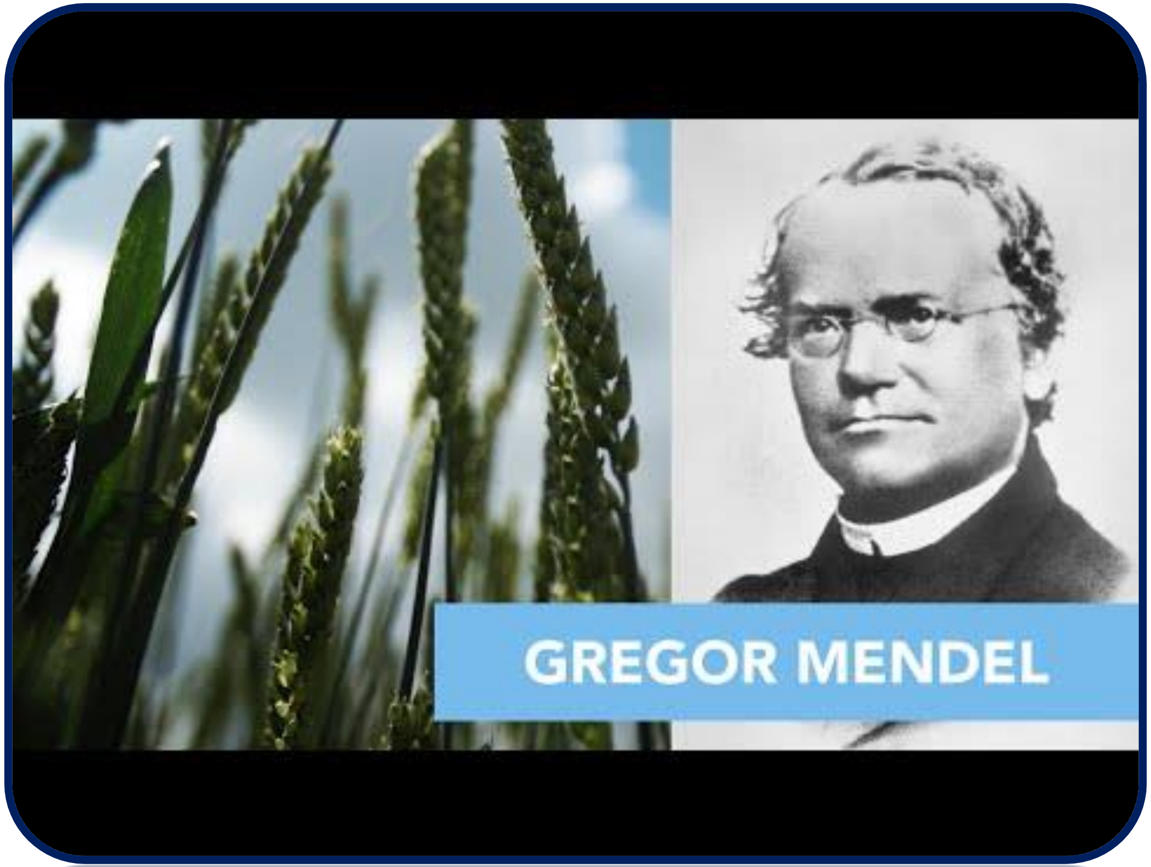
Lecture 7

Dr. Sameer Hasan Qari

المملكة العربية السعودية
وزارة التعليم
جَامِعَةُ أمّ القُرى
الكلية الجامعية بالجموم
قسم الاحياء

Mendelian & Non Mendelian Genetics

Mendelian Genetics



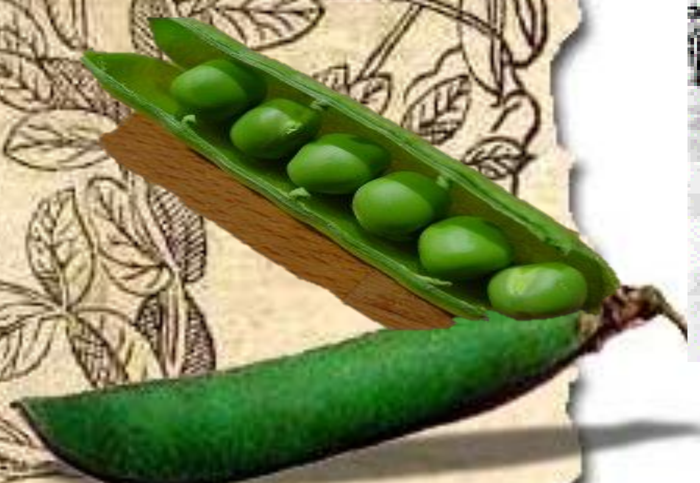
Mendel's Law of Segregation

- Austrian monk born in 1822
- in monastery known for research and teaching
- after his death (1884) acknowledgment of his discoveries in 1900

Gregor J. Mendel, O.S.A., experimental garden (35x7 meters) in the grounds of the Augustinian Monastery in Old Brno. Its appearance before 1922. *Courtesy of Villanova University Archives.*

The Monastery Garden with the greenhouse which Gregor J. Mendel, O.S.A., had built in 1870. Its appearance before

1902. *Courtesy of Villanova University.*



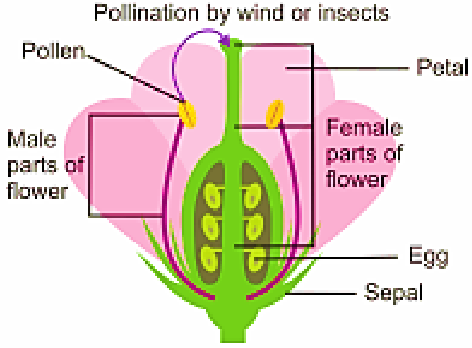
Experiments with Pea Plants

Pea plants have several advantages for genetics.

- Pea plants are available in many varieties with distinct heritable features (characters) with different variants (traits).
  - Seed coat colour (gray or white)- Seed shape (round or wrinkled) - Seed colour (yellow or green)
  - Pod colour (green or yellow) - Pod shape (inflated or constricted)
  - Stem length (tall or dwarf)
- Each pea plant has male (stamens) and female (carpal) sexual organs.
- In nature, pea plants typically self-fertilize, fertilizing ova with their own sperm.
- However, Mendel could also move pollen from one plant to another to cross-pollinate plants.

Pollination

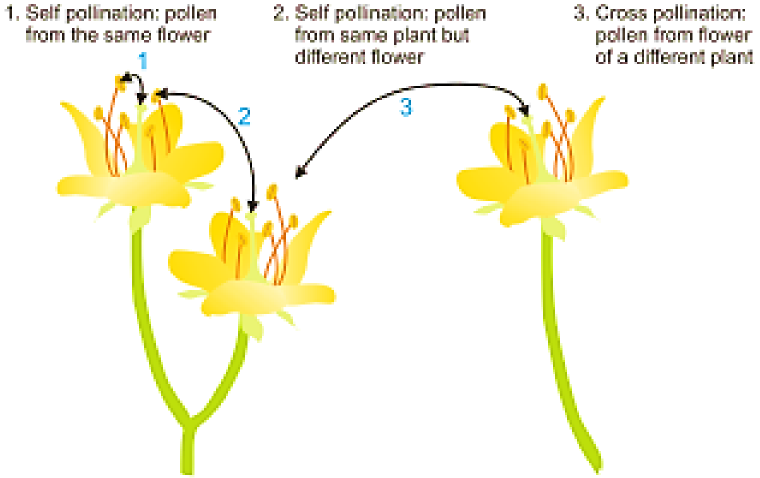
Pollination: Pollination is the **movement of pollen from the anther (male part) to the stigma (female part) of the flower** which contains the egg.



Types of Pollination

Self Pollination -

Pollen is transferred to the stigma of same flower or flowers borne to same plant.

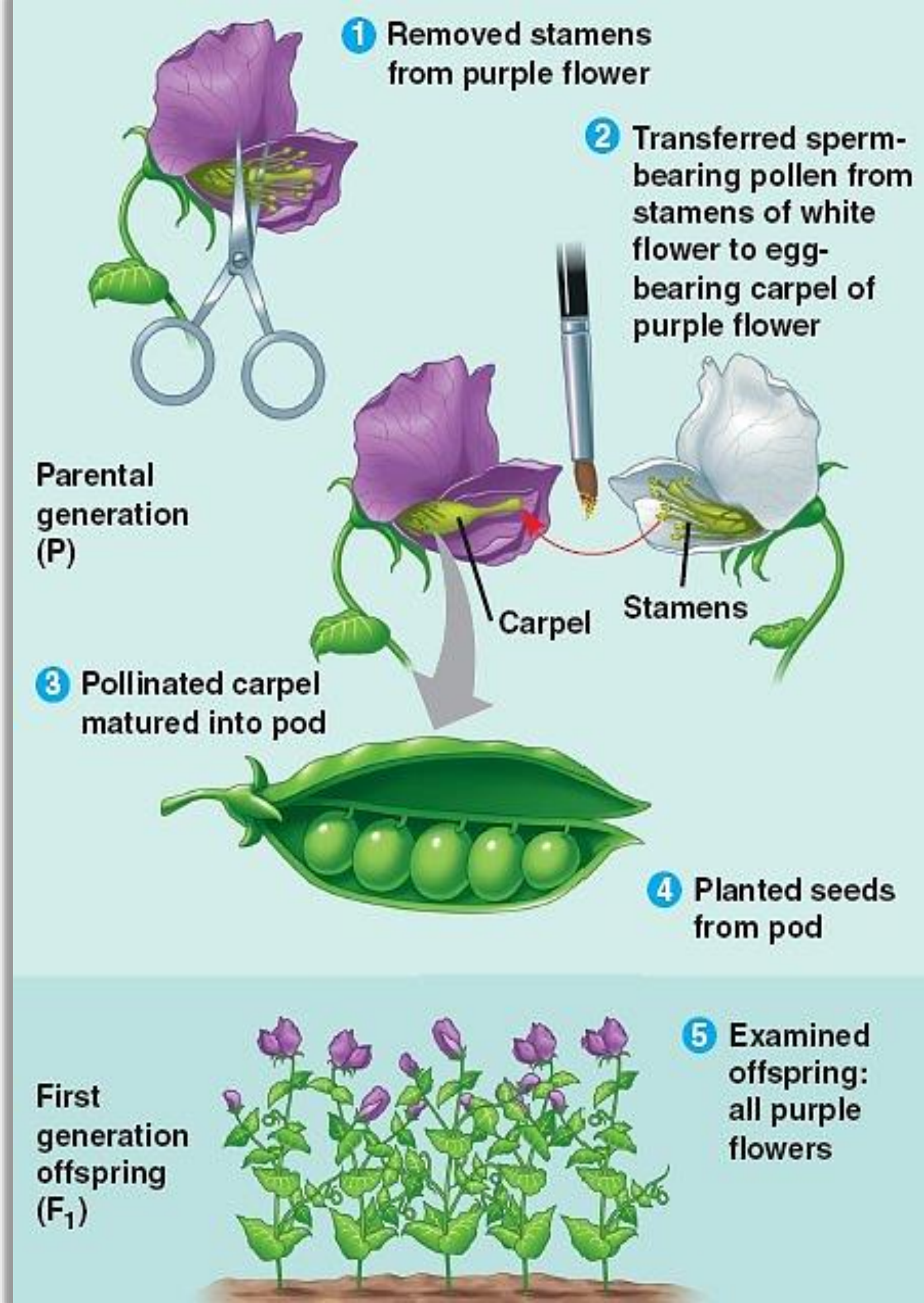


Cross Pollination -

Pollen is transferred to flowers of other plants of the same species.

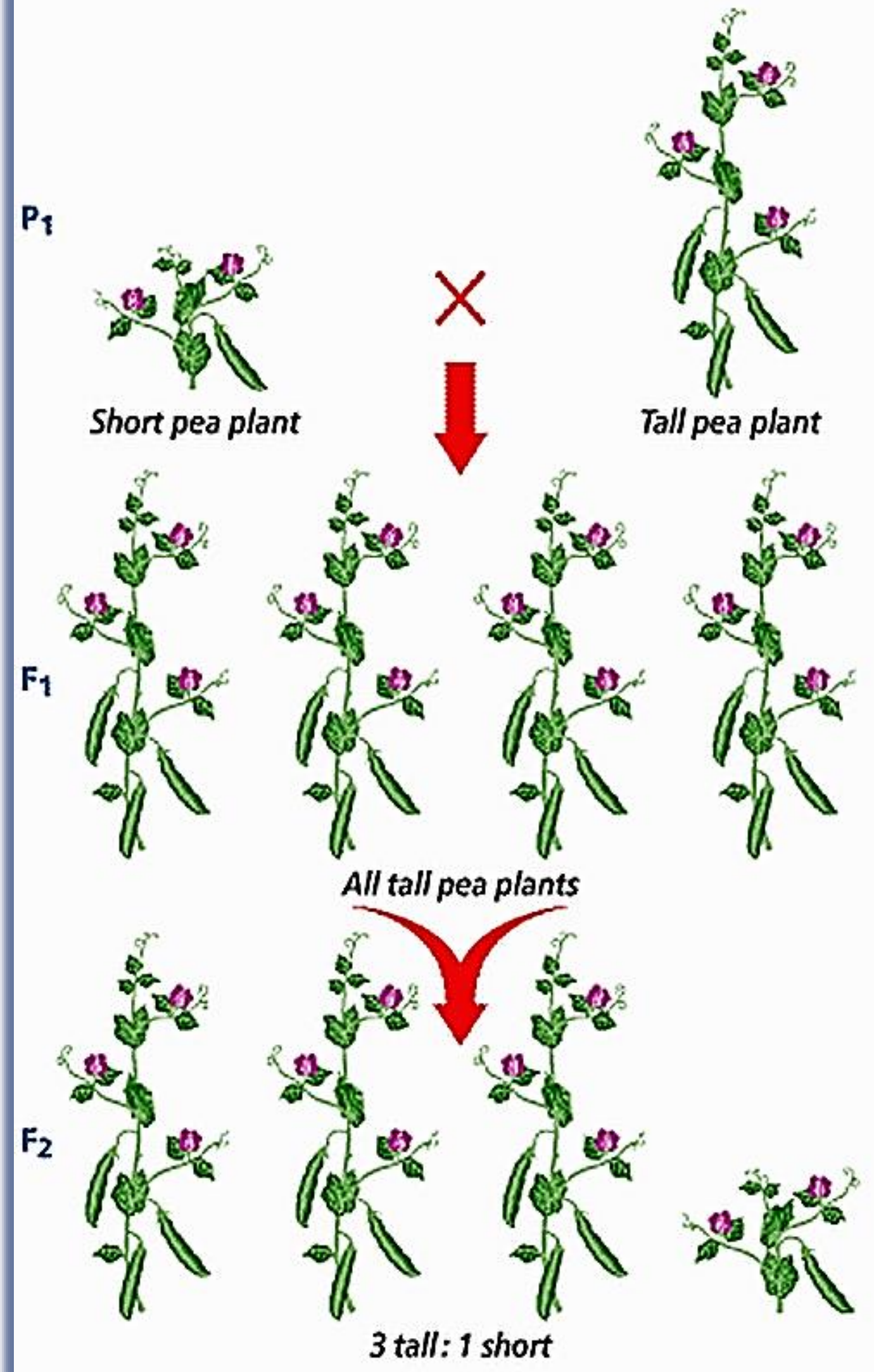
Cross-Pollination

- To hybridize 2 varieties of pea plants, Mendel used an artist's brush.
- He transferred pollen from a true breeding tall stem to the carpel of a true breeding short stem.



Tracking heritable characteristics

- Mendel tracked heritable characters for 3 generations.
- When F_1 hybrids were allowed to self-pollinate a 3:1 ratio of the 2 varieties occurred in the F_2 generation.



Cross-Pollination sympols

- **P** = parental generation
- **F1** = first filial generation (offspring or children)
- **F2** = second filial generation (grandchildren)

<u>Mendel's Cross pollination</u>

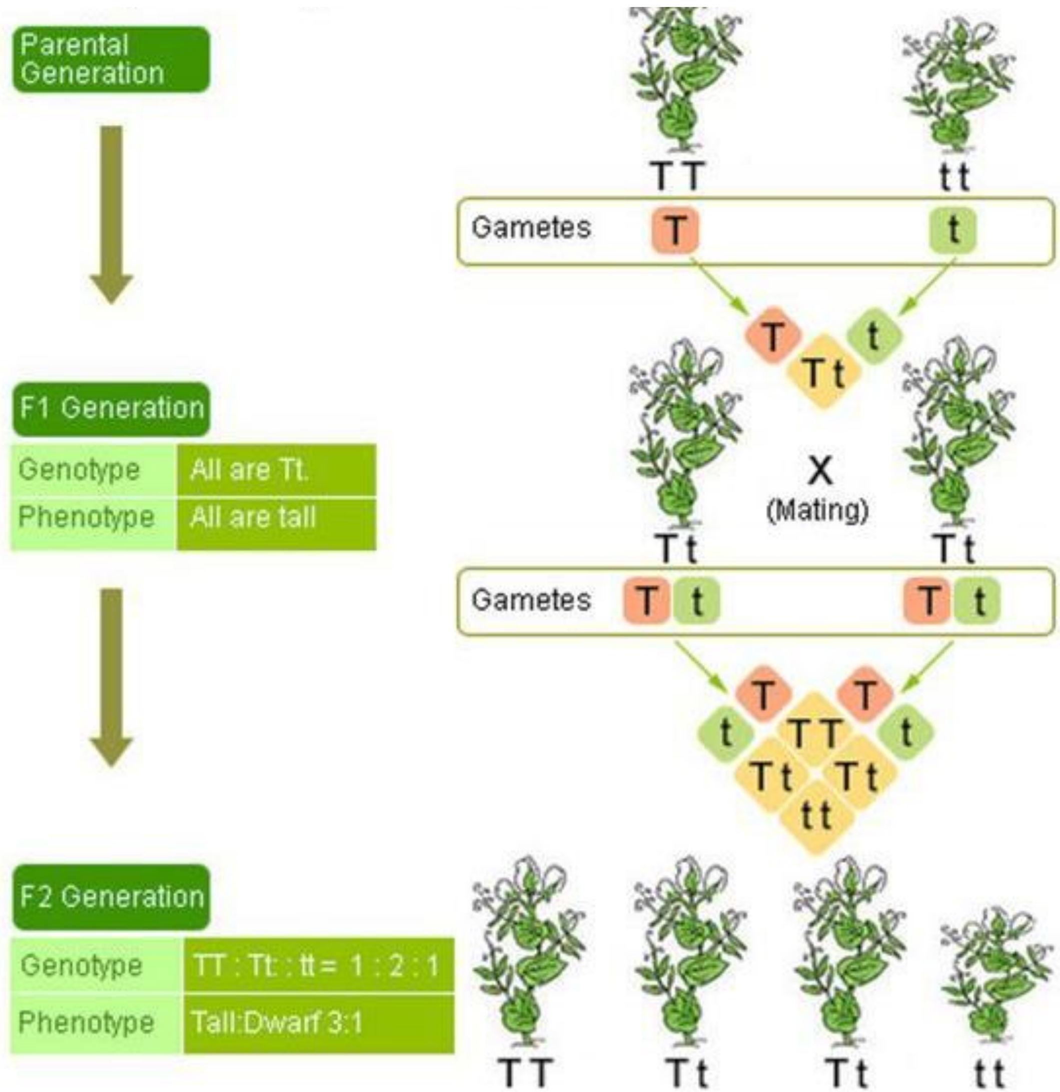
P two different parents, example: trait = height; alleles = tall or short

F1 offspring are all heterozygous plants. He used F1 as parents

<u>Heterozygotes</u> have different alleles.

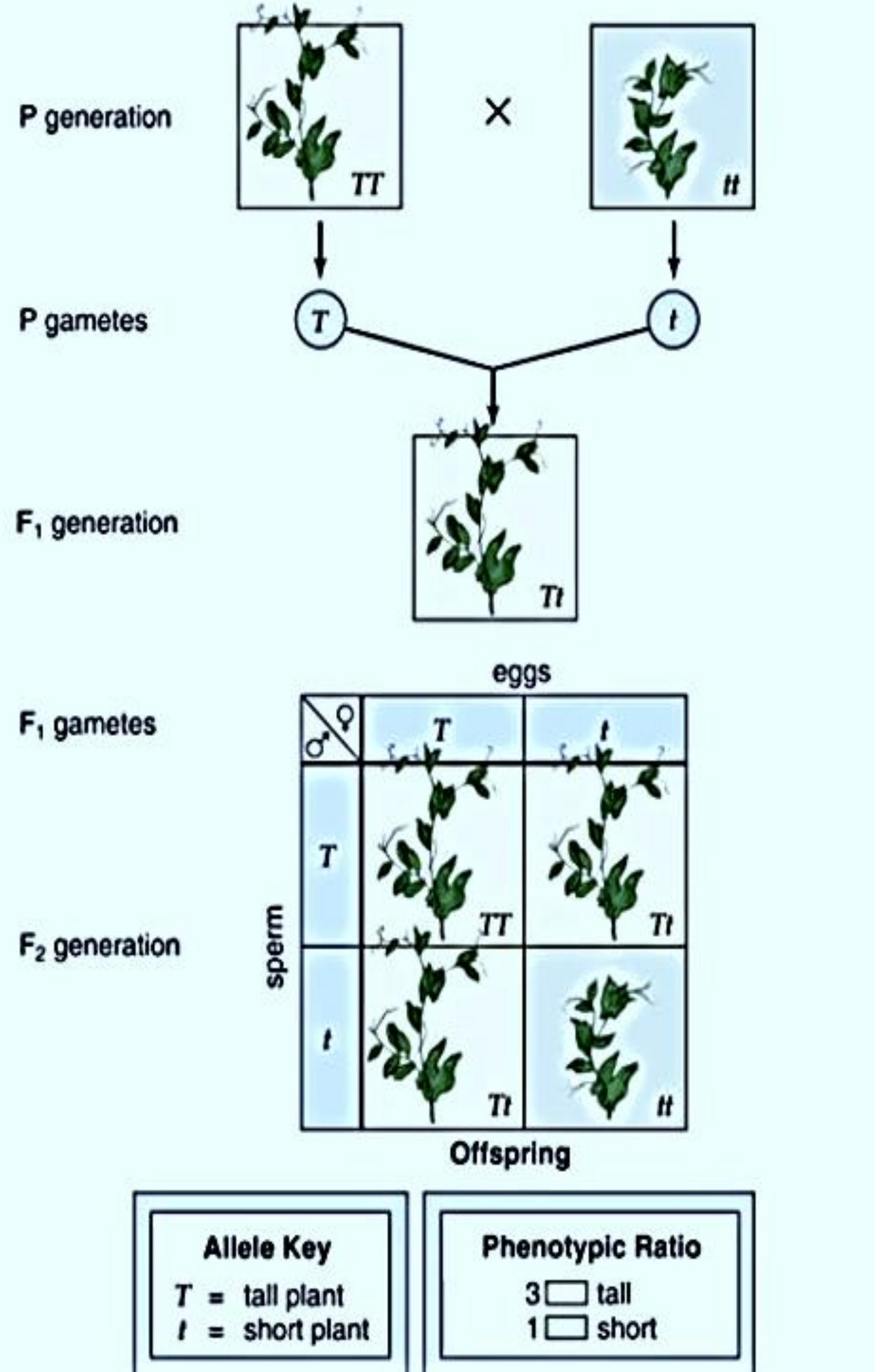
<u>Homozygotes</u> have the same alleles.

F2 'grandchildren' have ratios of 3 out of 4 dominant trait or 75% and 1 out of 4 recessive trait or 25%.



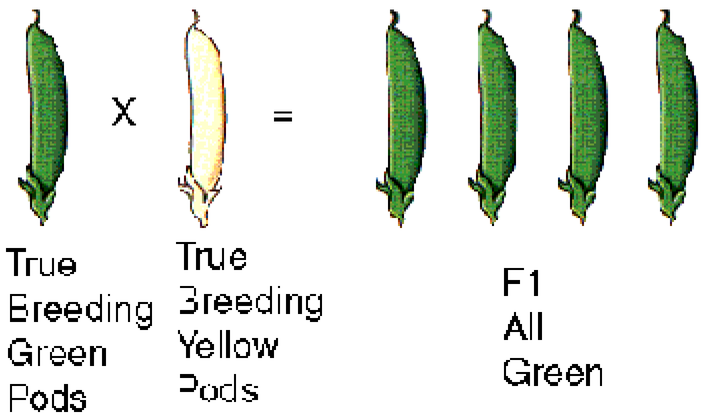
Mendel's law of segregation

- By carrying out these **monohybrid** crosses, Mendel determined that **the 2 alleles for each character segregate during gamete production.**
- Mendel discovered this c.1860.
- DNA was not discovered until 1953.

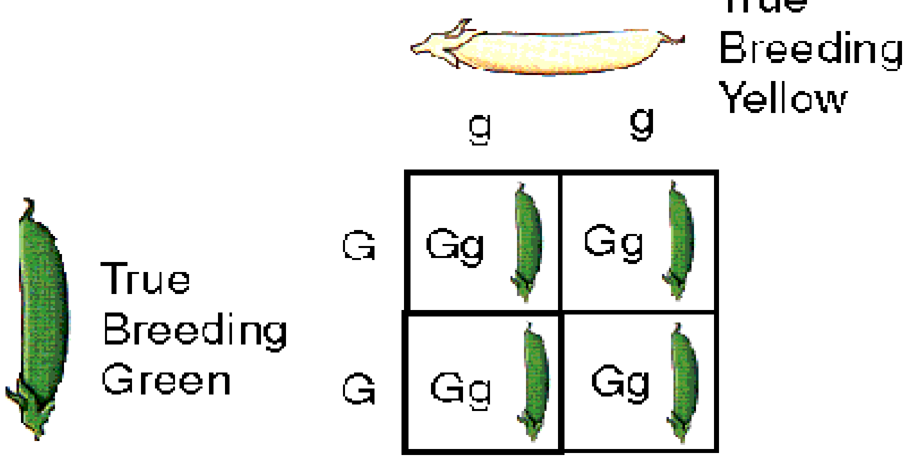


Other Example: Pods color

F1



- cross-pollination between true breeding green and yellow pods
- all F1 green

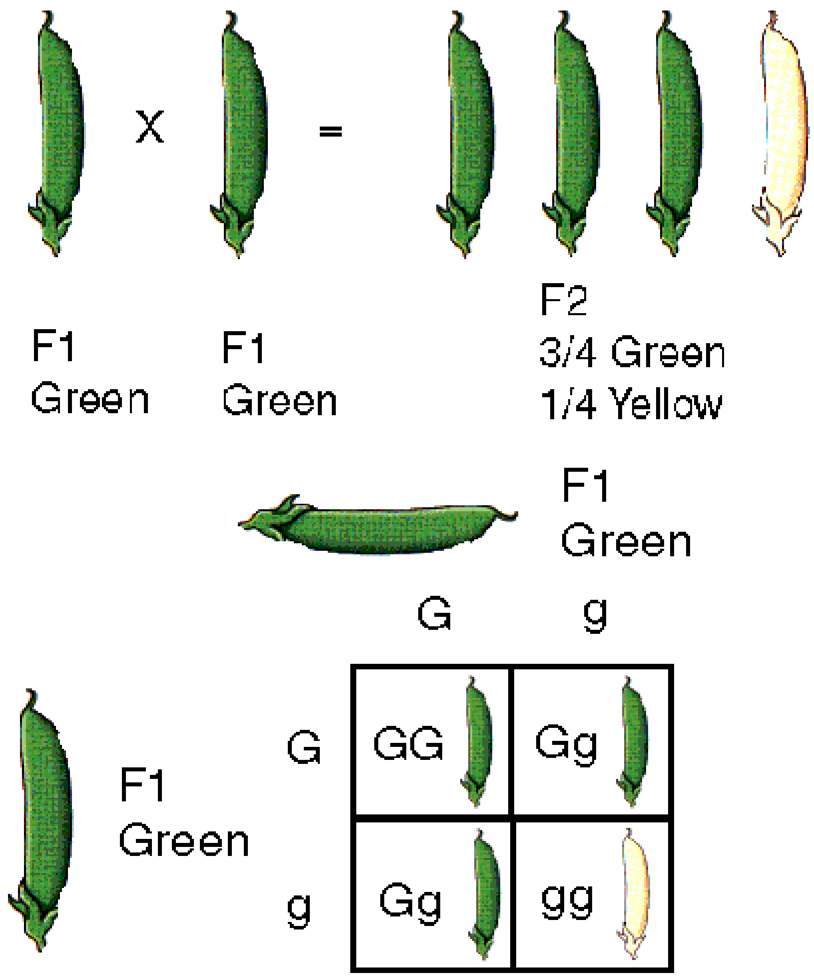


F1 Generation

Gg = heterozygous

Other Example: Pods color

F2



- self-pollination of green F1 plants
- ¾ in F2 green, ¼ yellow
- 3 : 1 ratio in pod colour in F2

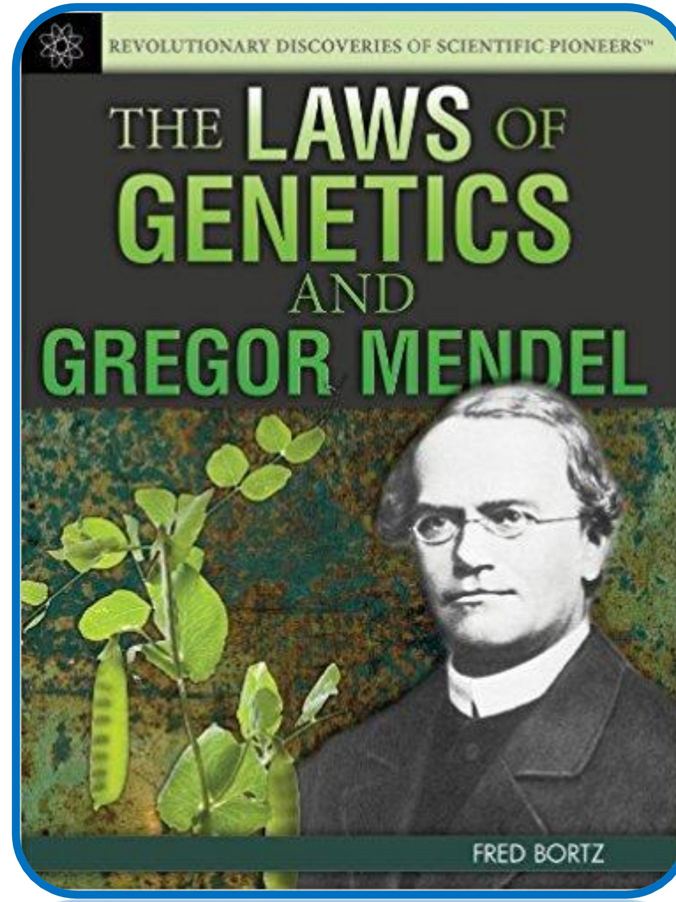
G = dominant = green

g = recessive = yellow

GG, gg = homozygous

Mendel's Generalization

- Alternative versions of genes account for variations in inherited characters
- A capital letter represents a **dominant** allele, and a lowercase letter represents a **recessive** allele
- For each character, an organism inherits two genes, one from each parent
  - two gametes form somatic cells
  - one allele comes from the **mother** and the other one from the **father**
- If the two alleles differ:
  - dominant allele is fully expressed in the organism's appearance (**phenotype)**
  - recessive allele has no noticeable effect on the organism's appearance (**genotype**)

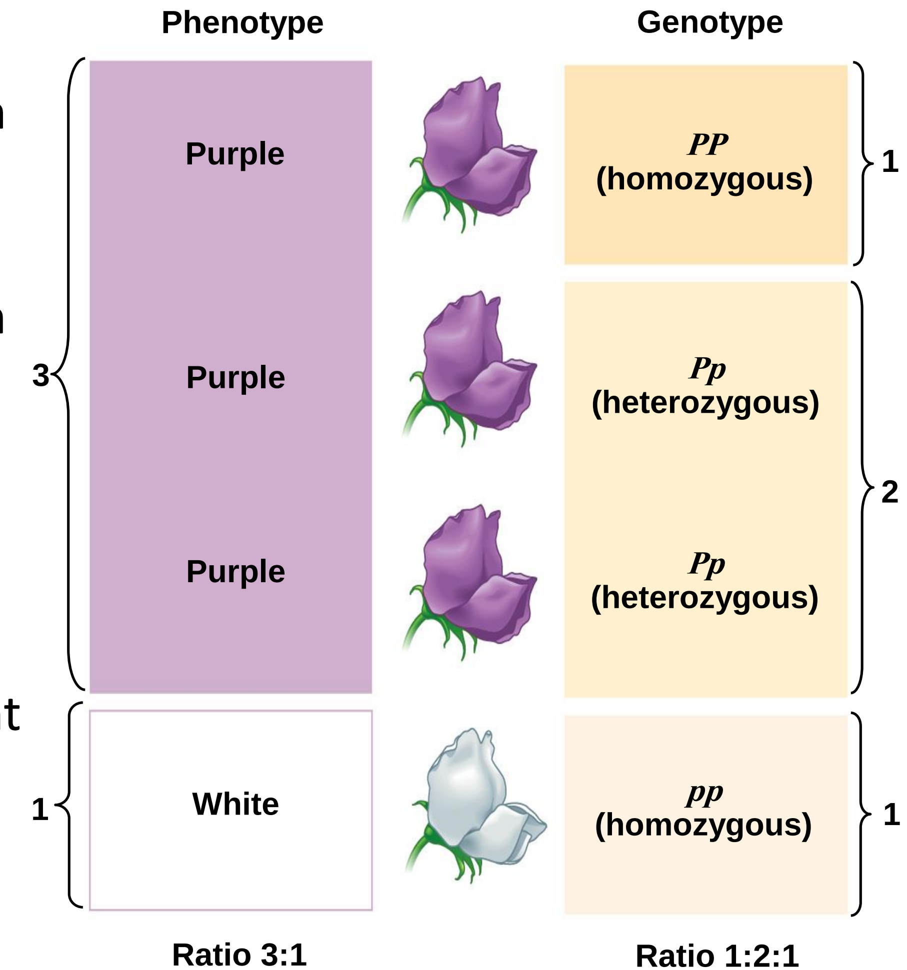


Useful Genetic Vocabulary

- 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

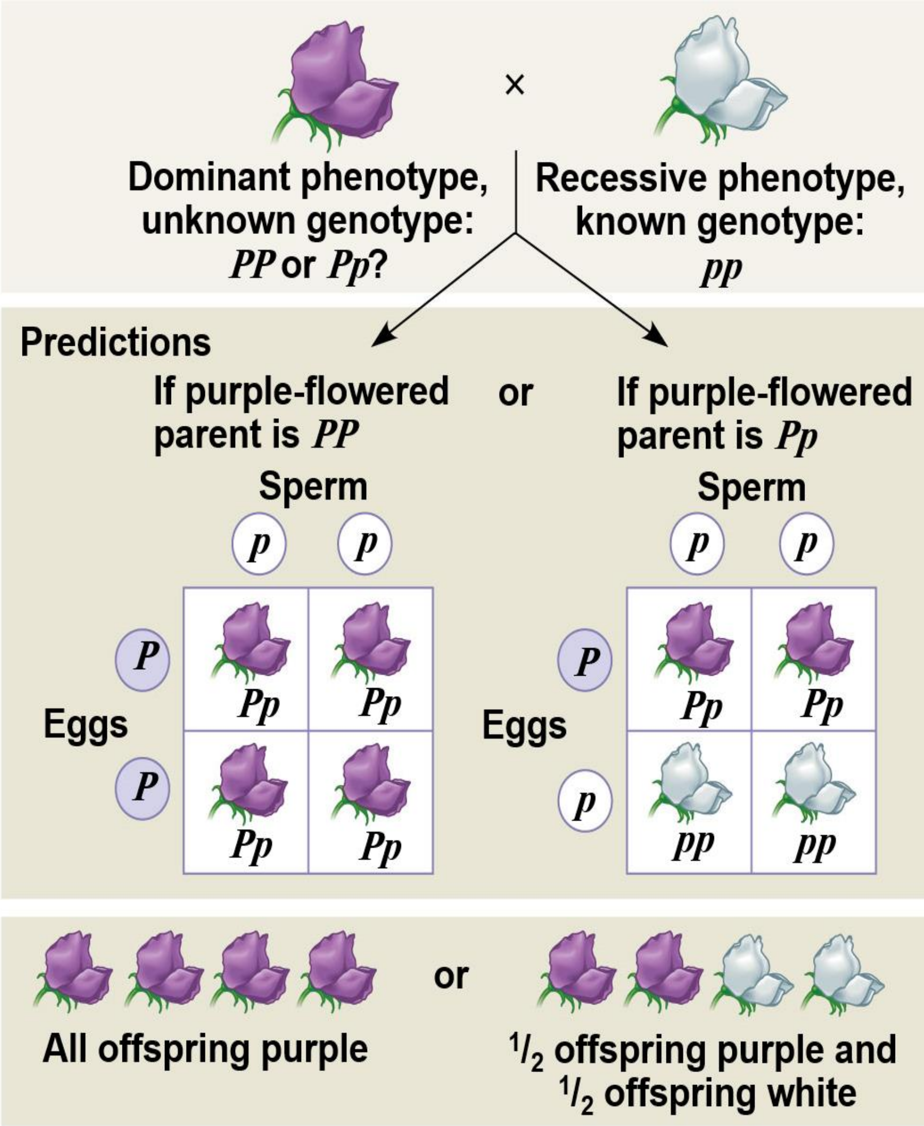
Useful Genetic Vocabulary

- Because of the different effects of **dominant** and **recessive** alleles, an organism's traits do not always reveal its genetic composition
- Therefore, we distinguish between an organism's **phenotype**, or physical appearance, and its **genotype**, or genetic makeup
- In the example of tall tree in pea plants, TT and Tt plants have the same phenotype (Tall) but different genotypes



The Testcross

- How can we tell the genotype of an individual with the dominant phenotype?
- Such an individual could be either homozygous dominant or heterozygous
- The answer is to carry out a testcross: breeding the mystery individual with a homozygous recessive individual
- If any offspring display the recessive phenotype, the mystery parent must be heterozygous



Mendel's law of Independent Assortment

monohybrids

- Mendel derived the law of segregation by following a **single character**
- The F1 offspring produced in this cross were **monohybrids**, individuals that are heterozygous for one character
- A cross between such heterozygotes is called a **monohybrid cross**

dihybrids

- Mendel identified his second law of inheritance by following **two characters** at the same time
- Crossing two true-breeding parents differing in two characters produces **dihybrids** in the F1 generation, heterozygous for both characters
- A dihybrid cross, a cross between F1 dihybrids, can determine whether two characters are transmitted to offspring as a package or **independently**

The law of independent assortment:

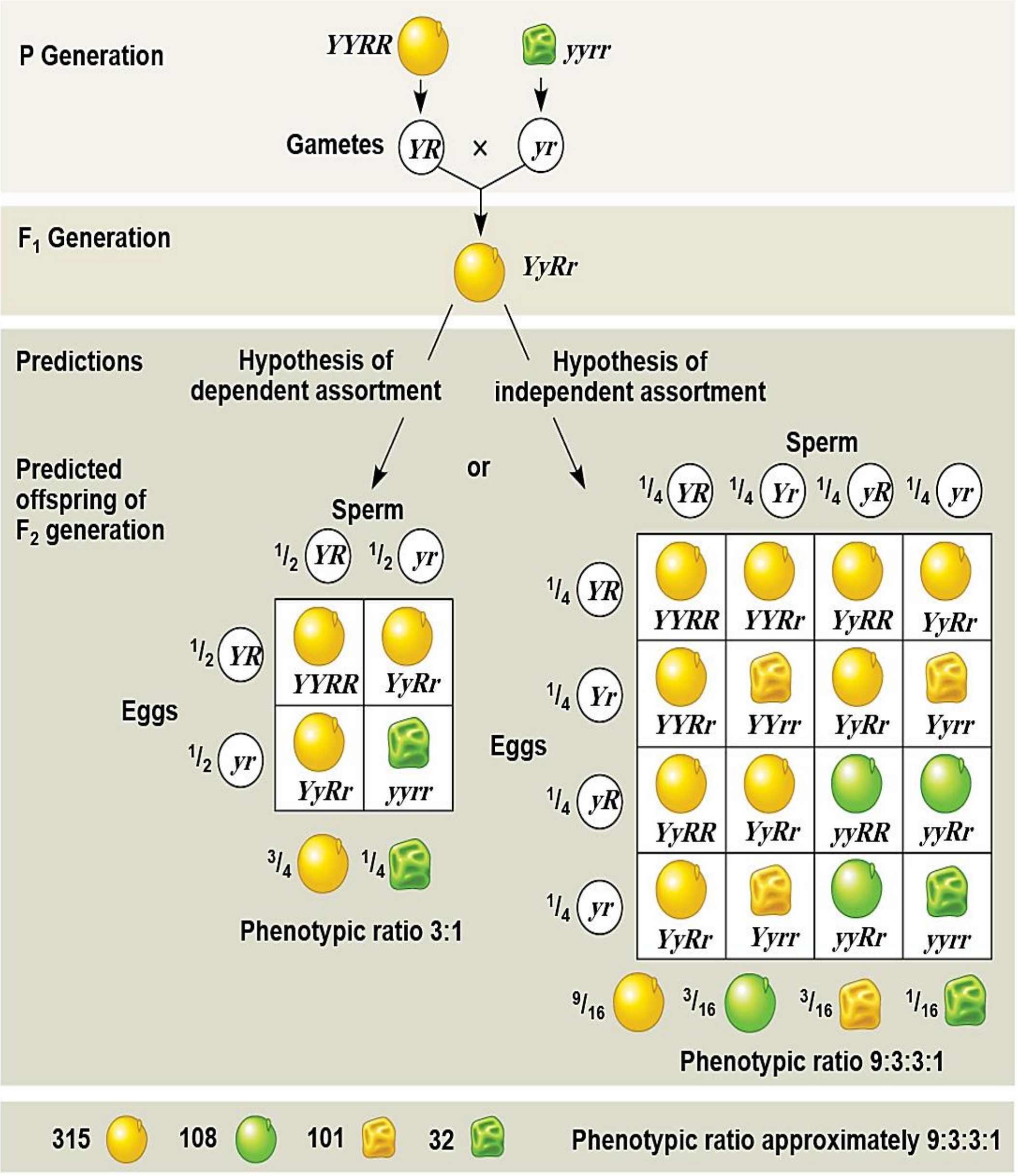
states that each pair of alleles segregates independently of each other pair of alleles during gamete formation

(Strictly speaking, this law applies only to genes on different, nonhomologous chromosomes or those far apart on the same chromosome)

But

Genes located near each other on the same chromosome tend to be inherited together

law of Independent Assortment

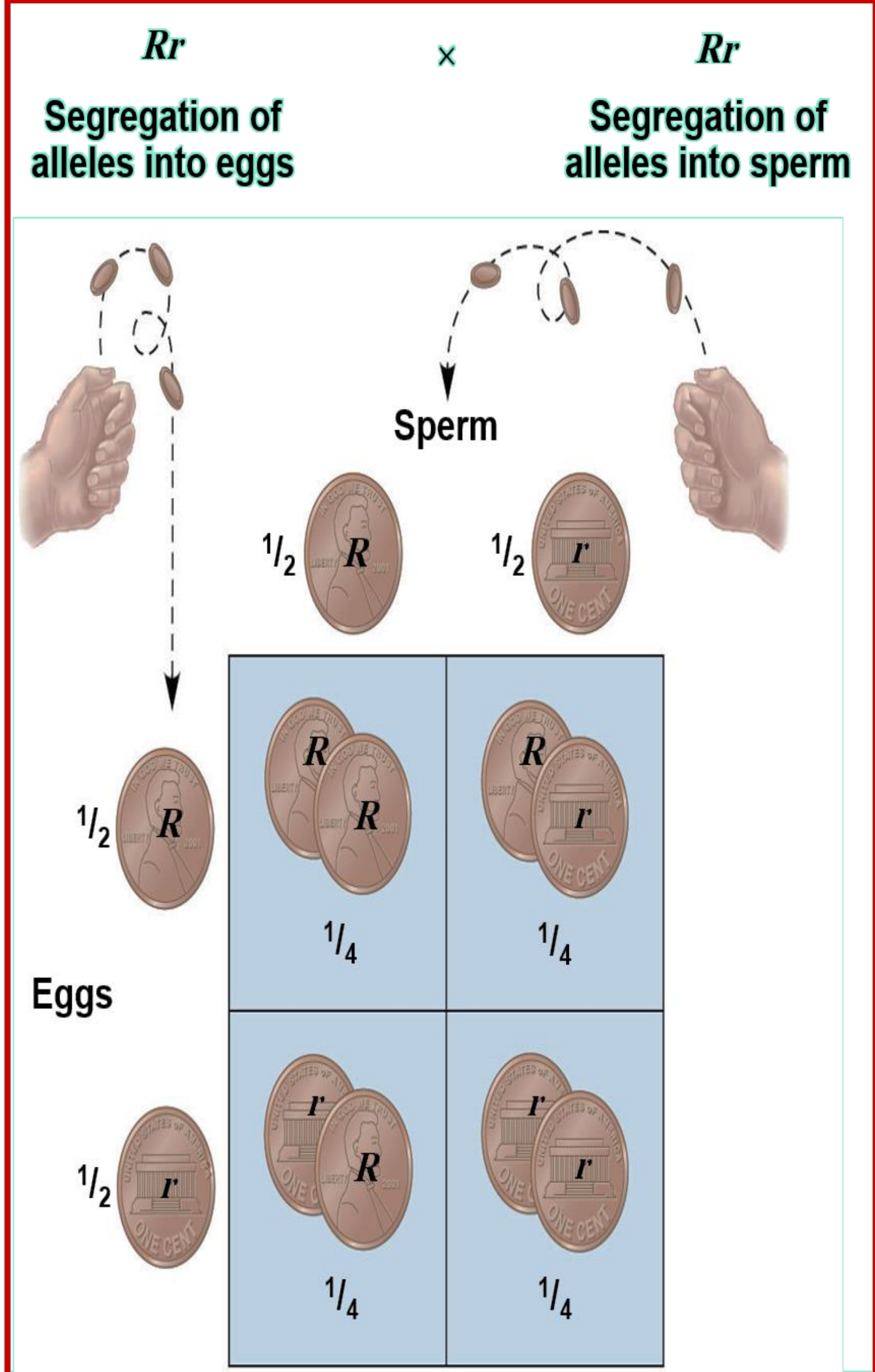


2: The laws of probability govern Mendelian inheritance

- Mendel's laws of segregation and independent assortment reflect the rules of probability
- When tossing a coin, the outcome of one toss has no impact on the outcome of the next toss
- In the same way, the alleles of one gene segregate into gametes independently of another gene's alleles

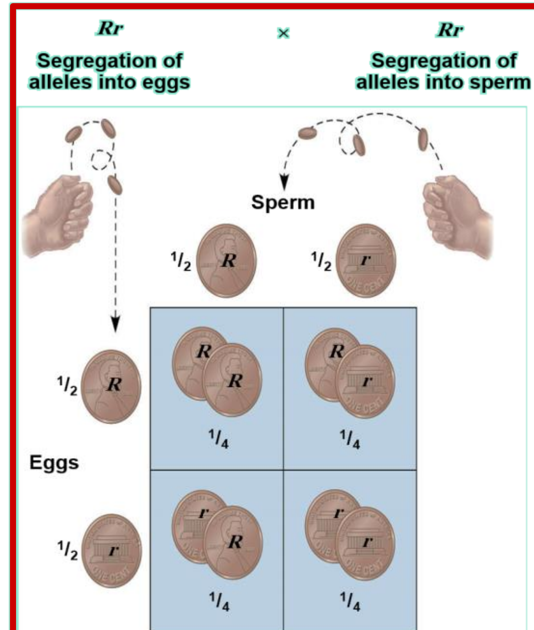
Mendel’s laws & The laws of probability

- The multiplication rule states that the probability that two or more independent events will occur together is the product of their individual probabilities
- Probability in an F1 monohybrid cross can be determined using the multiplication rule
- Segregation in a heterozygous plant is like flipping a coin: Each gamete has a chance of carrying the dominant allele and a chance of carrying the recessive allele



Solving Complex Genetics Problems with the Rules of Probability

- We can apply the multiplication and addition rules to **predict** the outcome of crosses involving multiple characters
- A **dihybrid** or other **multicharacter cross** is equivalent to **two or more** independent **monohybrid crosses** occurring simultaneously
- In **calculating** the chances for various **genotypes**, each **character is considered separately**, and **then the individual probabilities are multiplied**



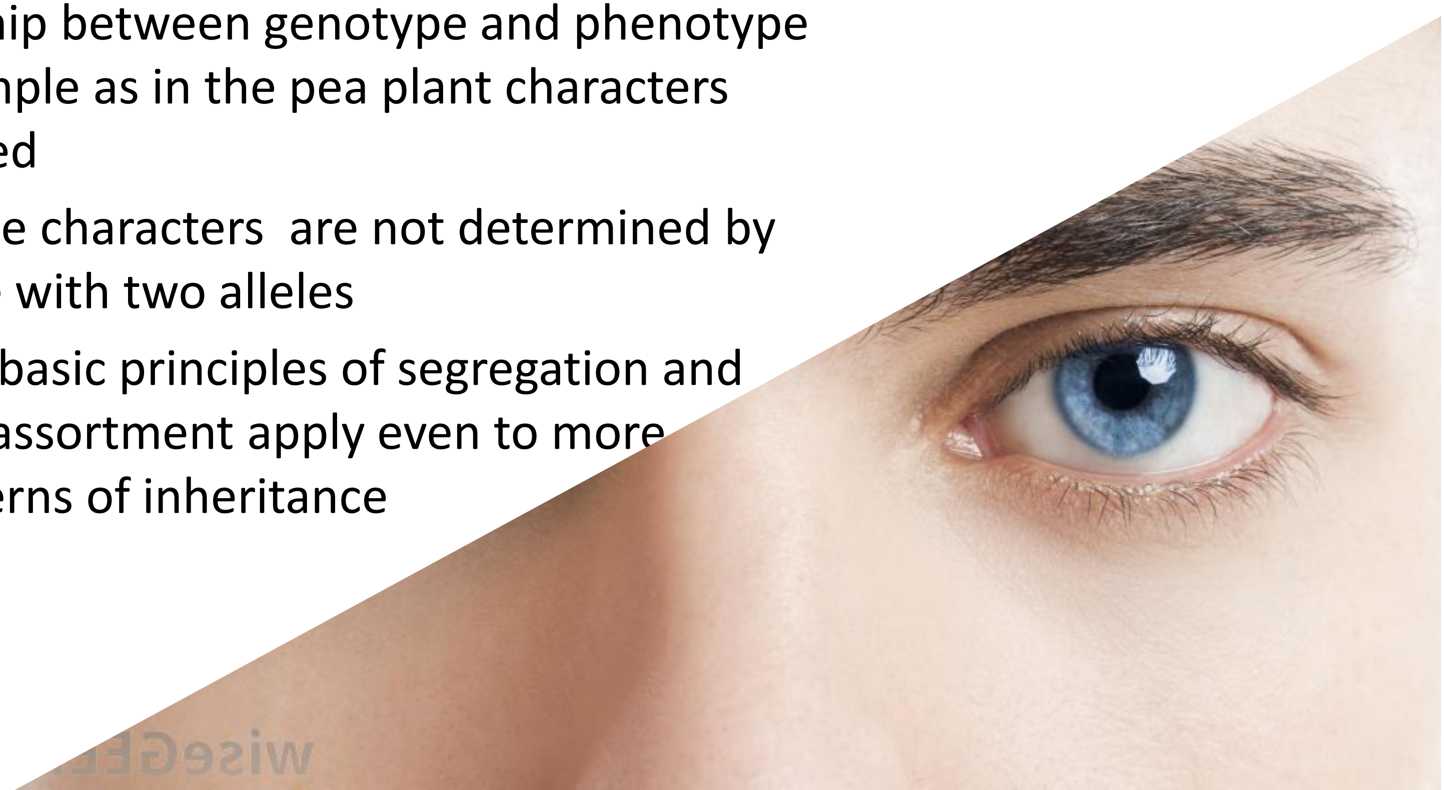
Probability of *YYRR* = $^1/_4$ (probability of *YY*) × $^1/_4$ (*RR*) = $^1/_{16}$

Probability of *YyRR* = $^1/_2$ (*Yy*) × $^1/_4$ (*RR*) = $^1/_8$

Must be know

Inheritance patterns are often more complex than predicted by simple Mendelian genetics

- The relationship between genotype and phenotype is rarely as simple as in the pea plant characters Mendel studied
- Many heritable characters are not determined by only one gene with two alleles
- However, the basic principles of segregation and independent assortment apply even to more complex patterns of inheritance



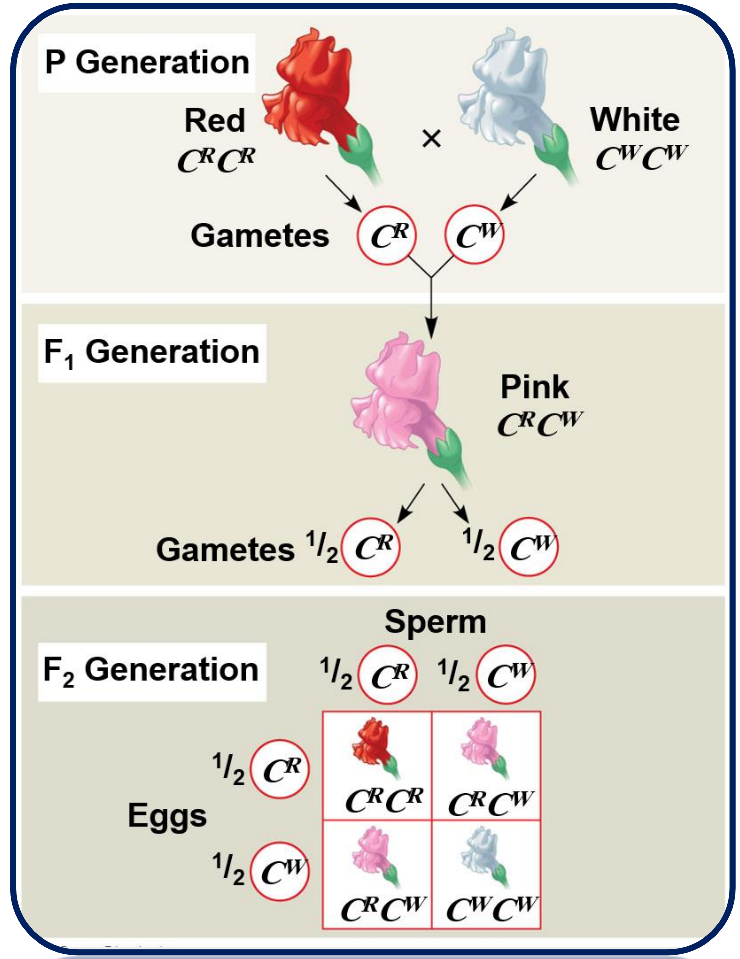
Non Mendelian Genetics for a Single Gene

- Inheritance of characters by a single gene may deviate from simple Mendelian patterns in the following situations:
  - When alleles are not completely dominant or recessive
  - When a gene has more than two alleles
  - When a gene produces multiple phenotypes

Degrees of Dominance

- **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are identical
- **In incomplete dominance**, the phenotype of F1 hybrids is somewhere between the phenotypes of the two parental varieties
- In codominance, two dominant alleles affect the phenotype in separate, distinguishable ways

Definition of incomplete dominance : The phenotype of the heterozygote is intermediate between two homozygotes.



Multiple Alleles (Codominance)

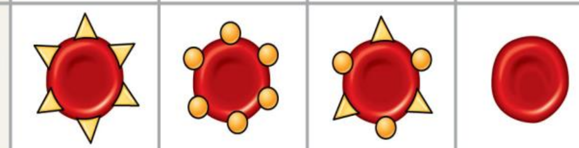
- Most genes exist in populations in more than two allelic forms
- For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme (I) that attaches A or B carbohydrates to red blood cells: IA, IB, and i.
- The enzyme encoded by the IA allele adds the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

Definition of Codominance : Two nonidentical alleles of a pair express two different phenotype in the heterozygote.

(a) The three alleles for the ABO blood groups and their carbohydrates

Allele	I^A	I^B	i
Carbohydrate	A △	B ○	none

(b) Blood group genotypes and phenotypes

Genotype	I^AI^A or I^Ai	I^BI^B or I^Bi	I^AI^B	ii
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

The ABO blood system

Genotypes	Phenotypes (Blood types)
$I^A I^A$	A
$I^A I^B$	AB
I^Ai	A
$I^B I^B$	B
I^Bi	B
ii	O

Note:

- Blood types A and B have two possible genotypes – homozygous and heterozygous.
- Blood types AB and O only have one genotype each.

Pleiotropy

- Most genes have multiple phenotypic effects, a property called pleiotropy
- For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as cystic fibrosis and **sickle-cell disease**

Definition of pleiotropy: A single gene affects more than one phenotype

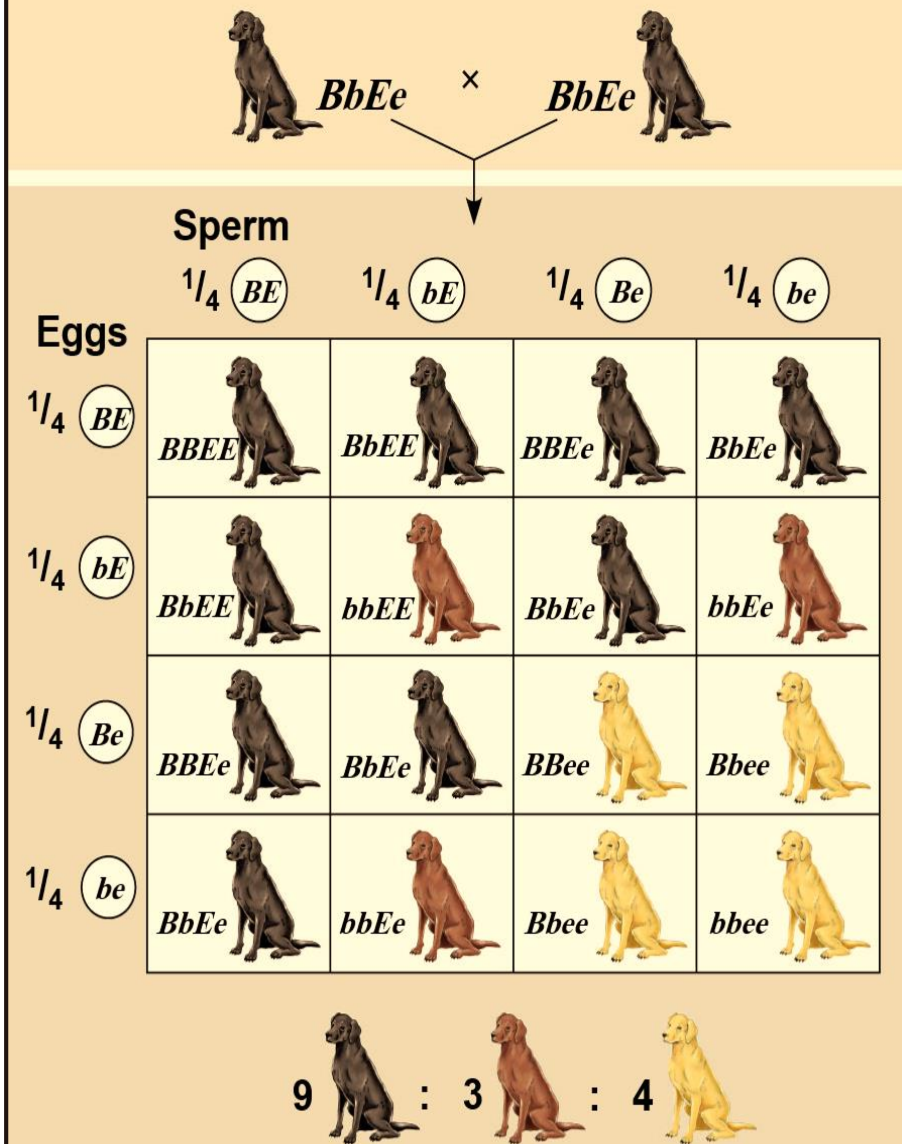
Extending Mendelian Genetics for Two or More Genes

- Some traits may be determined by two or more genes

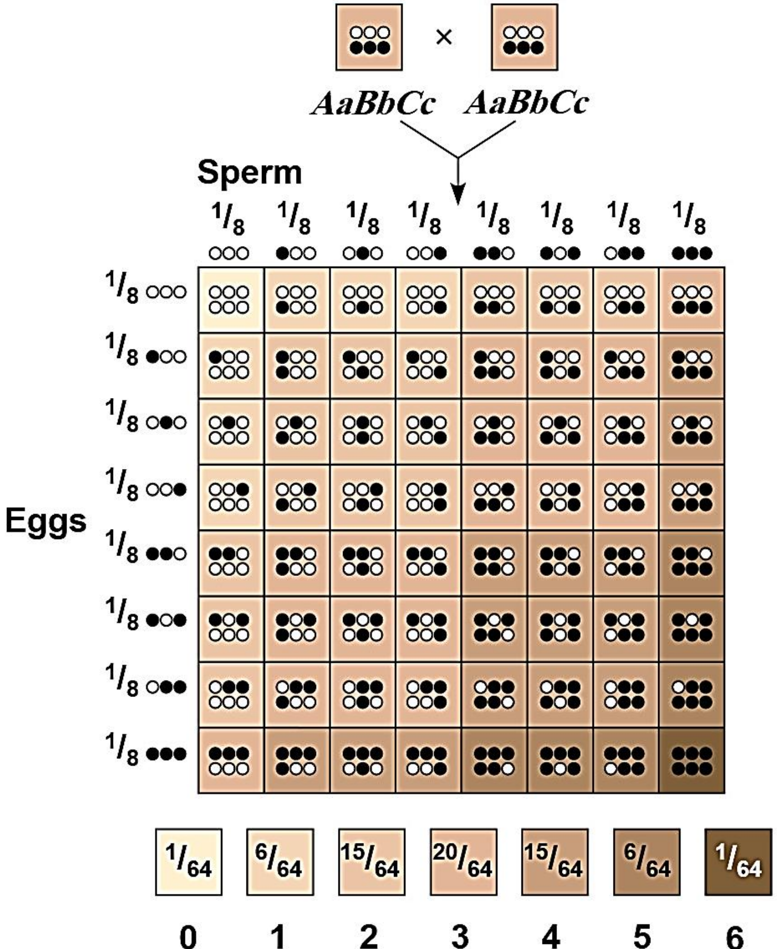
Epistasis

- In epistasis, a gene at one locus alters the phenotypic expression of a gene at a second locus
- For example, in Labrador retrievers and many other mammals, coat color depends on two genes
- One gene determines the pigment color (with alleles B for black and b for brown)
- The other gene (with alleles C for color and c for no color) determines whether the pigment will be deposited in the hair

Definition of Epistasis: Interaction of two or more gene pairs at different loci influence the same trait, but one allele has an overriding effect on the phenotype.



Polygenic Inheritance



, Inc.

Skin color in humans is an example of polygenic inheritance

Gene 1	dd	dD	dD	DD	Dd	Dd	DD
Gene 2	dd	dd	dD	Dd	Dd	DD	DD
Gene 3	dd	dd	dd	dd	DD	DD	DD
	Very light			Medium			Very dark
# of light "d" alleles	6	5	4	3	2	1	0
# of dark "D" alleles	0	1	2	3	4	5	6

Quantitative characters are those that vary in the population along a continuum

Quantitative variation usually indicates polygenic inheritance, an additive effect of two or more genes on a single phenotype

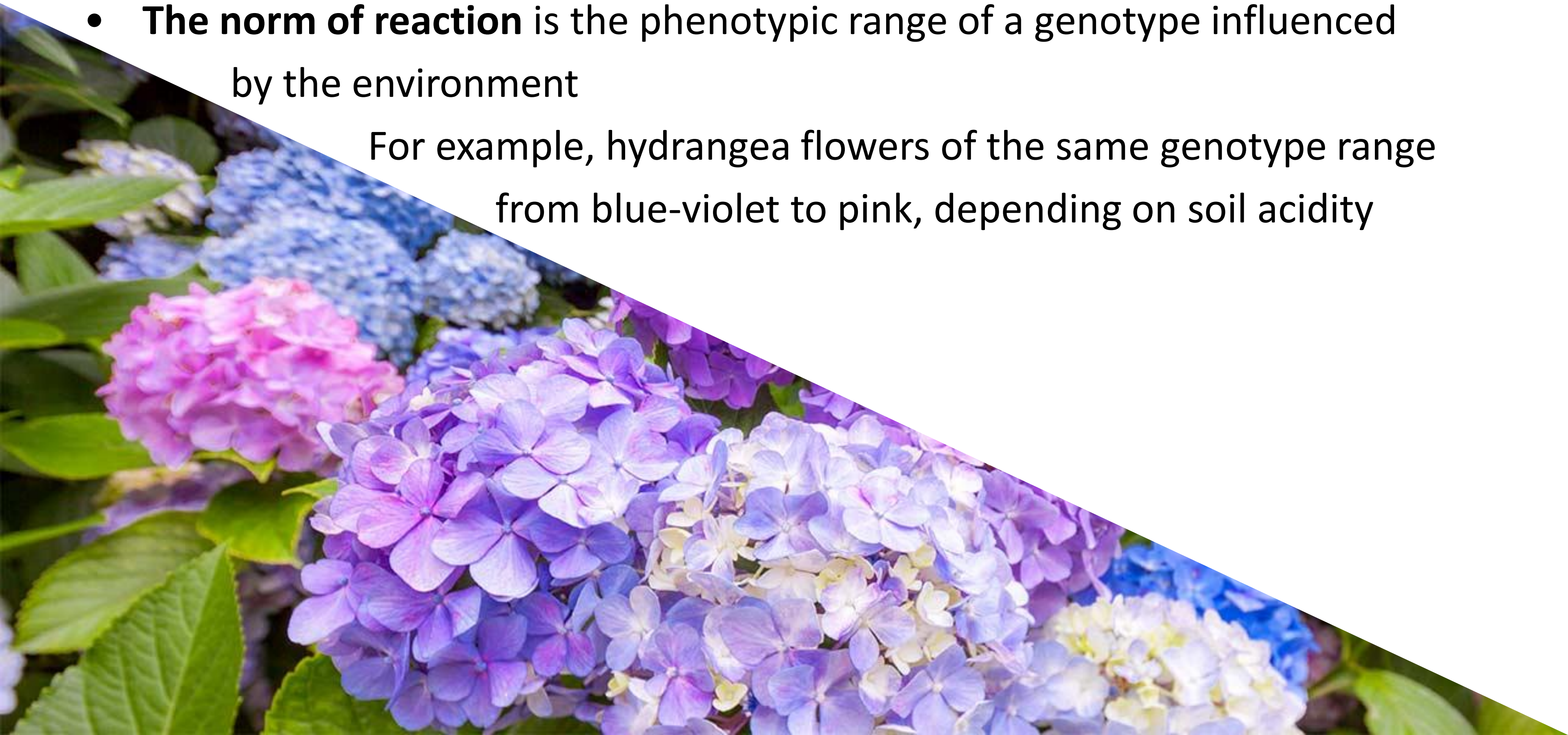
Definition of Polygenic: Continuous range of small differences in a given trait among all the individuals of a population due to inheritance of multiple alleles that affect the same trait.

Nature and Nutrition

The Environmental Impact on Phenotype

- Another departure from Mendelian genetics arises when the phenotype for a character **depends on environment** as well as genotype
- **The norm of reaction** is the phenotypic range of a genotype influenced by the environment

For example, hydrangea flowers of the same genotype range from blue-violet to pink, depending on soil acidity



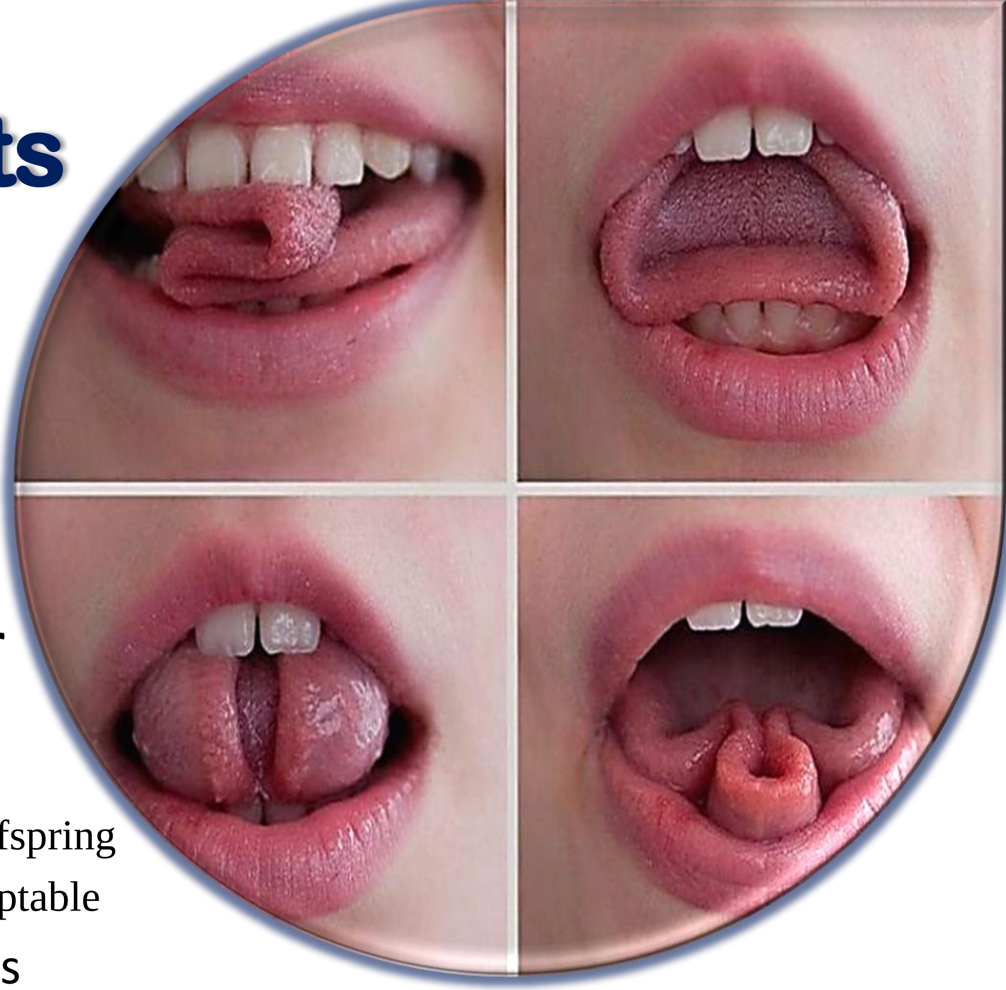
Nature and Nutrition

- Norms of reaction are generally broadest for **polygenic characters**
- Such characters are called multifactorial because genetic and environmental factors collectively influence phenotype

Integrating a Mendelian View of Heredity and Variation

- An organism's phenotype includes its physical appearance, internal anatomy, physiology, and behavior
- An organism's phenotype reflects its overall genotype and unique environmental history

Many human traits follow Mendelian patterns of inheritance

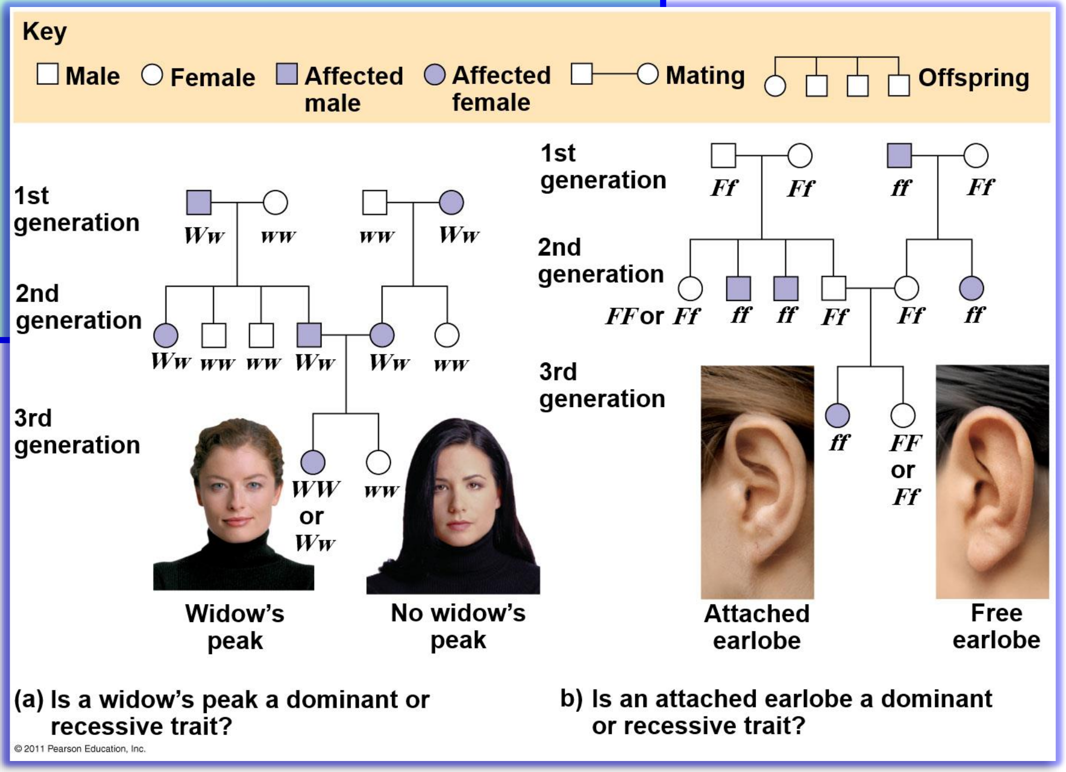


- **Humans are not good subjects for genetic research**
  - Generation time is too long
  - Parents produce relatively few offspring
  - Breeding experiments are unacceptable
- However, basic Mendelian genetics endures as the foundation of human genetics

So What? **Pedigree**

Pedigree Analysis

- A pedigree is a family tree that describes the interrelationships of parents and children across generations
- Inheritance patterns of particular traits can be traced and described using pedigrees



Test your Knowledge


End of Lecture
Good Luck!
See you
in next lecture…
Greetings of
DoctorQari
THE END
End Lecture 6

- Pedigrees can also be used to make predictions about future offspring
- We can use the multiplication and addition rules to predict the probability of specific phenotypes

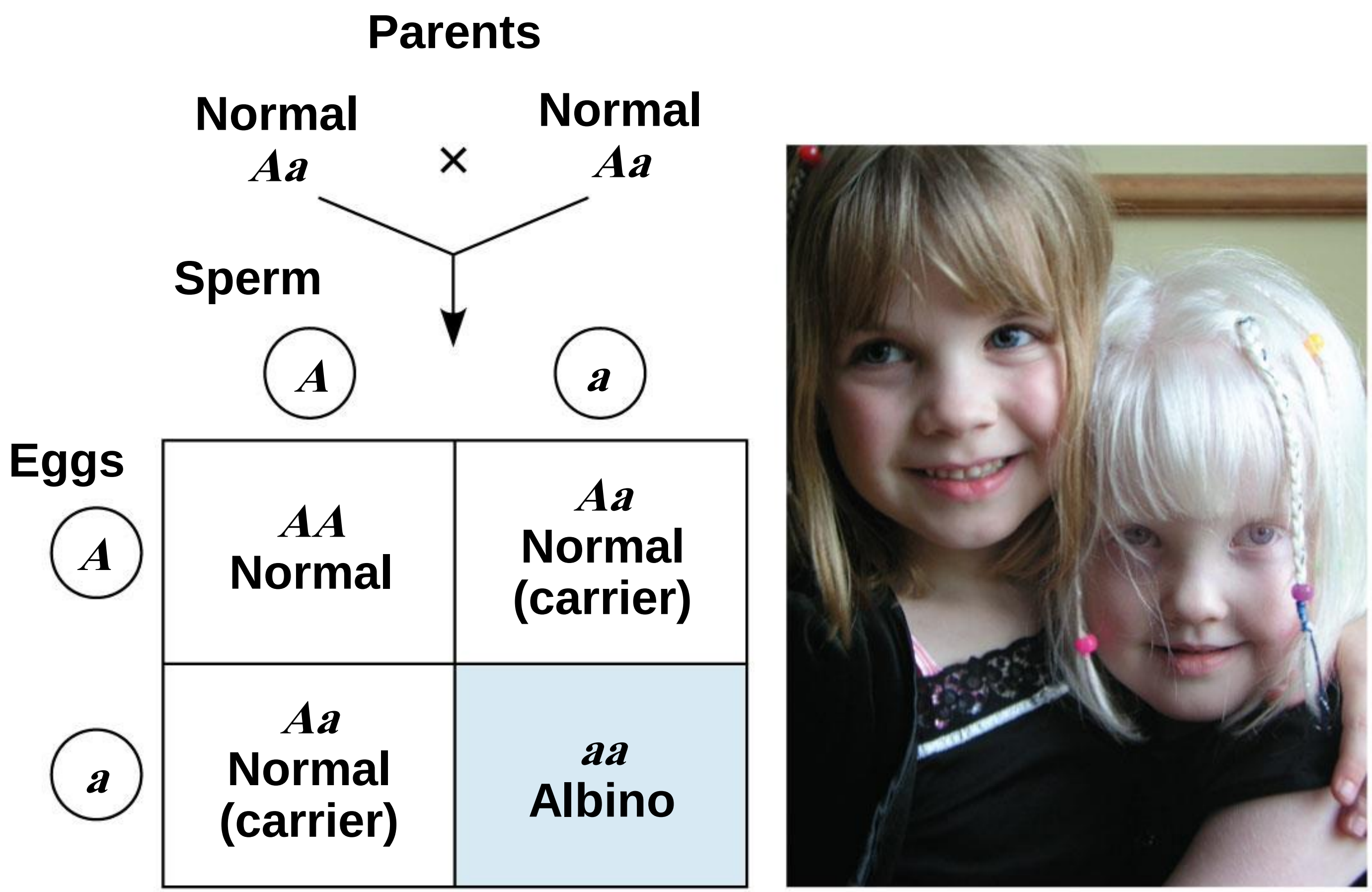
Recessively Inherited Disorders

- Many genetic disorders are inherited in a recessive manner
- These range from relatively mild to life-threatening

The Behavior of Recessive Alleles

- Recessively inherited disorders show up only in individuals homozygous for the allele
- Carriers are heterozygous individuals who carry the recessive allele but are phenotypically normal; most individuals with recessive disorders are born to carrier parents
- Albinism is a recessive condition characterized by a lack of pigmentation in skin and hair

Figure 14.16



Parents
Normal
Aa
×
Normal
Aa
Sperm
A
a
Eggs
A
a
AA
Normal
Aa
Normal
(carrier)
Aa
Normal
(carrier)
aa
Albino

- If a recessive allele that causes a disease is rare, then the chance of two carriers meeting and mating is low
- Consanguineous matings (i.e., matings between close relatives) increase the chance of mating between two carriers of the same rare allele
- Most societies and cultures have laws or taboos against marriages between close relatives

Sickle-Cell Disease: A Genetic Disorder with Evolutionary Implications

- Sickle-cell disease affects one out of 400 African-Americans
- The disease is caused by the substitution of a single amino acid in the hemoglobin protein in red blood cells
- In homozygous individuals, all hemoglobin is abnormal (sickle-cell)
- Symptoms include physical weakness, pain, organ damage, and even paralysis

Fig. 14-UN1

- Heterozygotes (said to have sickle-cell trait) are usually healthy but may suffer some symptoms
- About one out of ten African Americans has sickle cell trait, an unusually high frequency of an allele with detrimental effects in homozygotes
- Heterozygotes are less susceptible to the malaria parasite, so there is an advantage to being heterozygous