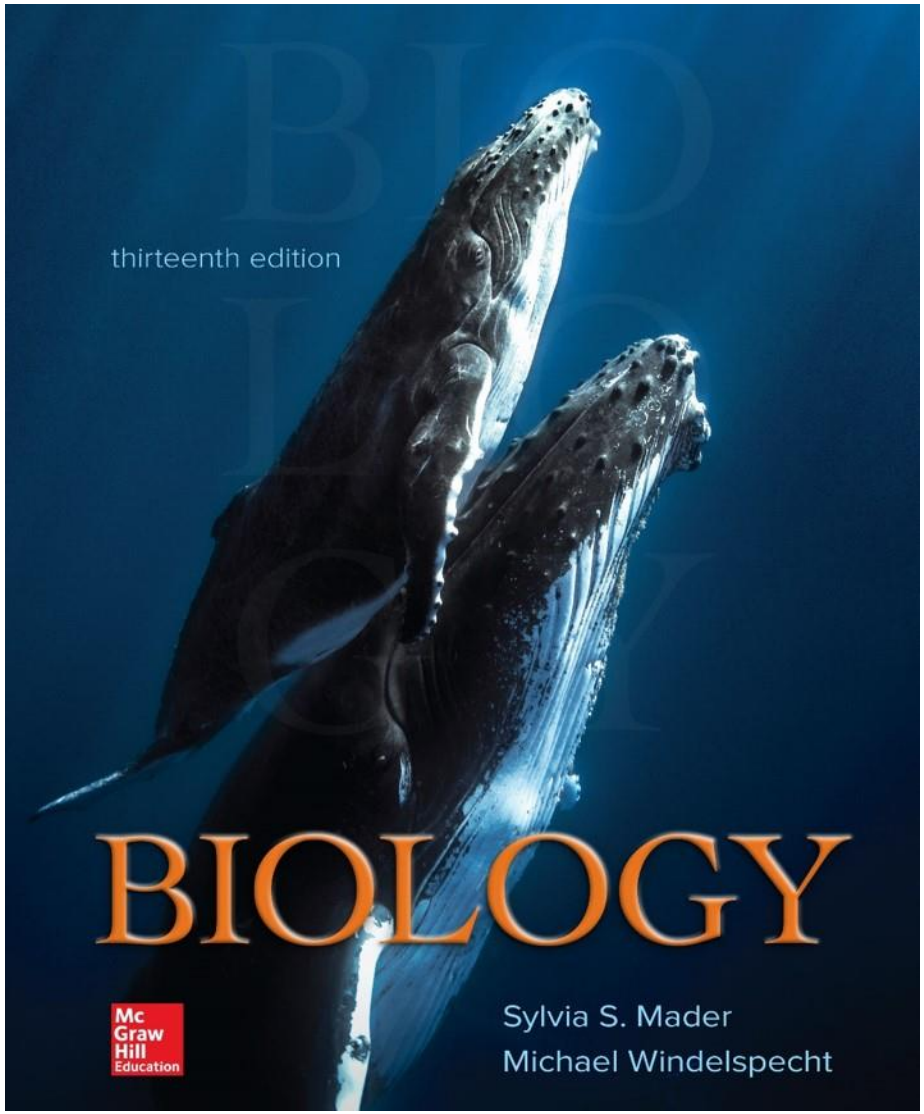


Biology

Sylvia S. Mader
Michael Windelspecht

Chapter 11
Mendelian Patterns of Inheritance
Lecture Outline

See separate FlexArt PowerPoint slides for all figures and tables pre-inserted into PowerPoint without notes.

Outline

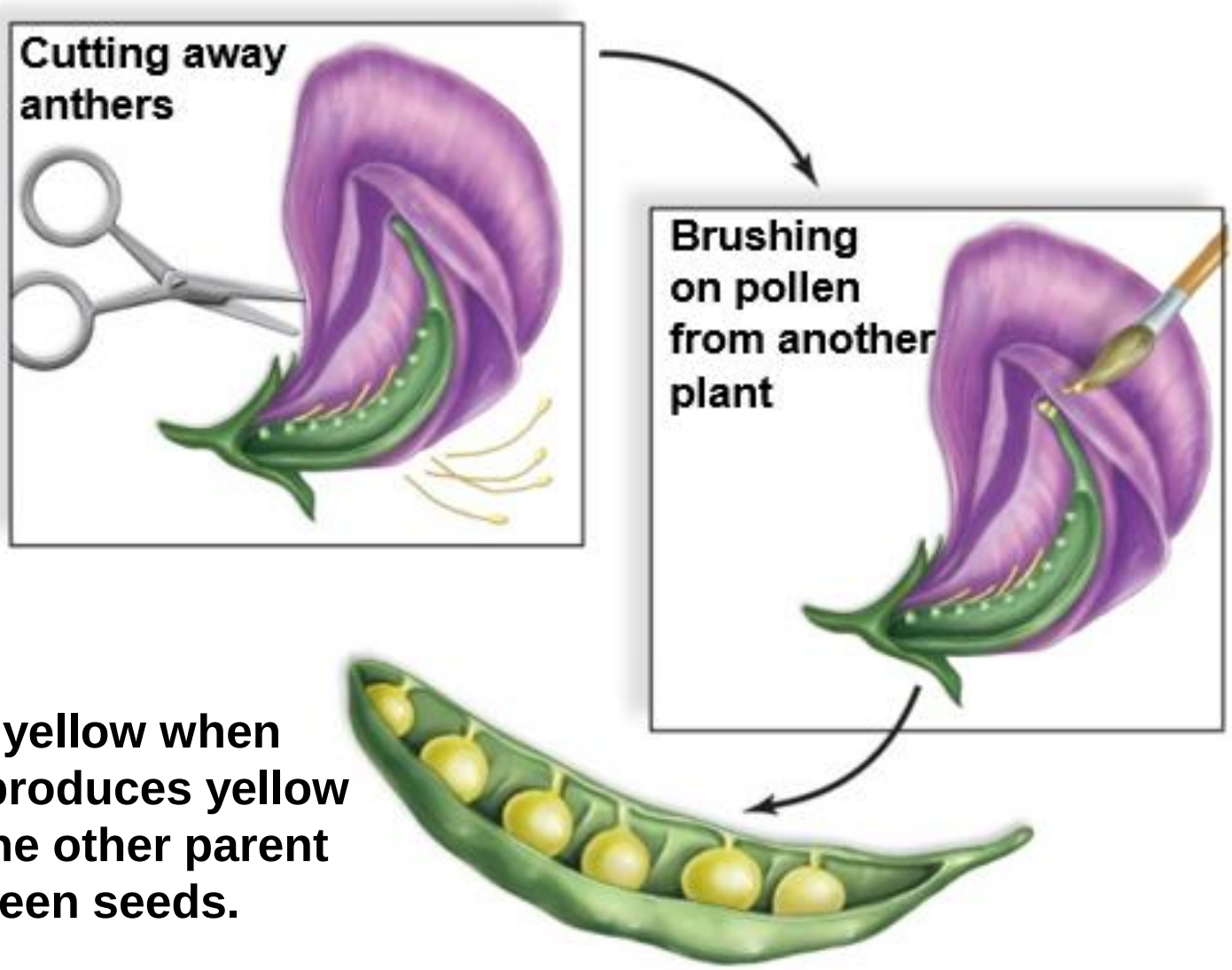
11.1 Gregor Mendel

11.2 Mendel's Laws

11.3 Mendelian Patterns of Inheritance and Human Disease

11.4 Beyond Mendelian Inheritance

Garden Pea Anatomy (2)



All peas are yellow when one parent produces yellow seeds and the other parent produces green seeds.

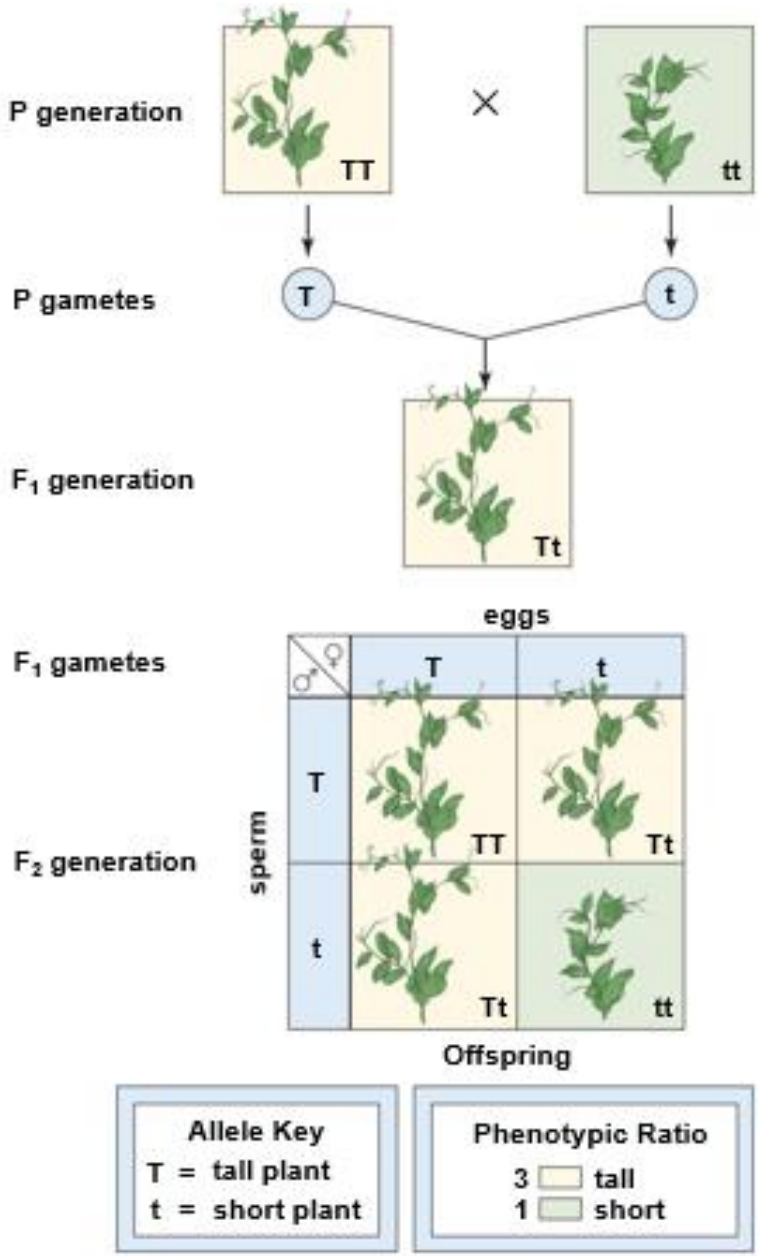
Jump to Garden Pea Anatomy (2) Long Description

11.2 Mendel's Laws

Mendel performed cross-breeding experiments.

- Used "true-breeding" (homozygous) plants
- Chose varieties that differed in only one trait (**monohybrid cross**)
- Performed reciprocal crosses
  - Parental generation = P
  - First filial generation offspring = F_1
  - Second filial generation offspring = F_2
- Formulated the **law of segregation**

Monohybrid Cross Done by Mendel (4)



Jump to Monohybrid Cross Done by Mendel (4) Long Description

Mendel's Laws (1)

Law of Segregation:

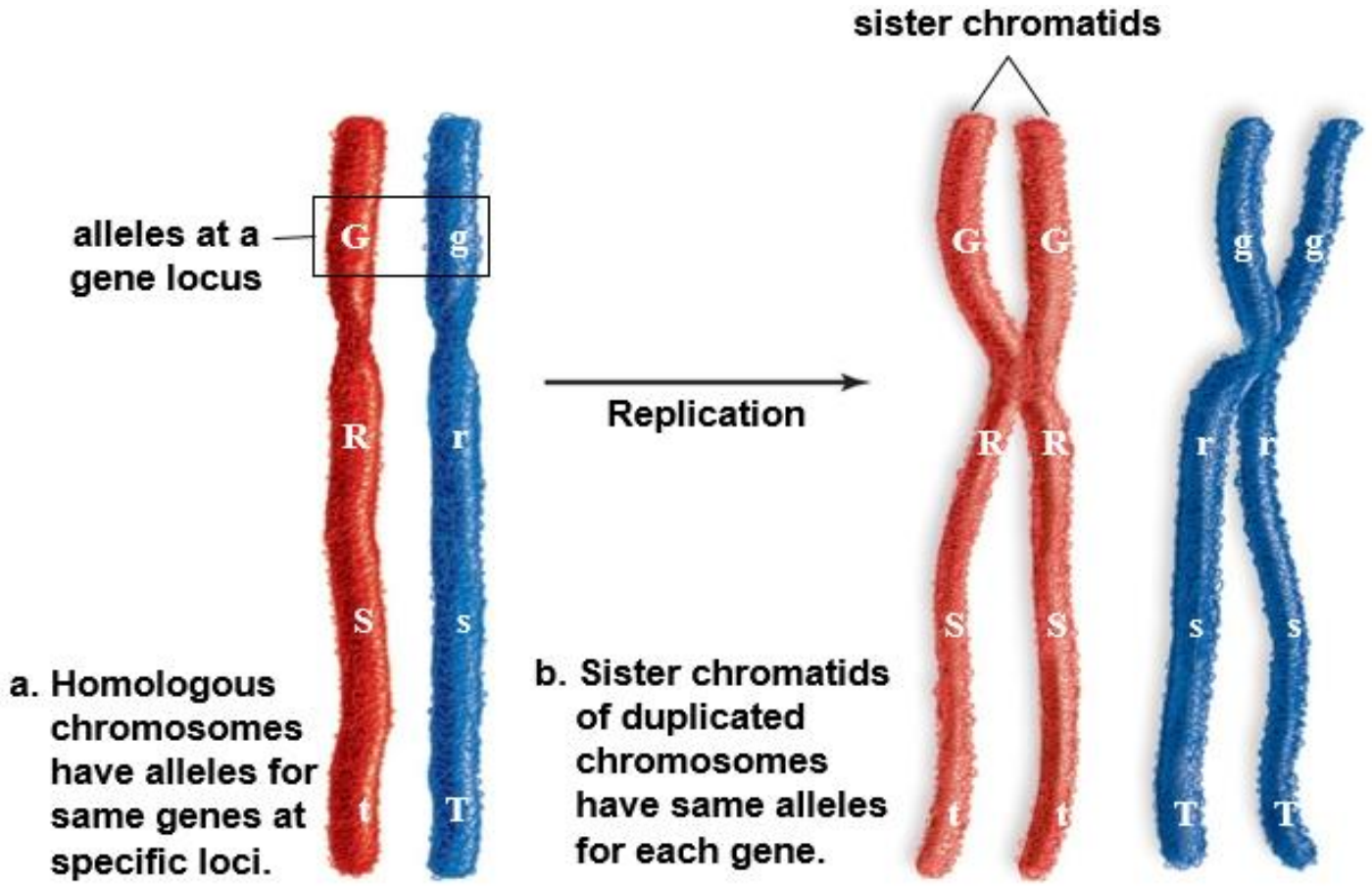
- Each individual has a pair of factors (alleles) for each trait.
- The factors (alleles) segregate (separate) during gamete (sperm and egg) formation.
- Each gamete contains only one factor (allele) from each pair of factors.
- Fertilization gives the offspring two factors for each trait.
- Results of the monohybrid cross: All F_1 plants were tall, disproving blending hypothesis.

Mendel's Laws (2)

Classical Genetics and Mendel's Cross:

- Each trait in a pea plant is controlled by two alleles (alternate forms of a gene).
- **Dominant allele** (capital letter) masks the expression of the **recessive allele** (lowercase).
- Alleles occur on a homologous pair of chromosomes at a particular gene locus.
  - **Homozygous** = identical alleles
  - **Heterozygous** = different alleles

Homologous Chromosomes



Jump to Homologous Chromosomes Long Description

Relationship Between Observed Phenotype and F_2 Offspring

Trait	Characteristics		F_2 Results		
	Dominant	Recessive	Dominant	Recessive	Ratio
Stem length	Tall 	Short 	787	277	2.84:1
Pod shape	Inflated 	Constricted 	882	299	2.95:1
Seed shape	Round 	Wrinkled 	5,474	1,850	2.96:1
Seed color	Yellow 	Green 	6,022	2,001	3.01:1
Flower position	Axial 	Terminal 	651	207	3.14:1
Flower color	Purple 	White 	705	224	3.15:1
Pod color	Green 	Yellow 	428	152	2.82:1
		Totals:	14,949	5,010	2.98:1

Jump to Relationship Between Observed Phenotype and F_2 Offspring Long Description

Mendel's Laws (3)

Genotype

- It refers to the two alleles an individual has for a specific trait.
- If identical, genotype is homozygous.
- If different, genotype is heterozygous.

Phenotype

- It refers to the physical appearance of the individual.

Mendel's Cross Viewed by Modern Genetics

The dominant and recessive alleles represent DNA sequences that code for proteins.

The dominant allele codes for the protein associated with the normal gene function within the cell.

The recessive allele represents a "loss of function."

During meiosis I, the homologous chromosomes separate.

- The two alleles separate from each other.
- The process of meiosis explains Mendel's law of segregation and why only one allele for each trait is in a gamete.

Mendel's Laws (4)

A **dihybrid cross** uses true-breeding plants differing in two traits.

Mendel tracked each trait through two generations.

- It started with true-breeding plants differing in two traits.
- The F_1 plants showed both dominant characteristics.
- F_1 plants self-pollinated.
- He observed phenotypes among F_2 plants.

Mendel formulated the **law of independent assortment**.

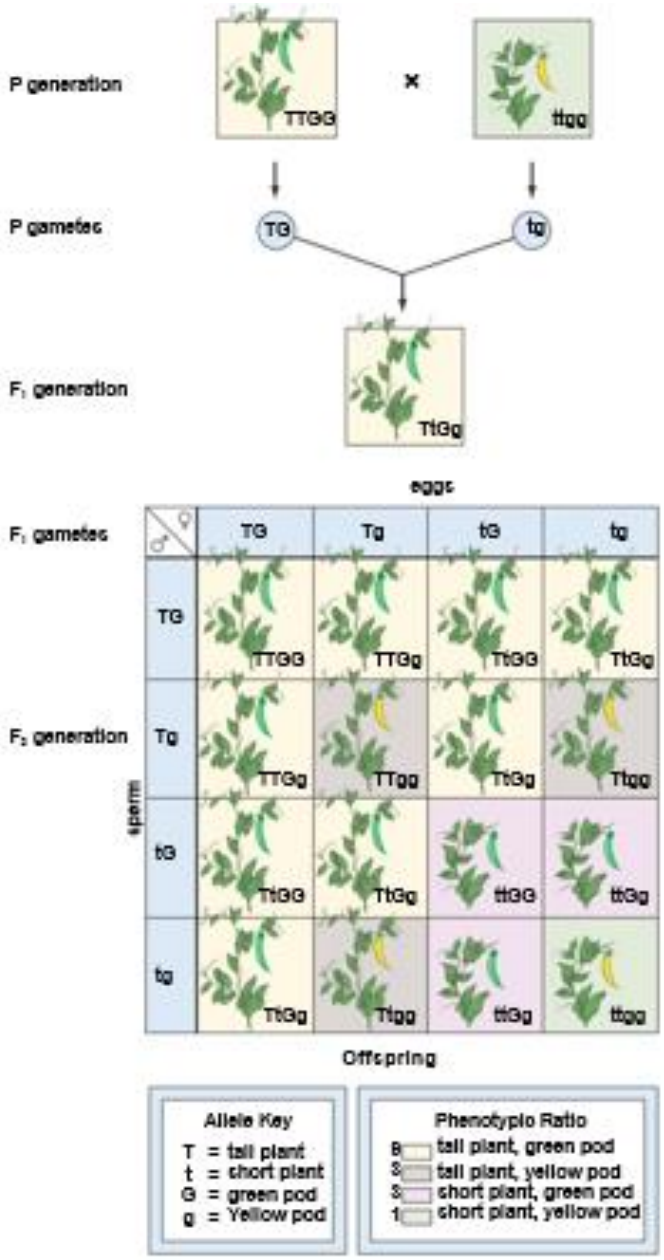
- The pair of factors for one trait segregate independently of the factors for other traits.
- All possible combinations of factors can occur in the gametes.

P generation is the parental generation in a breeding experiment.

F_1 generation is the first-generation offspring in a breeding experiment.

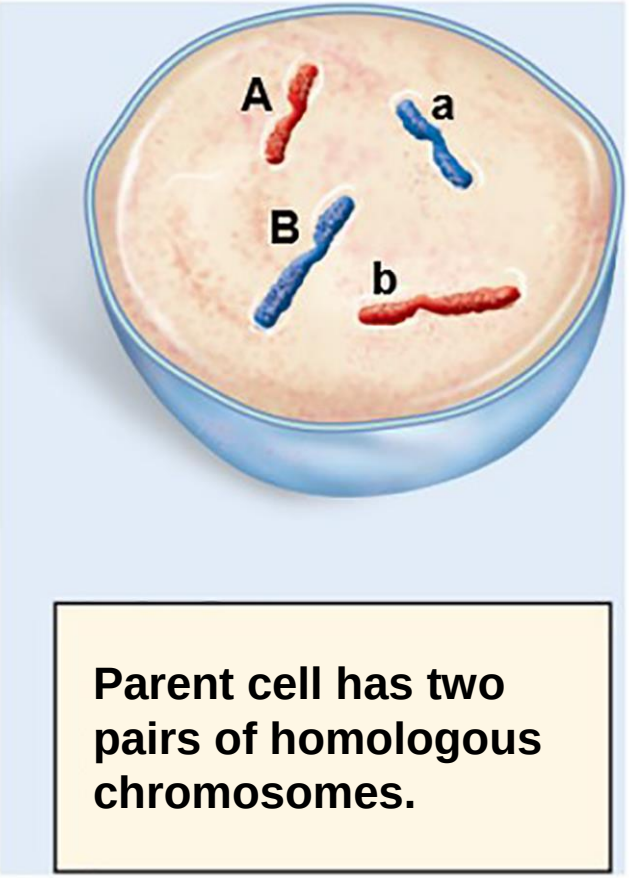
F_2 generation is the second-generation offspring in a breeding experiment.

Dihybrid Cross Done by Mendel

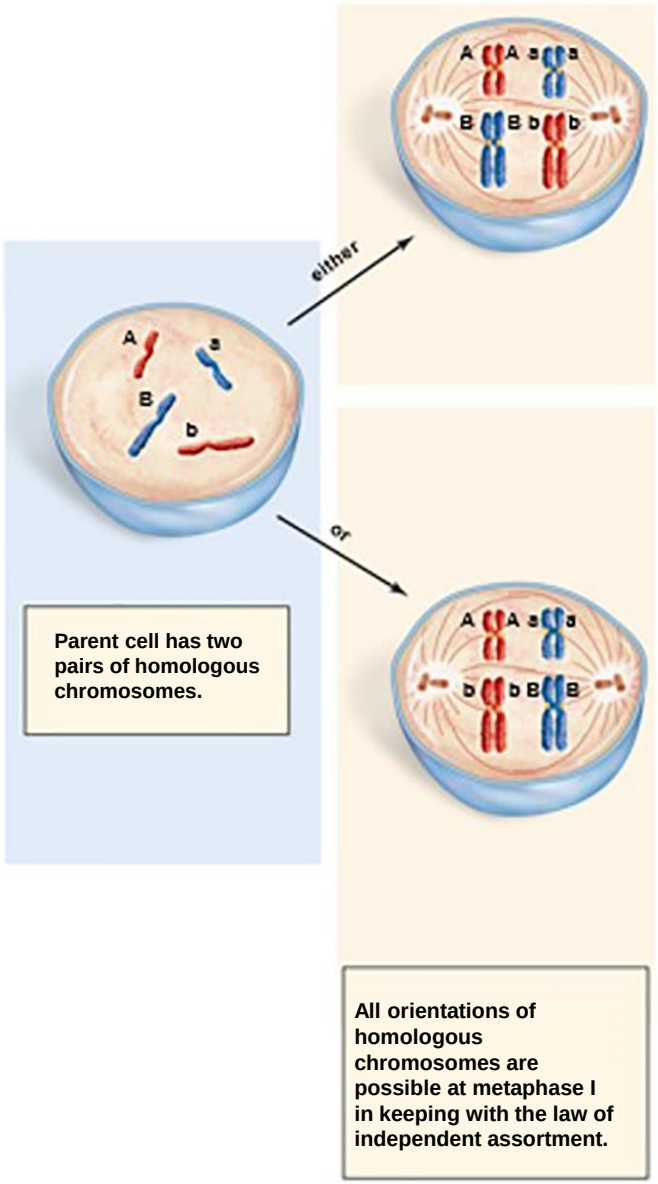


Jump to Dihybrid Cross Done by Mendel Long Description

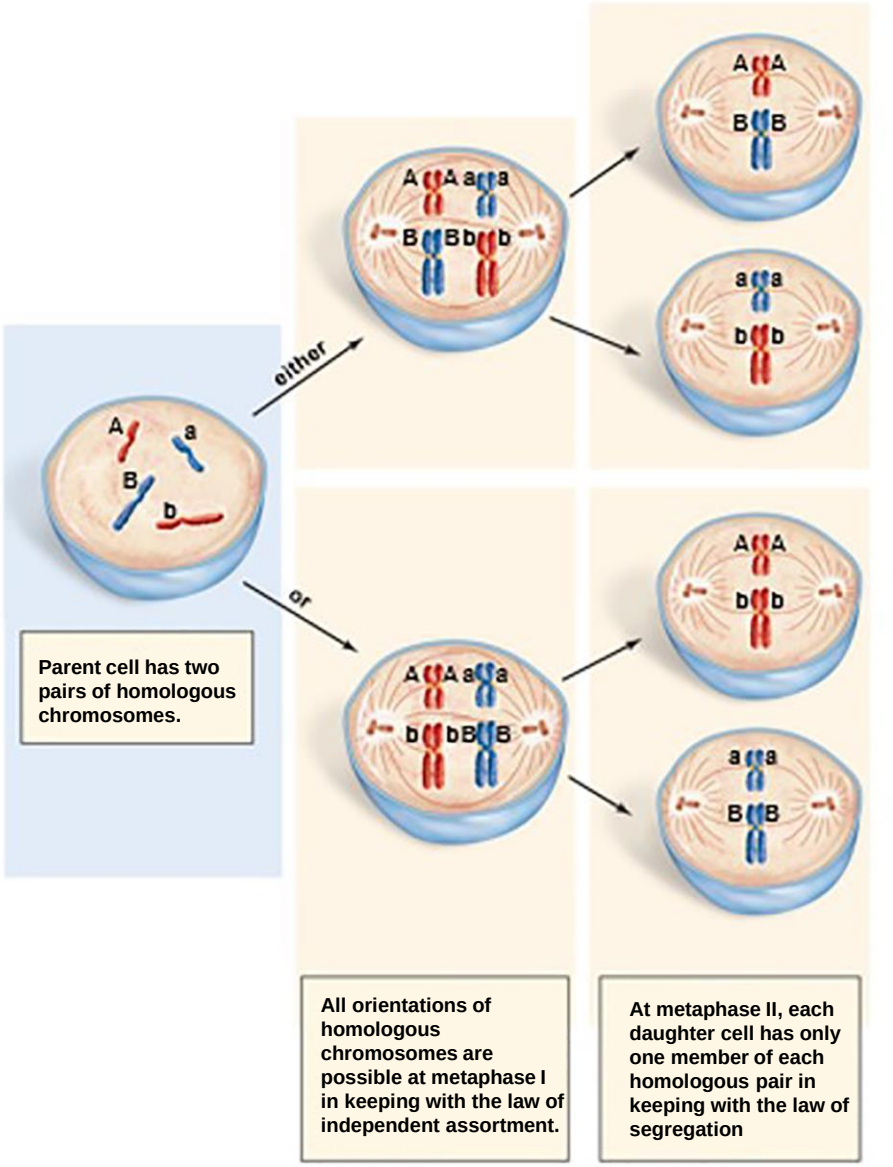
Independent Assortment and Segregation during Meiosis (1)



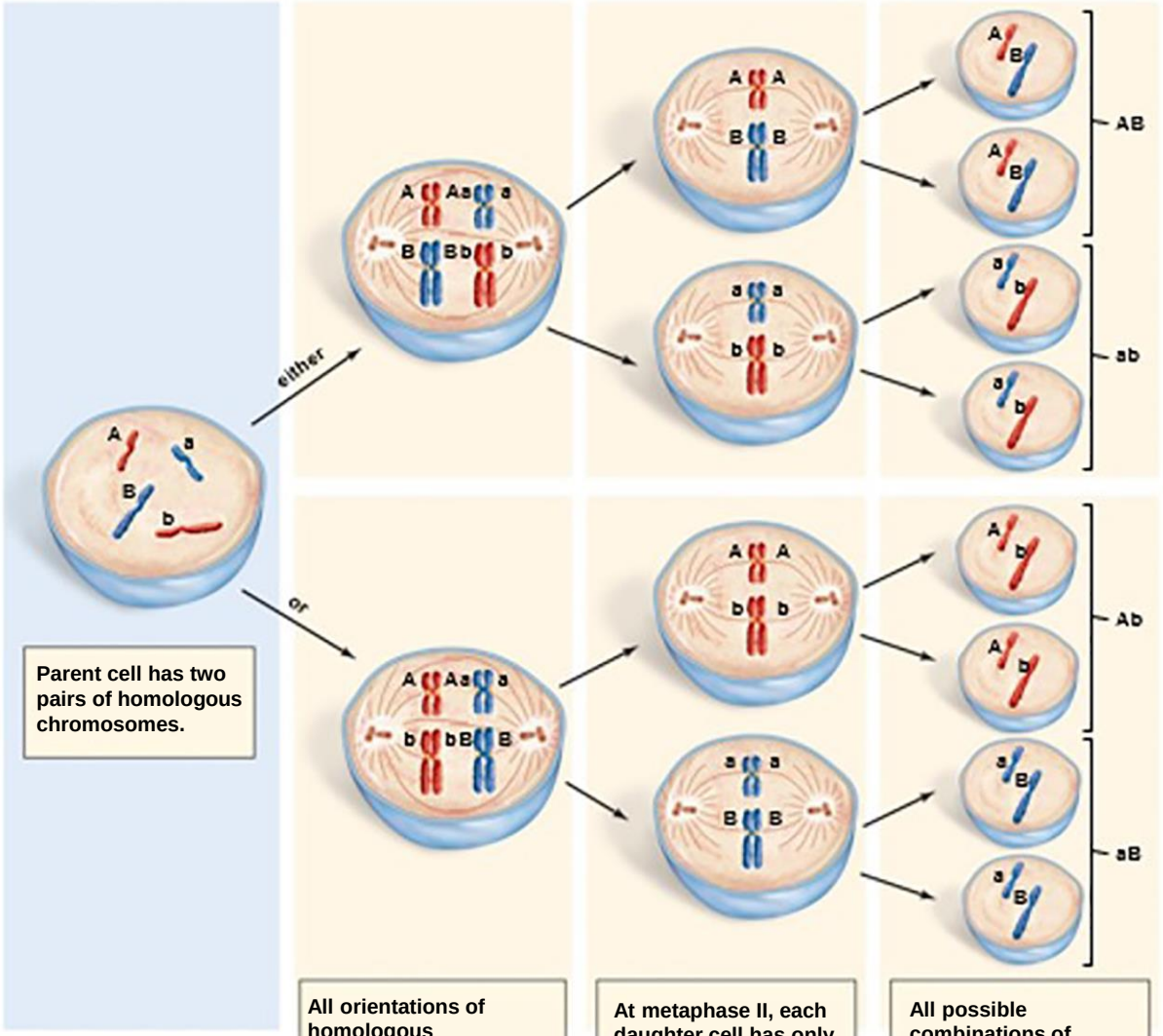
Independent Assortment and Segregation during Meiosis (2)



Independent Assortment and Segregation during Meiosis (3)



Independent Assortment and Segregation during Meiosis (4)



Jump to Independent Assortment and Segregation during Meiosis (4) Long Description

Mendel's Laws (5)

Punnett Square

- Table listing all possible genotypes resulting from a cross
  - All possible sperm genotypes are lined up on one side.
  - All possible egg genotypes are lined up on the other side.
  - All possible zygote genotypes are placed within the squares.

Mendel and the Laws of Probability (1)

Punnett Square

- It allows us to easily calculate probability of genotypes and phenotypes among the offspring.
- The Punnett square in the next slide shows a 50% (or $^1/_2$) chance.
  - The chance of $E = {}^1/_2$
  - The chance of $e = {}^1/_2$
- An offspring will inherit:
  - The chance of $EE = {}^1/_2 \times {}^1/_2 = {}^1/_4$
  - The chance of $Ee = {}^1/_2 \times {}^1/_2 = {}^1/_4$
  - The chance of $eE = {}^1/_2 \times {}^1/_2 = {}^1/_4$
  - The chance of $ee = {}^1/_2 \times {}^1/_2 = {}^1/_4$
- The sum rule allows us to add the genotypes that produce the identical phenotype to find out the chance of a particular phenotype.

Mendel and the Laws of Probability (2)

Punnett Square

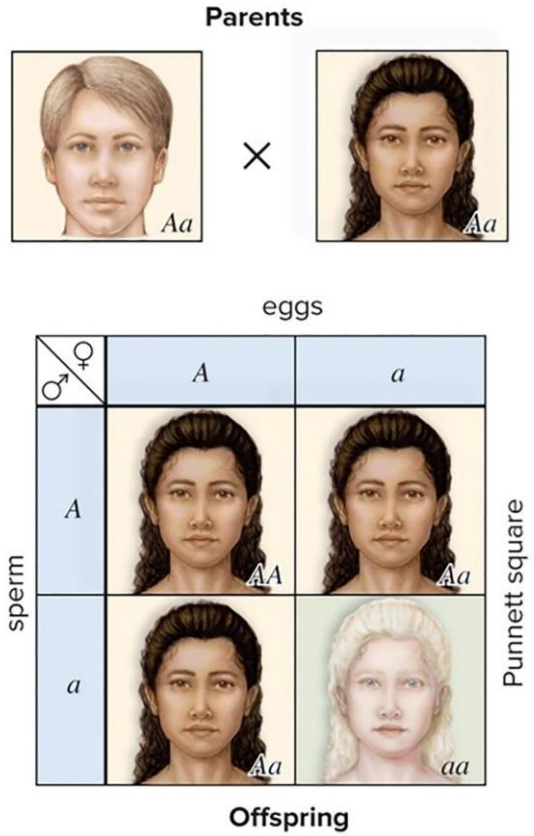
- It allows us to easily calculate probability of genotypes and phenotypes among the offspring.
- Punnett square in next slide shows a 50% (or $^1/_2$) chance.
- The chance of $A = {}^1/_2$
- The chance of $a = {}^1/_2$

An offspring will inherit

- The chance of $AA = {}^1/_2 \times {}^1/_2 = {}^1/_4$
- The chance of $Aa = {}^1/_2 \times {}^1/_2 = {}^1/_4$
- The chance of $aA = {}^1/_2 \times {}^1/_2 = {}^1/_4$
- The chance of $aa = {}^1/_2 \times {}^1/_2 = {}^1/_4$

The sum rule allows us to add the genotypes that produce the identical phenotype to find out the chance of a particular phenotype.

Punnett Square



Allele Key

A = Normal pigmentation
a = lack of pigmentation

Phenotypic Ratio

3 normal pigmentation
1 albino (no pigmentation)

Jump to Punnett Square Long Description

11.3 Mendelian Patterns of Inheritance and Human Disease

Genetic disorders are medical conditions caused by alleles inherited from parents.

Autosome is any chromosome other than a sex chromosome (X or Y).

Genetic disorders caused by genes on autosomes are called autosomal disorders.

- Some genetic disorders are autosomal dominant.
  - An individual with AA has the disorder.
  - An individual with Aa has the disorder.
  - An individual with aa does NOT have the disorder.
- Other genetic disorders are autosomal recessive.
  - An individual with AA does NOT have the disorder.
  - An individual with Aa does NOT have the disorder, but is a carrier.
  - An individual with aa DOES have the disorder.

11.4 Beyond Mendelian Inheritance

Some traits are controlled by **multiple alleles** (multiple allelic traits).

The gene exists in several allelic forms, but each individual only has two alleles.

ABO blood types

- The alleles:
  - antigen on red blood cells, anti-B antibody in plasma
  - antigen on red blood cells, anti-A antibody in plasma
  - *i* = Neither A nor B antigens on red blood cells, both anti-A and anti-B antibodies in plasma

The ABO blood type is also an example of **codominance**.

- More than one allele is fully expressed.
- Both and are expressed in the presence of the other.

ABO Blood Type

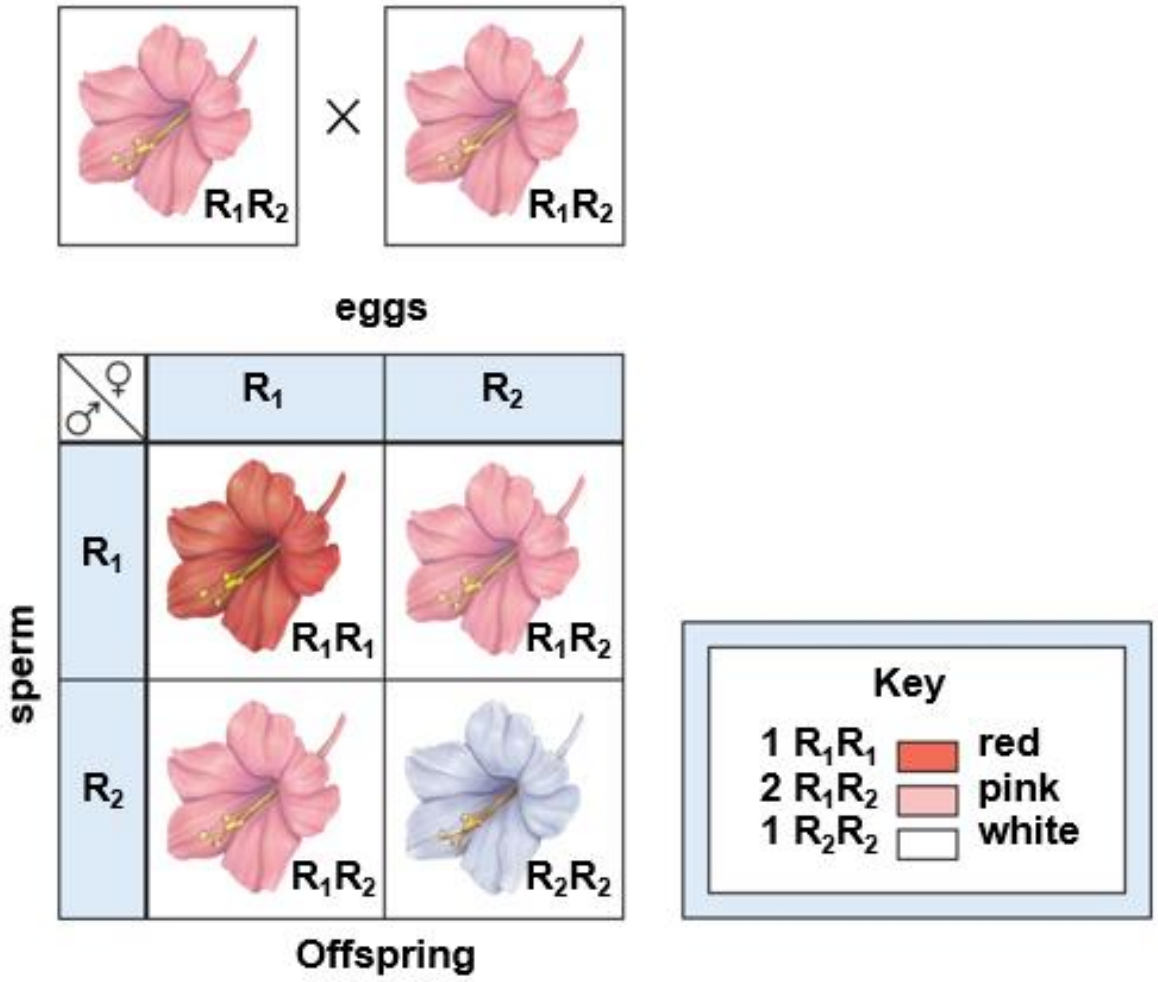
Phenotype	Genotype
A	$I^A I^A$, $I^A i$
B	$I^B I^B$, $I^B i$
AB	$I^A I^B$
O	*ii*

Beyond Mendelian Inheritance (1)

Incomplete Dominance

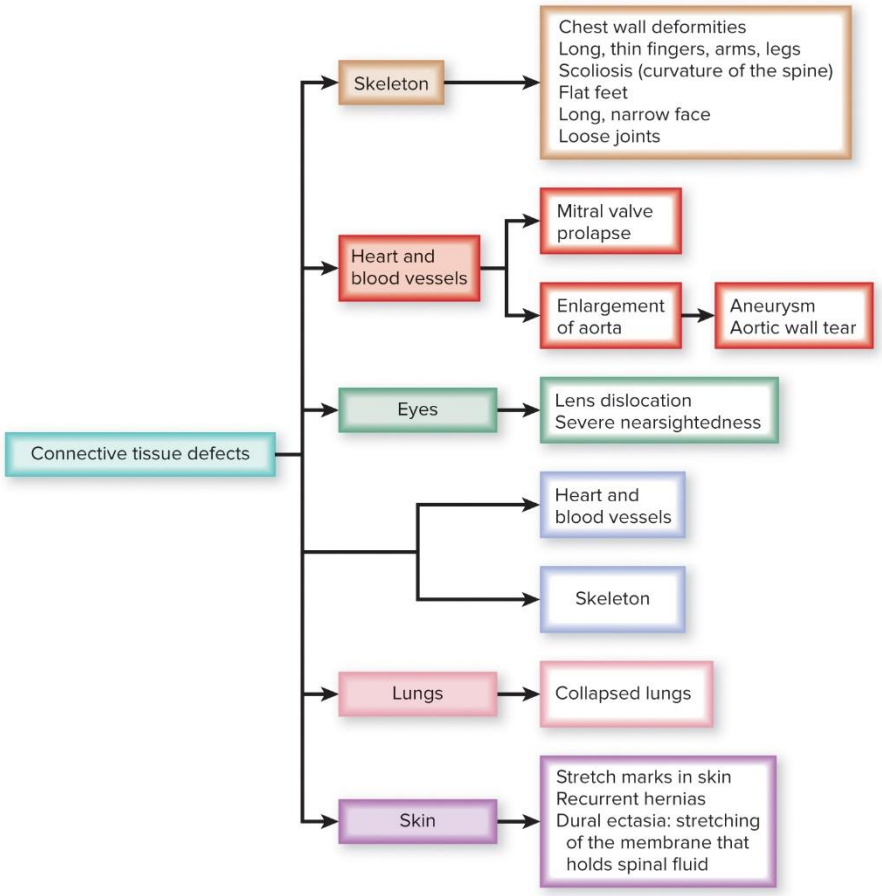
- Heterozygote has a phenotype intermediate between that of either homozygote.
  - Homozygous red has red phenotype.
  - Homozygous white has white phenotype.
  - Heterozygote has pink (intermediate) phenotype.
- Phenotype reveals genotype without a testcross.

Incomplete Dominance



Jump to Incomplete Dominance Long Description

Marfan Syndrome



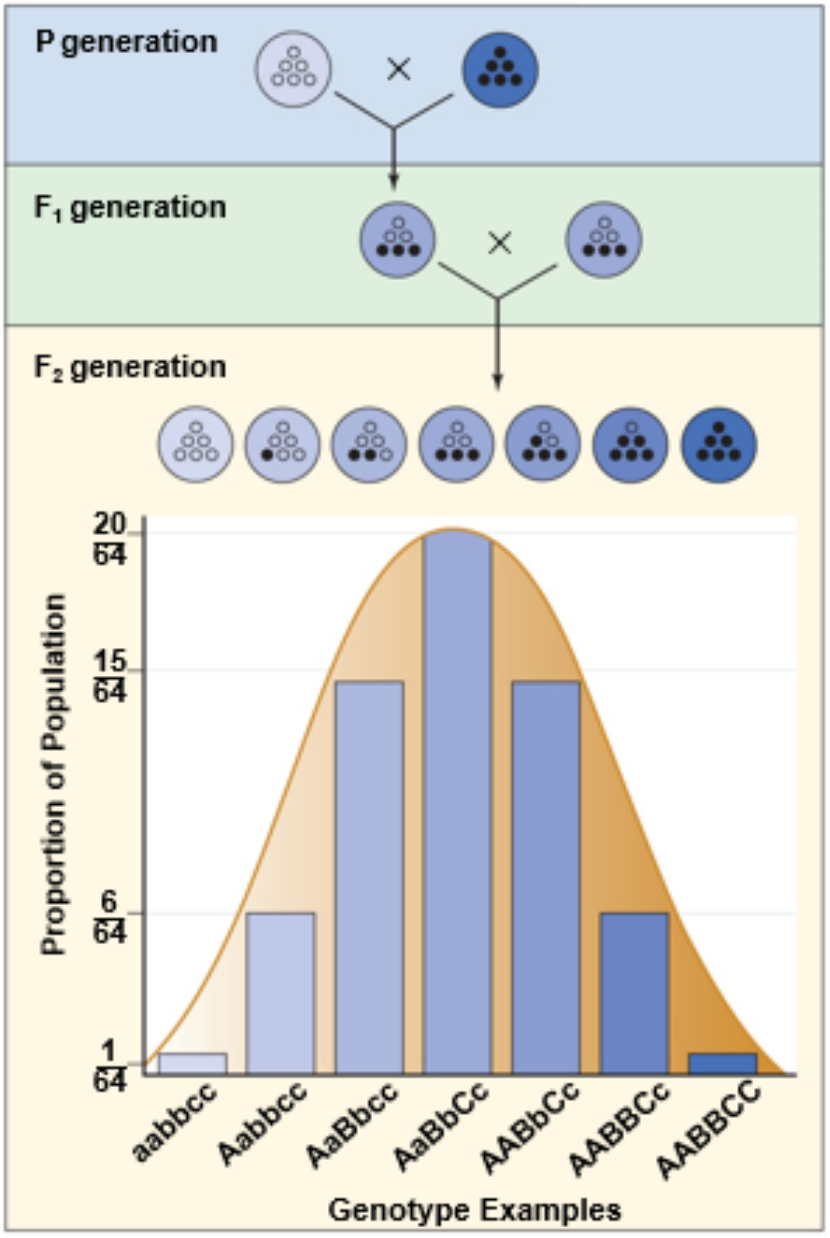
Jump to Marfan Syndrome Long Description

Beyond Mendelian Inheritance (5)

Polygenic Inheritance:

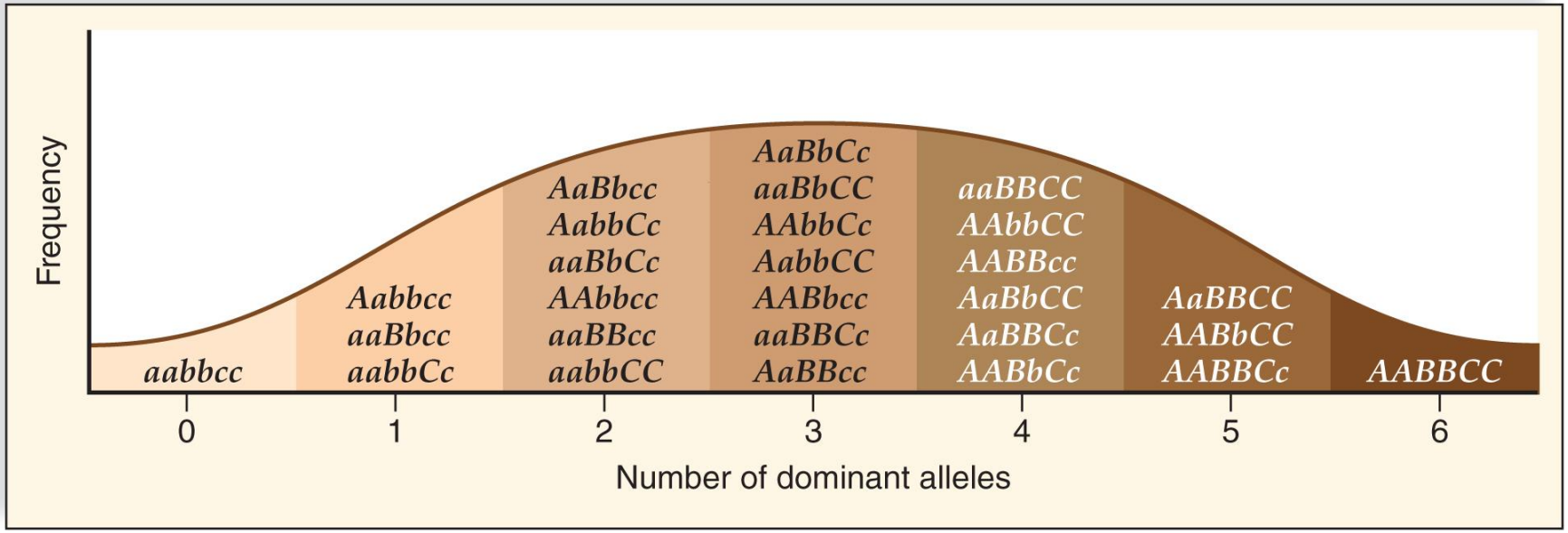
- Occurs when a trait is governed by two or more sets of alleles.
- Each dominant allele has a quantitative effect on the phenotype.
- These effects are additive.
- It results in continuous variation of phenotypes within a population.
- The traits may also be affected by the environment.
- Examples
  - Human skin color
  - Height
  - Eye color

Polygenic Inheritance



Jump to Polygenic Inheritance Long Description

Extending the Range of Mendelian Genetics (2)



Jump to Extending the Range of Mendelian Genetics (2) Long Description

Beyond Mendelian Inheritance (6)

X-Linked Inheritance

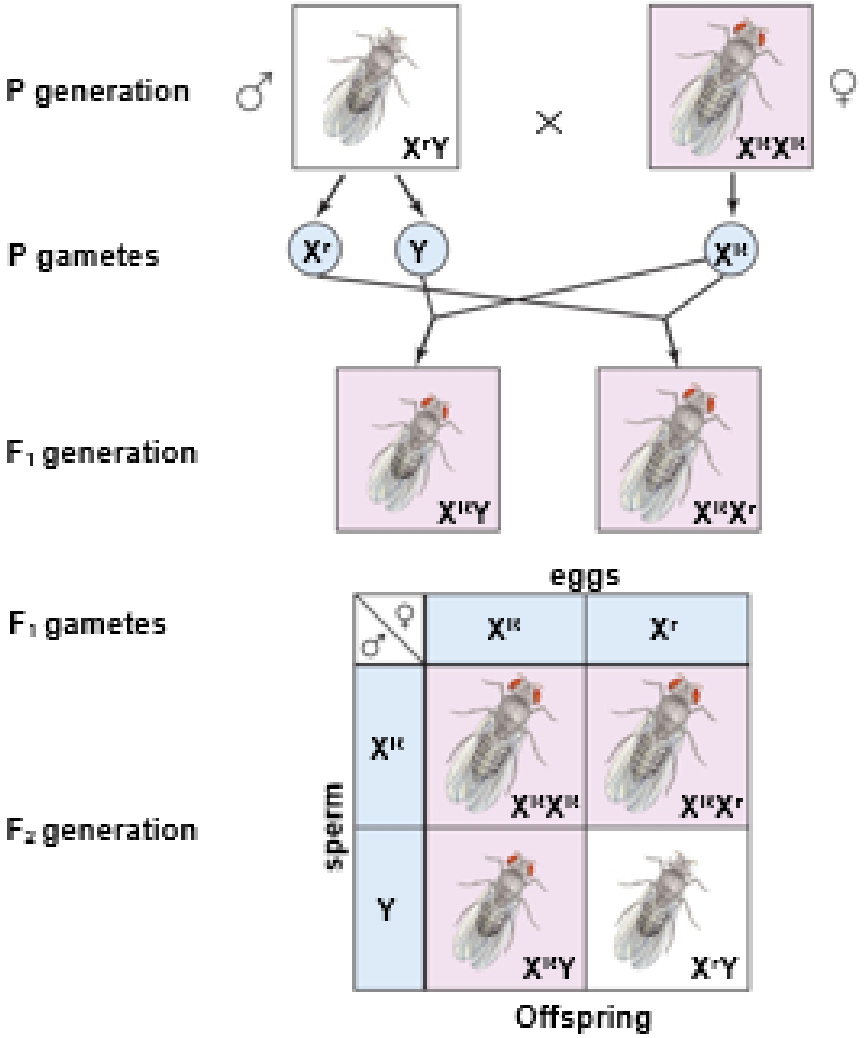
- In mammals
  - The X and Y chromosomes determine gender.
  - Females are XX.
  - Males are XY.

Extending the Range of Mendelian Genetics (1)

X-Linked Inheritance

- The term **X-linked** is used for genes that have nothing to do with gender.
  - X-linked genes are carried on the X chromosome.
  - The Y chromosome does not carry these genes.
  - It was discovered in the early 1900s by a group at Columbia University, headed by Thomas Hunt Morgan.
    - Performed experiments with fruit flies
      - They can be easily and inexpensively raised in simple laboratory glassware.
      - Fruit flies have the same sex chromosome pattern as humans.
      - Morgan's experiments with X-linked genes apply directly to humans.

X-Linked Inheritance



Jump to X-Linked Inheritance Long Description

Beyond Mendelian Inheritance (7)

Several X-linked recessive disorders occur in humans:

- Color blindness
  - The allele for the blue-sensitive protein is autosomal.
  - The alleles for the red- and green-sensitive pigments are on the X chromosome.
- Menkes syndrome
  - It is caused by a defective allele on the X chromosome.
  - It disrupts movement of the metal copper in and out of cells.
  - Phenotypes include kinky hair, poor muscle tone, seizures, and low body temperature.
- Muscular dystrophy
  - Causes wasting away of the muscle
  - It is caused by the absence of the muscle protein dystrophin.
- Adrenoleukodystrophy
  - It is an X-linked recessive disorder.
  - It is a failure of a carrier protein to move either an enzyme or very long chain fatty acid into peroxisomes.
- Hemophilia
  - It is an absence or minimal presence of clotting factor VIII or clotting factor IX.
  - An affected person’s blood either does not clot or clots very slowly.

Hemophilia and the Royal Families of Europe (1)

Hemophilia is called the bleeder's disease because the affected person's blood either doesn't clot correctly or doesn't clot at all.

People with hemophilia bleed internally and externally after injury.

Blood transfusions or clotting factor injections help with the disorder.

The pedigree shows why hemophilia is referred to as "the royal disease."

- Queen Victoria was the first of the royals to carry the gene.
- Eventually it was spread throughout the royal families of Europe through arranged marriages between the English, Spanish, Prussian, and Russian royal families.