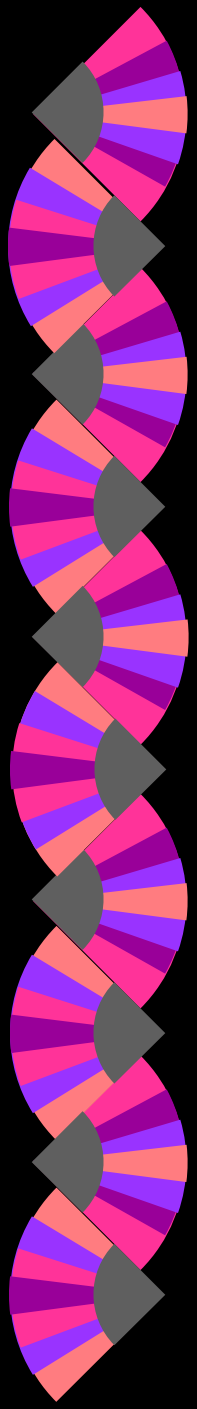
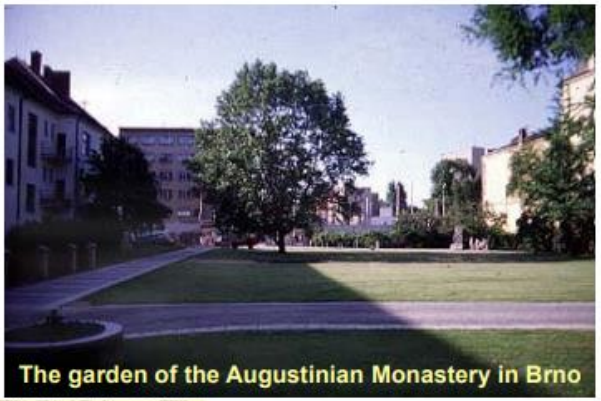
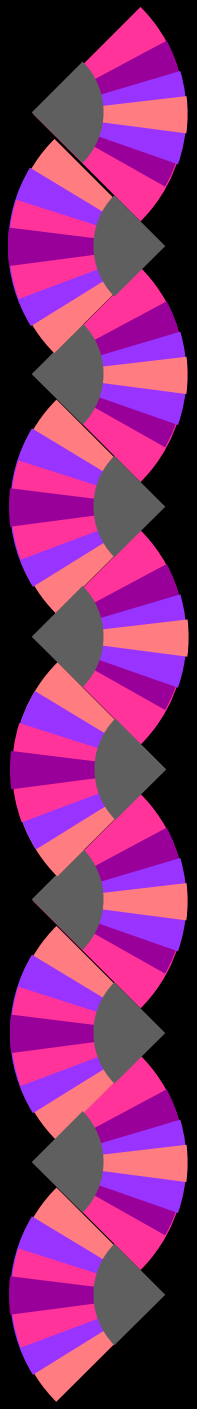


GENETICS

The History


The garden of the Augustinian Monastery in Brno


Friars of the Augustinian monastery in Brünn, in 1860-ies



- GENETICS - The study of the way animals & plants pass on to their offspring such as:

  eye color, hair color, height, body build, blood types, intelligence, gender, etc.

- HEREDITY - Characteristics that a child receives from both parents

What genetic principles account for the passing of traits from parents to offspring?

The "blending" hypothesis is the idea that genetic material from the two parents blends together (like blue and yellow paint blend to make green)

How about when one paint color is more in volume

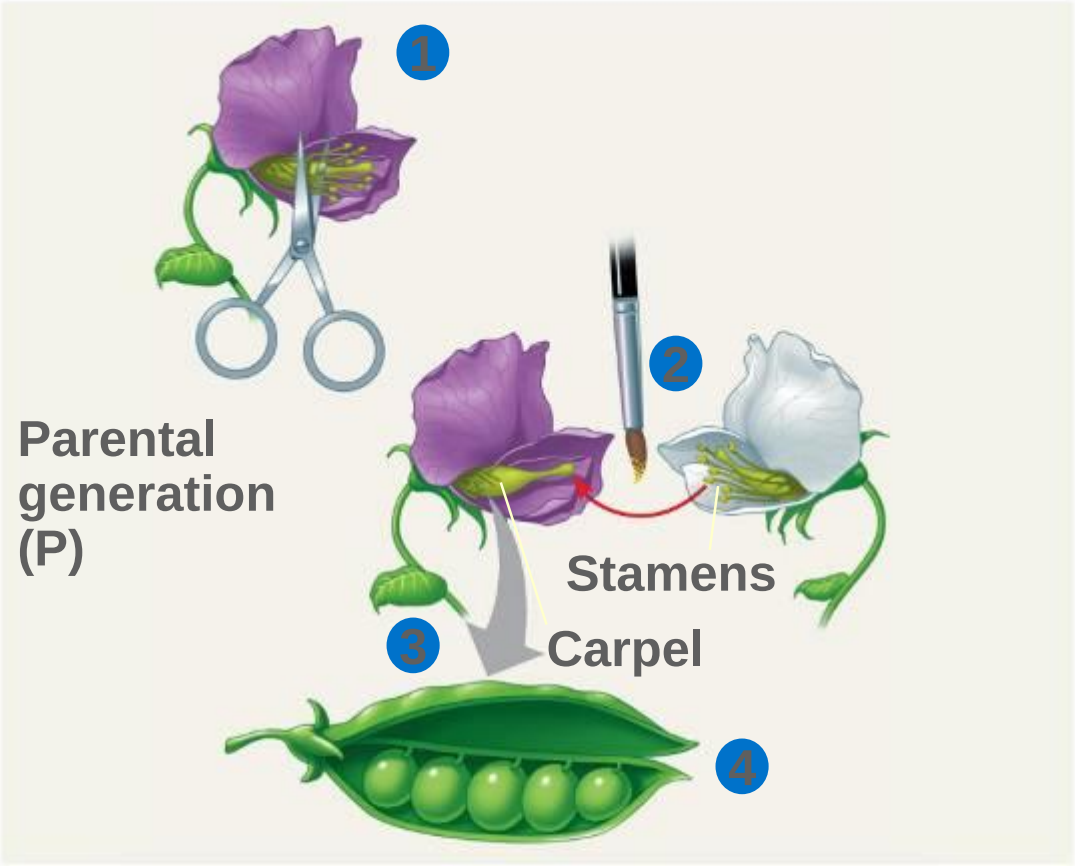
- The "particulate" hypothesis is the idea that parents pass on discrete heritable units (genes)
- This hypothesis can explain the reappearance of traits after several generations (Diabetes, eye colour)
- **Mendel** documented a particulate mechanism through his experiments with garden peas

Mendel did some experiments on garden pea: *Pisum sativum*

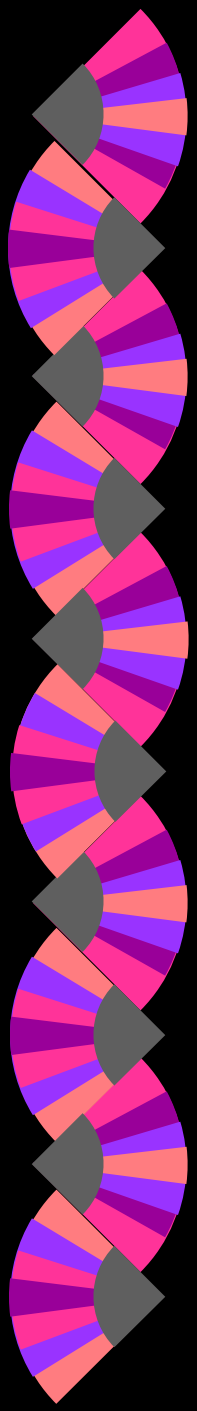
Advantages of pea plants for genetic study

- There are many varieties with distinct heritable features, or **characters** (such as flower color); character variants (such as purple or white flowers) are called **traits**
- Mating can be controlled to ensure the result's.
- Each flower has sperm-producing organs (stamens) and an egg-producing organ (carpel)
- Cross-pollination (fertilization between different plants) involves dusting one plant with pollen from another.

TECHNIQUE

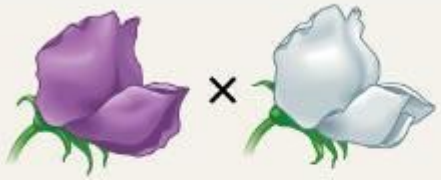


RESULTS



EXPERIMENT

P Generation (true-breeding parents)



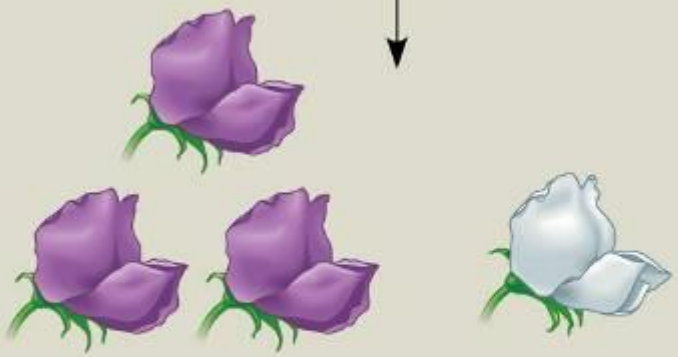
Purple flowers × White flowers

F_1 Generation (hybrids)

All plants had purple flowers

Self- or cross-pollination

F_2 Generation



705 purple-flowered plants

224 white flowered plants

The Law of Segregation

- When Mendel crossed contrasting, true-breeding white- and purple-flowered pea plants, all of the F_1 hybrids were purple
- When Mendel crossed the F_1 hybrids, many of the F_2 plants had purple flowers, but some had white
- Mendel discovered a ratio of about three to one, purple to white flowers, in the F_2 generation

Important

Why two colors in F2 generation ?

- First: alternative versions of genes account for variations in inherited characters
- For example, the gene for flower color in pea plants exists in two versions, one for purple flowers and the other for white flowers
- These alternative versions of a gene are now called **alleles**
- Each gene resides at a specific locus on a specific chromosome

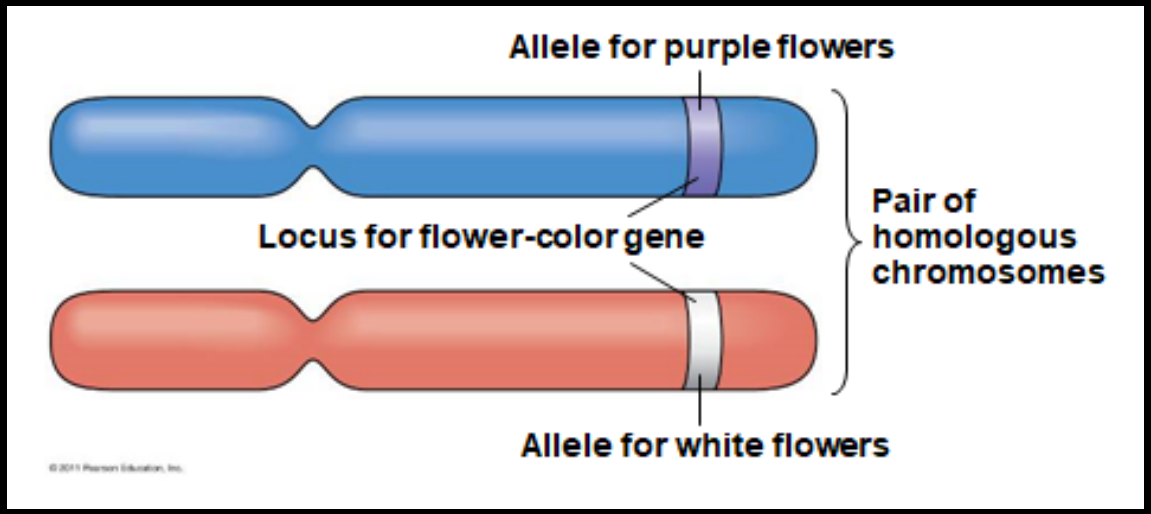
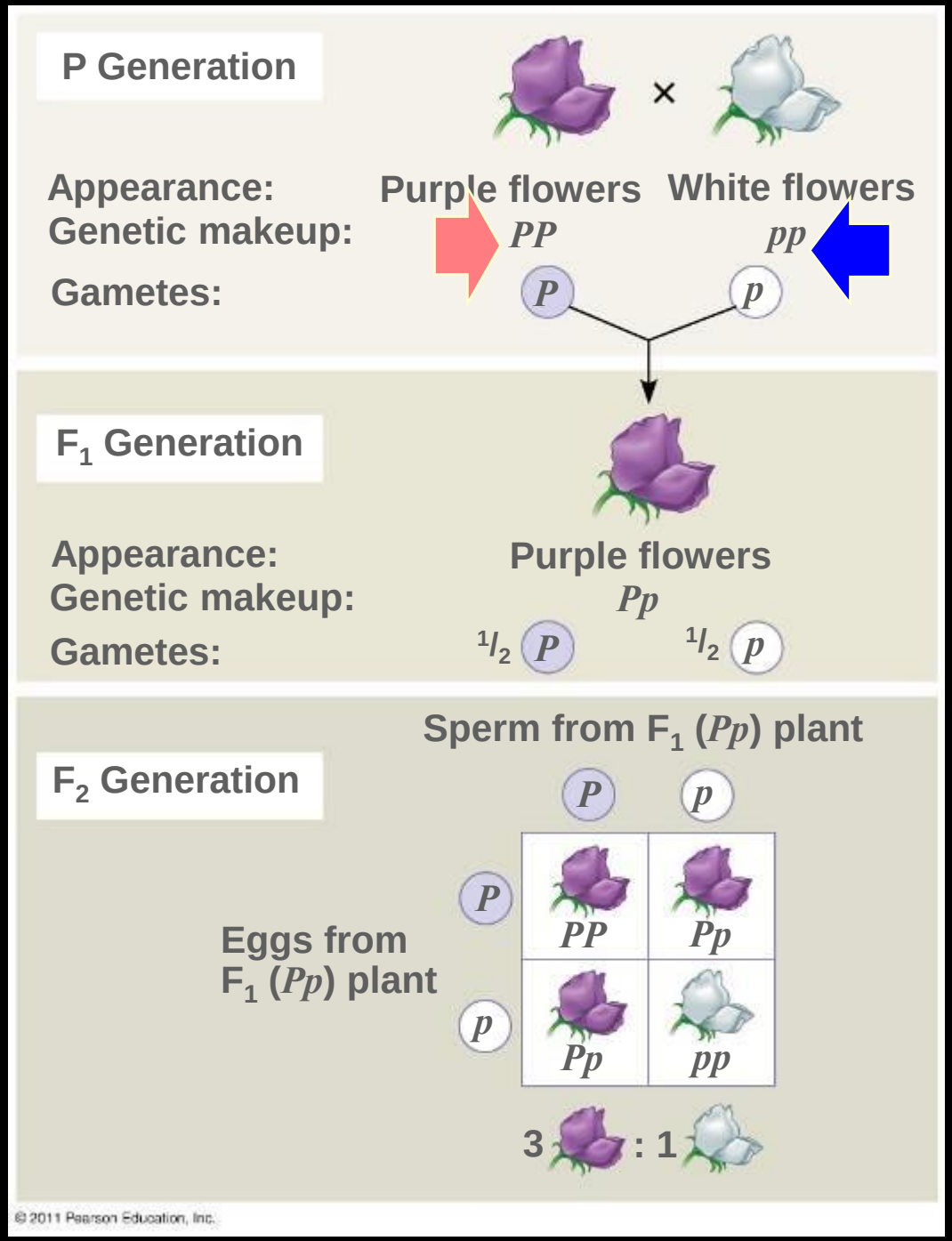


Figure 14.5-3

Lets understand again

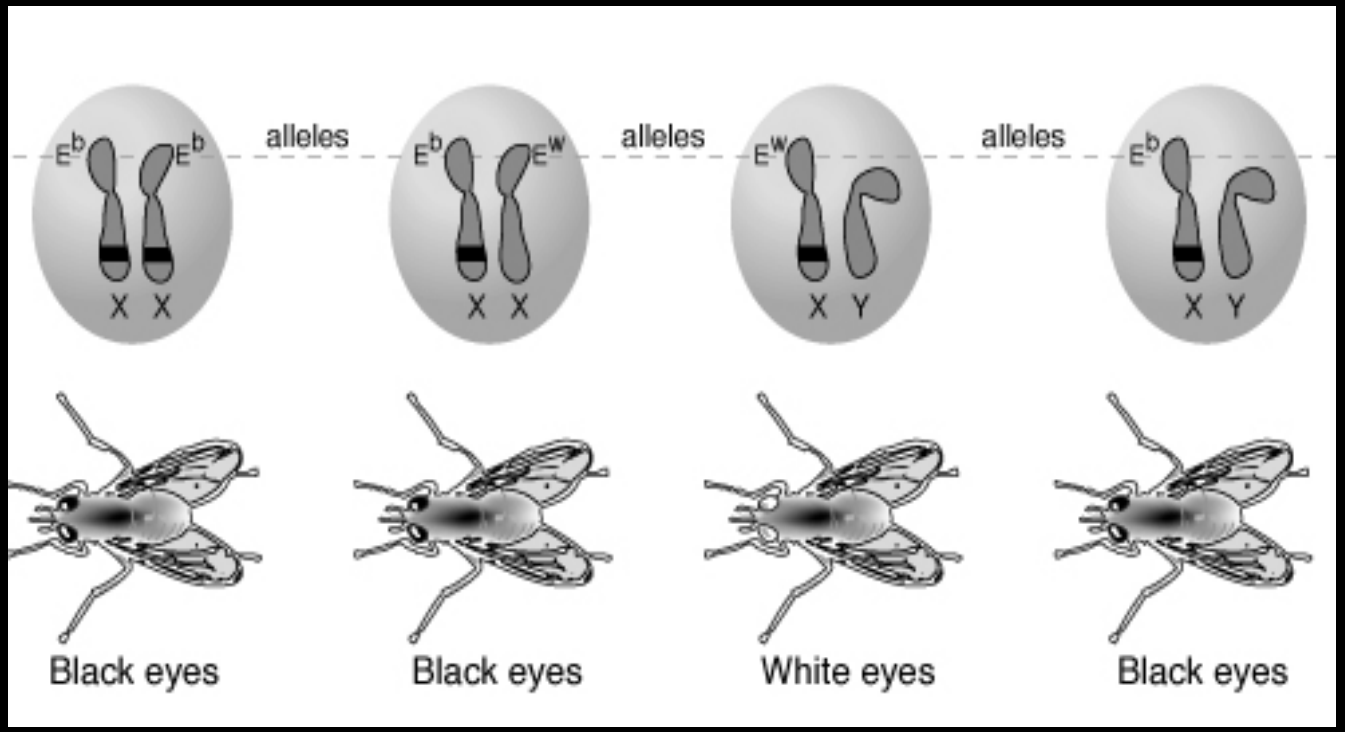


Useful Genetic Vocabulary

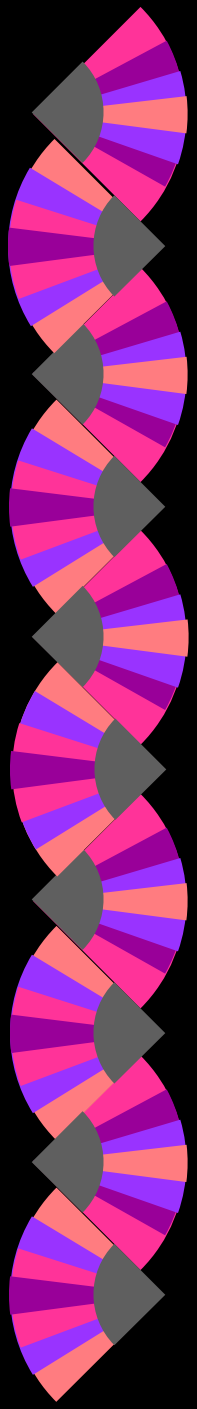
- An organism with two identical alleles for a character is said to be **homozygous** for the gene controlling that character
- An organism that has two different alleles for a gene is said to be **heterozygous** for the gene controlling that character
- Unlike homozygotes, heterozygotes are not true-breeding

Phenotype		Genotype	
Purple		*PP* (homozygous)	1
Purple		*Pp* (heterozygous)	2
Purple		*Pp* (heterozygous)	
White		*pp* (homozygous)	1
3			
1			
Ratio 3:1		Ratio 1:2:1	

Genotypes ⟷ Phenotypes (example)

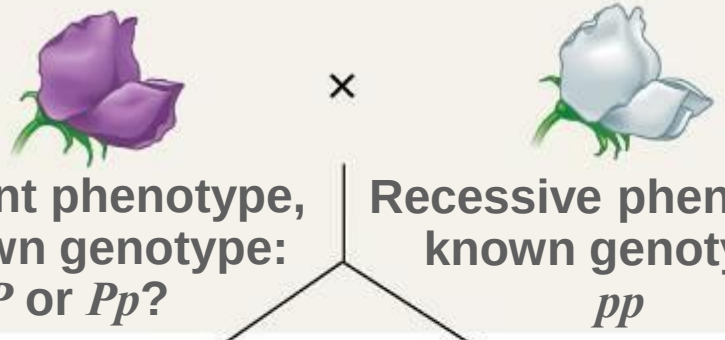


- E^b- *dominant* allele.
- E^w- *recessive* allele.

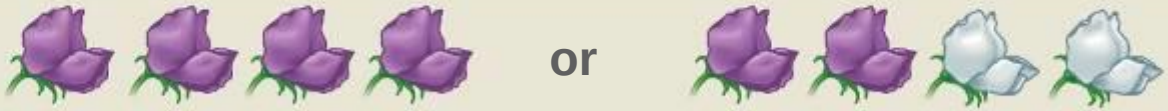


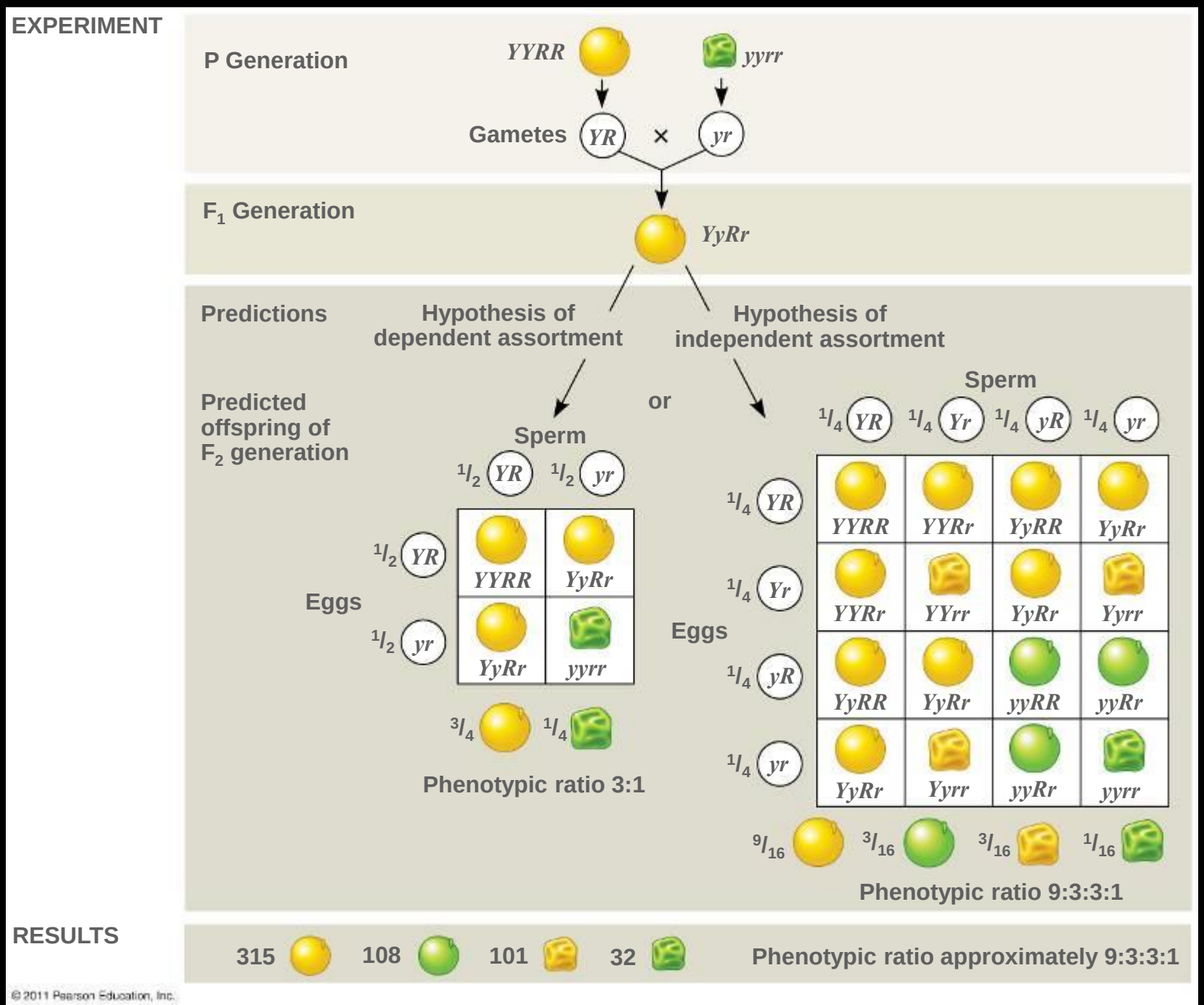
Another Experiment

TECHNIQUE



RESULTS





Important

Law	Definition
Law of segregation	During gamete formation, the alleles for each gene segregate from each other so that each gamete carries only one allele for each gene.
Law of independent assortment	Genes for different traits can segregate independently during the formation of gametes.
Law of dominance	Some alleles are dominant while others are recessive; an organism with at least one dominant allele will display the effect of the dominant allele.

By that time people start knowing that there are something which carry genetic information.

What was that ????

Mendals time
1882-1894

Gene term coined
By Wilhelm Johannsen
In 1905

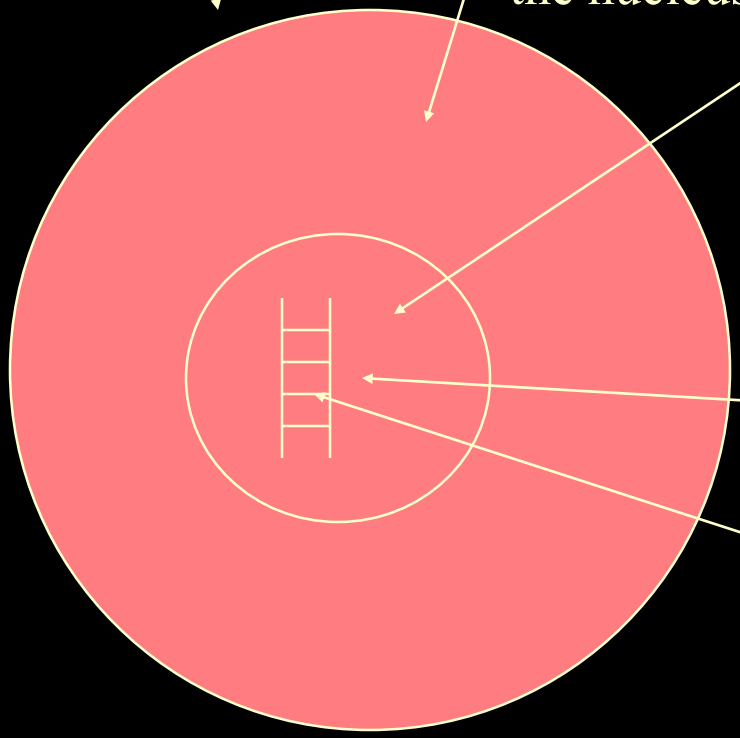
DNA molecular structure
1953

DNA was first isolated
by Friedrich Miescher in 1869

Any idea how Mendel's experiment is still in use to understand the a very important clinical condition

Where is the carrier molecule

- CELL: Basic unit of all living matter (Adult = over 10 trillion cells)



CYTOPLASM: Substance of a cell outside of the nucleus

NUCLEUS: Central point of cell / contains genetic coding for maintaining life systems and issuing commands for growth & reproduction

CHROMOSOMES:

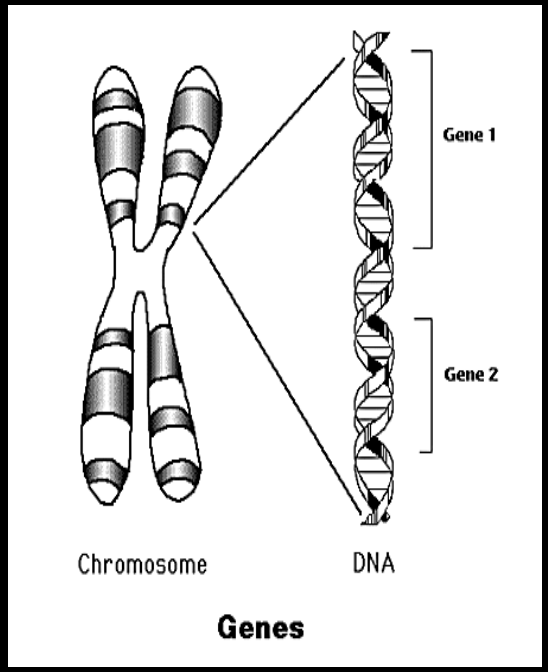
46 in each Nucleus (23 pairs)

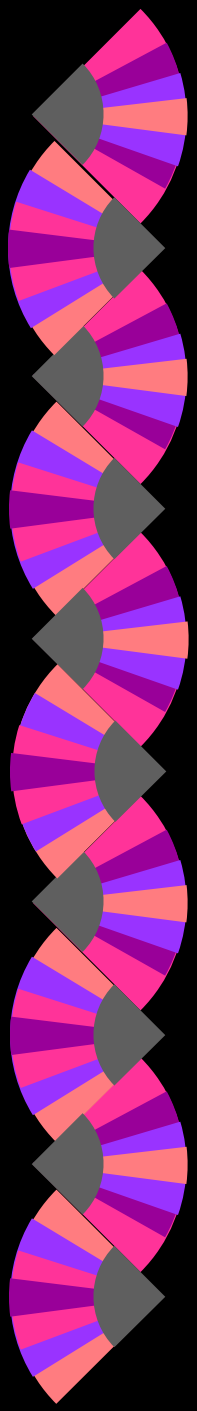
GENES: bands on chromosomes (thousands of genes)

DNA on genes (billions of DNA)

Genetic Information

- **Gene** – basic unit of genetic information. Genes determine the inherited characters.
- **Genome** – the collection of genetic information.
- **Chromosomes** – storage units of genes.
- **DNA** - is a nucleic acid that contains the genetic instructions specifying the biological development of all cellular forms of life

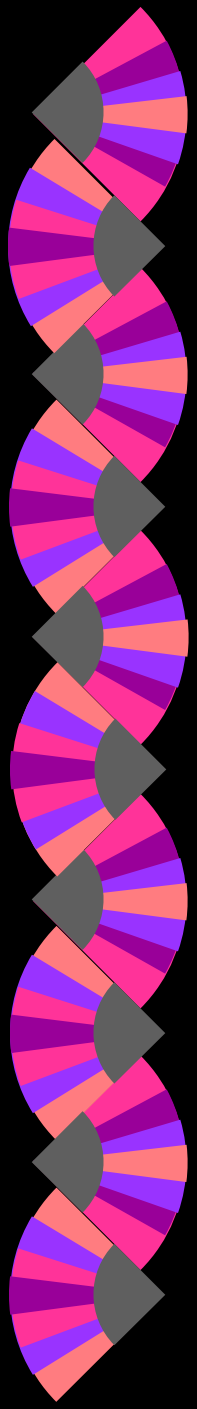




? ? ? ? ? ? ? ?

- How many chromosomes are there in each cell?
- 46 CHROMOSOMES or 23 PAIRS

- How many chromosomes are in Reproductive (egg & sperm) or Germ cells?
- 23 CHROMOSOMES
  - (combined = the 46 chromosomes)



CELL DIVISION

- MITOSIS: Cell divides by copying the DNA - cell splits - new cell with normal number of chromosomes (Cell growth & repair)

- MEIOSIS: Creates 1/2 sets of chromosomes
  - Women = 23 Men = 23 Combined = 46

- Female Sex Cells XX (Ovum or Egg)
- Male Sex Cells XY (Sperm)
- Baby Girl = XX Baby Boy = XY
  - Conception is the union of an OVUM and the SPERM

- DOMINATE Gene: More powerful - trait seen in person
- RECESSIVE Gene: Weaker and hides in the background. Trait can only determine when two of them are present - may show up in future generations.
- CARRIER: Has a recessive gene that is not visible
- SEX-LINKED: Mother passes the recessive X to son
  - Color-blind male receives the trait from his mother.
  - The mother is usually not color-blind herself.

- **B = BROWN eyes (dominate)** **b = BLUE eyes (recessive)**
- BB = BROWN eyes
- bb = BLUE eyes
- Bb = BROWN eyes but carry the recessive BLUE eye gene

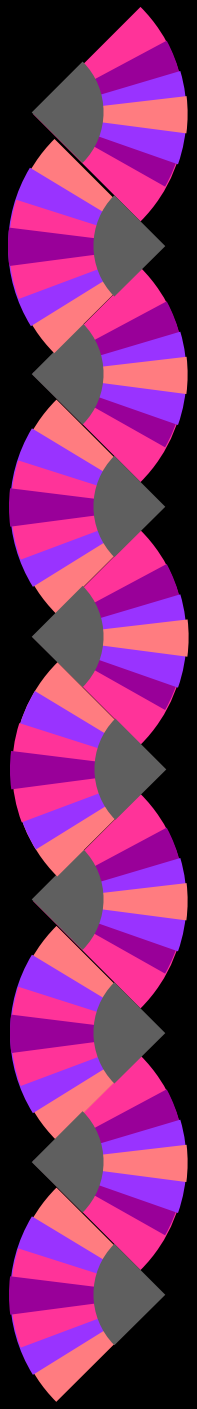
MULTIPLE BIRTHS

- ZYGOT: the cell that is formed when a sperm fertilized an egg (ovum)
- MONOZYGOT: Identical Twins 1 Egg + 1 Sperm
  - Fertilized ovum splits into 2 identical cells - Always the same gender
- DIZYGOT: Fraternal Twins 2 Eggs + different Sperm
  - Will look different - May be different or the same gender
- MULTIPLE BIRTHS: More than 2
  - May be identical, fraternal or both - May be different or the same gender
- CONJOINED (Siamese) TWINS: Ovum splits apart, but the separation is not completed. Babies are joined at some part of their bodies.

What 4 FACTORS may contribute to Multiple Births?

- 1) History in the family
- 2) Increased hormones naturally
  - More than 1 egg released
- 3) Fertility Drugs
  - More than 1 egg released
- 4) Age 32-36

- Likelihood of multiple pregnancies in the United States
  - Twins: Blacks- 1 in 73 Whites 1 in 93
  - Triplets: 1 in 10,000
  - Quadruples: 1 in 620,000



- Sex - Linked or X - Linked Defect: When an X-gene from the mother is faulty. There is a 50/50 chance of the child inheriting the disorder.
- Syndrome: When a group of signs and symptoms occur together and characterize a particular problem.
- Congenital Malformation: A condition that is present at birth.
- Multi-factorial Defects: Interaction of genes with other genes OR with environmental factors.
- Chromosomal Error: The fertilized egg cell that contains chromosomes in an abnormal number, structure or arrangement.