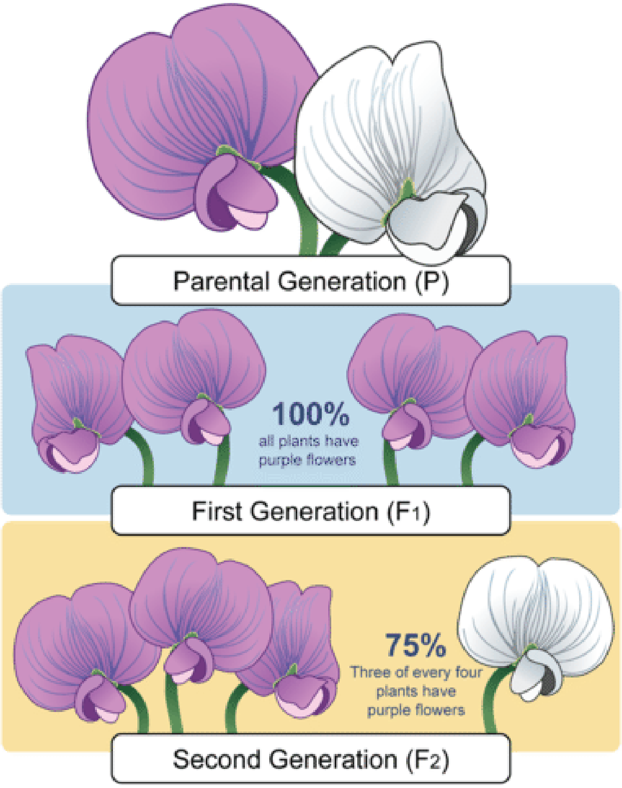
CLASSICAL GENETICS



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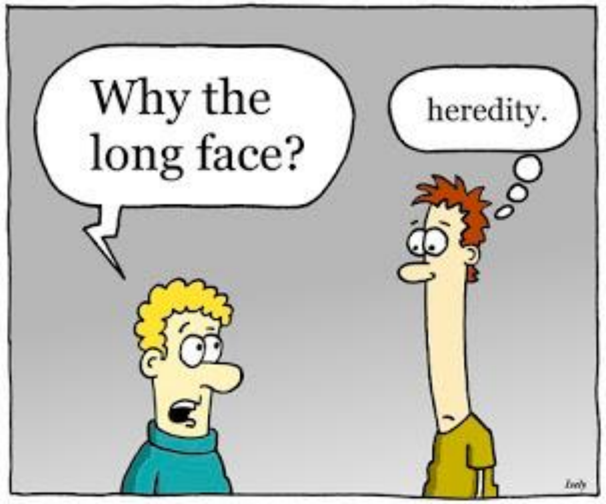
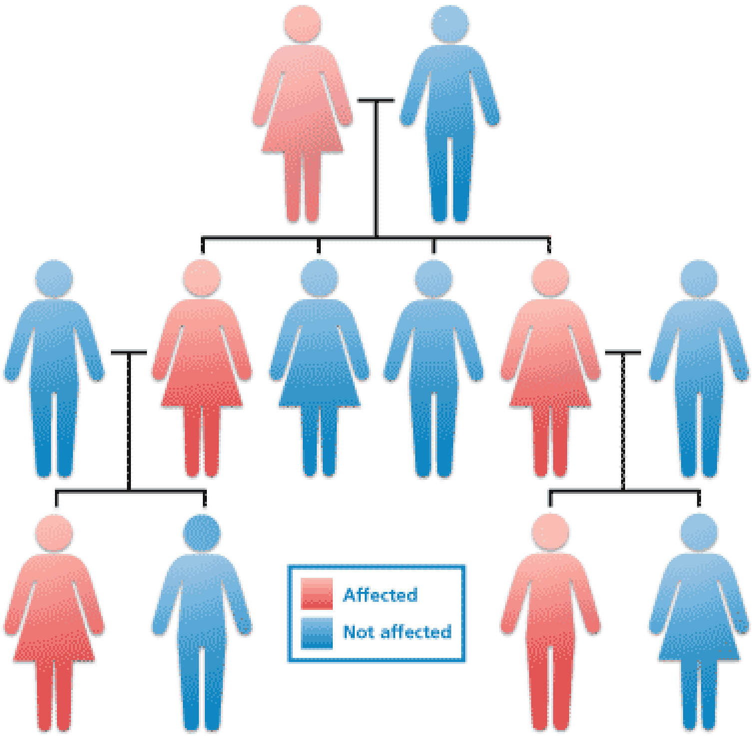
HEREDITY

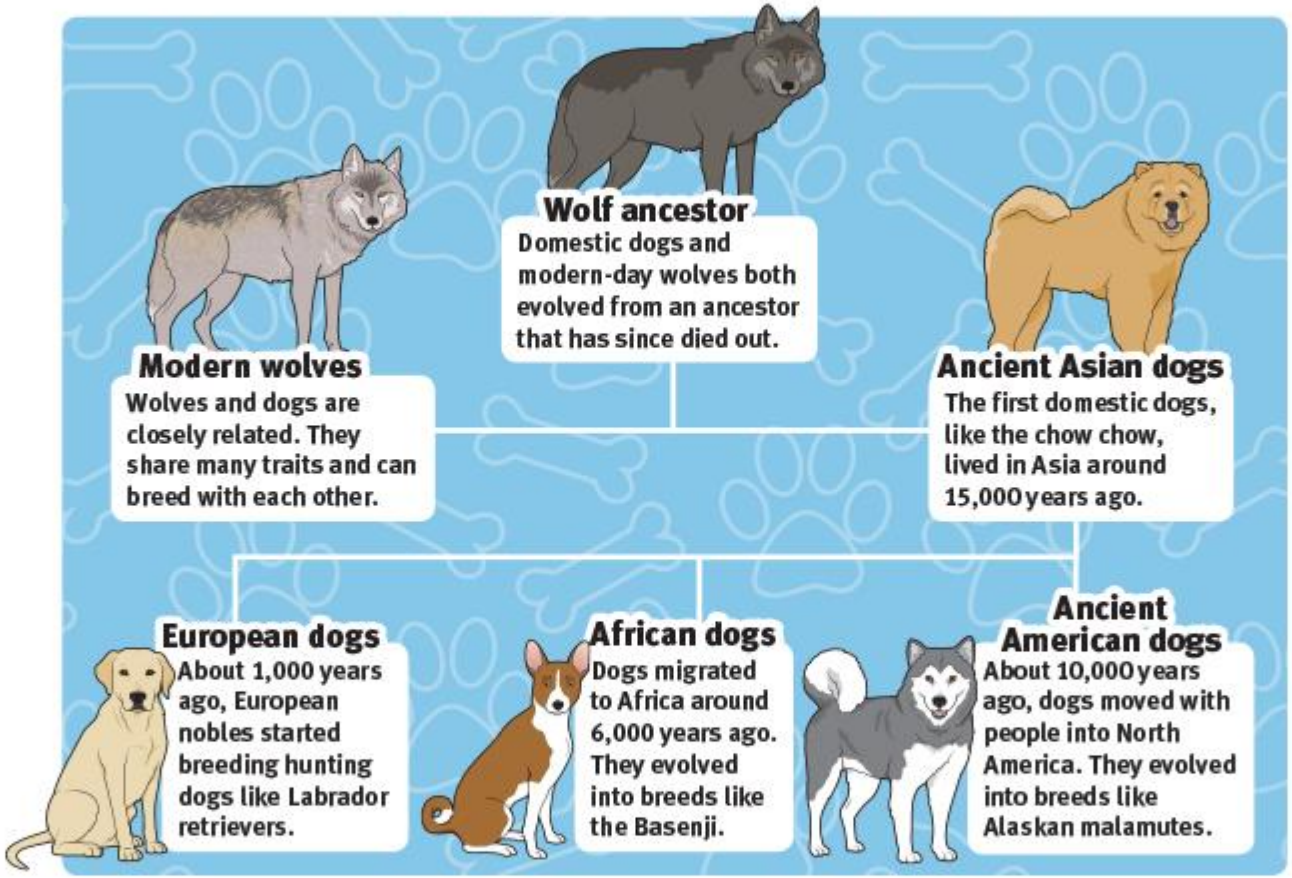
- Heredity- What we inherit from our parents!
- Our parents provided most of the information (in the sex cells) that governs our appearance, our activity, and our behavior → GENES.





Genetics is also seen as the study of Genes or genetic variation.

EARLY EVIDENCE

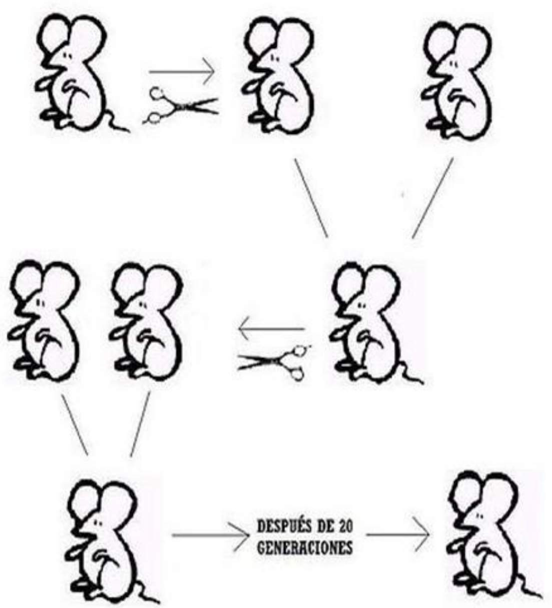


- Breeding and domestication
- ~ 8000-10,000 BC
- Camels, Horse, Oxen
- Various breeds of Wolves & Fox are derived over ages via artificial selection
- Evidence of plant breeding of wheat/ rice/ date palm

Key → Like the Trait → Breed!

EARLY IDEAS OF INHERITANCE

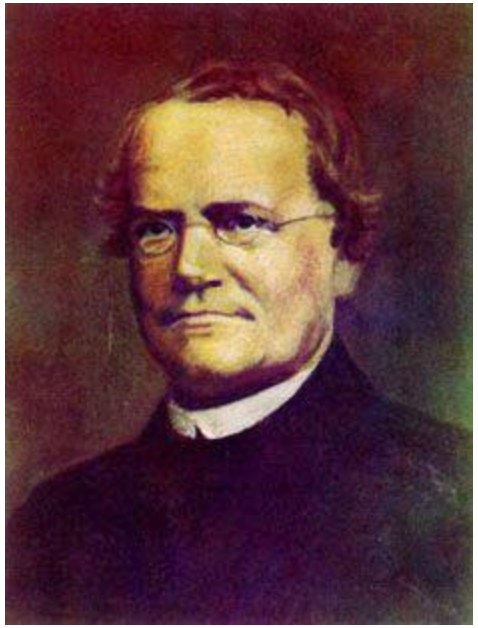
- Hippocrates (400BC)- seeds are produced by all body parts
- Formed the basis of Charles Darwin's proposed Theory of Inheritance:
- Jean Baptiste Lamarck- Theory of Pangenesis
- August Weissman:



- Weismann cut the tails off mice and bred them for over 20 generations. Each time the mice were born with tails.
- We now know that traits are passed on by genes which are <u>not</u> affected by life experiences.

Charles Darwin

MENDELIAN GENETICS

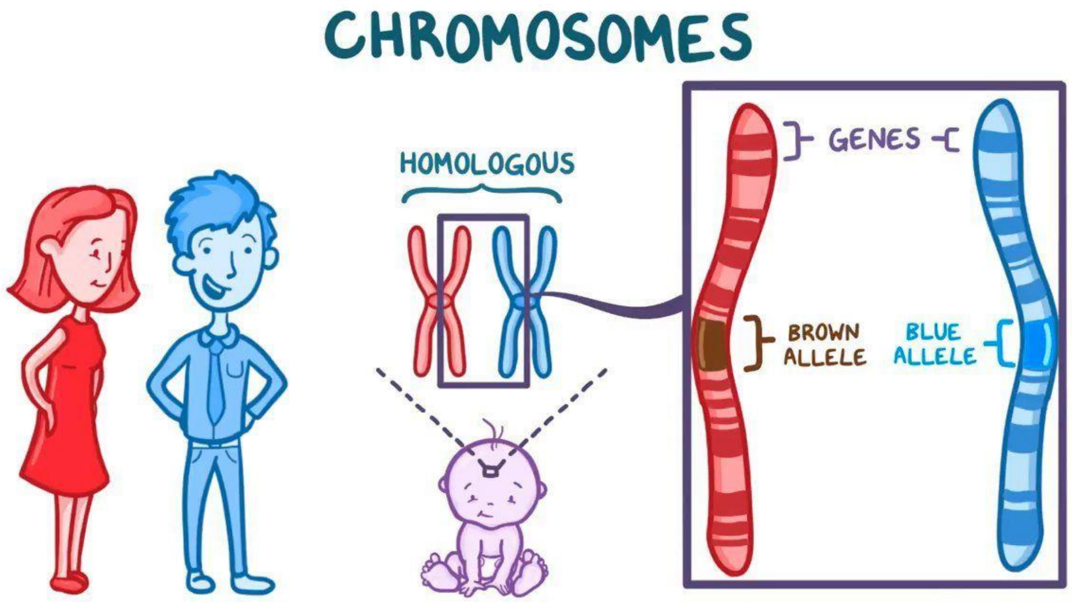


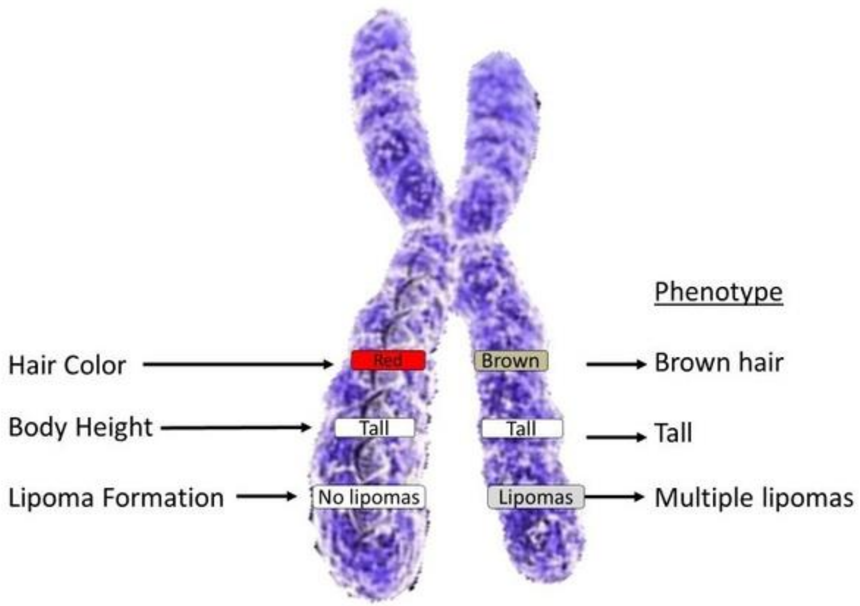
- Gregor Mendel- Father of Genetics
- Czech Republic born who later moved to Austria and became monk
- Studied in University of Vienna but did not receive degree
- Studied heredity in peas in his monastery, carefully choosing traits that did not appear to blend. Collected data from 1856 - 1865.
- Mendel was the first to follow single traits from generation to generation instead of trying to document and follow every trait in the plants.

GMI
GREGOR MENDEL INSTITUTE
OF MOLECULAR PLANT BIOLOGY

TERMINOLOGY

- Genes consist of a polynucleotide chain called **DNA.**
- It carries recipe to make **Protein.**
- DNA is organized into **chromosomes**.



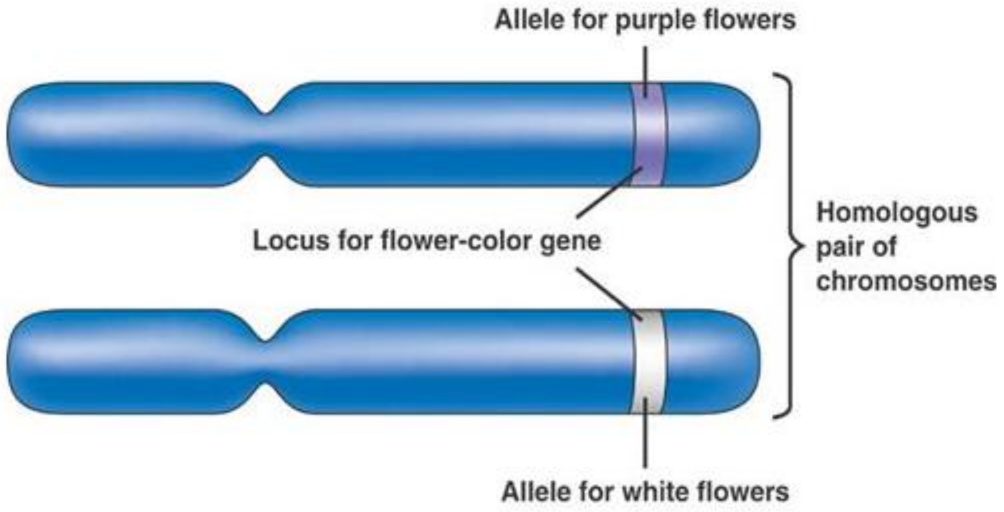


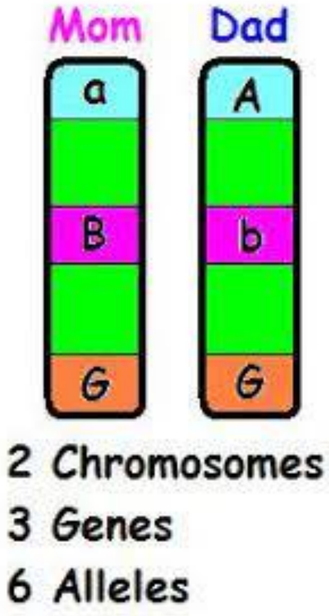
– **Characteristic** (Character) / Gene
  - a heritable feature that varies among individuals (e.g.eye colour, flower colour, etc.)

– **Trait** / Allele
  - Each variant of a characteristic (e.g. blue vs. brown eyes, purple vs. white flowers)

GENE AND ALLELE

- Gene: a heritable factor that controls a specific characteristic
  - Located in specific places on chromosomes
  - Humans have roughly 20,000-25,000 genes.
- Alleles: alternative forms of gene
- Example:
  - Gene: eye color
  - Alleles: blue vs. brown

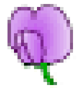
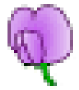
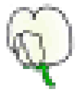




GENOTYPE VS PHENOTYPE

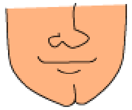
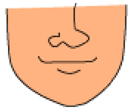
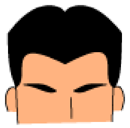
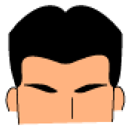
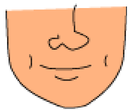
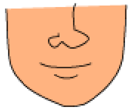
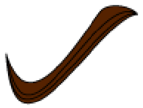
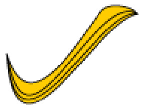
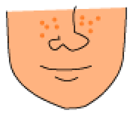
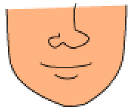
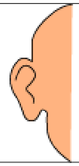
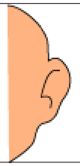
- The **genotype** is the genetic makeup of an organism.
  - If an individual has two identical alleles of a certain gene, the individual is **homozygous** for the related character.
  - If an individual has two different alleles of a certain gene, the individual is **heterozygous** for the related character.
- The **phenotype** is the appearance of an organism.
  - Thus, genotype determines phenotype.

Genotype vs. Phenotype

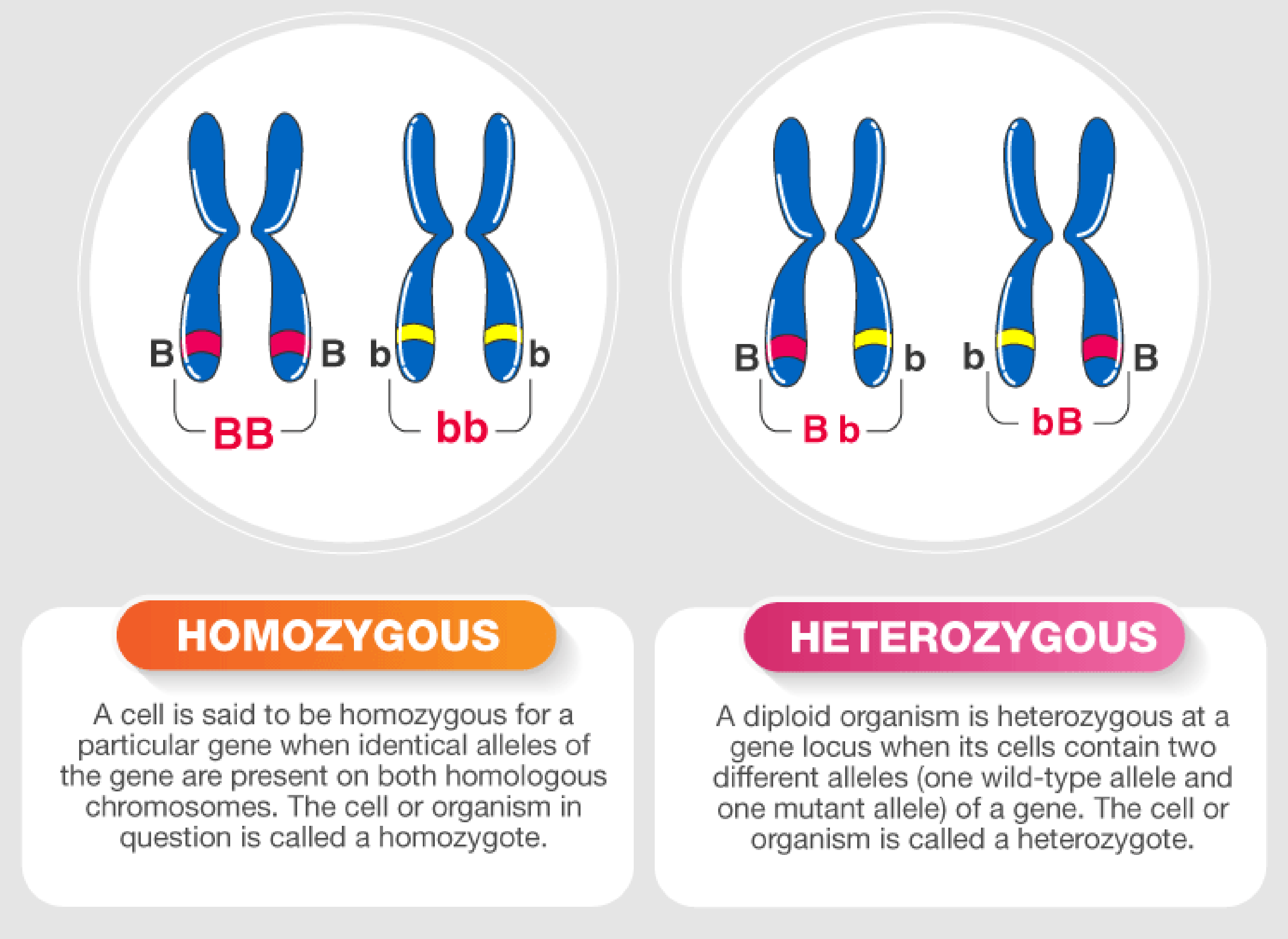
Genotype	Phenotype
BB Homozygous dominant	
Bb Heterozygous	
bb Homozygous recessive	

Both alleles contribute to the genotype

PHENOTYPE

Dominant Gene		Recessive Gene	
Cleft Chin		No Cleft	
Widow's Peak		No Widow's Peak	
Dimples		No Dimples	
Brown/Black Hair		Blonde Hair	
Freckles		No Freckles	
Brown Eyes		Gray/Blue Eyes	
Free Earlobe		Attached Earlobe	

GENOTYPE


B
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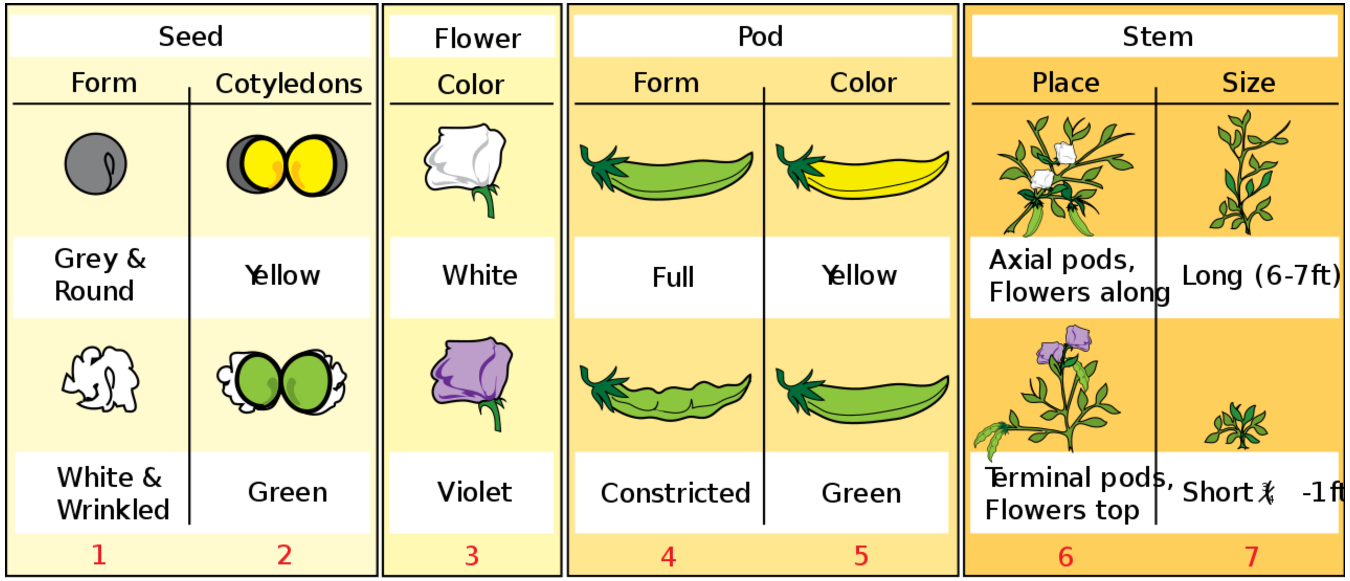
HOMOZYGOUS
A cell is said to be homozygous for a particular gene when identical alleles of the gene are present on both homologous chromosomes. The cell or organism in question is called a homozygote.
HETEROZYGOUS
A diploid organism is heterozygous at a gene locus when its cells contain two different alleles (one wild-type allele and one mutant allele) of a gene. The cell or organism is called a heterozygote.

MENDEL AND THE PEAS

Mendel's Law of Genetics:

- Law of Dominance
- Law of Segregation
- Law of Independent Assortment



MENDEL AND THE PEAS

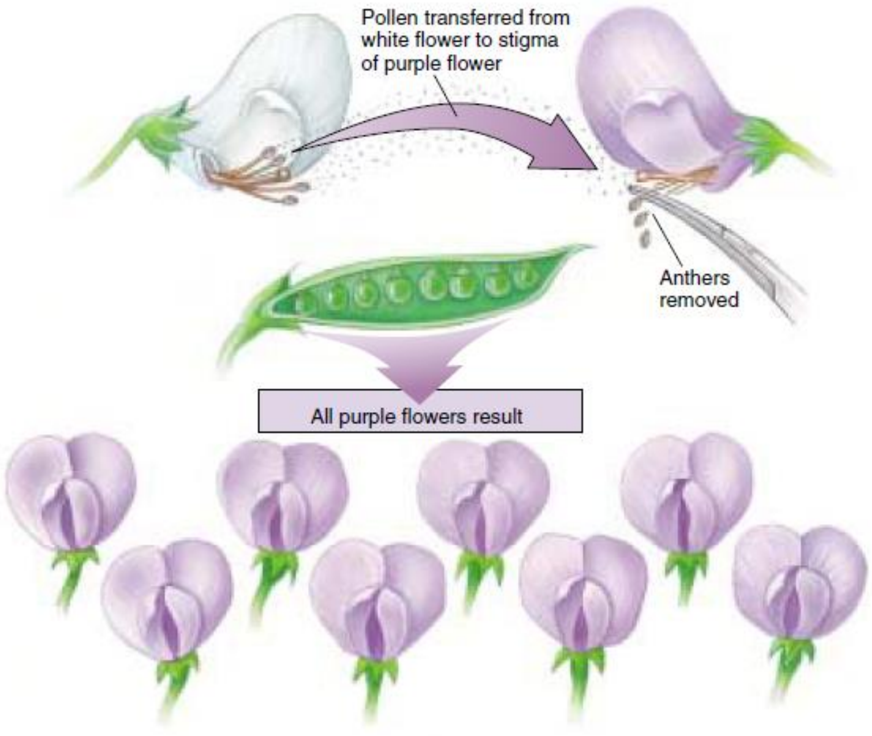
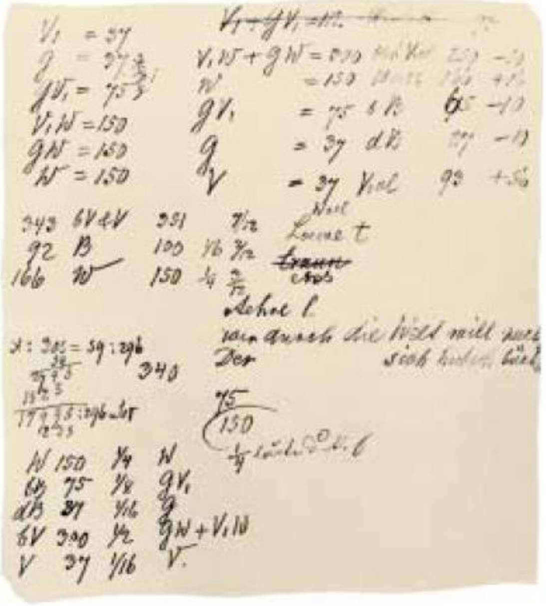
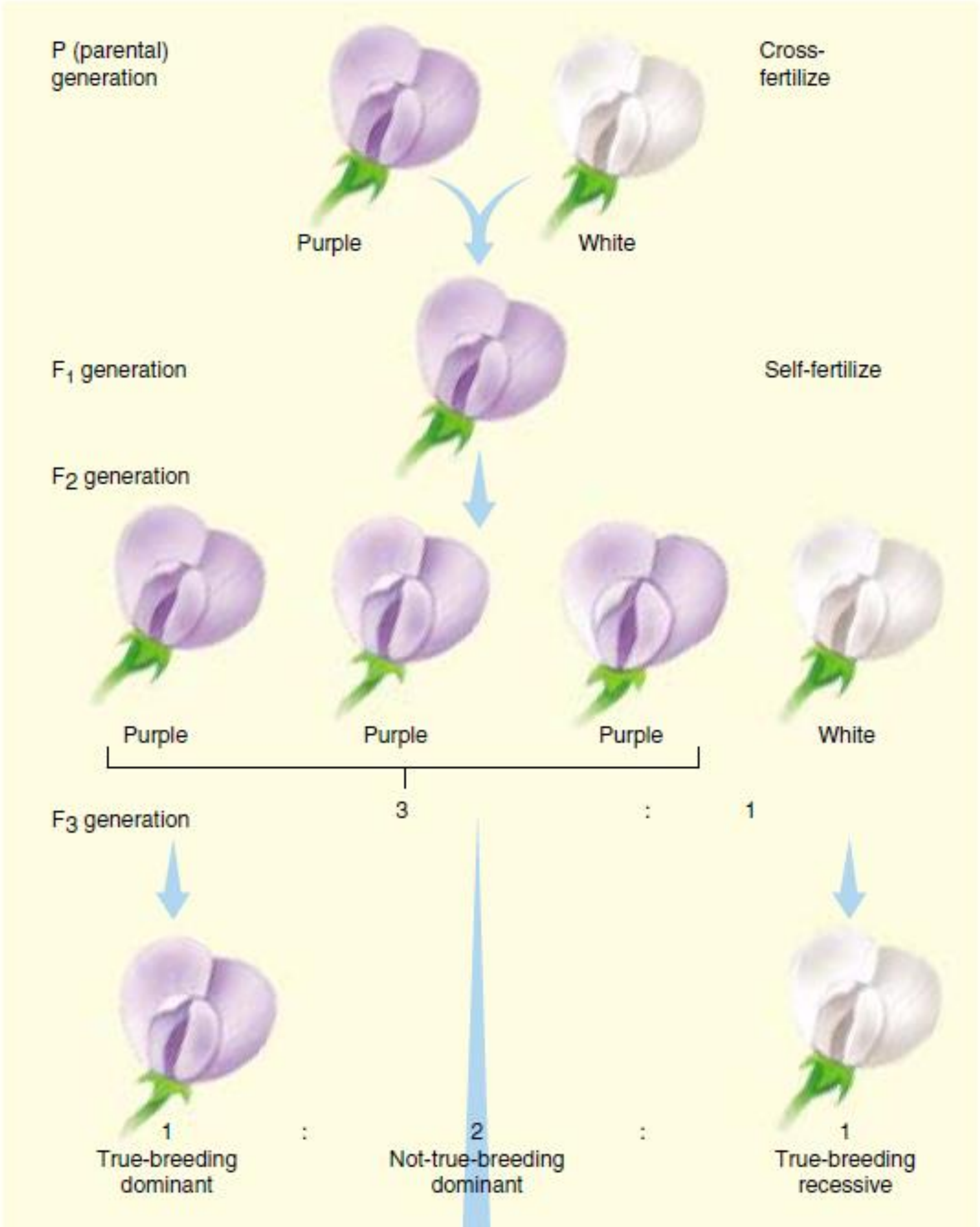


FIGURE 13.8
How Mendel conducted his experiments. Mendel pushed aside the petals of a white flower and collected pollen from the anthers. He then placed that pollen onto the stigma (part of the carpel) of a purple flower whose anthers had been removed, causing cross-fertilization to take place. All the seeds in the pod that resulted from this pollination were hybrids of the white-flowered male parent and the purple-flowered female parent. After planting these seeds, Mendel observed the pea plants they produced. All of the progeny of this cross had purple flowers.



GENERATIONS

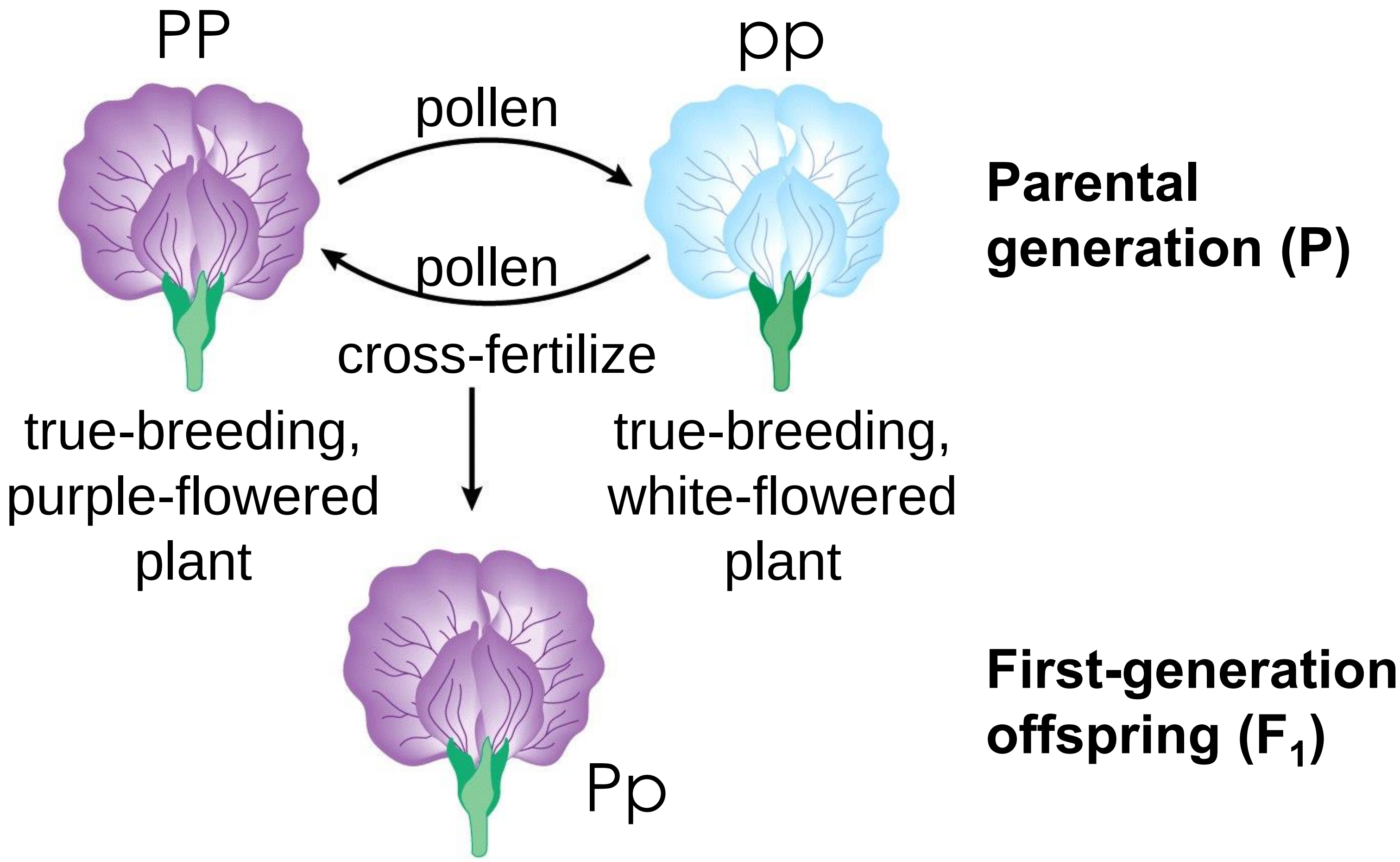


GAMETE FORMATION

LAW OF DOMINANCE

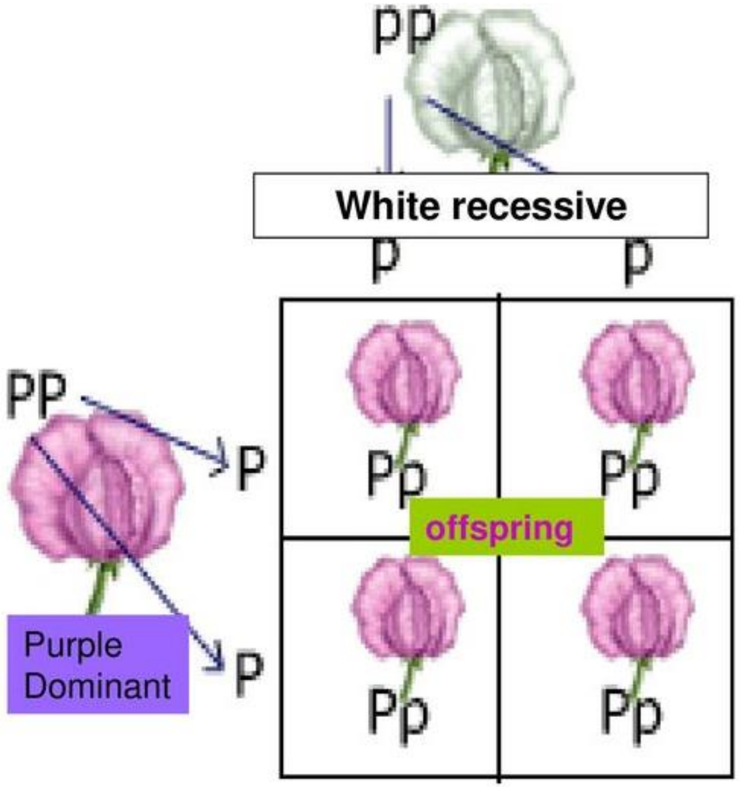
Law of purity of gametes:

- Traits are controlled by two factors that can be called "dominant" or "recessive."
- A "dominant" trait shows if the offspring inherits at least one dominant factor from one parent.
- A "recessive" trait shows only if the offspring inherits two recessive factors, one from each parent.

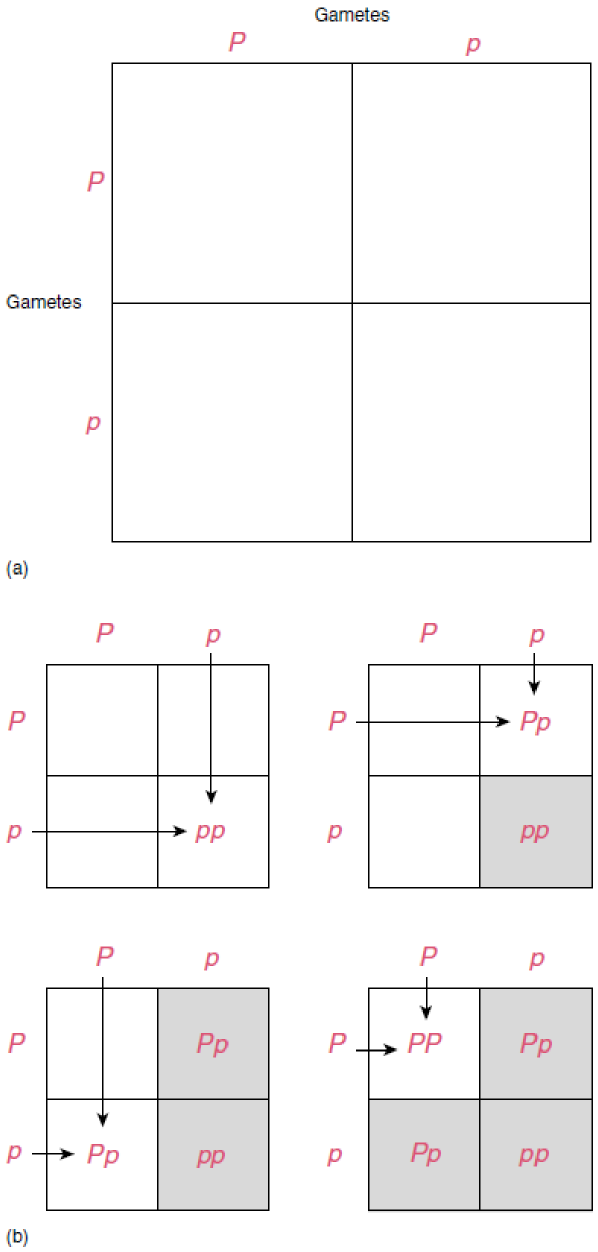


LAW OF DOMINANCE

- We use a punnett square to show the results:
- Two purebred parents are crossed
- One parent has PP it has purple flowers
- The other has pp white flowers
- A dominant trait is a capital letter P
- A recessive trait is a small letter p
- The offspring are all purple. Because they have one dominant gene P. Even though they have a p gene.



PUNNETT SQUARE

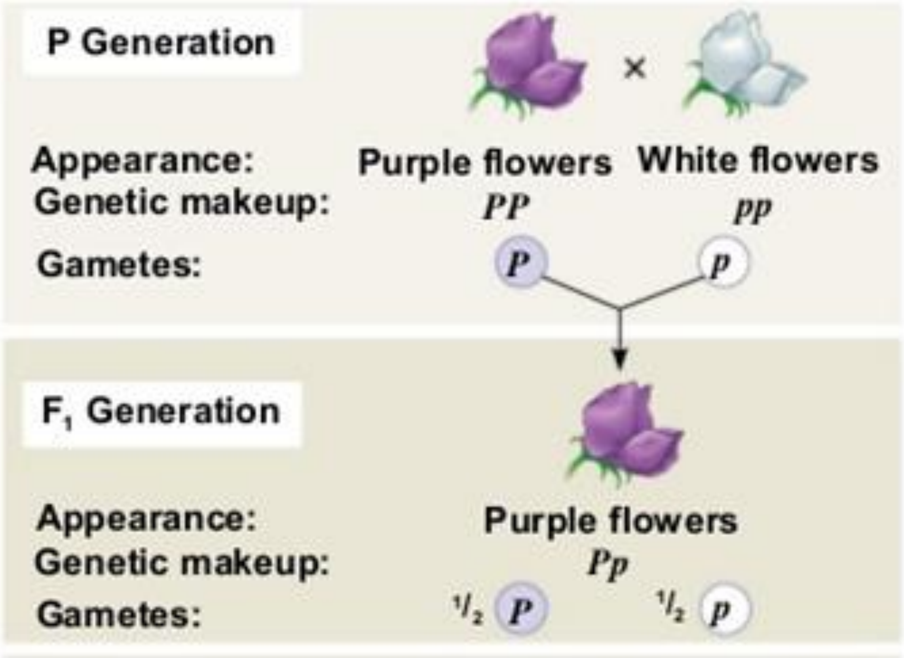


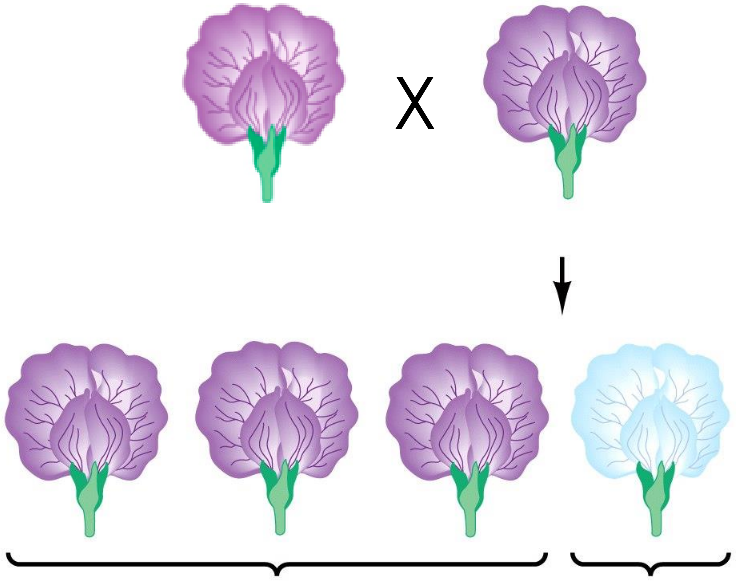
A Punnet square is one way to predict the outcome of a cross by showing all the possible combinations of all the possible gametes.

BLOOD GROUP

LAW OF SEGREGATION

- Each individual has a pair of factors controlling each trait, one inherited from each biological parent.
- During the formation of gametes (sex cells) these two factors separate. Only one ends up in each sex cell.



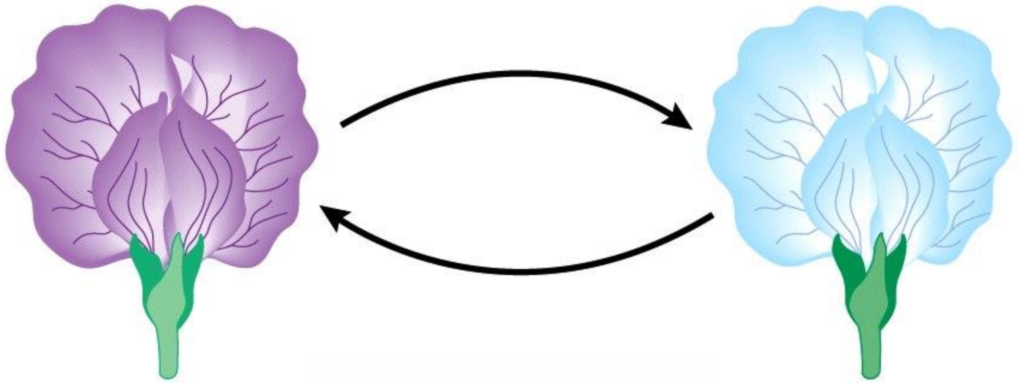


In this cross between two purple-flowered pea plants, one-quarter of the offspring have white flowers.

Which color is dominant?

The purple-flowered trait is dominant because each an individual who inherits at least one copy of the purple allele (P) shows the purple phenotype.

The white-flowered trait is recessive because an individual must inherit two copies of the white allele (p) to show the white phenotype.



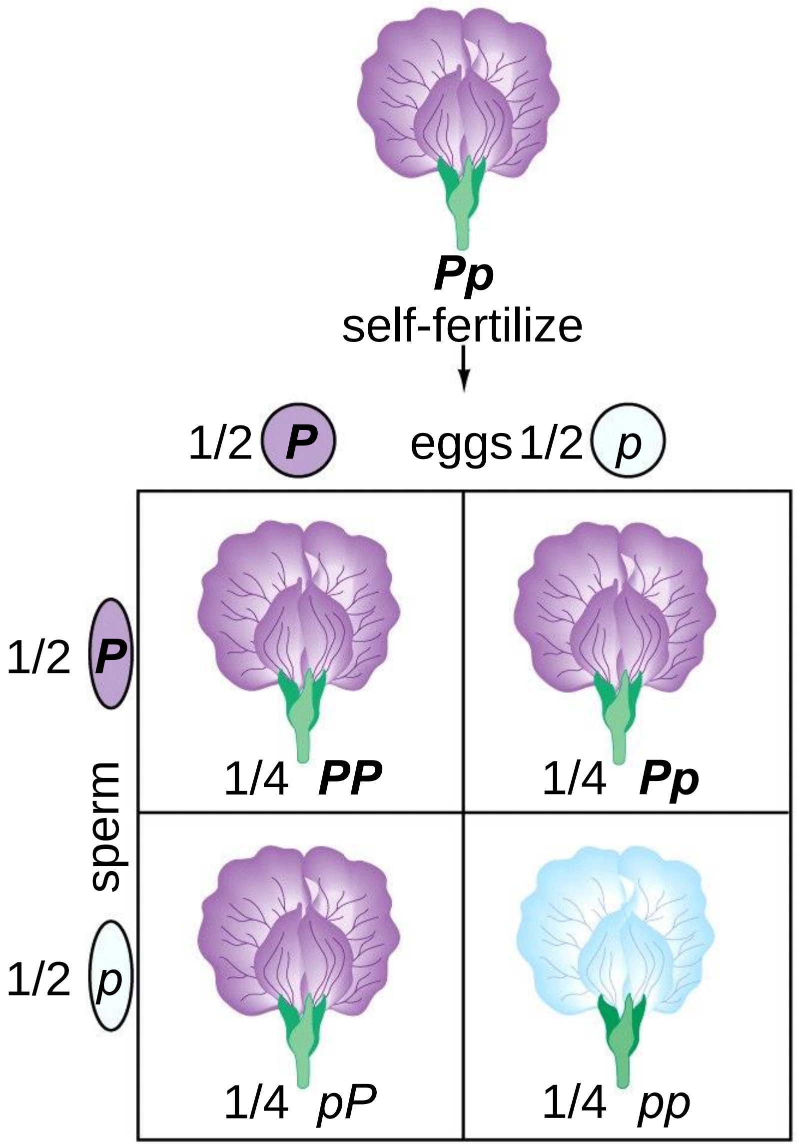
genotypes:	PP or Pp	pp
phenotype	purple	white

Same letter, different case = same gene, different allele

LAW OF SEGREGATION

- Chromosomes, which carry genes, separate from one another during gamete formation.
- Chromosomes sort independently of one another during gamete formation, but each gamete gets ONE of each kind of chromosome.

Phenotype- 3 Purple: 1 White
Genotype- PP: Pp: pP: pp
Genotype- 1:2:1



Punnett Square

LAW OF SEGREGATION

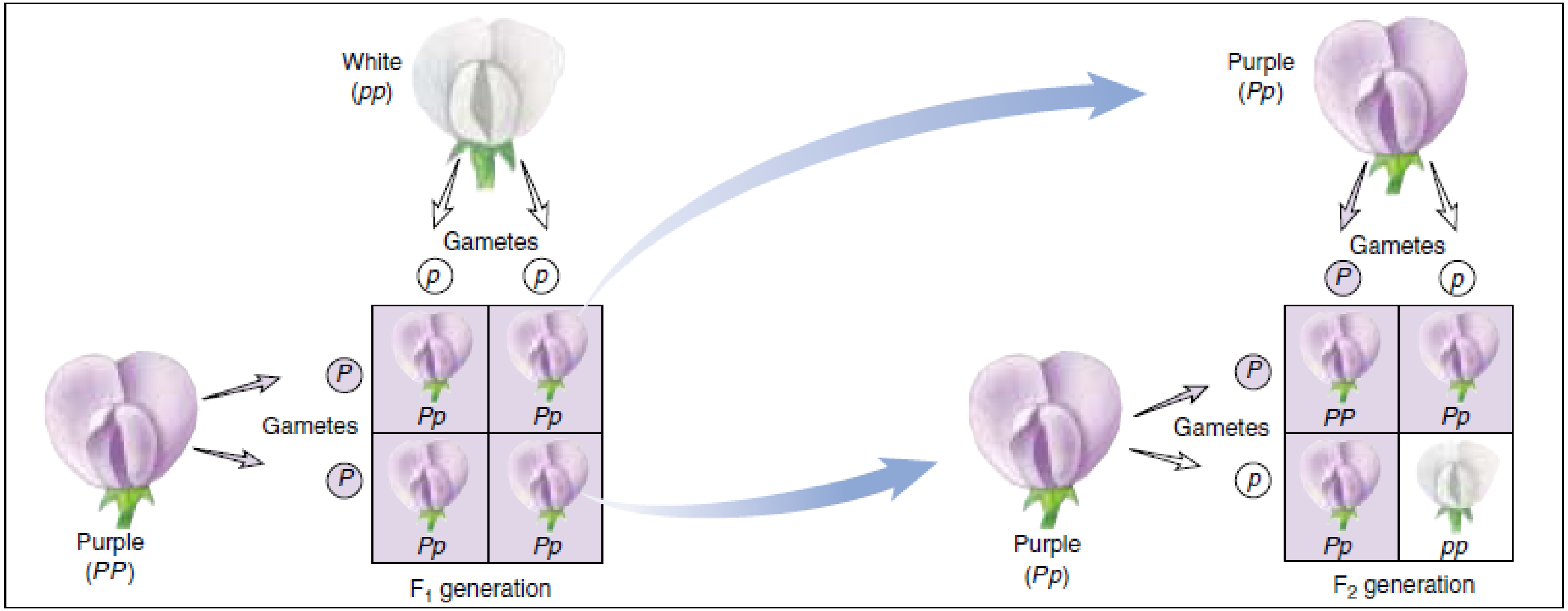
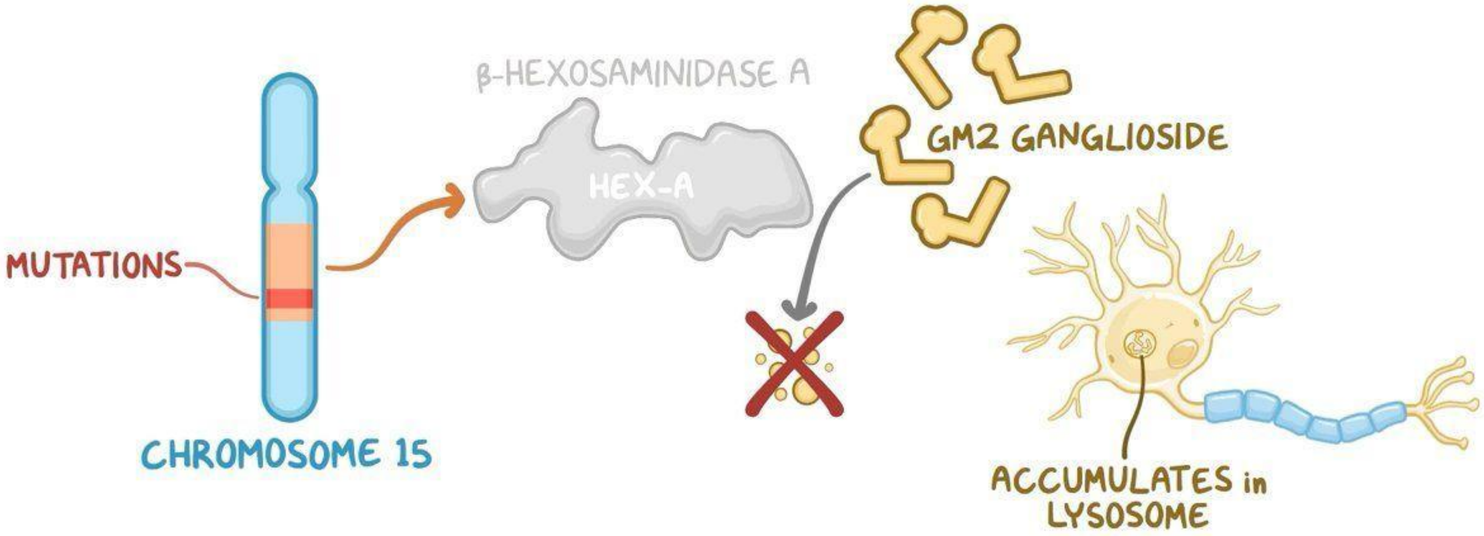


FIGURE 13.14
Mendel's cross of pea plants differing in flower color. All of the offspring of the first cross (the F_1 generation) are *Pp* heterozygotes with purple flowers. When two heterozygous F_1 individuals are crossed, three kinds of F_2 offspring are possible: *PP* homozygotes (purple flowers); *Pp* heterozygotes (also purple flowers); and *pp* homozygotes (white flowers). Therefore, in the F_2 generation, the ratio of dominant to recessive phenotypes is 3:1. However, the ratio of genotypes is 1:2:1 (1 *PP*: 2 *Pp*: 1 *pp*).

TAY-SACHS DISEASE (TSD)

* LYSOSOMAL STORAGE DISORDER



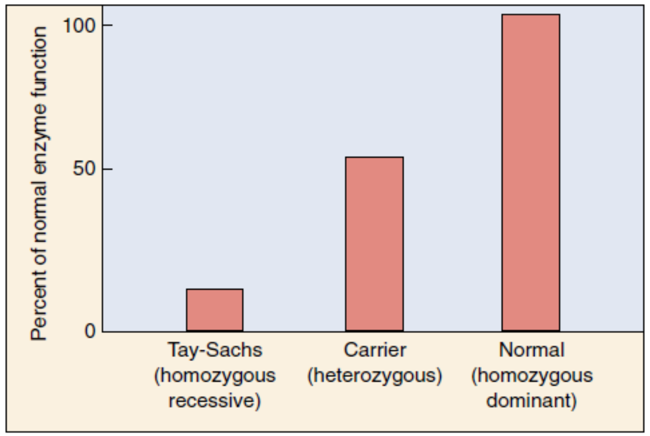
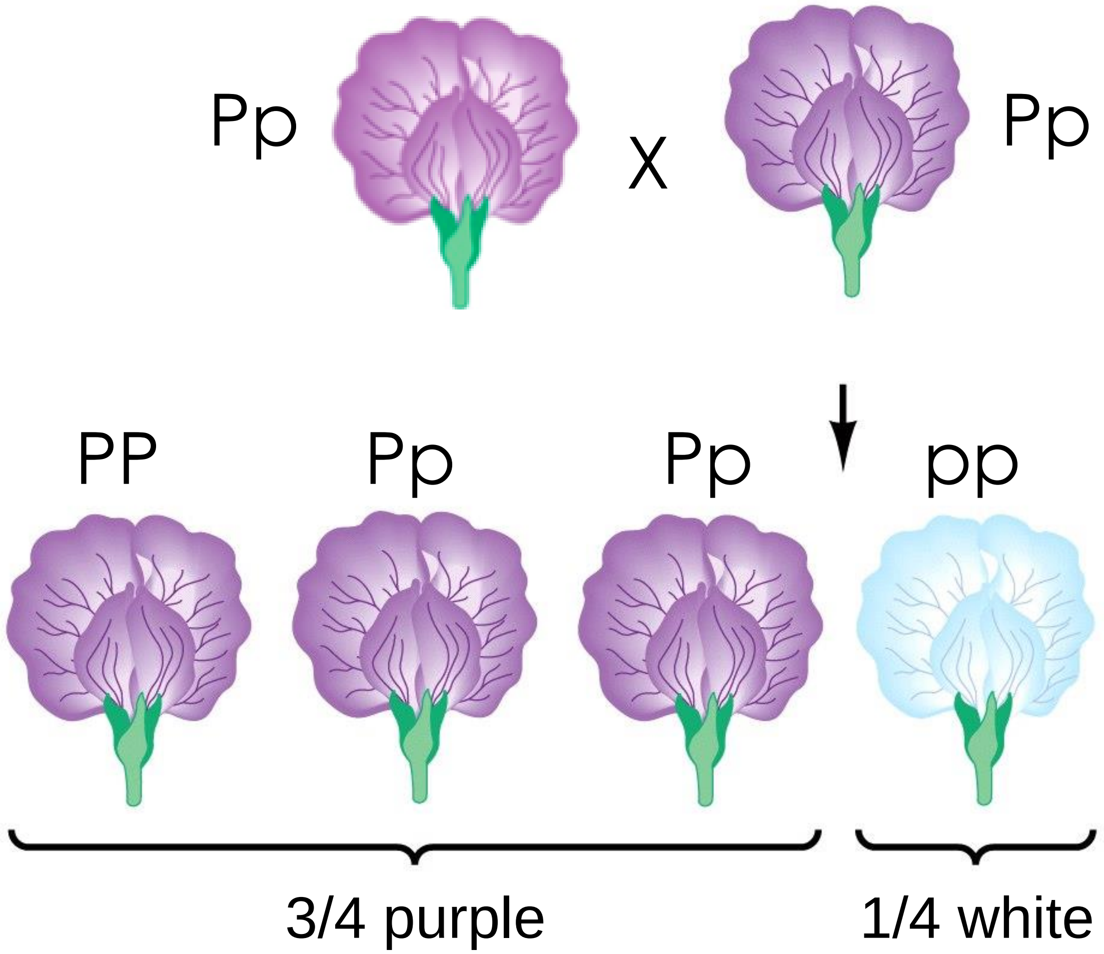


FIGURE 13.22
Tay-Sachs disease. Homozygous individuals (*left bar*) typically have less than 10% of the normal level of hexosaminidase A (*right bar*), while heterozygous individuals (*middle bar*) have about 50% of the normal level—enough to prevent deterioration of the central nervous system.

A genetic metabolic disorder caused by deficiency of the enzyme hexosaminidase A (hex-A) that results in a failure to process a lipid called GM2 ganglioside that accumulates in the brain and other tissues.

LAW OF SEGREGATION

Offspring of the F1 generation (the hybrids) may be purple-flowered if they inherit at least one factor for purple flowers, or may be white flowered if they inherit the white factor from both parents.



First-generation offspring (F_1)

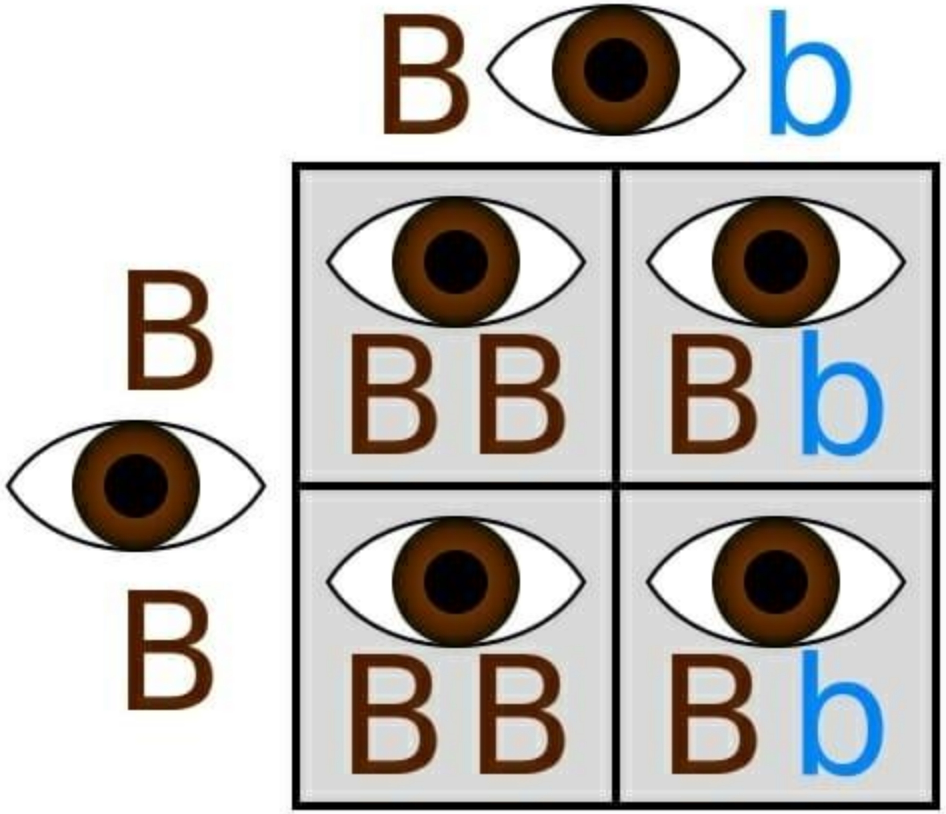
Second-generation offspring (F_2)

TEST:

- Brown Eye (BB) X Brown Eye (Bb)
- Hint bb= Blue Eye

TEST:

- Brown Eye (BB) X Brown Eye (Bb)
- Hint bb= Blue Eye



TEST CROSS

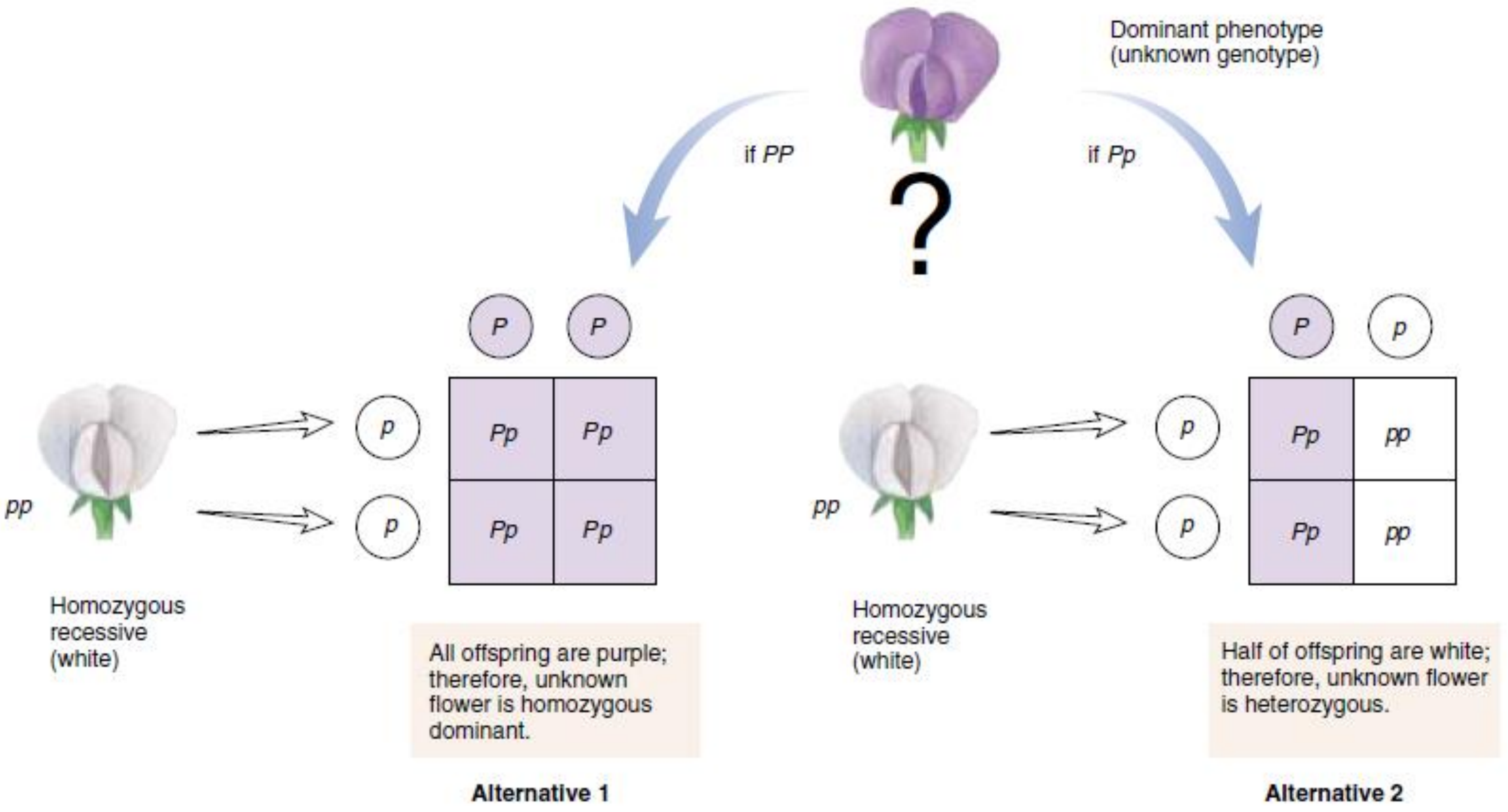
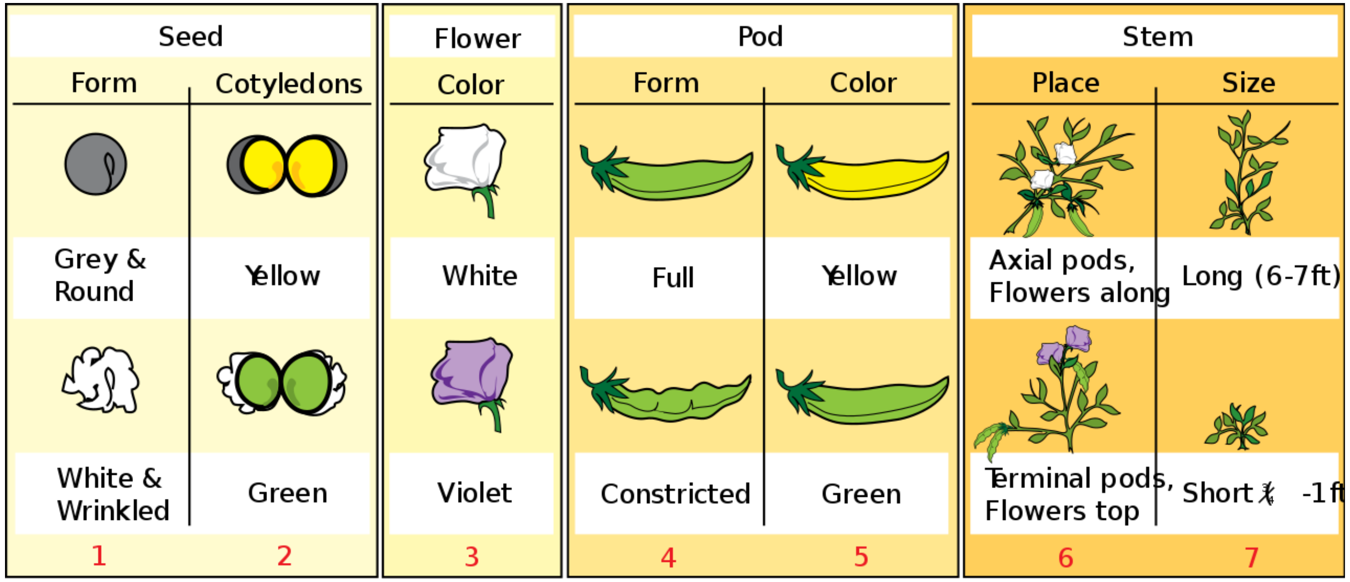


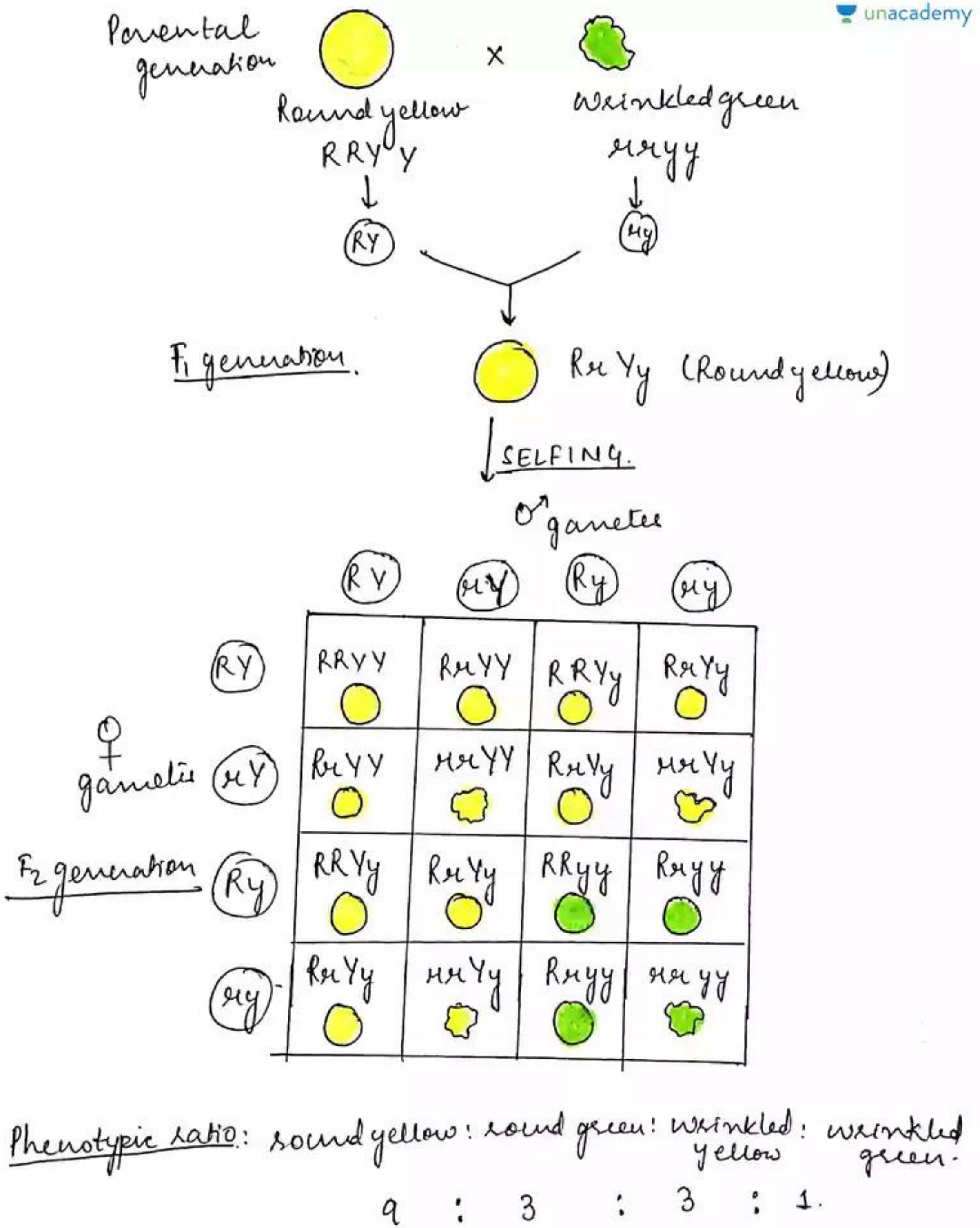
FIGURE 13.15
A testcross. To determine whether an individual exhibiting a dominant phenotype, such as purple flowers, is homozygous or heterozygous for the dominant allele, Mendel crossed the individual in question with a plant that he knew to be homozygous recessive, in this case a plant with white flowers.

LAW OF INDEPENDENT ASSORTMENT

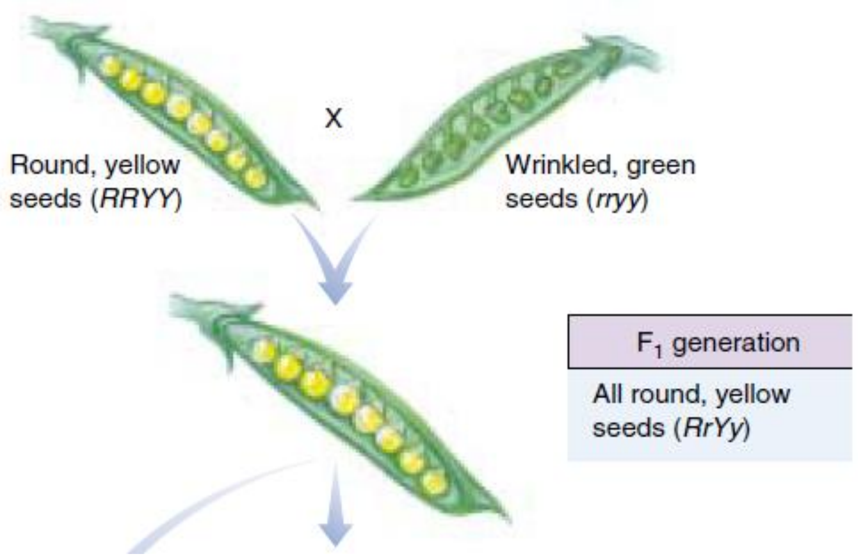
- When genetic factors segregate in the gametes, they segregate independently of one another. A dominant allele for one trait does not guarantee inheritance of a dominant allele for a different trait.



LAW OF INDEPENDENT ASSORTMENT

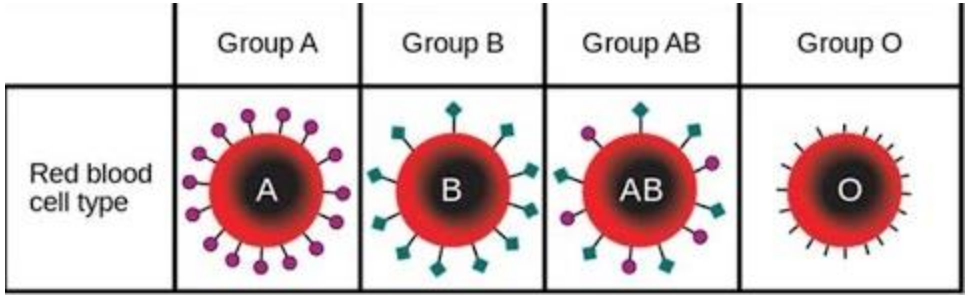


LAW OF INDEPENDENT ASSORTMENT



CODOMINANCE

- Most genes exist in populations in more than two allelic forms
- For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme (I) that attaches A or B carbohydrates to red blood cells: I^A, I^B, and i.
- The enzyme encoded by the I^A allele adds the A carbohydrate, whereas the enzyme encoded by the I^B allele adds the B carbohydrate; the enzyme encoded by the i allele adds neither

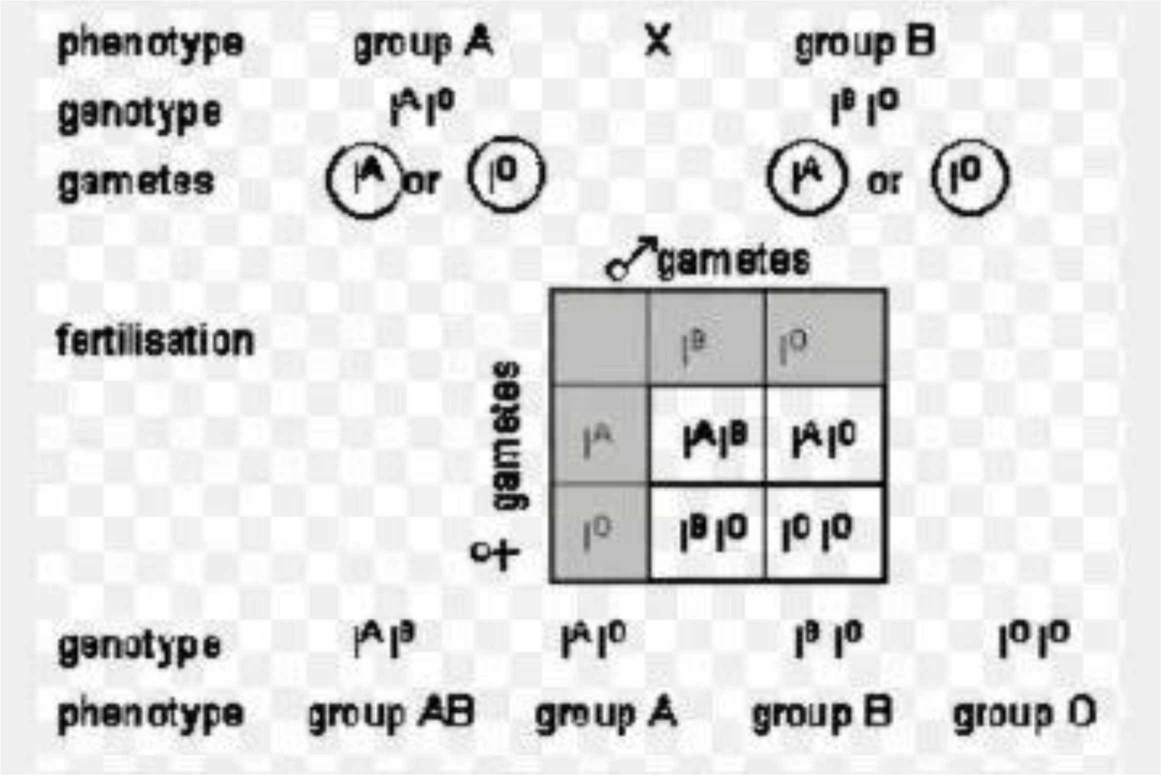


Blood type	Genotype	
A	I^A, I^O I^A, I^A	AO AA
B	I^B, I^O I^B, I^B	BO BB
AB	I^A, I^B	AB
O	I^O I^O	OO

MULTIPLE ALLELES

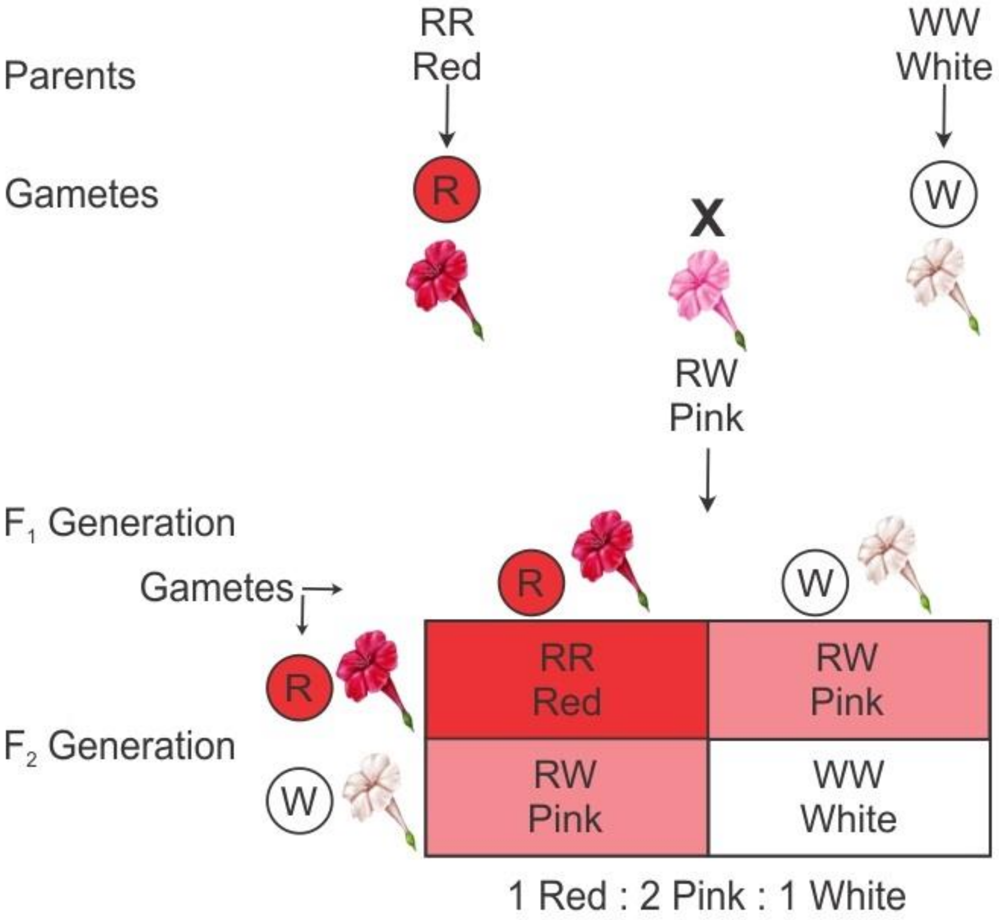
phenotype	group A	X	group B
genotype	$I^A I^O$		$I^B I^O$

MULTIPLE ALLELES

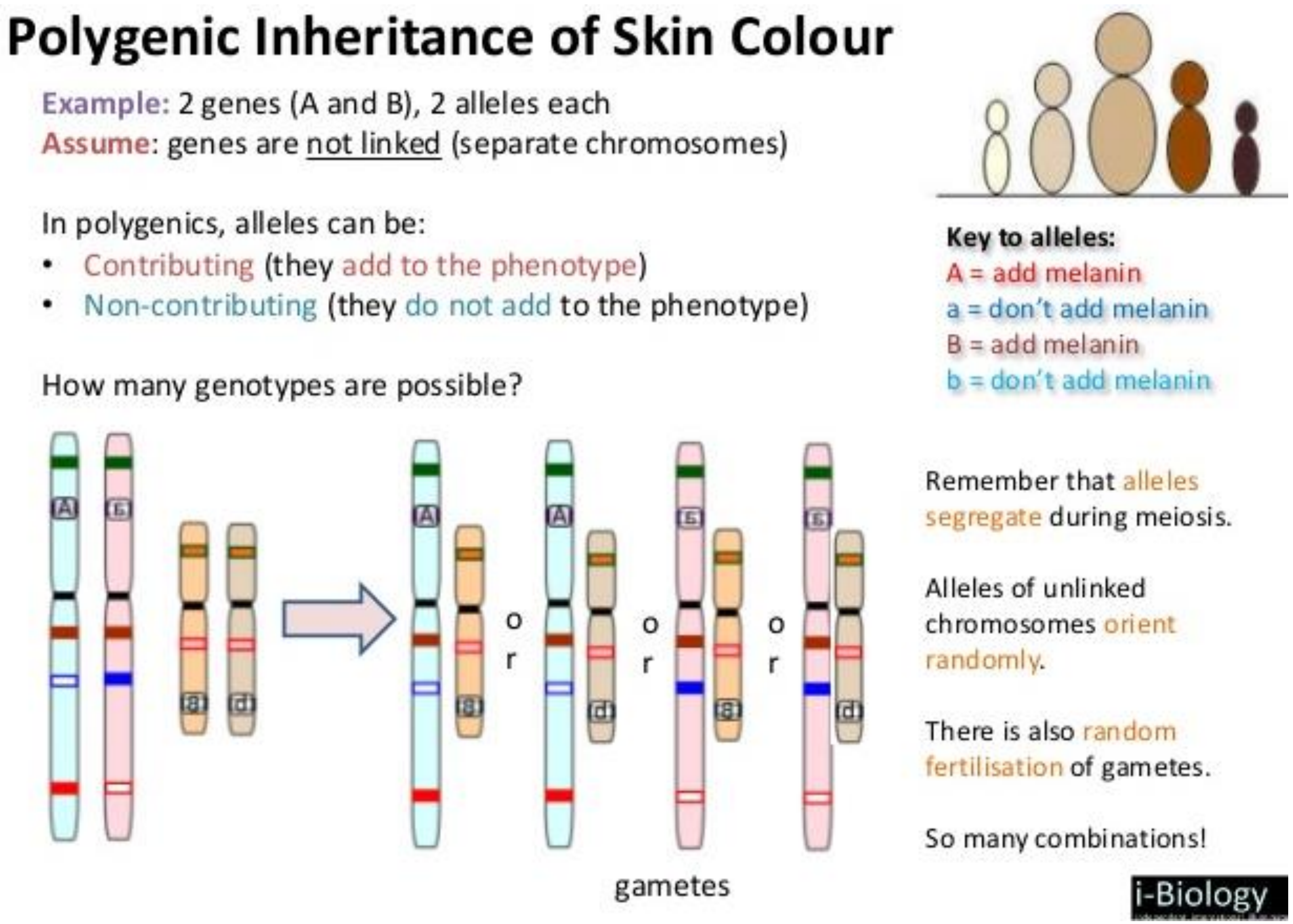


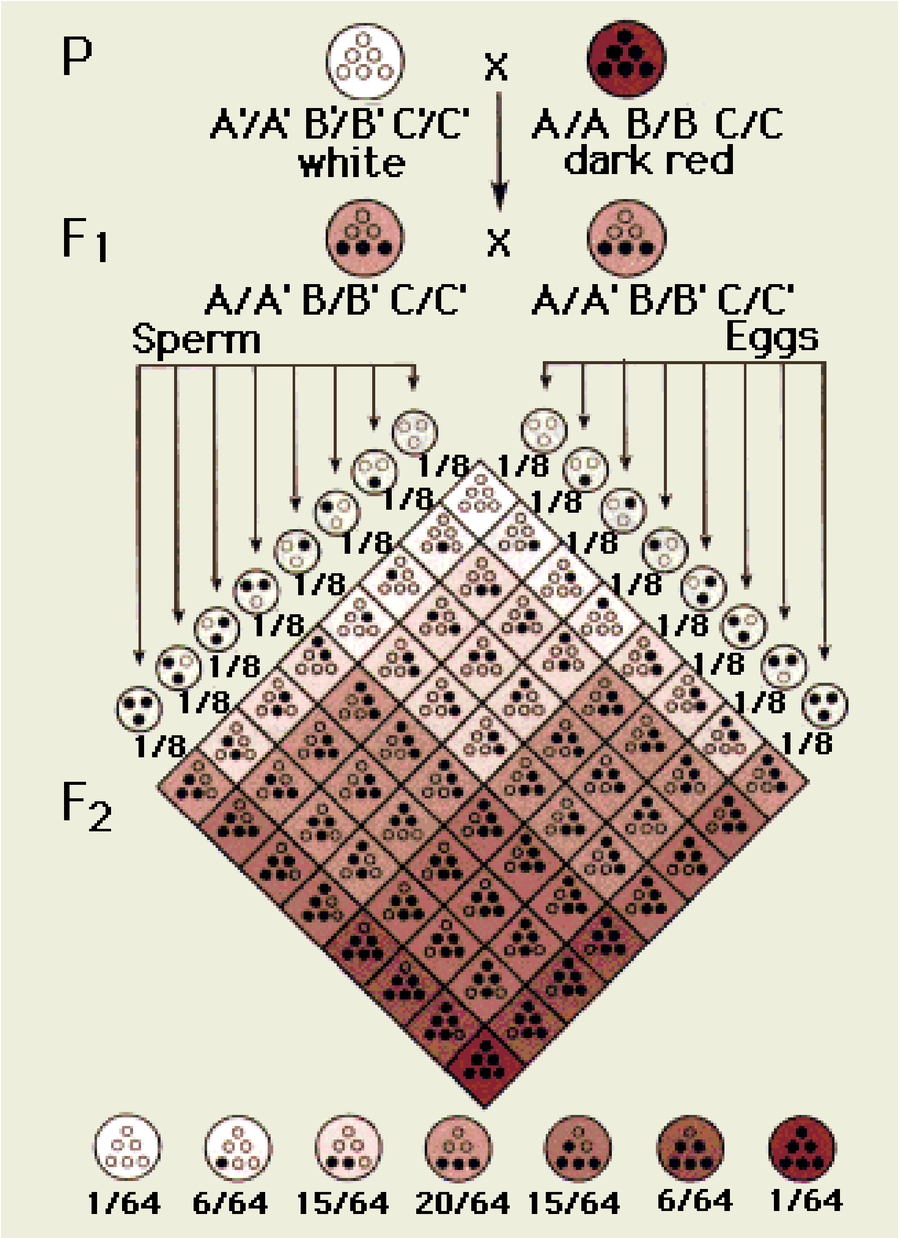
INCOMPLETE DOMINANCE

- **Defined**: Neither allele is completely dominant
- Heterozygous = blended appearance
- More than 2 different phenotypes can occur
- Ex: Many flower colors



POLYGENIC INHERITANCE





HIGHLIGHTS OF SOME DISCOVERIES

- 1900 - Mendel's work was rediscovered by Hugo de Vries after 16 years post Mendel's death
- 1902 - Sutton proposed that genes were located on chromosomes.
- 1944 - The genetic material was found to be DNA.
- 1953 - Watson and Crick propose a model for the structure of DNA that also suggests a means for its faithful replication.
- 1966 - How DNA worked to control the activities of the cell had all been worked out [DNA → RNA → protein]

HIGHLIGHTS OF SOME DISCOVERIES

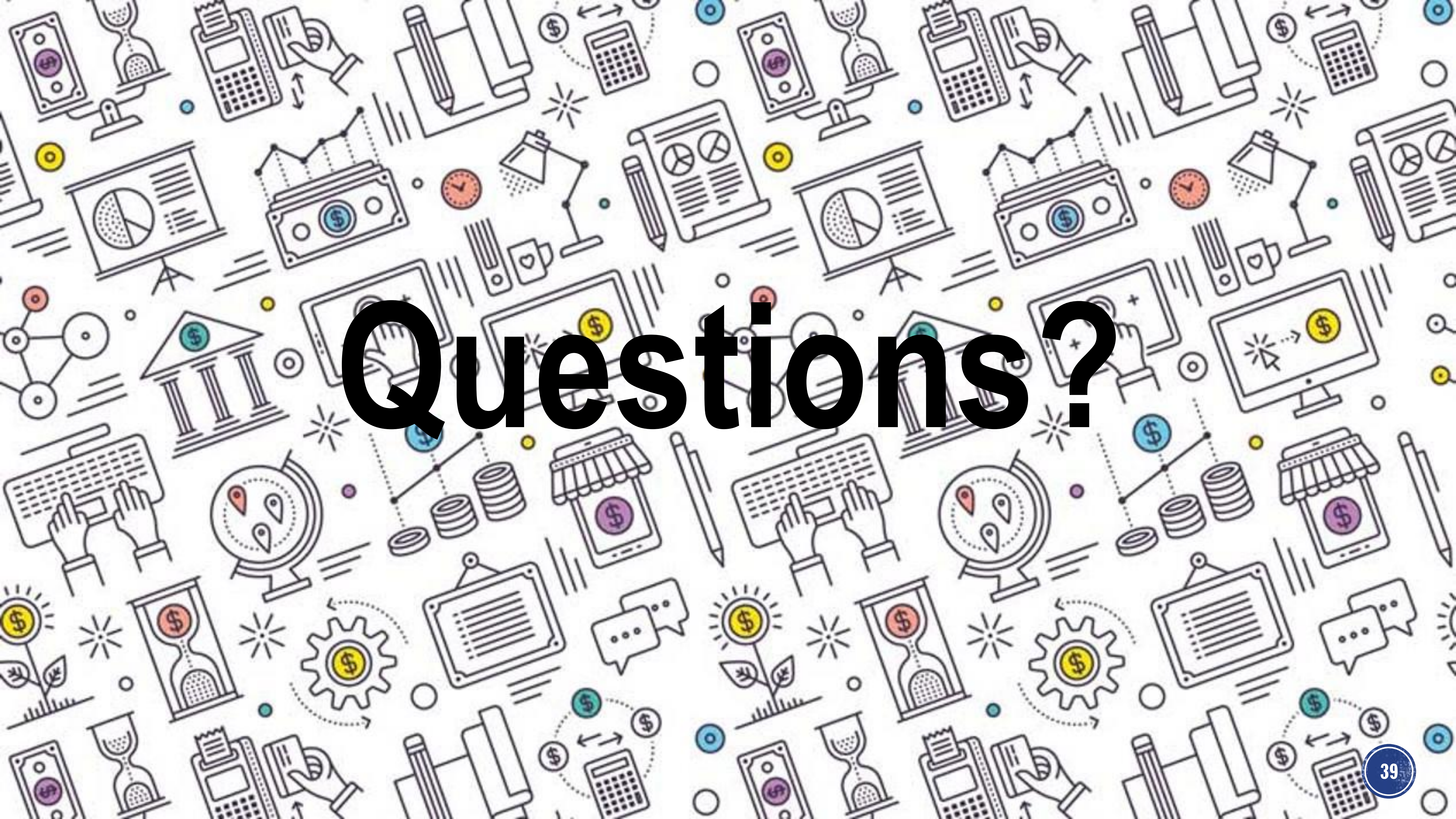
- 1973 – Recombinant DNA molecules formed.
- 1977 – Sequencing of DNA achieved.
- 1983 – PCR technique developed.
- 1990 – First successful gene therapy.
- 1995 – The Human Genome Project (HGP) gets underway.
- 2003 – HGP essentially completed.

HOW MUCH DNA (& GENES) DO CELLS HAVE?

Species	Genes	DNA (bp)
E. coli (bacterium)	4,400	4,600,000
Yeast cell	6,000	12,000,000
Roundworm	19,000	97,000,000
Fruit fly	13,600	165,000,000
Rice plant	55,000	466,000,000
Gallus gallus (Chicken)	23,000	1,000,000,000
Rat	30,000	2,750,000,000
Homo sapiens (human)	**25,000**	**3,200,000,000**
Amoeba proteus (amoeba)	?	290,000,000,000

3.2 Billion bp

75 million base pairs for 25,000 genes (ie. 2% of total DNA). The rest of the DNA is referred to as junk DNA.



Questions?