Mendelian Genetics

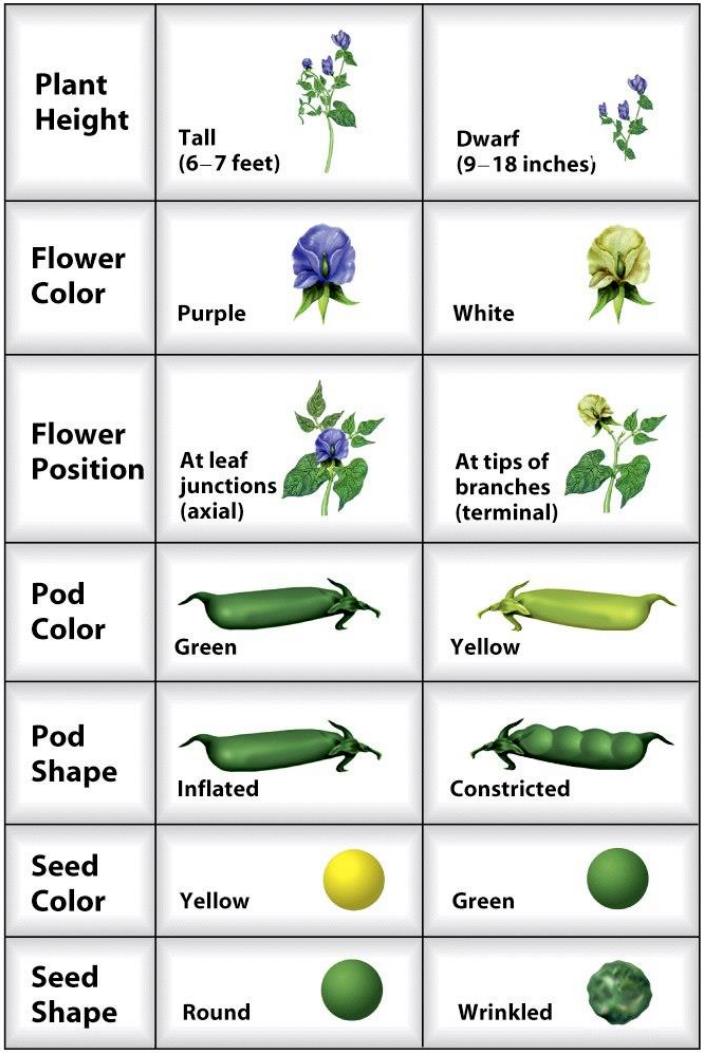
Gregor Mendel

- Raised on farm and understood the value of plant breeding.
- At 21, entered priesthood and studied plant breeding in a monastery in the Czech Republic.
- Loved to read especially about natural sciences and was aware of Darwin's findings.
- Studied the inheritance of traits in pea plants.

- Considered the **FATHER OF GENETICS!!!!!**

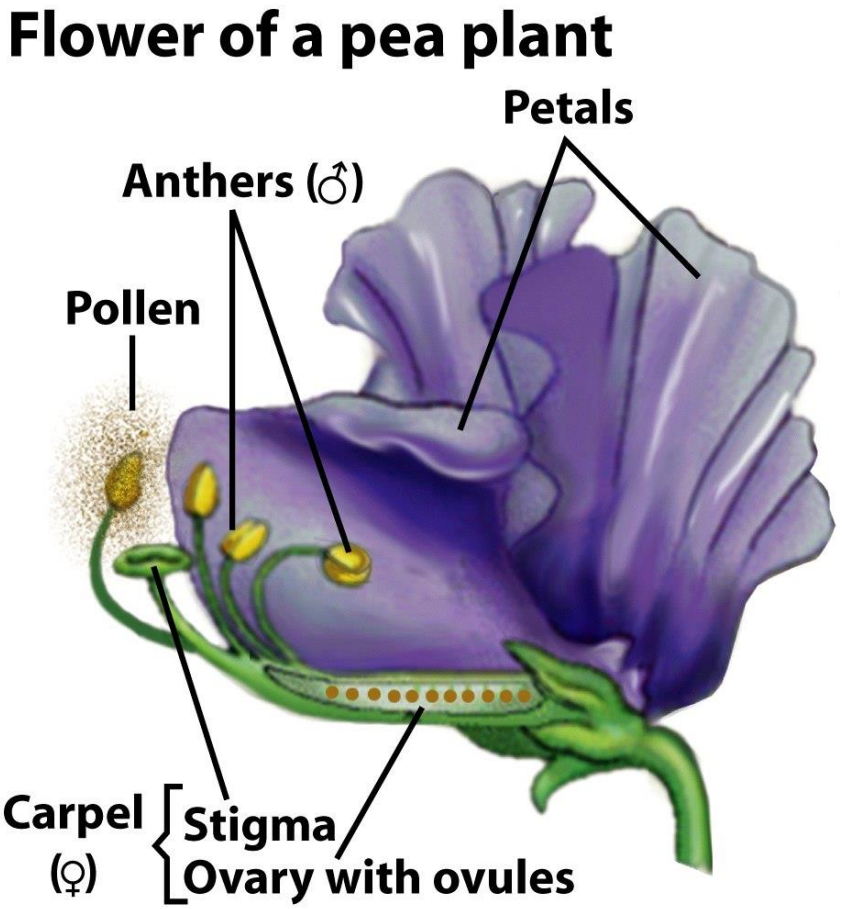
Gregor Mendel

- Studied traits that occur in distinct forms.
- Developed true-breeding varieties
  - When bred amongst themselves these plants produced offspring identical to the parent for that trait.
- Used mathematical analysis in his studies.



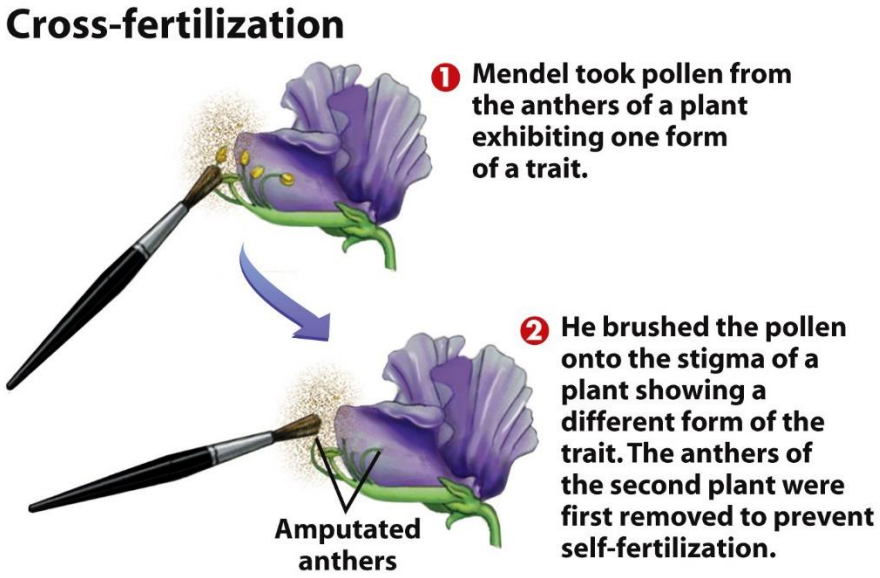
Definitions

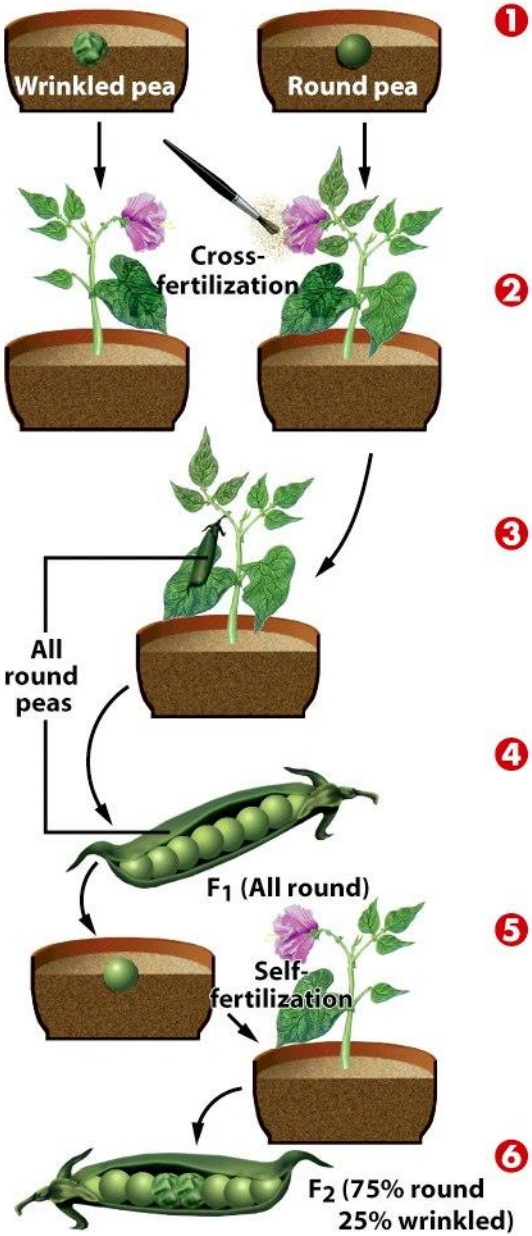
- Gametes: reproductive cells produced by sexually reproducing organisms.
  - Two types:
    - **male gametes = sperm**
      - **In plants: contained in pollen**
    - **Female gametes = eggs**
      - **In plants: contained in ovules**
      - **Ovules contained in carpels**



Fertilization

- Fertilization: fusion of egg and sperm
  - Self-fertilized: fusion of sperm and egg from same plant
  - Cross fertilized: fusion of egg and sperm from two different plants
    - **Produced hybrids**
- F_1: first generation
- F_2: second generation





1. Mendel grew pea plants from true-breeding wrinkled-seeded and true-breeding round-seeded varieties.

2. When these plants matured, he used pollen taken from the anthers of the wrinkled-seeded variety to fertilize the plant grown from the round-seeded variety. He removed the anthers of the round-seeded plant to prevent self-fertilization.

3. The carpel of the round-seeded plant matured into a pea pod containing all round peas. These peas were the seeds of the F_1 generation.

4. Mendel planted the F_1 peas and allowed them to grow into mature plants.

5. These plants were allowed to self-fertilize. Pollen from the anthers of the F_1 plants fell directly onto the tops of the carpels of the same flower.

6. The carpels of the F_1 plants matured into pea pods containing peas—the seeds of the F_2 generation. About 75% of these peas were round and about 25% were wrinkled.

Mendel's Interpretations

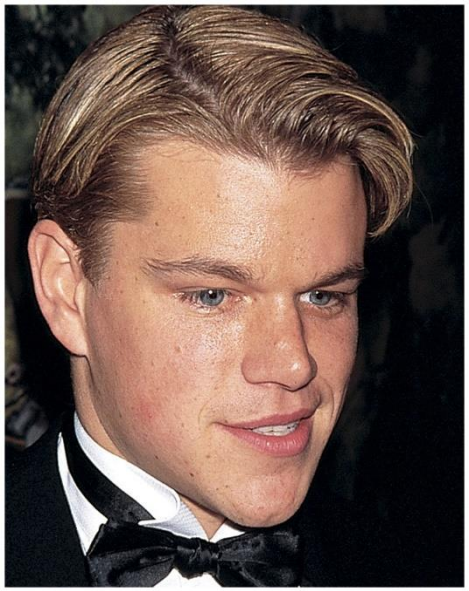
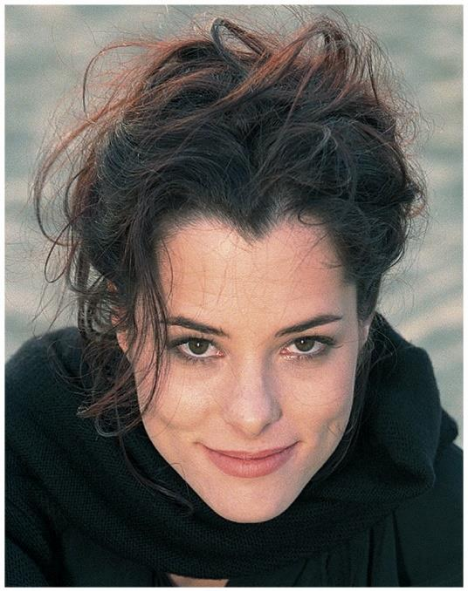
- Modified to incorporate today's vocabulary
  - Genes: the hereditary information that determines a single trait
  - Alleles: alternate forms of a gene
- When an organism inherits two identical alleles for a trait, organism is said to be homozygous for the trait.
- When an organism inherits to different alleles for one trait, the organism is called heterozygous for the trait.

Why were all the seeds in the F_1 generation round?

- Dominant vs. Recessive Allele
  - Dominant: an allele that is expressed whenever it is present
  - Recessive: an allele that is masked whenever the dominant allele is present.
- Dominant and recessive alleles influence an organism's phenotype

Genotype and Phenotype

- Genotype: Genetic makeup of an individual. It is determined by the alleles present for each trait.
- Phenotype: Physical appearance of a trait. It is the expression of the genotype.

Can you look at someone and determine their genotype?

- Testcross is used to determine the genotype of an individual
  - Testcross: crossing an organism with unknown genotype with one that is homozygous recessive for the trait.
  - Example:
    - Plant that has round (R) seeds crossed with one that is homozygous recessive (rr) for wrinkled seeds

R? x rr

Using Punnett Squares to Predict the Inheritance of Sickle Cell Anemia

Sickle Cell Anemia

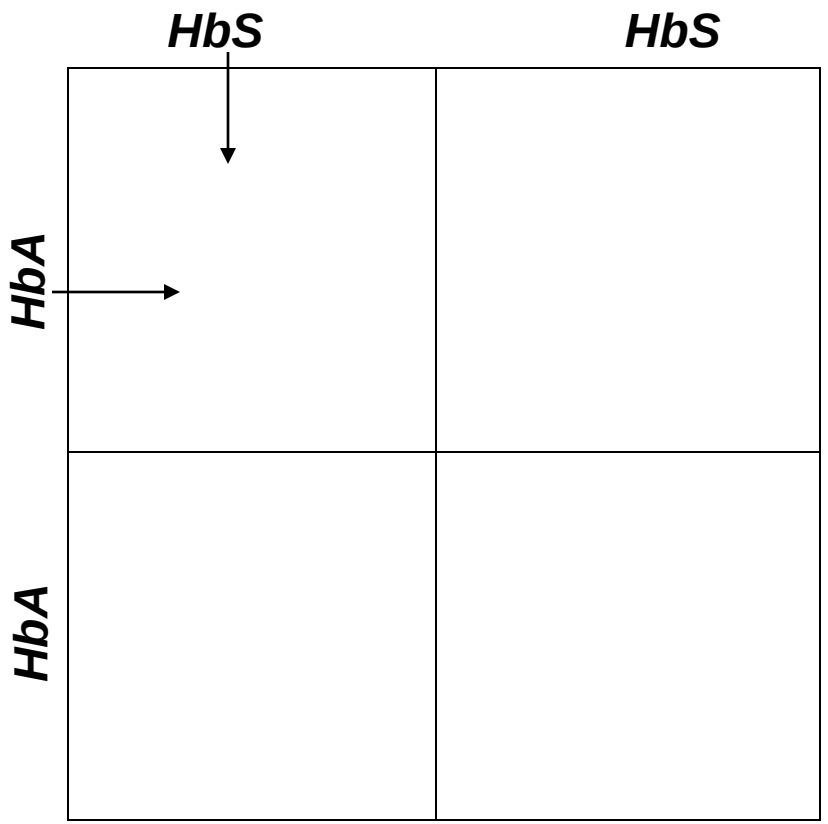
- Due to presence of recessive allele denoted by *HbS.*
- Causes distortion of red blood cells into long, thin sickles.
- Causes pain, tissue damage, and cells to rupture prematurely.
- Carriers: have heterozygous genotypes and do not manifest symptoms.

Punnett Squares

- Used to predict possible offspring genotypes
- Place alleles for each parent on each side

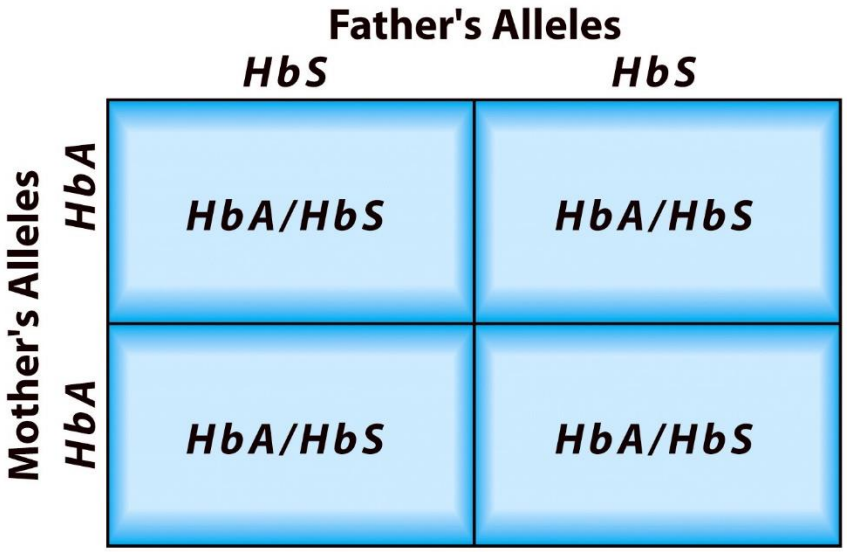
Mother's genotype Father's genotype

HbA/ HbA ***x HbS/ HbS***

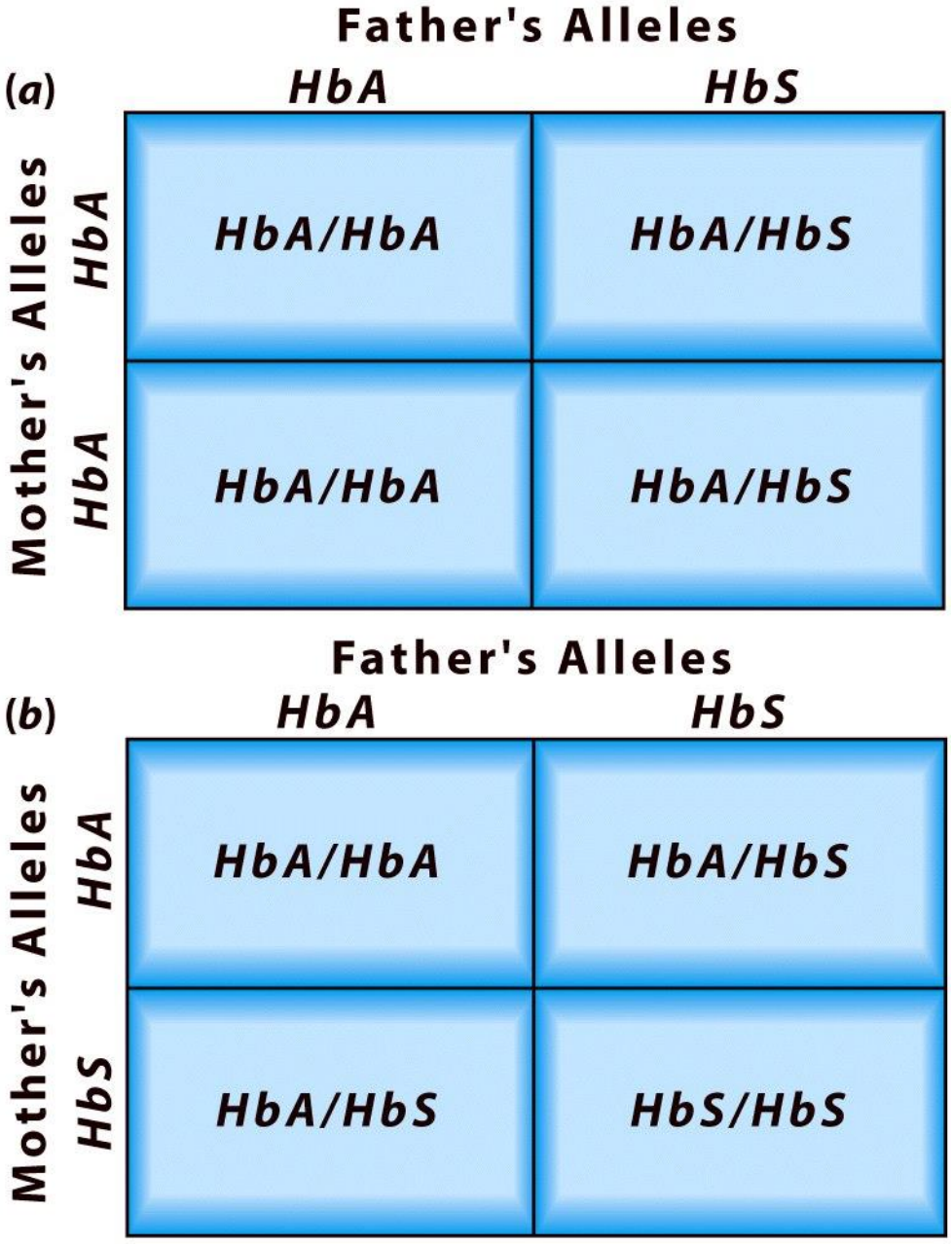


Predict inheritance

- Given parents' genotypes, you can predict offspring's genotypes and phenotypes
- *HbS/ HbS = homozygous recessive results in sickle cell anemia*
- *Both HbA/ HbS (heterozygous) and HbA/ HbA (homozygous dominant) are normal.*



What are the genotypic and phenotypic ratios for this cross?

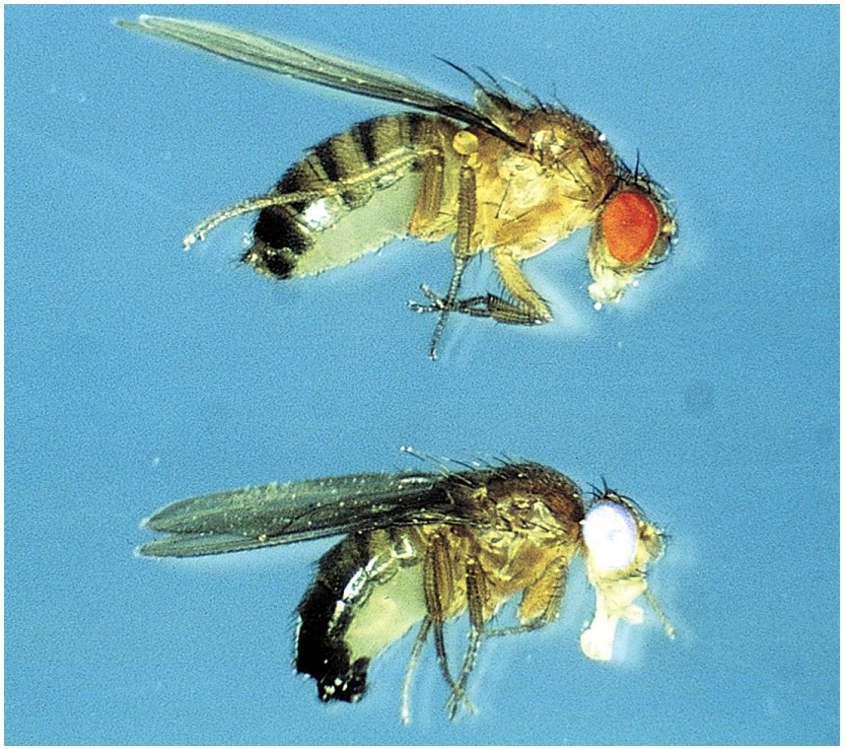

(a)
Father's Alleles
HbA
HbS
Mother's Alleles
HbA
HbA
HbA/HbA
HbA/HbS
HbA/HbA
HbA/HbS
(b)
Father's Alleles
HbA
HbS
Mother's Alleles
HbA
HbS
HbA/HbA
HbA/HbS
HbA/HbS
HbS/HbS

Mendel's Laws

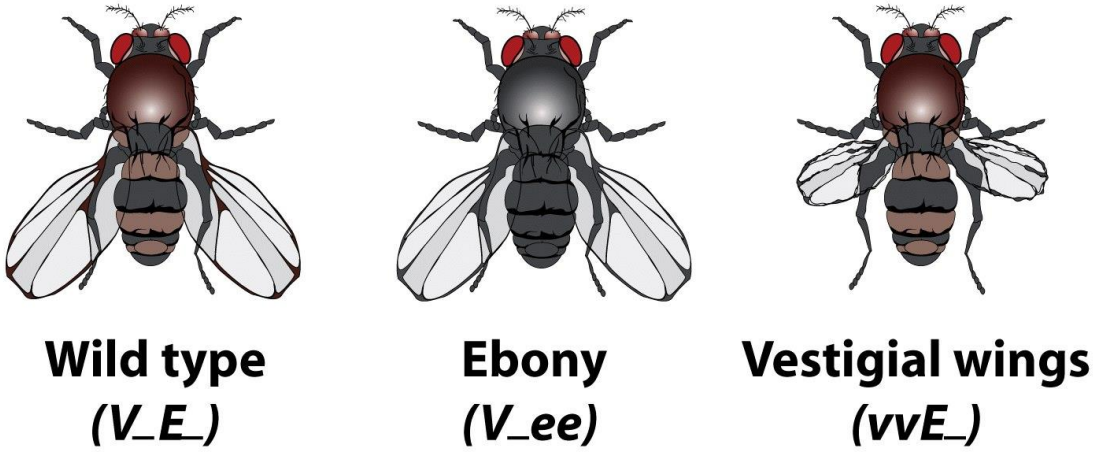
- Based on Mendel's data, he formulated two laws:
  - Law of Segregation: A parent contributes only one of its alleles for a trait to each offspring.
  - If parent is heterozygous for a trait, the particular allele donated to the offspring is random.

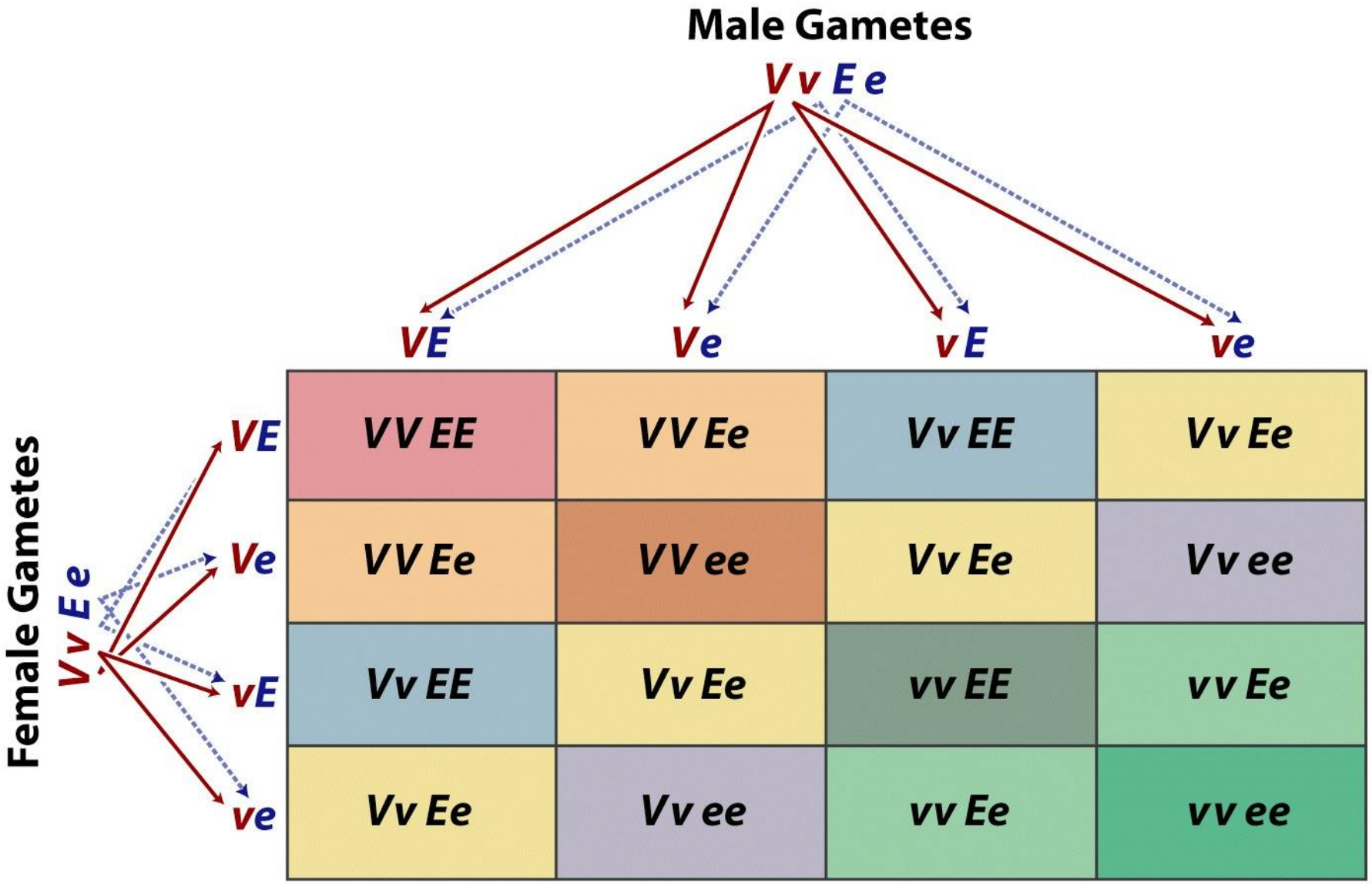
Mendel's Laws

- Law of Independent Assortment: Alleles of one gene are passed to offspring independently of the alleles of other genes.
  - Applies to the inheritance of two or more genes simultaneously.



- Can use the inheritance of traits in fruit flies to illustrate the law of independent assortment.
- Dihybrid cross
  - A fertilization in which the parents differ in two distinct traits or characteristics.




Male Gametes
V v E e
VE
Ve
vE
ve
Female Gametes
V v E e
VE
Ve
vE
ve
VV EE
VV Ee
Vv EE
Vv Ee
VV Ee
VV ee
Vv Ee
Vv ee
Vv EE
Vv Ee
vv EE
vv Ee
Vv Ee
Vv ee
vv Ee
vv ee

Male Gametes

Female Gametes	*VE*	*Ve*	*vE*	*ve*
VE	Wild type 	Wild type 	Wild type 	Wild type 
Ve	Wild type 	Broad wing ebony body 	Wild type 	Broad wing ebony body 
vE	Wild type 	Wild type 	Vestigial wing striped body 	Vestigial wing striped body 
ve	Wild type 	Broad wing ebony body 	Vestigial wing striped body 	Vestigial wing ebony body 

Probability

- Can be used to better understand segregation and independent assortment.
- Look at coins first…

Why Aren't Members of the Same Species Identical?

- Sources of Genetic Variation
  - Law of Independent Assortment
  - Mutation

Law of Independent Assortment

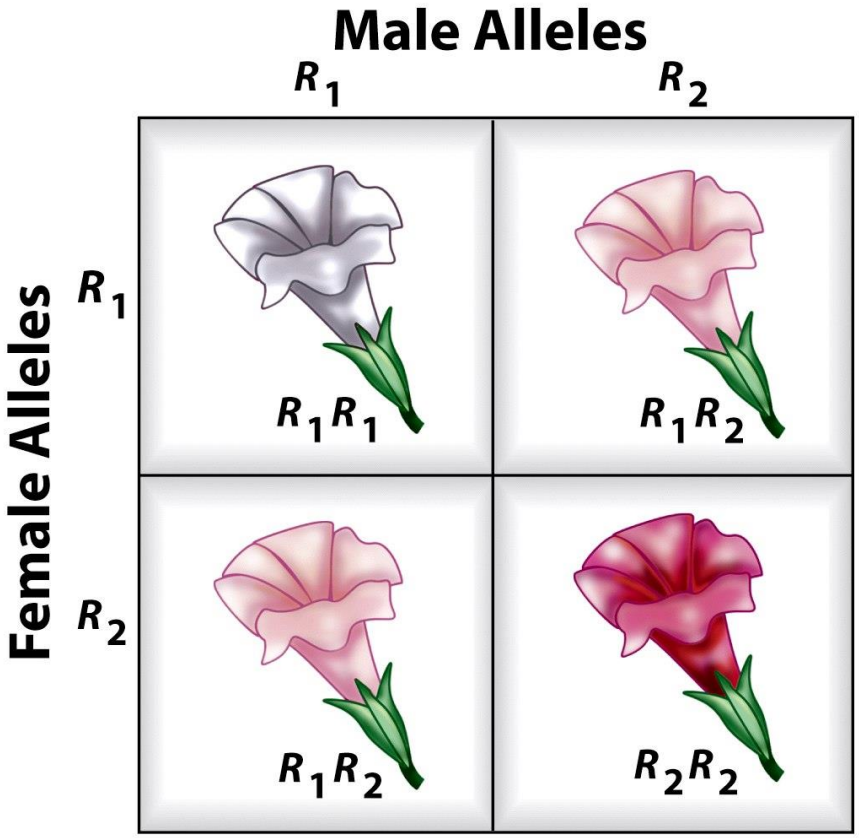
- Allows for new gene combinations or genetic recombination
- Can mathematically predict the possible combinations
  - Number of possible genotypes = 2^n

  where n = the number of genes or traits considered
  - Example: considering 100 traits:
    - **$2^{100} = 1.26765 \times 10^{30}$**

Do Mendel's Laws Always Apply?

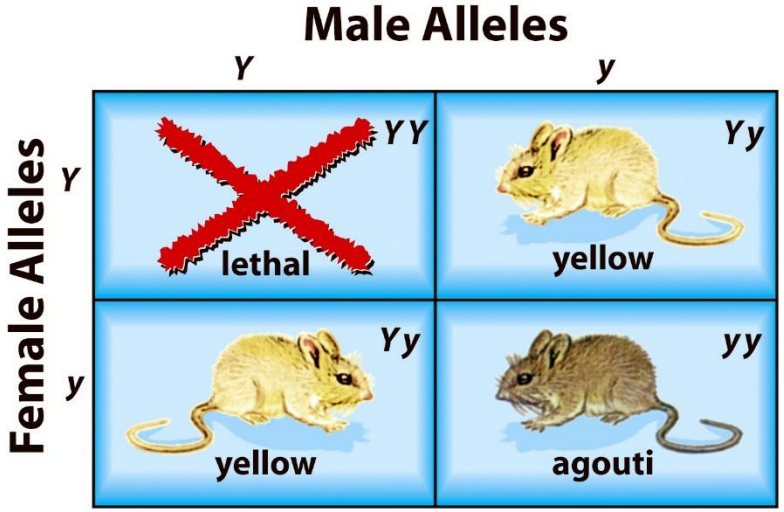
Incomplete Dominance

- A condition where all three genotypes are expressed.
- Phenotypic level: contradicts Mendel's conclusions.
- Genotypic level: consistent with Mendel's laws.



Lethality

- A condition in which the inheritance of a lethal combination of alleles results in death of the organism.
- Lucien Cuenot studied inheritance of coat color in mice.
- Huntington’s disease.



Pleiotropy

- A single gene affects two or more traits.

- Examples:
  - One gene affects whether seed coat is round/wrinkled
  - Cat fur

Polygenic vs. Monogenic inheritance

- Polygenic: A trait affected by many genes.
  - Examples: Height, weight, skin color
- Monogenic: Traits determined by single gene with two alleles.
  - Examples: Flower color in four-o'clock plants