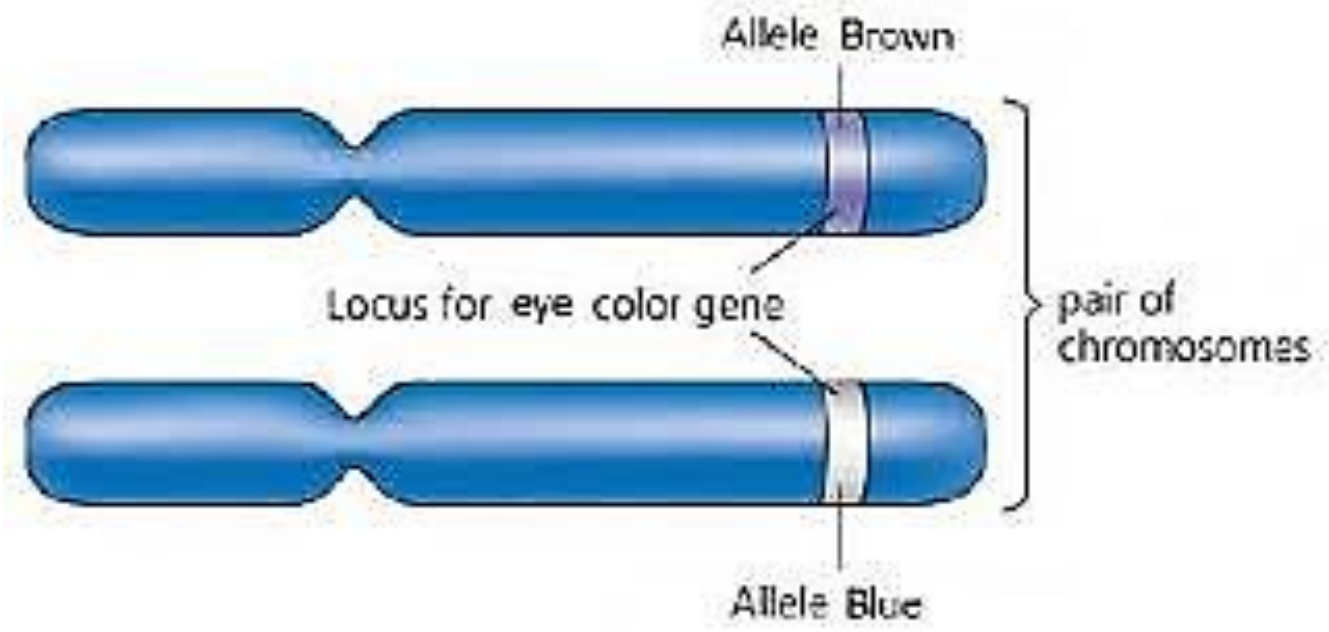
- Different molecular forms of the same gene
- Arise by mutation
- Each allele is passed from each parent to the offspring



Locus

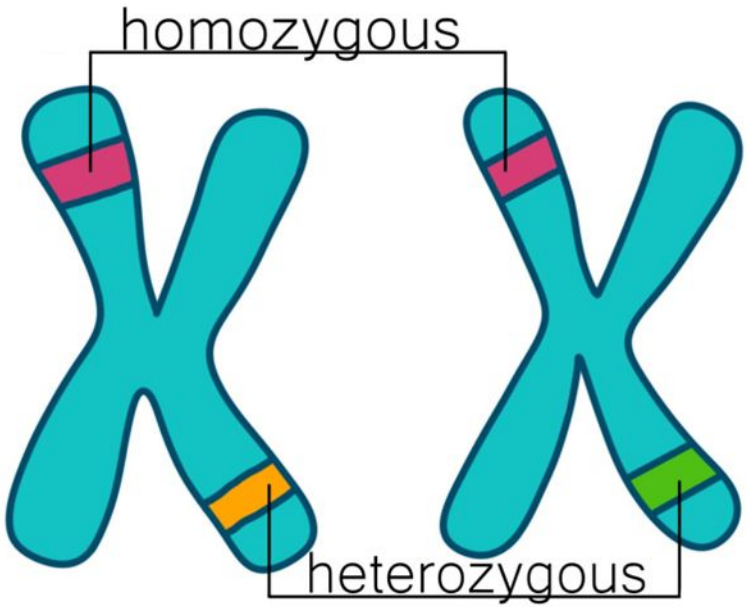
- Each gene has a specific location (locus) on a chromosome

- Alleles are located in the same locus of a chromosome



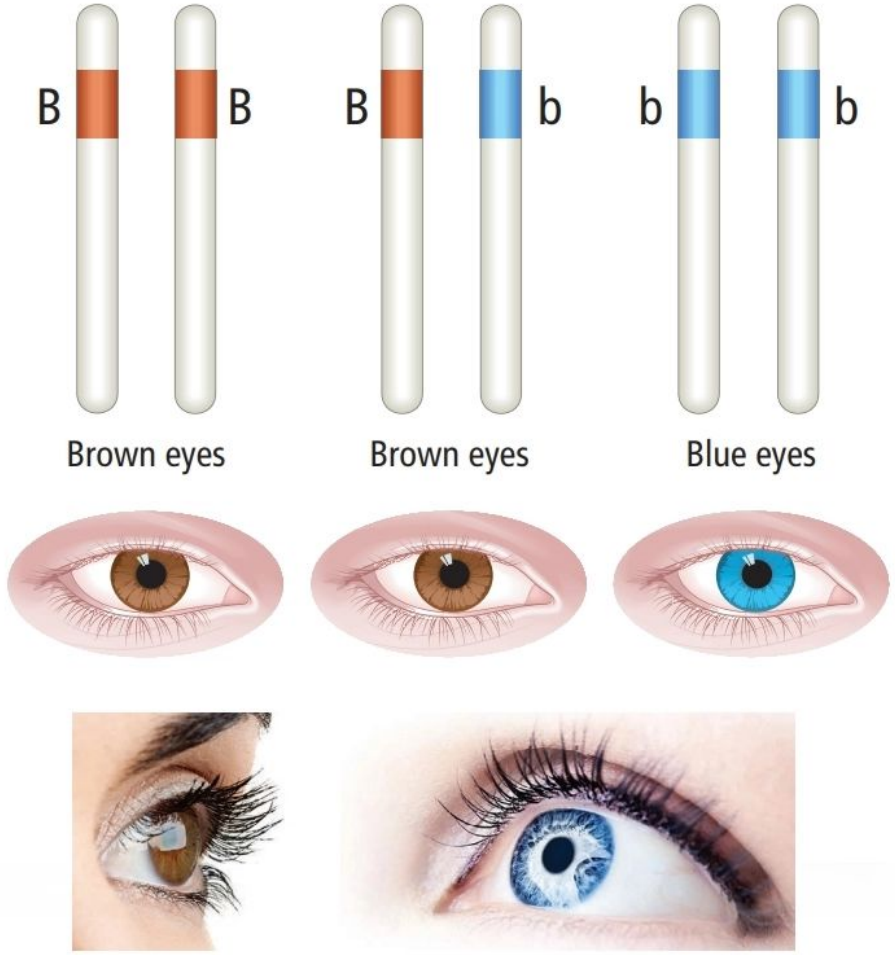
Allele combinations

- Homozygous
  - having two identical alleles at
  - *AA* or *aa*
- Heterozygous
  - having two different alleles at
  - *Aa*



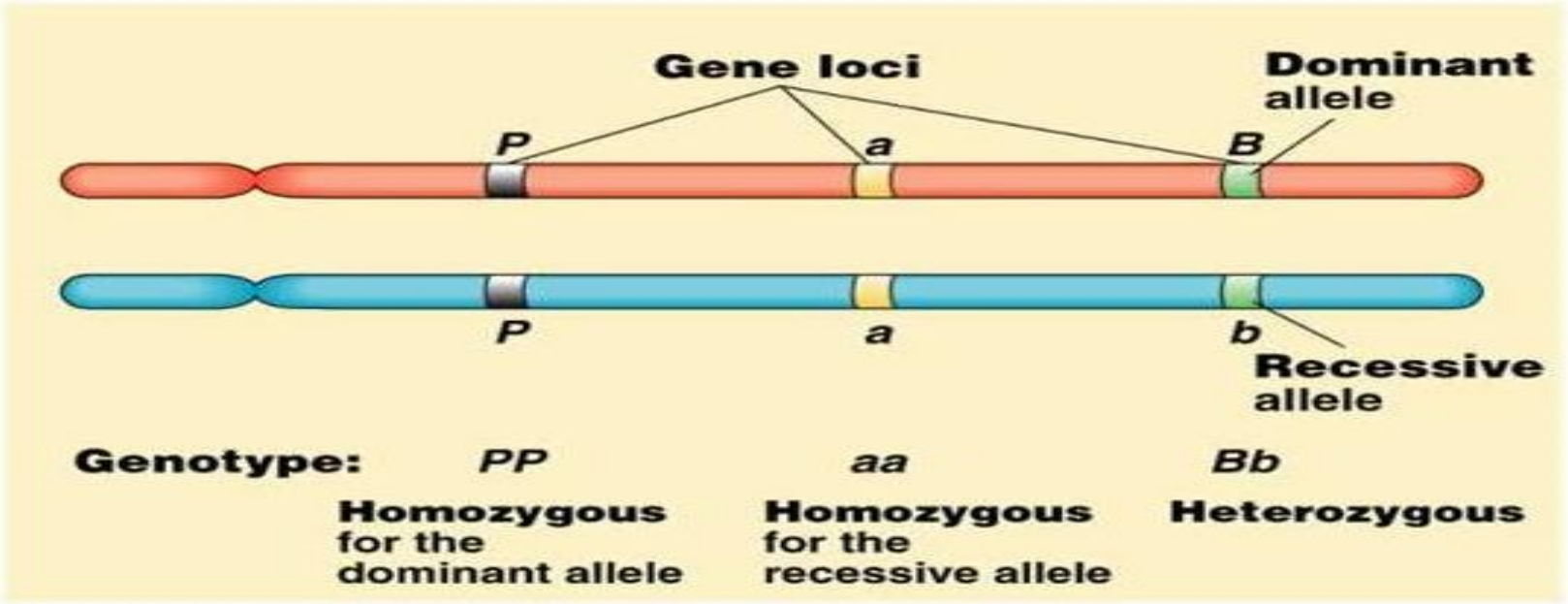
Dominant & Recessive Alleles

- A dominant allele is always expressed and hence masks a recessive allele that is paired with it.
- Recessive alleles are only expressed when both alleles are recessive.
- Dominant alleles are written in capital letters (B) and recessive alleles are written in small letters (b).



Examples

Allele	Term	Result
1 dominant + 1 dominant	Homozygous dominant (BB)	Dominant trait is expressed
1 recessive + 1 recessive	Homozygous recessive (bb)	Recessive trait is expressed
1 dominant + 1 recessive	Heterozygous (Bb)	Dominant trait is expressed



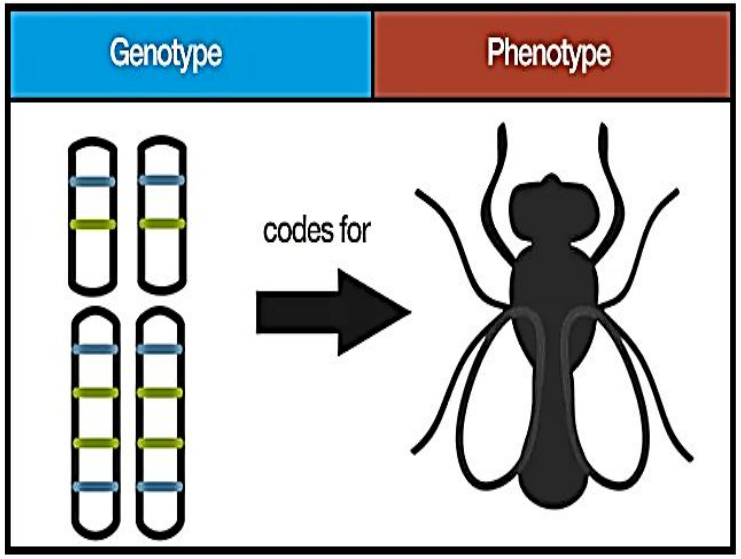
Genotype & Phenotype

- Genotype refers to particular genes an individual carries

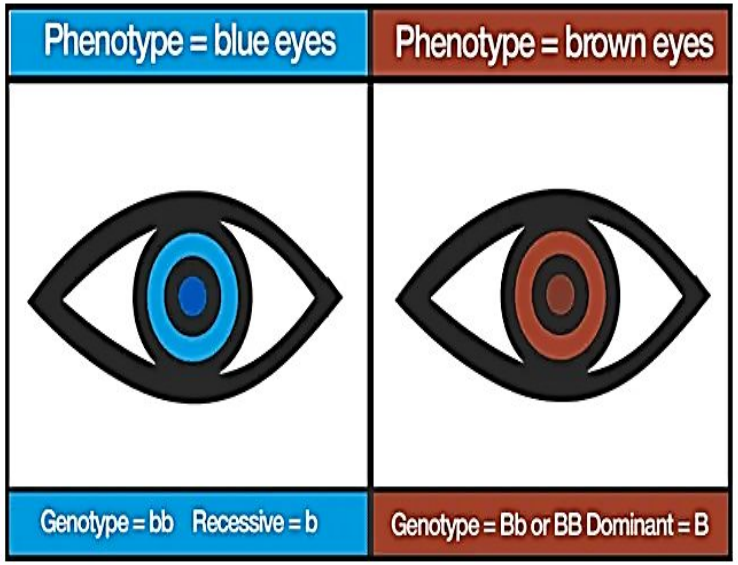
- Phenotype refers to an individual's observable traits

- We cannot always determine genotype by observing phenotype: Homozygous dominant or Heterozygous?

Genotype is the genetic makeup of the organism

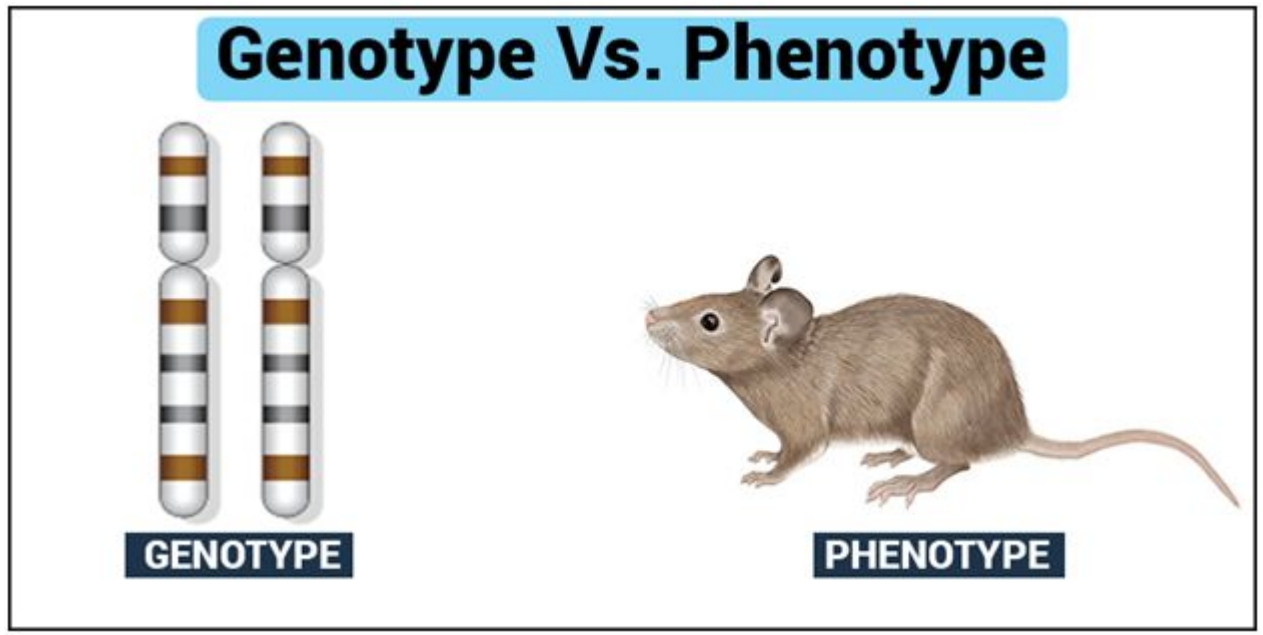


Phenotype is the physical apperance of the organism



GENOTYPE VERSUS PHENOTYPE

GENOTYPE	PHENOTYPE
Genotype is the genetic makeup of an organism	Phenotype is the morphology, properties and behavior of an organism
Can be determined by observing DNA by genotyping methods	Can be determined by observing outward characters
Completely depends on the gene sequences	Depends on the genotype and environmental factors
Inherited by the offspring	Not inherited by the offspring
Consists of all hereditary information that is the expressed and suppressed genes	Consists of expressed genes



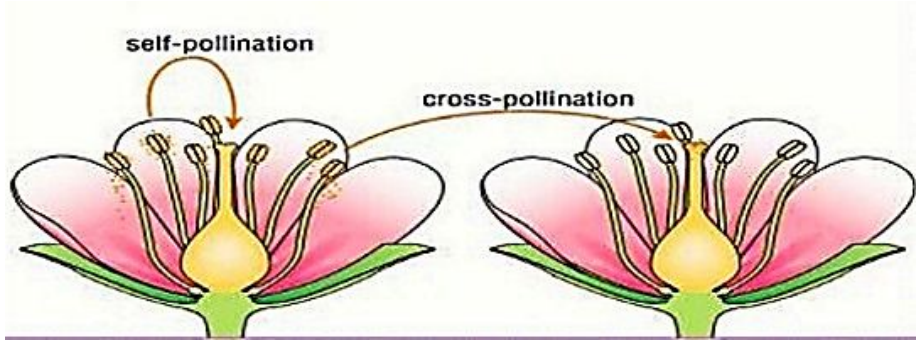
Gregor Mendel

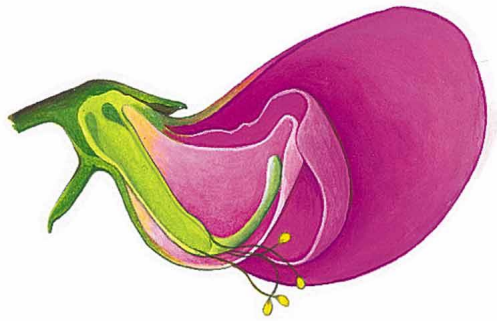
- Strong background in plant breeding and mathematics.
- Using pea plants, found indirect but observable evidence of how parents transmit genes to offspring.
- Particulate theory: physical traits are inherited as ‘particles’

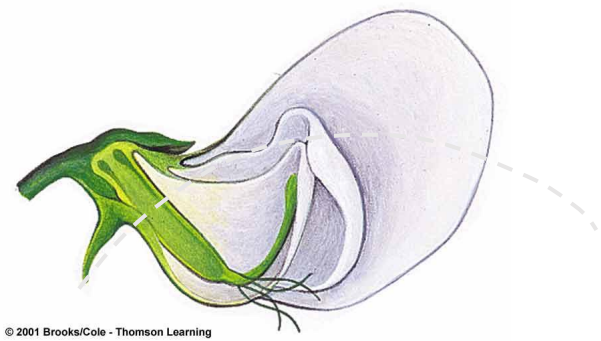
Father of genetics!

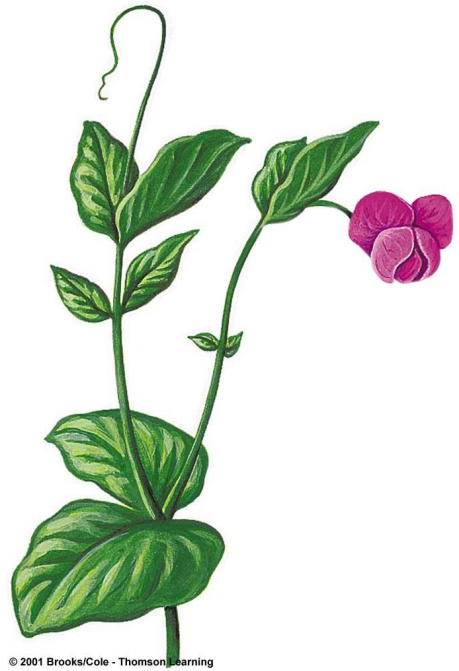
The Garden Pea Plant

- Self-pollinating
- True breeding (different alleles not normally introduced)
- Can be experimentally cross-pollinated



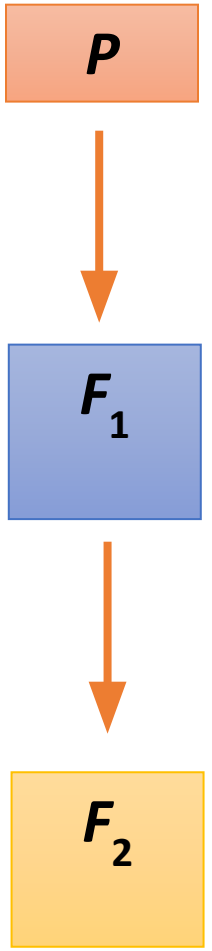






Tracking Generations

- Parental generation mates to produce:
- First-generation offspring mate to produce:
- Second-generation offspring



Monohybrid cross

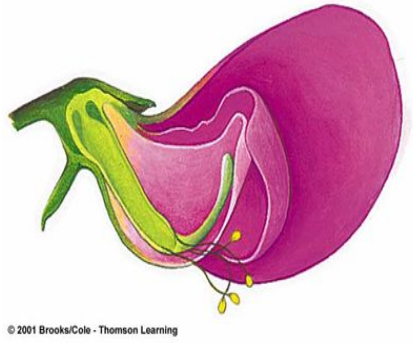
- **A mix of genetics between two homozygous genotypes:**
- **Either completely dominant or completely recessive**

- **Mono: one**
- **Hybrid: combination of two organisms**

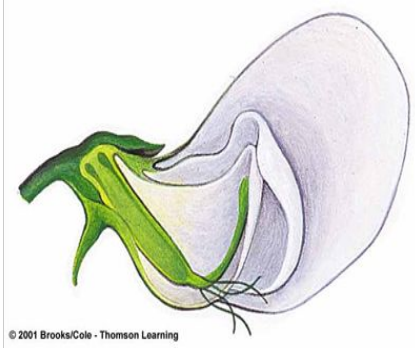
- **For garden pea plants, Mendel observed the colour of pea plants**

Female gametes

Male gametes	*a*	*a*
A	*Aa*	*Aa*
A	*Aa*	*Aa*



Purple: AA

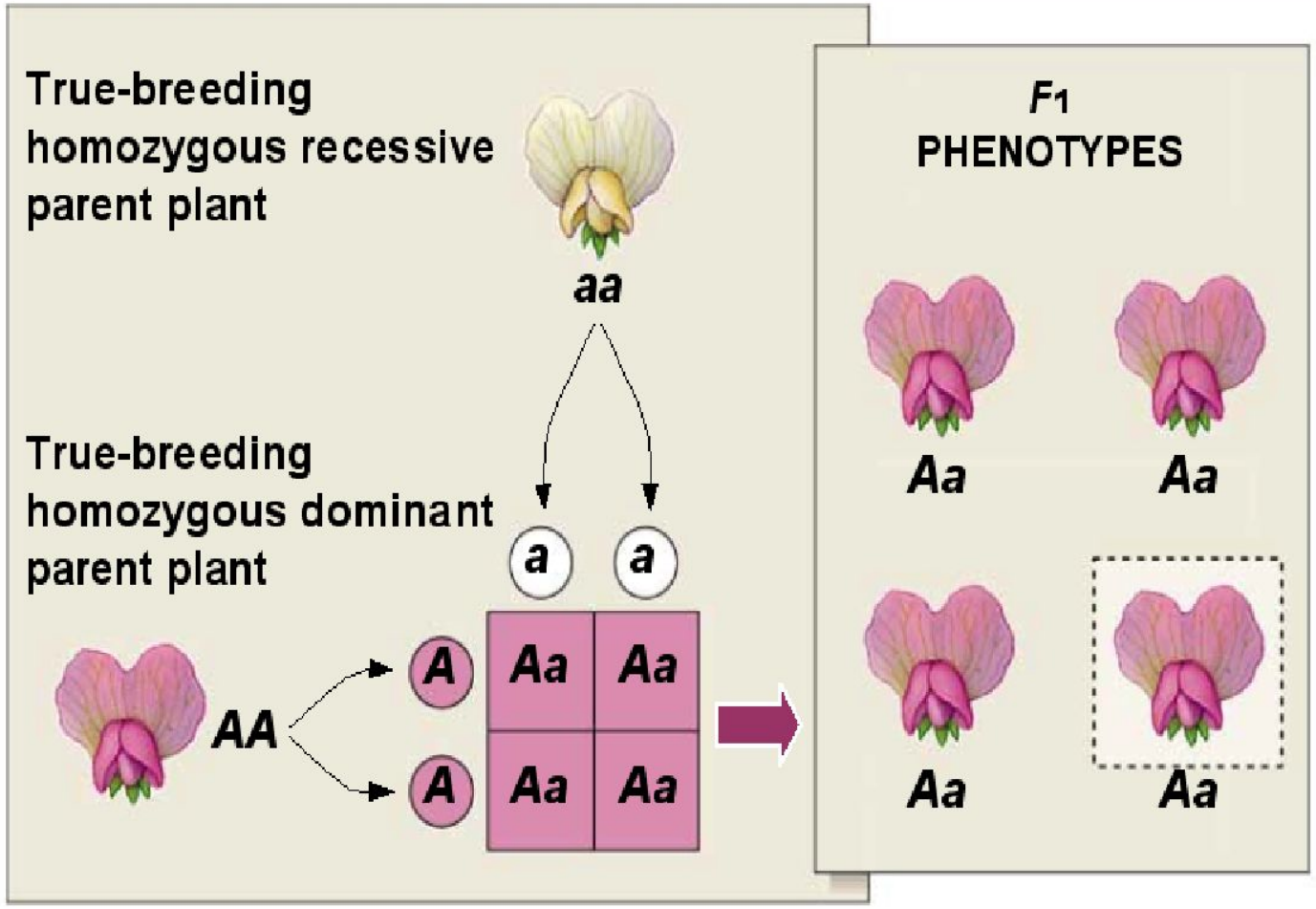


White: aa

F_1 Results of One Monohybrid Cross

Result

All offspring were **heterozygous dominant (Aa)**

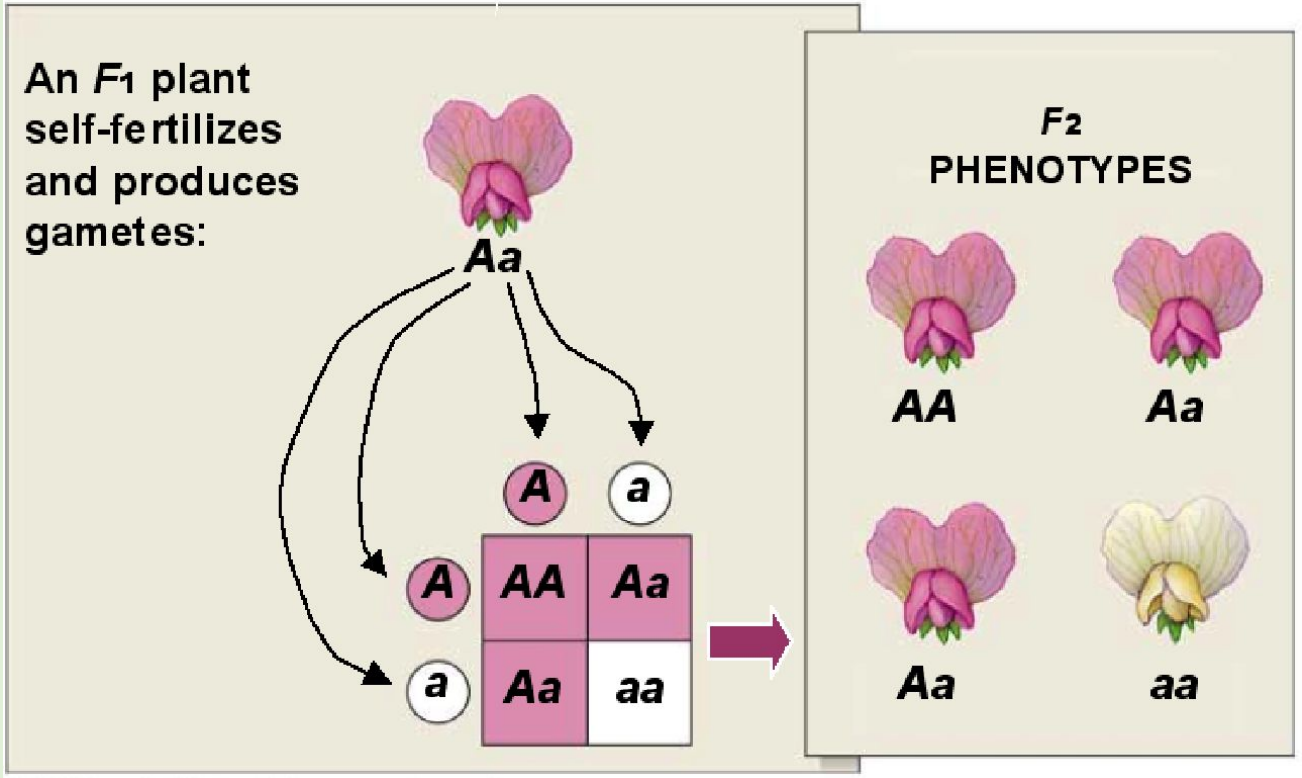


F_2 Results of Monohybrid Cross

Result

- 2 offspring were **heterozygous dominant (Aa)**
- 1 offspring was **homozygous dominant (AA)**
- 1 offspring was **homozygous recessive (aa)**

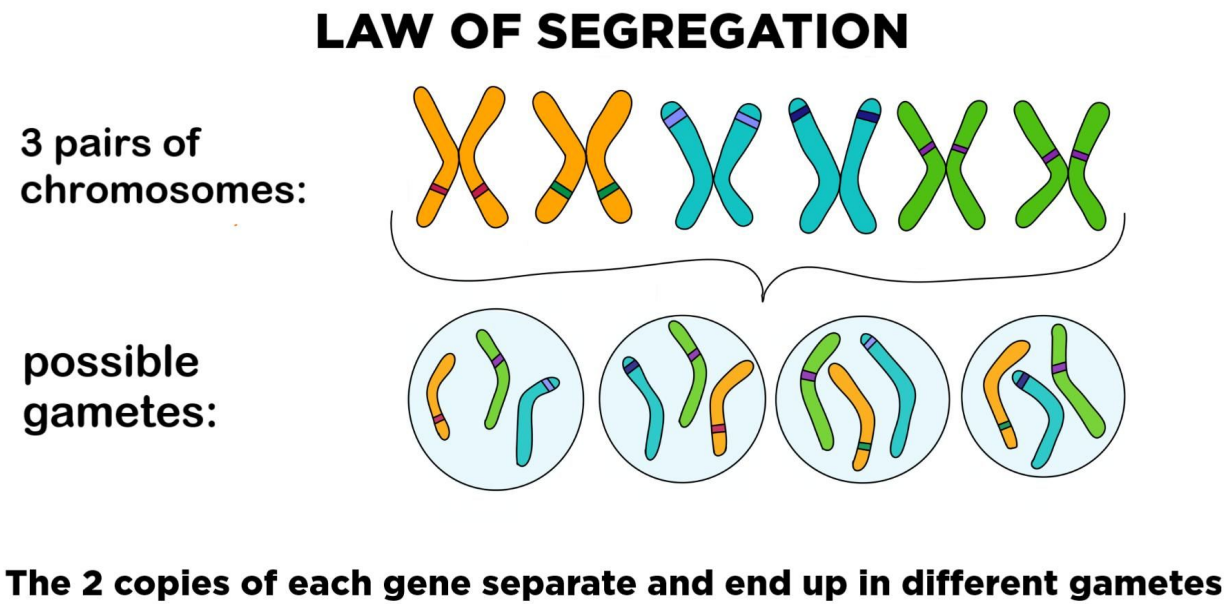
- Phenotypic ratio – 3 dominant : 1 recessive



Mendel's Theory of Segregation

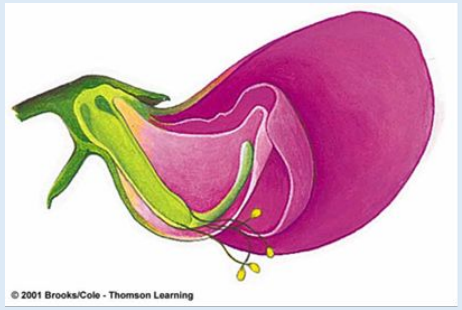
- An individual inherits a unit of information (allele) about a trait from each parent.

- During gamete formation, the alleles segregate from each other.

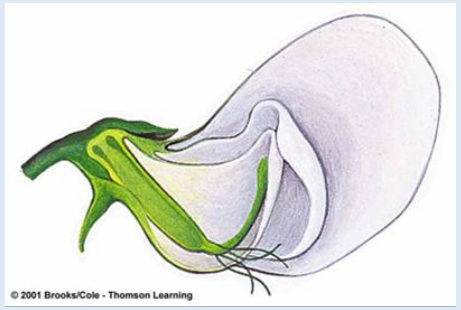


Dihybrid Cross

- Experimental cross between individuals that are homozygous for different versions of **two** traits.
- Di = two
- Hybrid = combination of two organisms



Purple: AA



White: aa

+

Tall: BB

Dwarf: bb

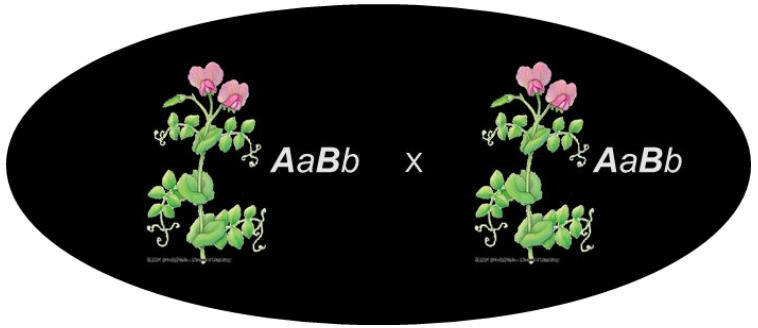
F_1 Results of Mendel's Dihybrid Crosses

AABB x ***aabb***

	ab	***ab***	***ab***	***ab***
AB	***AaBb***	***AaBb***	***AaBb***	***AaBb***
AB	***AaBb***	***AaBb***	***AaBb***	***AaBb***
AB	***AaBb***	***AaBb***	***AaBb***	***AaBb***
AB	***AaBb***	***AaBb***	***AaBb***	***AaBb***

- **All plants displays the dominant form of both traits (Purple and tall)**
- **We now know:**
  - **All plants inherited one allele for each trait from each parent**
  - **All plants were heterozygous (*AaBb*)**

Phenotypic Ratios in F_2



If the two traits are coded for by genes on separate chromosomes, sixteen allele combinations are possible:

	1/4 AB	1/4 Ab	1/4 aB	1/4 ab
1/4 AB	1/16 AABB	1/16 AABb	1/16 AaBB	1/16 AaBb
1/4 Ab	1/16 AABb	1/16 AAbb	1/16 AaBb	1/16 Aabb
1/4 aB	1/16 AaBB	1/16 AaBb	1/16 aaBB	1/16 aaBb
1/4 ab	1/16 AaBb	1/16 Aabb	1/16 aaBb	1/16 aabb

Four Phenotypes:

- Tall, purple-flowered (9/16)
- Tall, white-flowered (3/16)
- Dwarf, purple-flowered (3/16)
- Dwarf, white-flowered (1/16)

Phenotypic ratio: 9:3:3:1

Mendel's Law of Independent Assortment

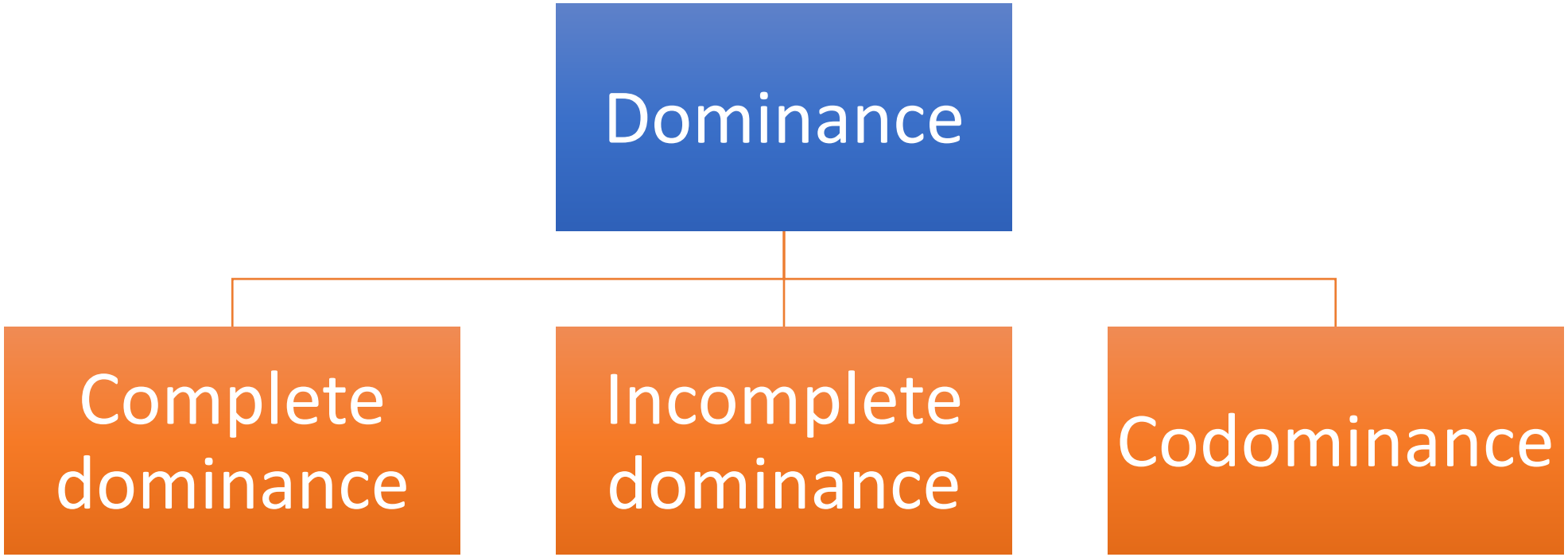
- Mendel concluded that the two "units" for the first trait were to be assorted into gametes independently of the two "units" for the other trait.

- Members of each pair of homologous chromosomes are sorted into gametes at random during meiosis.

MENDEL'S FIRST LAW
VERSUS
MENDEL'S SECOND LAW

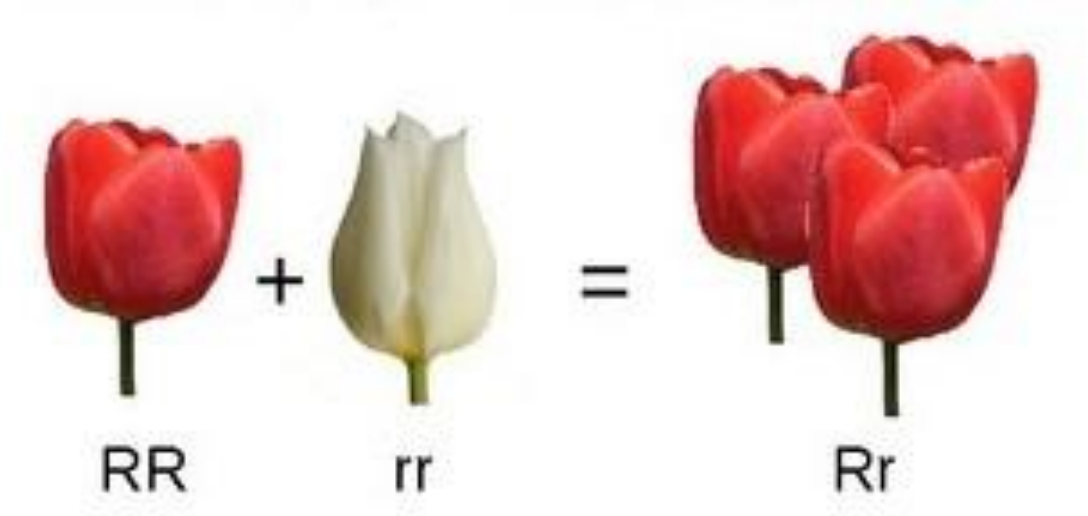
MENDEL'S FIRST LAW	MENDEL'S SECOND LAW
A principle that describes the separation of the two copies of each hereditary factor during the production of gametes	A principle that describes the independent assortment of alleles of different genes during the formation of gametes
Also called the law of segregation	Also called the law of independent assortment
Uses a monohybrid cross	Uses a dihybrid cross
Ratio of offspring is 3:1	Ratio of offspring is 9:3:3:1

Dominance Relations



Complete dominance

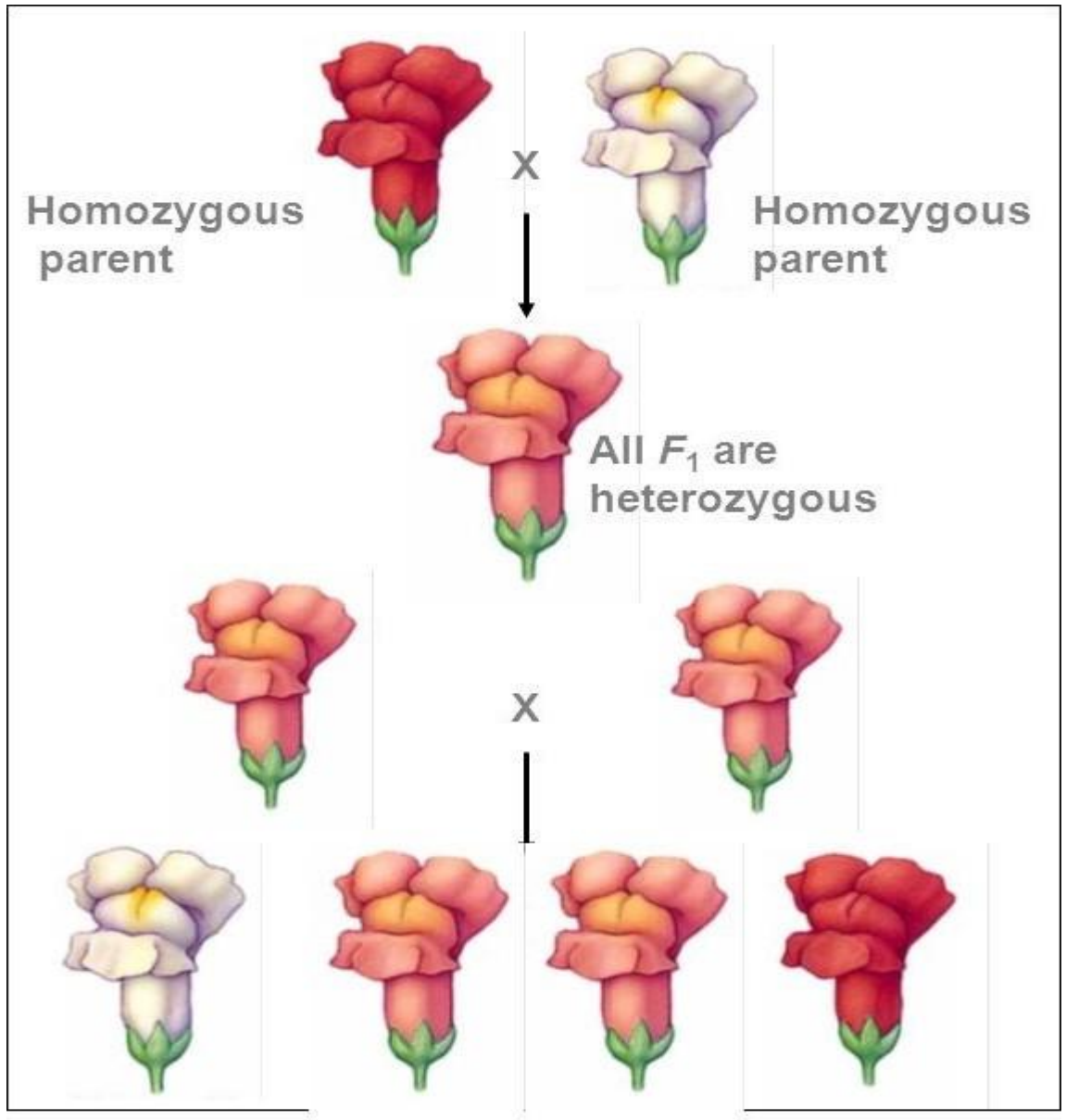
- Dominance in heterozygous condition where the dominant allele completely masks the effects of the recessive allele.



Incomplete dominance

- Dominant allele does not completely mask the effect of the recessive allele.
- Heterozygous phenotype is somewhere between that of two homozygotes.
- Blend of both traits

Flower colour in Snapdragons



Codominance

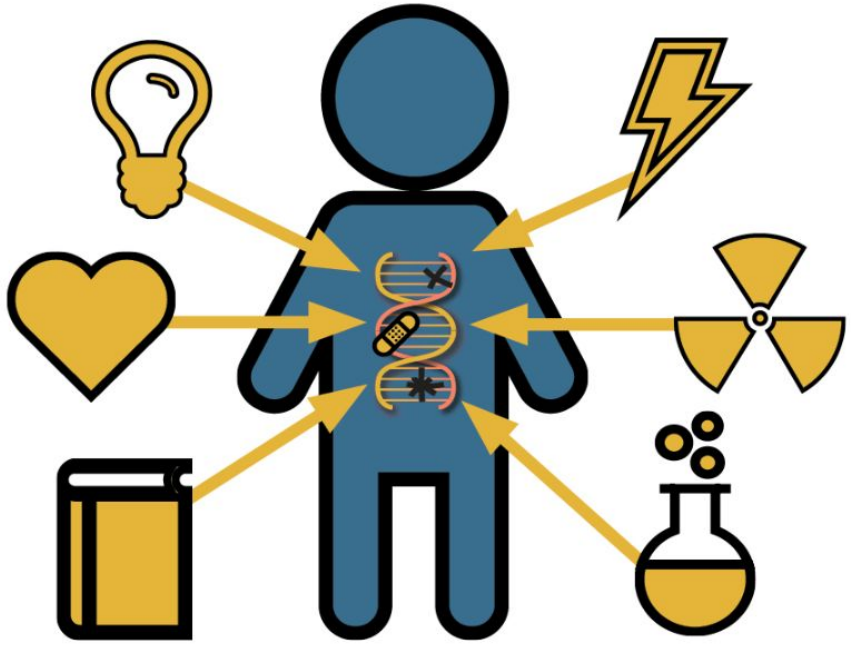
- Both alleles of a particular trait are fully expressed.
- Non-identical alleles specify two phenotypes that are both expressed in heterozygotes.
- Offspring has a phenotype that shows combination of both traits.

Epigenetics

- Epigenetics is the genetic control of a living organism through factors **other than the DNA sequence**

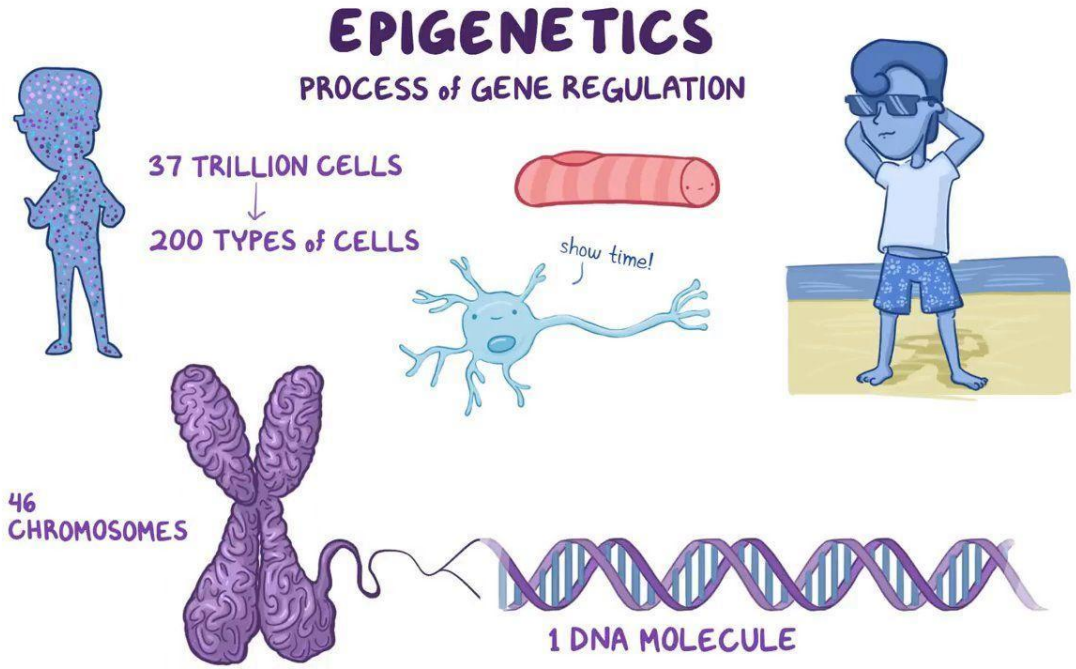
- During development, DNA accumulates chemical marks that affect how much of a gene is expressed. These chemical marks are called **epigenomes**

- Epigenetic changes can also switch certain genes ‘on’ or ‘off’!



How Epigenetics Works?

- All our cells have the same DNA, so why do we even have different cells?



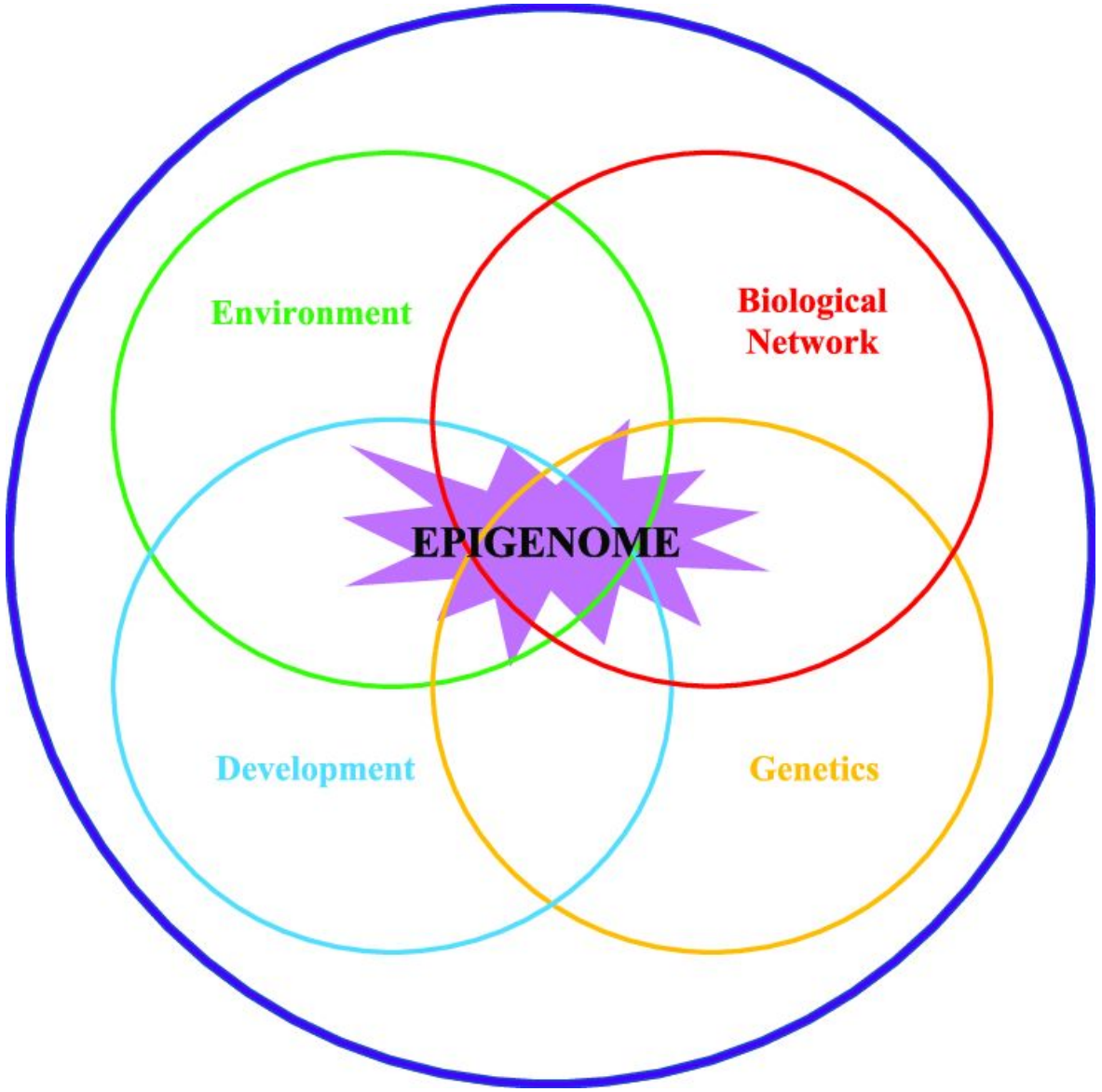
- Why are genotypically identical twins *not* phenotypically identical?

- What are the factors that cause people to develop differently?

Factors Affecting Epigenomes

Four key factors:

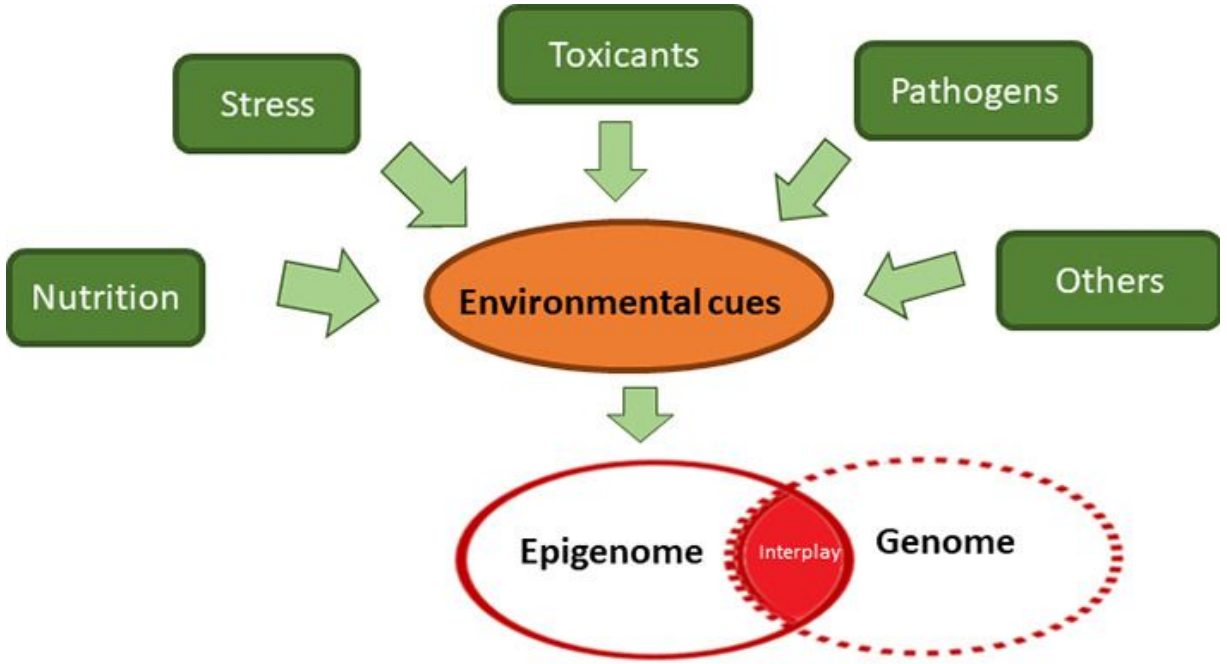
1. Genetics
2. Environment
3. Biological Networks
4. Development



Genetics and Environment

- The protein structure and arrangement of genes themselves can sometimes influence how the chemical marks are distributed across the DNA

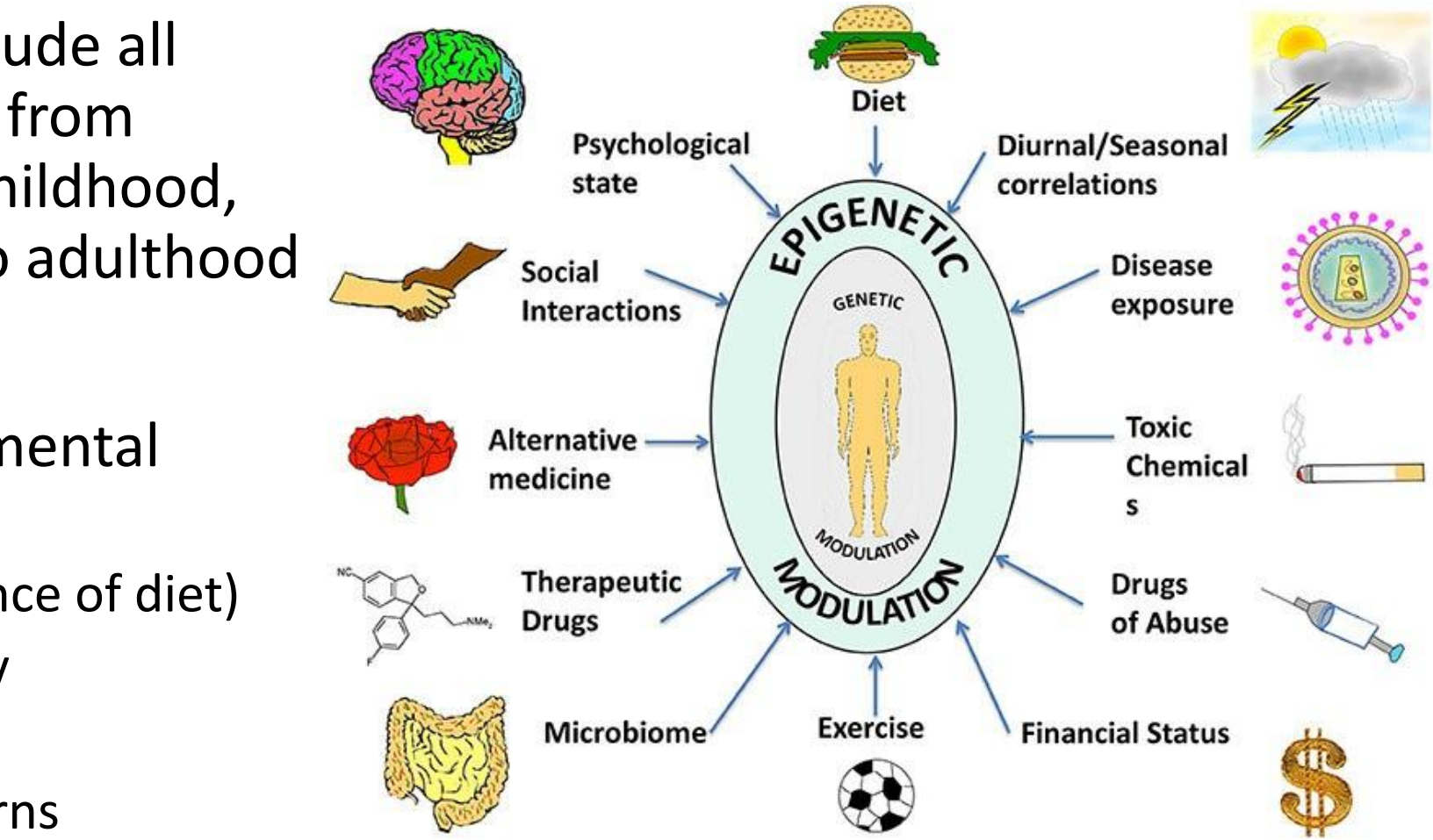
- Environmental factors consist of '<u>outside</u>' factors that can have an impact on genes. These can be:
  - Pollutants
  - Allergens
  - Heavy metals
  - Chemical toxins
  - Electromagnetic radiation



Developmental Factors

- Development can include all stages of life, starting from embryonic, to early childhood, all the way through to adulthood

- Examples of developmental factors are:
  - Nutrient intake (balance of diet)
  - Stress and psychology
  - Smoking and alcohol
  - Social behavior patterns



Case study – Identical Twins, Different behaviour?

- Suppose there is a pair of identical twins – they share the same genome, and thus have the same genotype

- However, one twin likes to read books and spend time on the computer growing up, while the other likes to play musical instruments and prefers to spend more time outdoors.

- <u>How is this difference explained through epigenetics?</u>