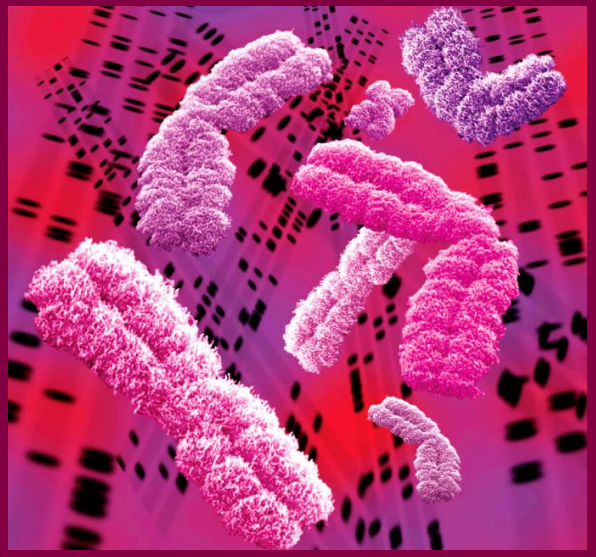
BIOLOGY I

Chapter 14:
MENDEL AND THE GENE IDEA



Evelyn I. Milian

Instructor

GENETICS: *Summary of Basic Terms*

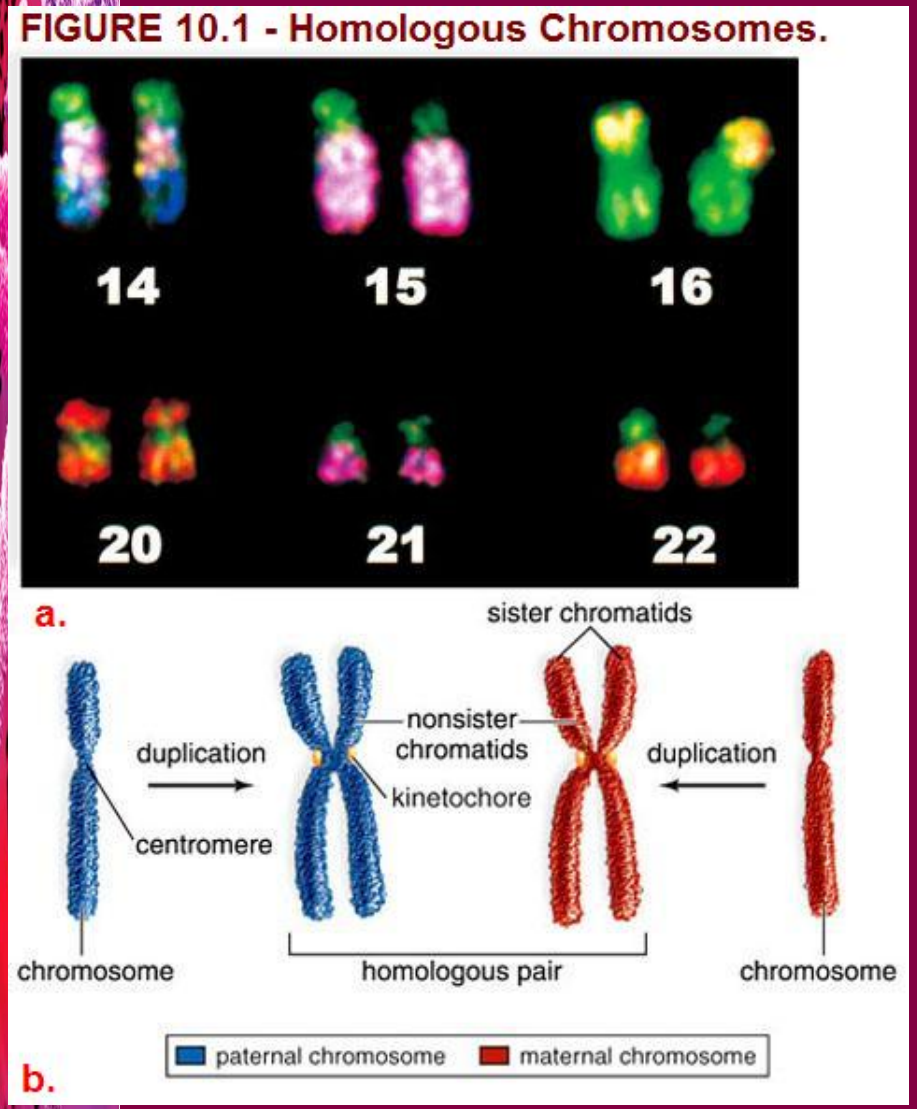
(* *Note: Some of these terms have been discussed in previous chapters, others are new. Study them in the context of the topics and figures*.)

Genetics	The science that studies **heredity** and **gene function**. ***Heredity*** is the **transmission of traits** from one generation to the next. ✓Genetics includes the study of what **genes** are, how they carry information, how the information is expressed, and how they are replicated and passed or transmitted to subsequent generations of cells or other organisms **(heredity)**. Genetic studies both genetic similarities and **genetic variation**, the differences between parents and offspring (descendants, or progeny) or among individuals of a population.
Gene	Unit of hereditary information consisting of a **segment of DNA** (deoxyribonucleic acid, a molecule of nucleotides) that encodes a functional product, usually a protein. DNA is the genetic material of cells. All information for the structure and function of an organism is coded in its genes.
Genome	The complete genetic information in a cell or organism (all genes).

GENETICS: *Summary of Basic Terms*

Chromosome 	The cellular threadlike structure that contains the **genetic material** of cells (in the nucleus of an eukaryotic cell, or the nucleoid region of prokaryotic cells). Each chromosome consists of one very long **DNA molecule** and **associated proteins**. In other words, chromosomes contain the genes.
Homologous chromosomes	Chromosomes that synapse (pair) during meiosis. Remember: ✓**Meiosis** is the type of cellular division that is part of sexual reproduction and produces 4 daughter cells with one of each kind of parental chromosome (these cells are not identical). One homologous chromosome is inherited from the father, the other from the mother. ✓Homologous chromosome pairs have the same length, centromere position, and shape. ✓They carry information about the same genetic traits, although this information is not necessarily identical.
Sex chromosomes	Pair of chromosomes responsible for **determining the sex** of an individual. ✓In humans, the sex chromosomes in females are **XX**, and those in males are **XY**.
Autosomes	Chromosomes that are **not directly involved in determining sex**, as opposed to a sex chromosome.

Homologous Chromosomes



- In diploid body cells, the chromosomes occur in pairs called **homologous chromosomes**.

a. In this micrograph of stained chromosomes from a human cell, the pairs have been numbered.

b. These chromosomes are duplicated and each one is composed of two **chromatids**. The **sister chromatids** contain the exact same genes; the nonsister chromatids contain genes for the same traits (e.g., type of hair, color of eyes), but they may differ in that one could “call for” dark hair and eyes and the other for light hair and eyes.

GENETICS: *Summary of Basic Terms*

Gamete	**Reproductive cell** (also called sex cell or germ cell): • **egg:** female reproductive cell. • **sperm:** male reproductive cell. •Gametes unite during sexual reproduction, or fertilization, to produce a cell called **zygote**.
Haploid (n)	Cell or individual that has only **one** of each type of chromosome (in other words, having only one set of unpaired chromosomes). ✓ **Gametes** (reproductive cells such as sperm and egg) are haploid.
Diploid (2n)	Cell or individual with **two** of each type of chromosome. ✓ **Somatic cells** (all the body cells of the organism, which are not reproductive cells) are diploid.

Animation: *Gamete Production*

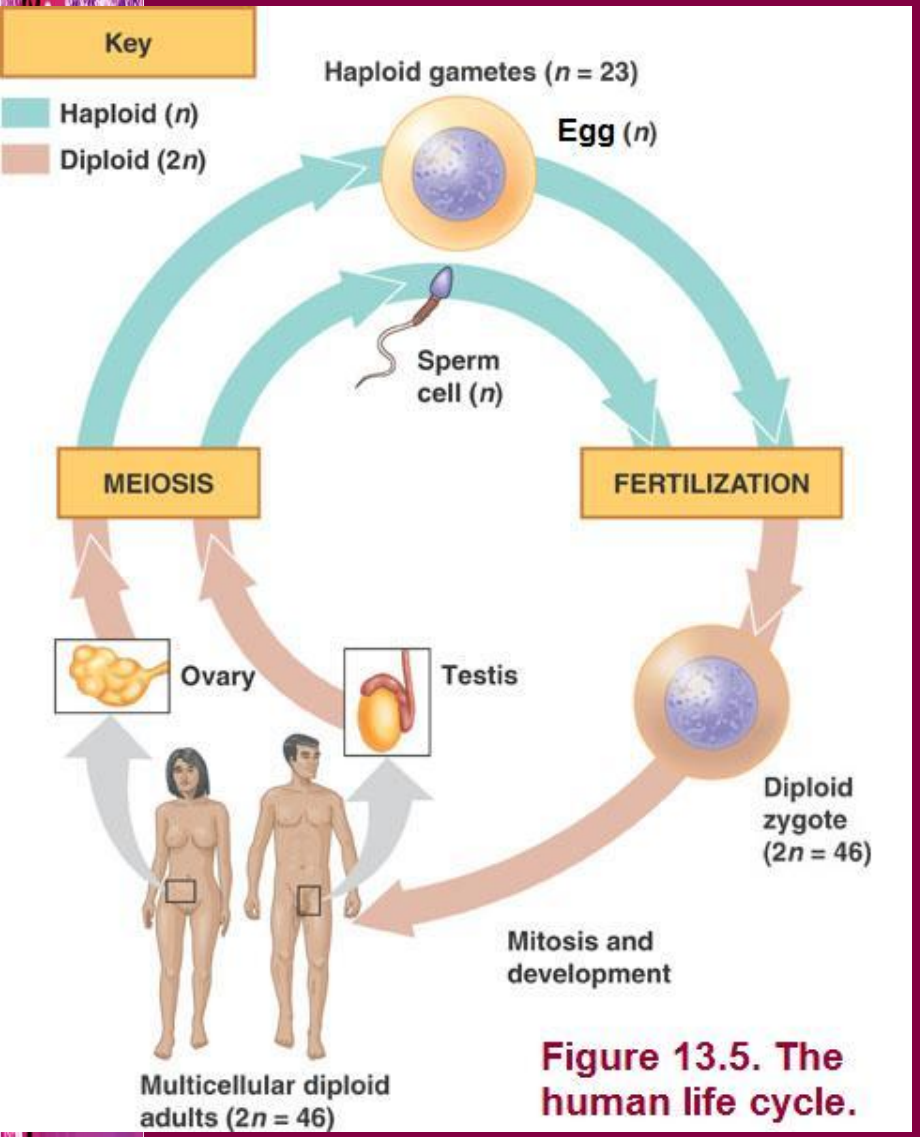


Figure 13.5. The human life cycle.

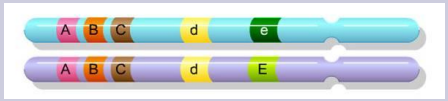
- ..\BIOLOGY-SOLOMON-Images\chapter11\Animations\gamete_producing.html (These animations can only be seen during class. You can search for similar animations in the internet or the book's website.)

- **The Human Life Cycle:** In each generation, the number of chromosome sets doubles at **fertilization**, but is halved during **meiosis**. For humans, the number of chromosomes in a haploid cell is 23, consisting of one set (*n* = 23); the number of chromosomes in the diploid zygote and all somatic cells arising from it is 46, consisting of two sets (2*n* = 46).

GENETICS: *Summary of Basic Terms*

Genotype	**Genes** of an organism for a particular trait or traits. The genetic makeup of an organism.
Phenotype	The **traits** (characteristics) of an organism; it can be defined as outward **appearance** (such as flower color), as **behavior,** or in **molecular** or **physiological** terms (such as glycoproteins on red blood cells). The phenotype is the *visible expression* of a genotype.
Wild type	An individual with the normal (**most common**) phenotype. Phenotype or genotype that is characteristic of the **majority** of individuals of a species in a natural environment.

GENETICS: *Summary of Basic Terms*

Allele 	***Alternative*** form or version of a gene. ✓ Alleles occur at the same locus in homologous chromosomes (chromosomes that are alike and pair in meiosis). Alleles specify slightly different versions of the same trait.
Locus (***loci*** in plural)	The specific physical **location** of a given gene within a chromosome.
Homozygous	Possessing two **identical** alleles for a given gene or a particular trait or character. A ***homozygote*** is also called a ***true breeding*** organism.
Heterozygous	Possessing two **different** alleles for a given gene or a particular trait or character. A ***heterozygote*** is also called a ***hybrid*** organism.

GENETICS: *Summary of Basic Terms*

Dominant allele	Allele that is fully expressed, and **masks** the expression of the other allele (the recessive allele). ✓ The dominant allele is the one that is expressed (it exerts its phenotypic effect) in a **heterozygote**. ✓ It is identified with a ***uppercase (capital) letter***, usually the first letter of the trait.
Recessive allele	Allele whose phenotypic effect **is not observed** in a heterozygote; its expression **is masked** by a dominant allele. ✓ The recessive allele is expressed (exerts its phenotypic effect) *only in the* ***homozygote***. ✓ It is identified by the same letter as the dominant allele, but ***lowercase*** (small).

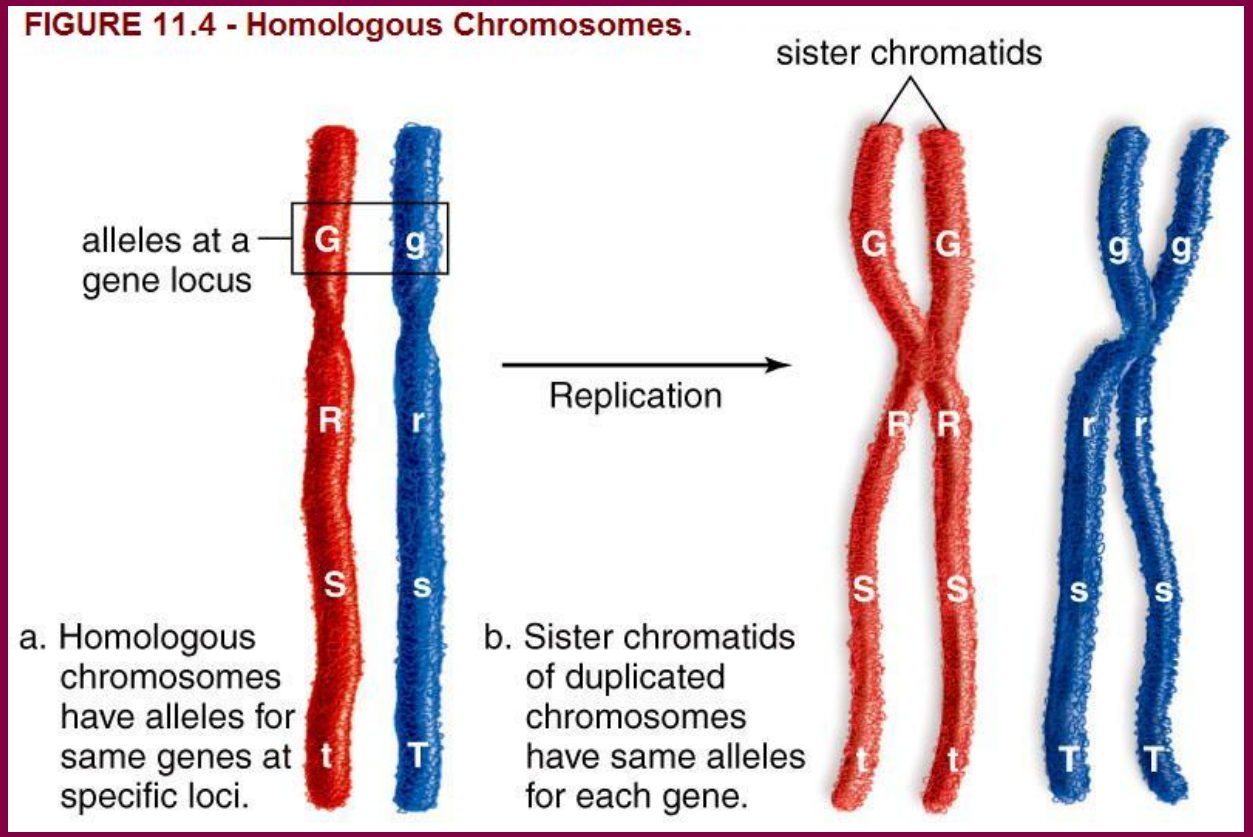
GENETICS: *Summary of Basic Terms*

True-breeding	A true-breeding organism, sometimes also called **pure-bred**, is an organism having certain biological traits which are passed on to all subsequent generations when bred with another true-breeding organism for the same traits. In other words, to "breed-true" means that two organisms with a particular, heritable phenotype produce only offspring with that (same) phenotype. ✓It also refers to plants that produce offspring of the **same variety** when they self-pollinate (the offspring are like the parent plants and like each other). ✓For example, a plant with purple flowers is true-breeding if the seeds produced by self-pollination all give rise to plants that also have purple flowers.
Hybrid	The offspring that results from the mating, or crossing, of two true-breeding varieties of the same species (**hybridization**), or from the mating of individuals from two different species.
Monohybrid	An organism that is heterozygous with respect to a **single** gene of interest. A cross between two monohybrids is called a ***monohybrid cross*.**
Dihybrid	An organism that is heterozygous with respect to **two** genes of interest. A cross between dihybrids is called a ***dihybrid cross***.

GENETICS: *Summary of Basic Terms*

P generation	**Parental generation**; parent individuals from which offspring are derived; P stands for "parental".
F_1 generation	**First filial generation**; the first offspring in a series of genetic crosses (produced from mating two parents). (Filial comes from the Latin *filius* = sons and daughters).
F_2 generation	**Second filial generation**; offspring resulting from interbreeding of the F_1 generation (produced from mating two F_1 individuals).

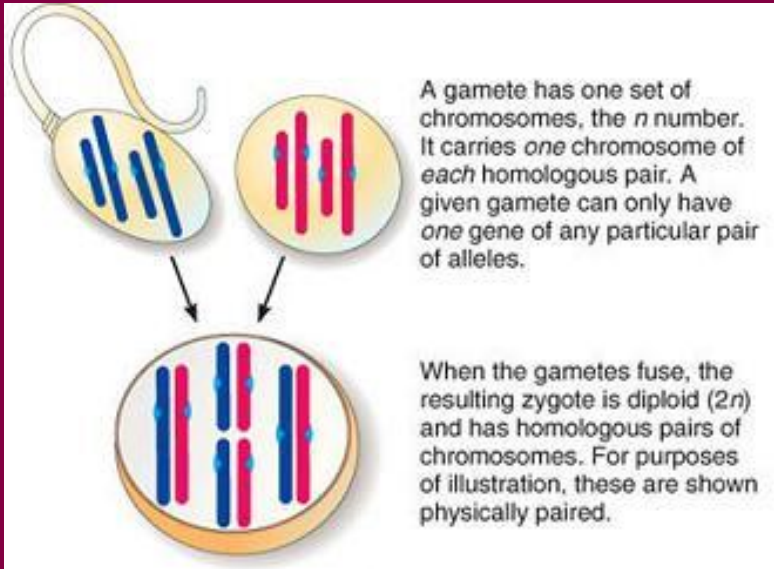
Homologous Chromosomes and Alleles



a) The letters represent ***alleles***; that is, alternate forms of a gene. Each allelic pair, such as ***Gg*** or ***Tt***, is located on homologous chromosomes at a particular gene *locus*.

b) Sister chromatids carry the same alleles in the same order.

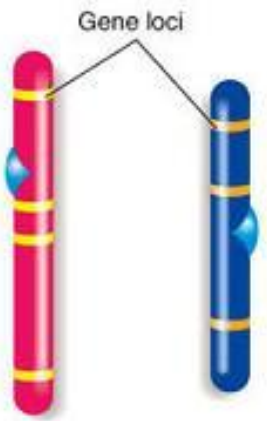
Figure 11-5. Gene loci and their alleles.



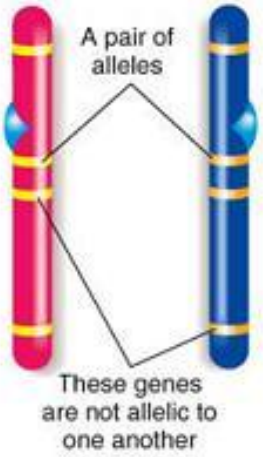
A gamete has one set of chromosomes, the *n* number. It carries *one* chromosome of *each* homologous pair. A given gamete can only have *one* gene of any particular pair of alleles.

When the gametes fuse, the resulting zygote is diploid ($2n$) and has homologous pairs of chromosomes. For purposes of illustration, these are shown physically paired.

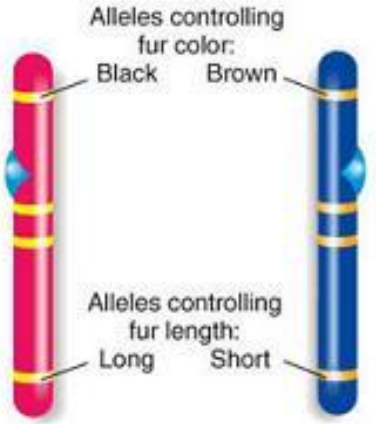
(a) One member of each pair of homologous chromosomes is of maternal origin (*red*), and the other is paternal (*blue*).



(b) These chromosomes are nonhomologous. Each chromosome is made up of hundreds or thousands of genes. A locus is the specific place on a chromosome where a gene is located.

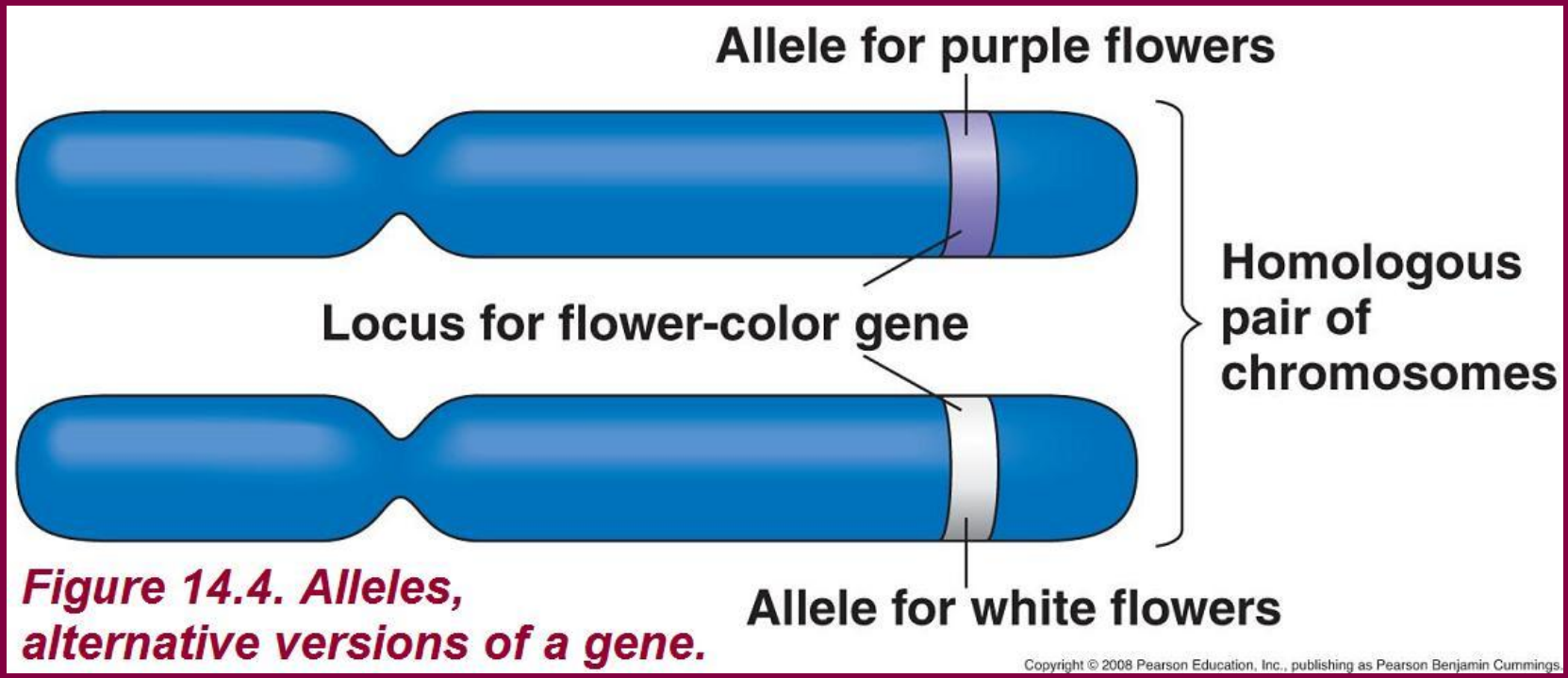


(c) These chromosomes are homologous. Alleles are members of a gene pair that occupy corresponding loci on homologous chromosomes.



(d) Alleles govern the same character but do not necessarily contain the same information.

Alleles, alternative versions of a gene



- A ***somatic*** (body) cell has two copies of each chromosome (forming a homologous pair) and thus two alleles of each gene, which may be identical or different. This figure depicts a homologous pair of chromosomes in a hybrid pea plant. The chromosome with an allele for purple flowers was inherited from one parent, and that with an allele for white flowers from the other parent.

Animation: Genetic Terms

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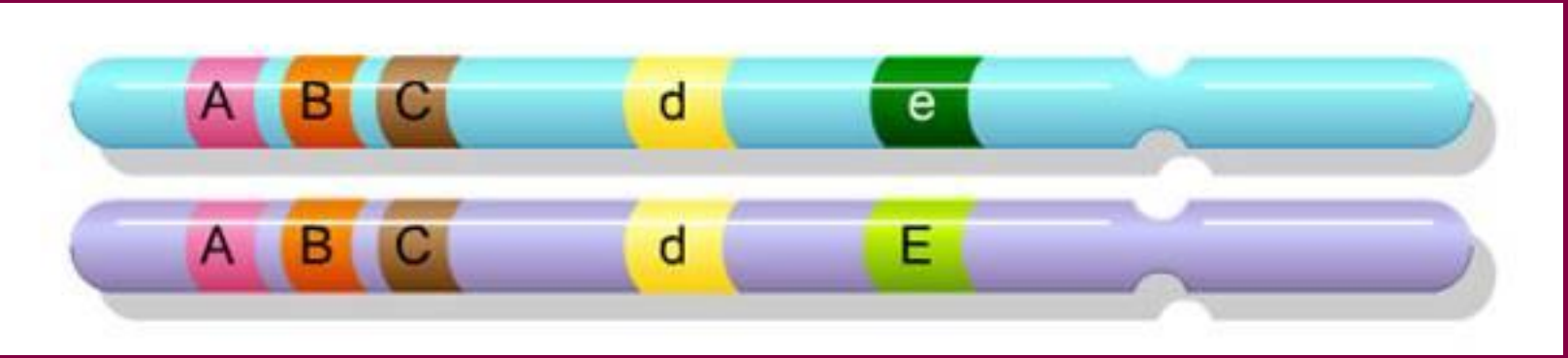


GREGOR MENDEL: Father of Genetics

- Mendel, an austrian monk, discovered the basic principles or **laws or principles of heredity** by breeding garden peas during the mid-1800s.
- Mendel showed that parents pass on to their offspring discrete genes that retain their identity through the generations.
- Mendel's work was ignored during his lifetime, and not published until early in the twentieth century when scientists revisited his research.

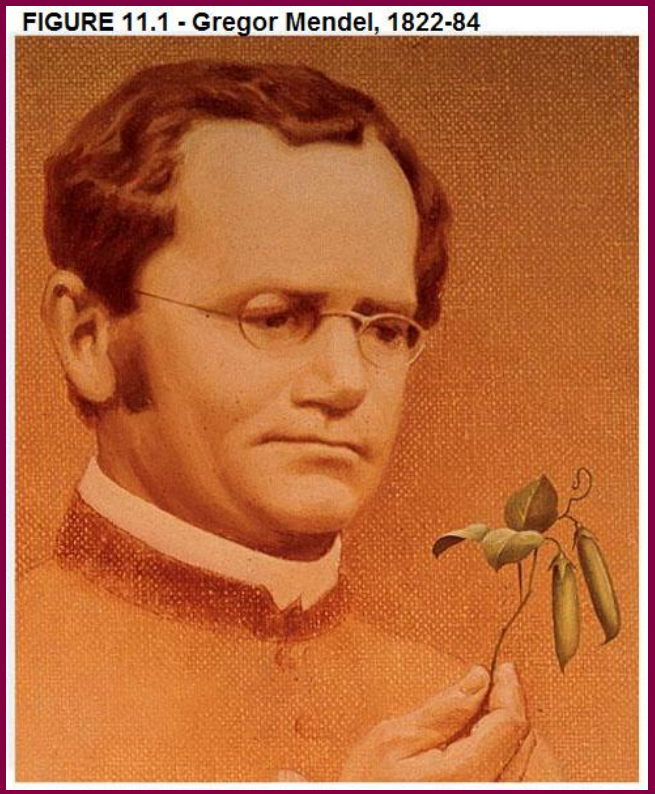


FIGURE 11.1 - Gregor Mendel, 1822-84

Mendel proposed a "particulate" hypothesis of inheritance: the gene idea. This model states that parents pass on discrete heritable units—**genes**—that retain their separate identities in offspring.

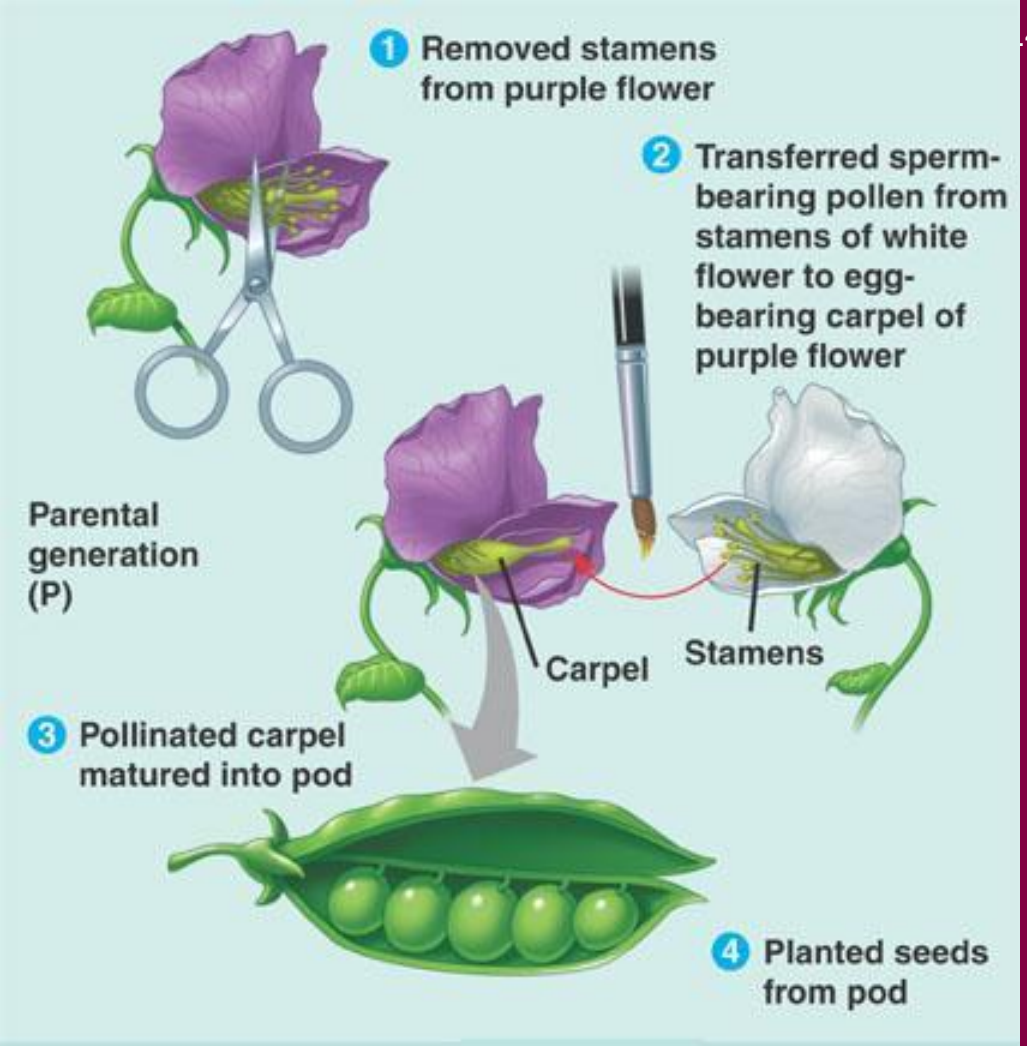


Figure 14.2

Mendel's Experiments

- Mendel crossed pea plants that varied in flower color.
- In the garden pea, ***Pisum sativum***, pollen grains contained in the anther produce sperm, and ovules in the ovary contain eggs. When Mendel performed crosses, he brushed pollen from one plant onto the stigma of another plant. After sperm fertilized eggs, the ovules developed into seeds (peas).
- ***Results:*** When pollen from a white flower was transferred to a purple flower, the first-generation hybrids **all** had **purple flowers**.

Mendel's Experiment

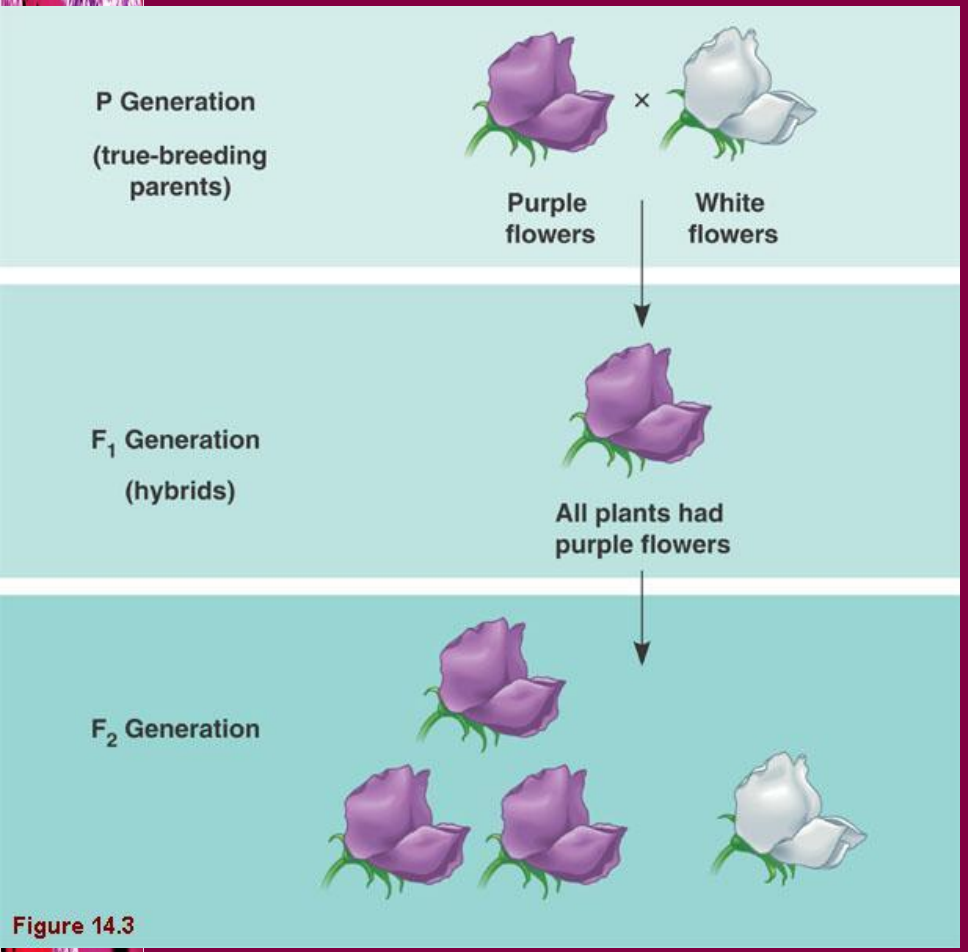


Figure 14.3

*** The white flower color did not disappear, but the purple color dominated.**

- Mendel crossed **true-breeding** purple-flowered pea plants and white-flowered pea plants. The resulting **F_1 hybrids** were allowed to self-pollinate or were cross-pollinated with other F_1 hybrids. Flower color was then observed in the F_2 generation.
- ***Results*:** Both purple-flowered plants and white-flowered plants appeared in the **F_2** generation.
- In Mendel's experiment 705 plants had purple flowers, and 224 had white flowers, showing a ratio of approximately **3 purple: 1 white**.
- This showed that purple flower color is a ***dominant*** trait and white flower color is a ***recessive*** trait.

Punnett Square Method for a One-Trait Cross

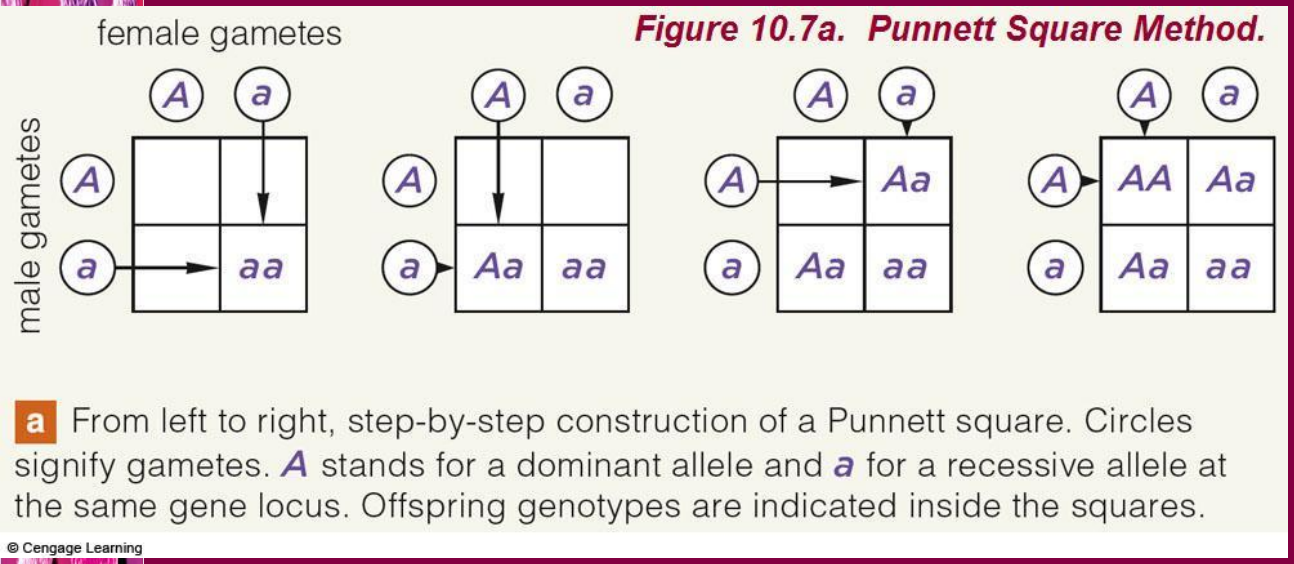
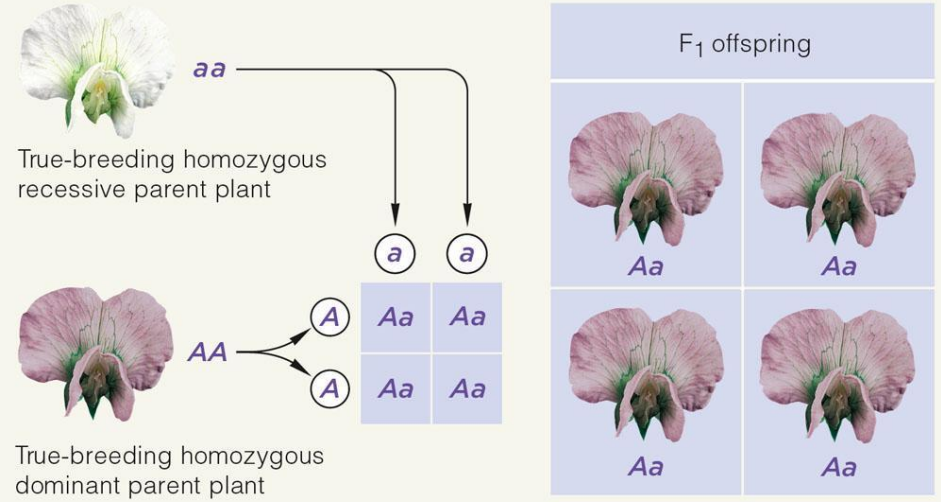


Figure 10.7a. Punnett Square Method.

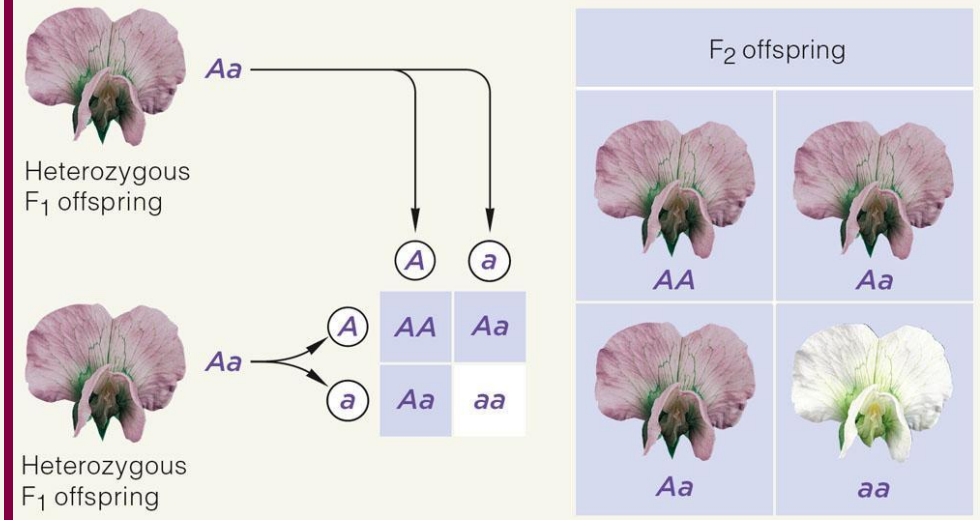
a From left to right, step-by-step construction of a Punnett square. Circles signify gametes. ***A*** stands for a dominant allele and ***a*** for a recessive allele at the same gene locus. Offspring genotypes are indicated inside the squares.

Possible Event	Probable Outcome
Sperm *A* meets egg *A*	→ ¼ *AA*
Sperm *A* meets egg *a*	→ ¼ *Aa*
Sperm *a* meets egg *A*	→ ¼ *Aa*
Sperm *a* meets egg *a*	→ ¼ *aa*



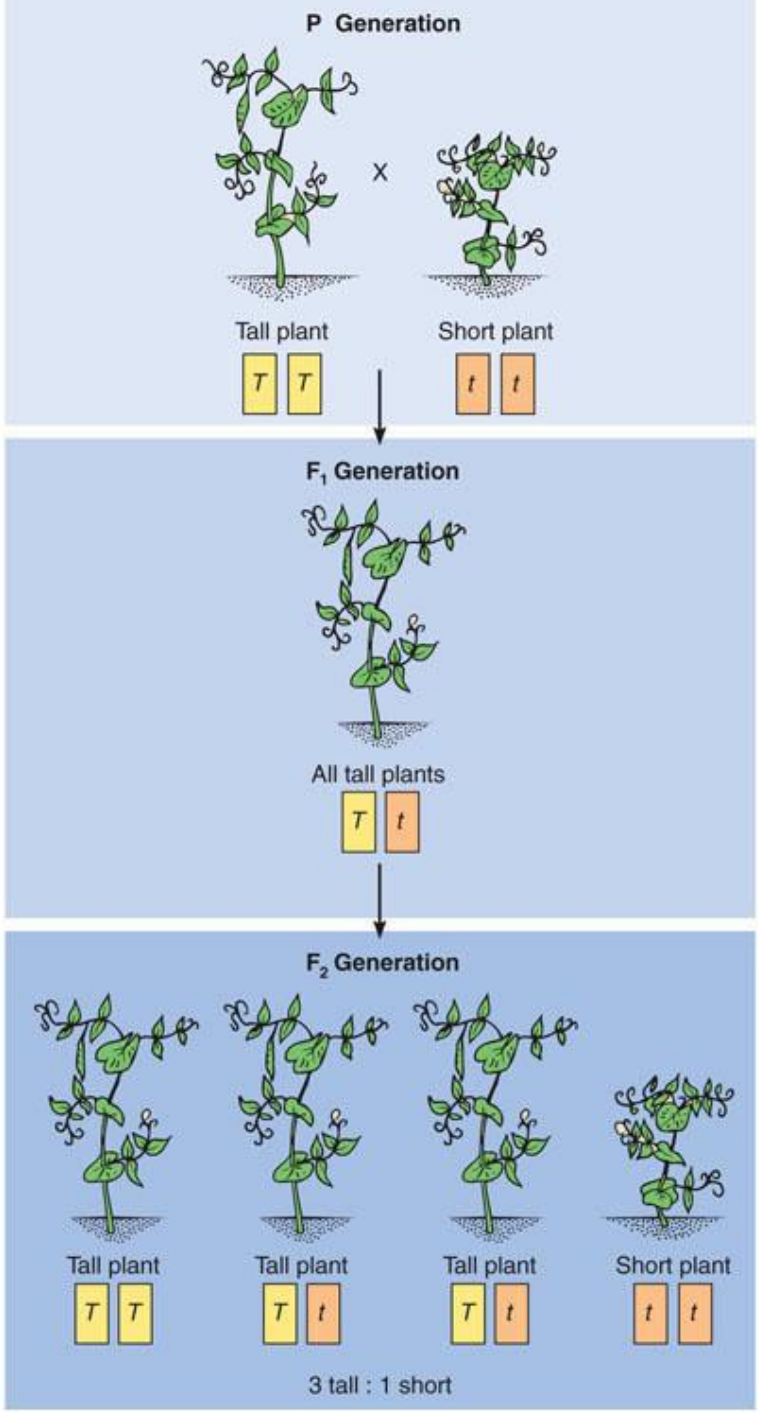
b Cross between two plants that breed true for different forms of a trait.

Figure 10.7b. Punnett Square Method.



c Cross between heterozygous F_1 offspring.

Figure 10.7c. Punnett Square Method.



One of Mendel's Pea Crosses

- When the F_1 generation of tall pea plants is self-pollinated, what phenotypes appear in the F_2 generation?
- Although only the "factor" (gene) for tall height is expressed in the F_1 generation, Mendel hypothesized that *the factor for short height is not lost*. He predicted that the short phenotype would reappear in the F_2 generation.
- Mendel crossed true-breeding tall pea plants with true-breeding short pea plants, yielding only tall offspring in the F_1 generation. He then allowed these F_1 individuals to self-pollinate to yield the F_2 generation.
- **Conclusion:** The F_2 generation included 787 tall and 277 short plants, which resulted in a ratio of about **3:1** (3 tall: 1 short; see figure).

Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant:Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Seed color	Yellow 	×	Green 	6022:2001	3.01:1
Seed shape	Round 	×	Wrinkled 	5474:1850	2.96:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

Mendel's Experiments

- When observing flower color, Mendel reasoned that the **heritable factor** for white flowers *did not disappear* in the F_1 plants, but was somehow **hidden or masked** when the purple-flower factor was present. Purple flower color is a ***dominant*** trait and white flower color is a ***recessive*** trait.
- Mendel observed the same pattern of inheritance in six other characters.
- All of the crosses produced approximately a **3:1 ratio**.
  - For example: 787/277 = 3/1.

Mendel's Model of Inheritance: The Law or Principle of Segregation

- Mendel developed a model to explain the **3:1 inheritance pattern** that he consistently observed among the $\mathbf{F_2}$ offspring in his pea experiments.
  1. **Alternative versions of genes (factors)** account for variations in inherited characteristics (traits).
  2. For each character, an organism inherits **two alleles or factors**, one from each parent.
  3. If the two alleles at a locus differ, then one, the **dominant allele**, determines the organism's appearance; the other, the **recessive allele**, has no noticeable effect on the organism's appearance.
  4. **The law of segregation:** The two alleles or genes for a heritable character segregate (separate) from one another during gamete formation and end up in different gametes.

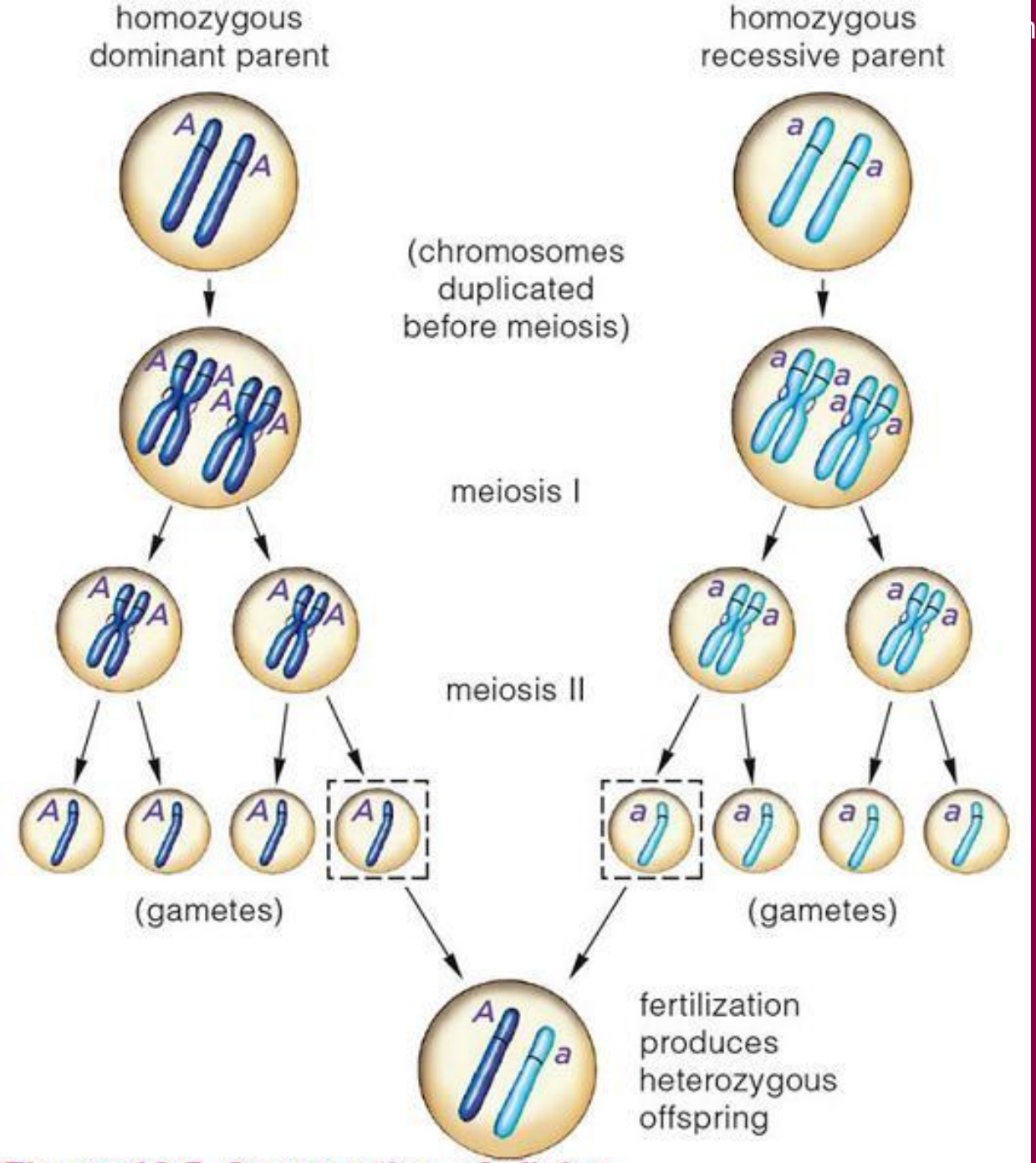
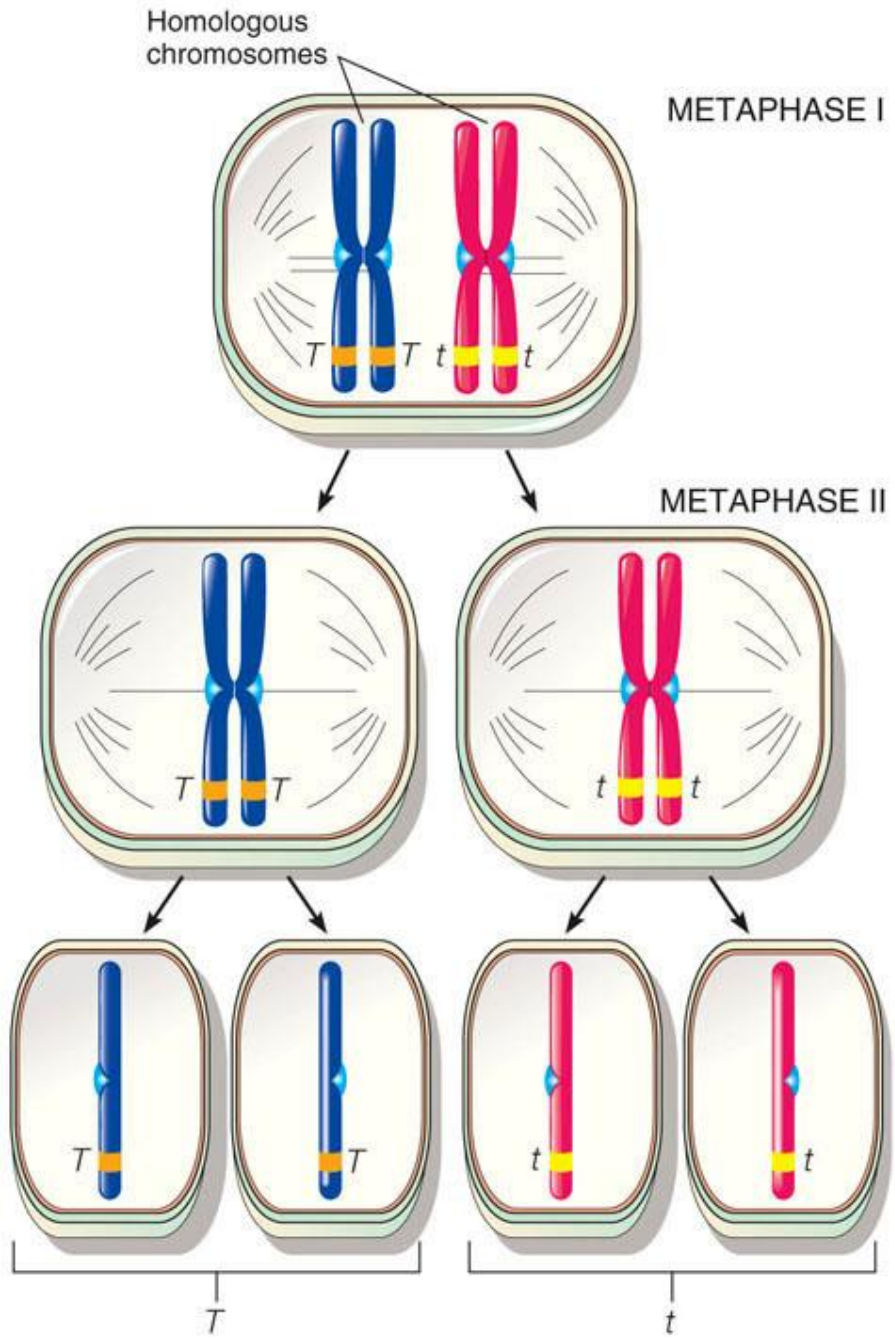


Figure 10.5. Segregation of alleles.

Mendel's Law or Theory of Segregation

- One gene of a pair segregates from the other gene in a monohybrid cross.
- Two parents that breed true for two versions of a trait produce only heterozygous offspring.



Mendel's Model of Inheritance: The Principle of Segregation

- Mendel's ***law or principle of segregation*** is related to the events of **meiosis**: The separation of homologous chromosomes during **meiosis** results in the **segregation of alleles**.
- Note that half of the gametes carry ***T*** and half carry ***t*.**

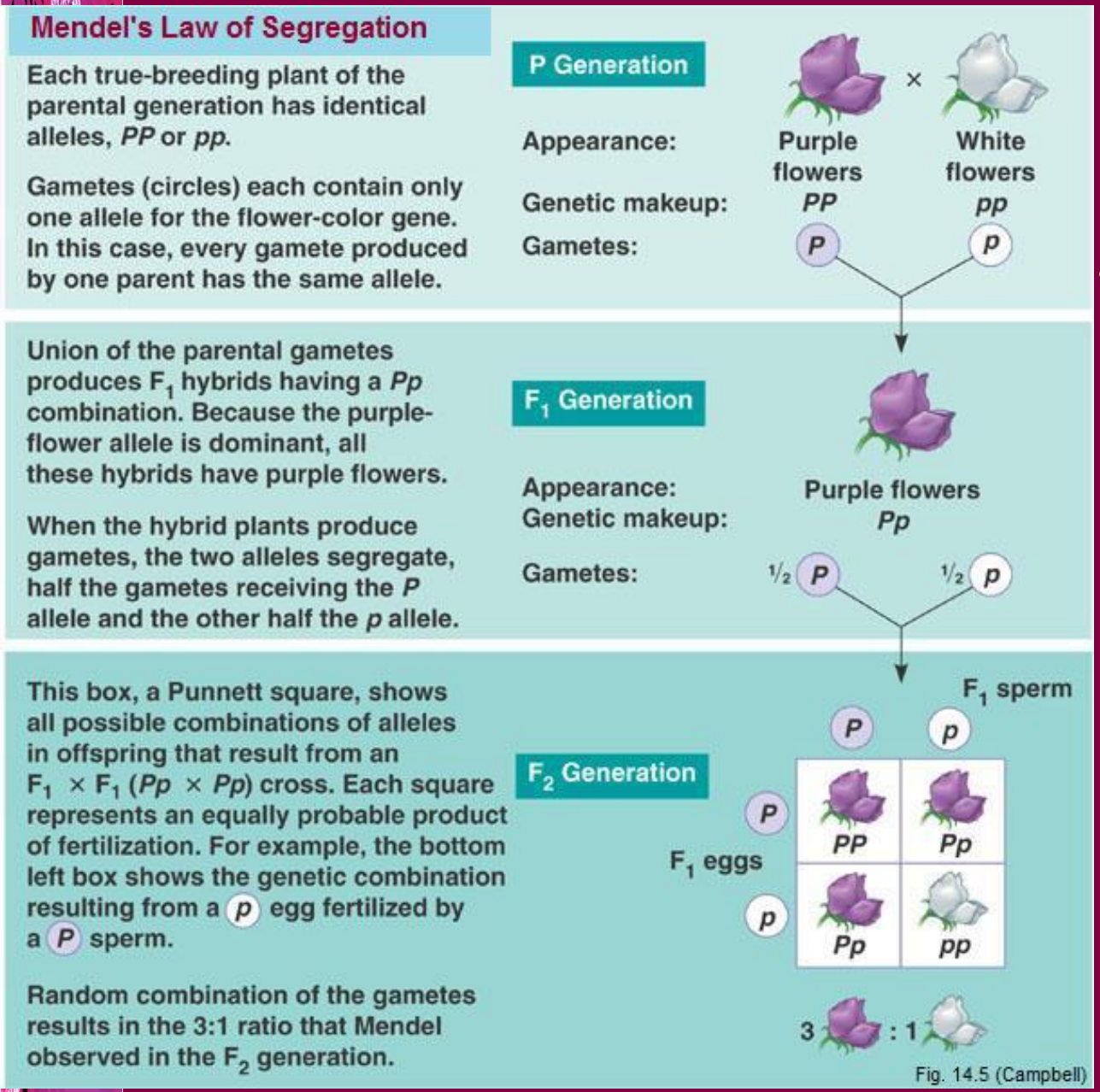


Fig. 14.5 (Campbell)

Mendel's Laws or Principles of Heredity: *The Law of Segregation*

- Each individual has **two alleles or factors** for each genetic trait, and the factors **segregate** (separate) during the formation of gametes (in meiosis).
  - In other words, each gamete receives only one of each parent's pair of genes for each trait.

Phenotype versus Genotype

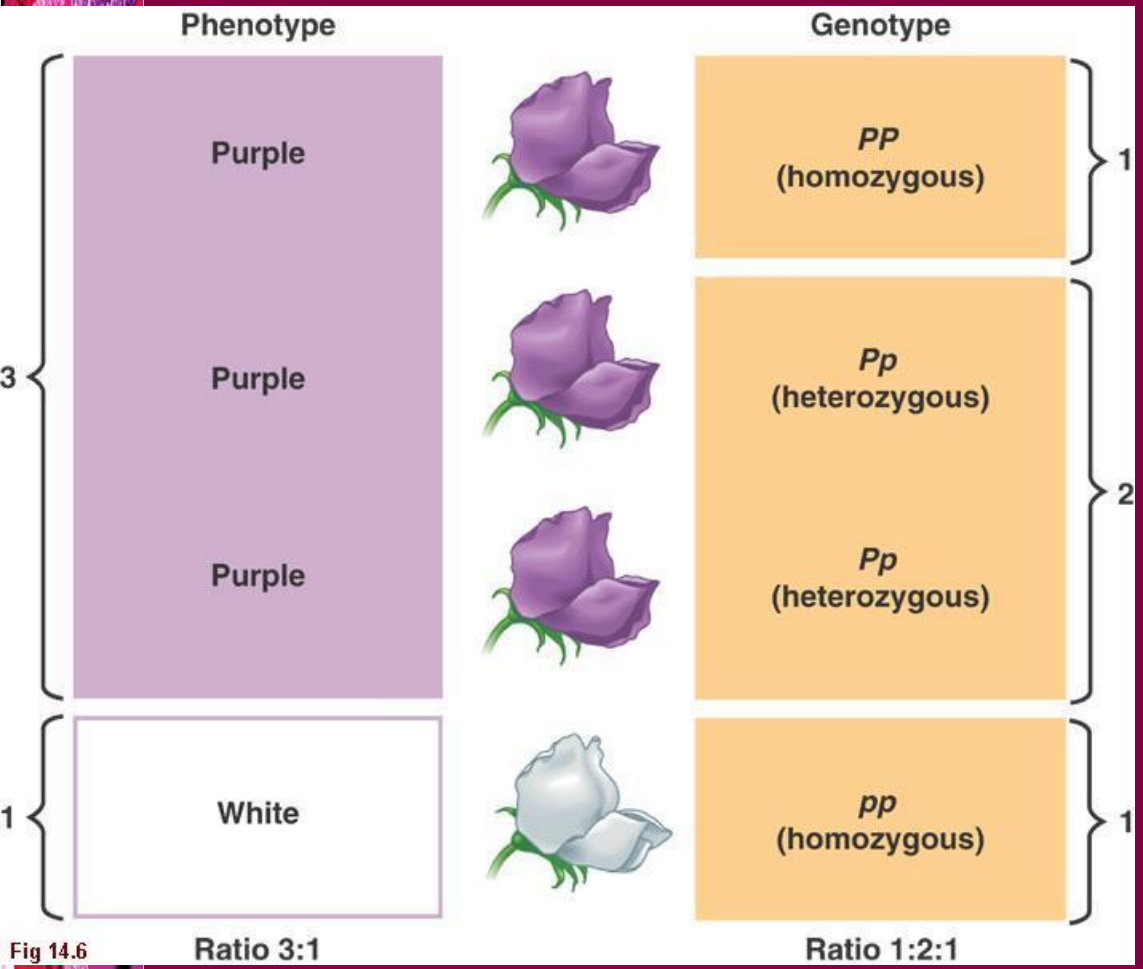


Fig 14.6

- Grouping F_2 offspring from a cross for flower color according to phenotype results in the typical **3:1 phenotypic ratio**. In terms of genotype, however, there are actually two categories of purple-flowered plants, ***PP*** **(homozygous)** and ***Pp*** **(heterozygous)**, giving a **1:2:1 genotypic ratio**.

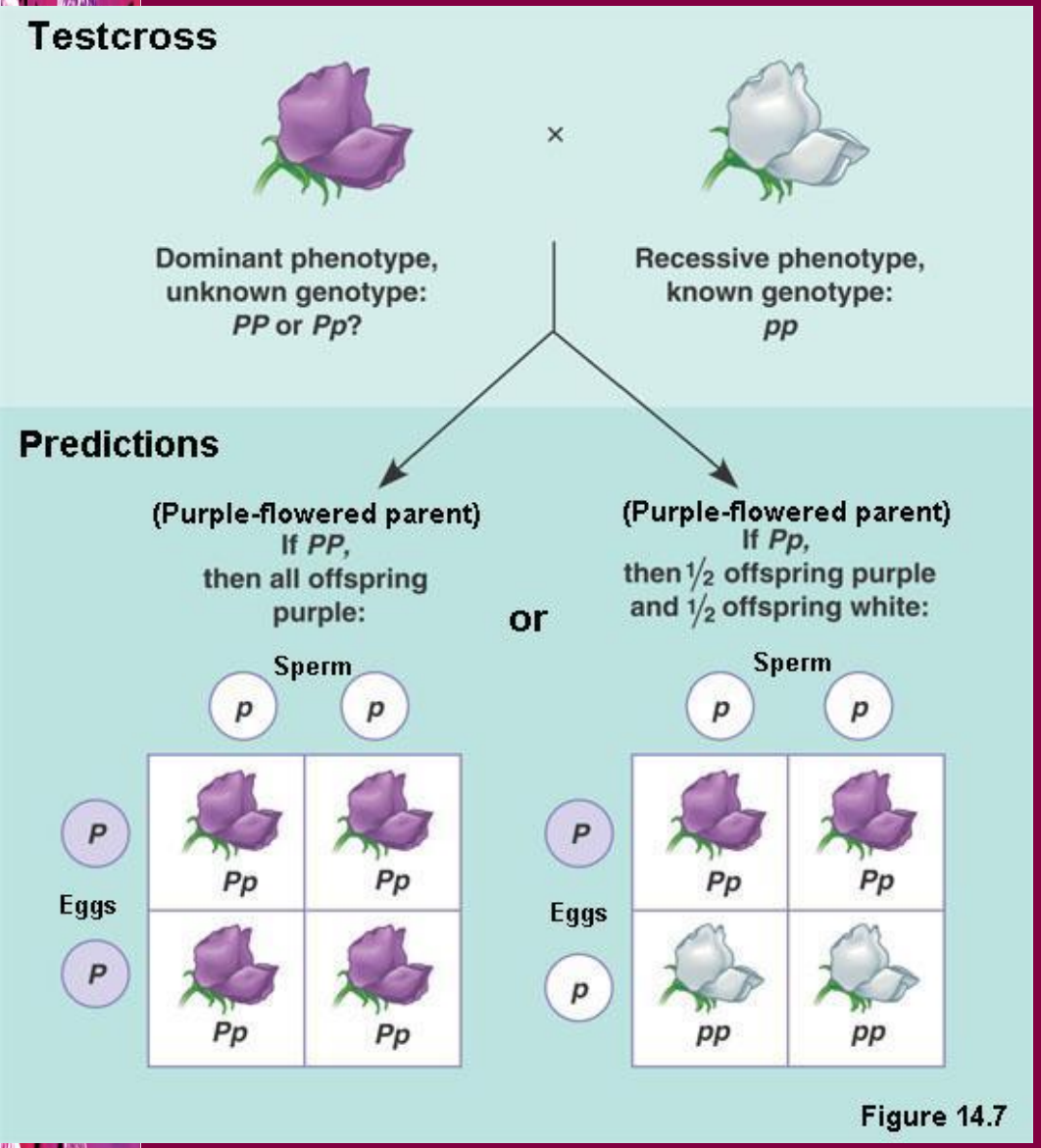


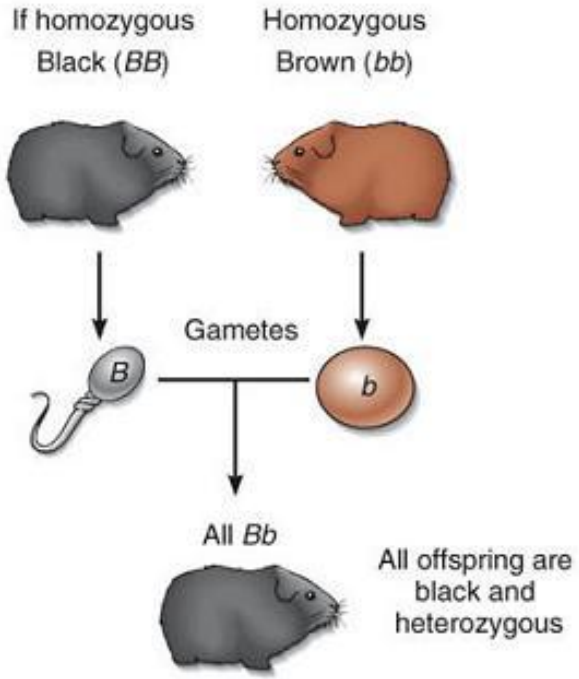
Figure 14.7

The Testcross

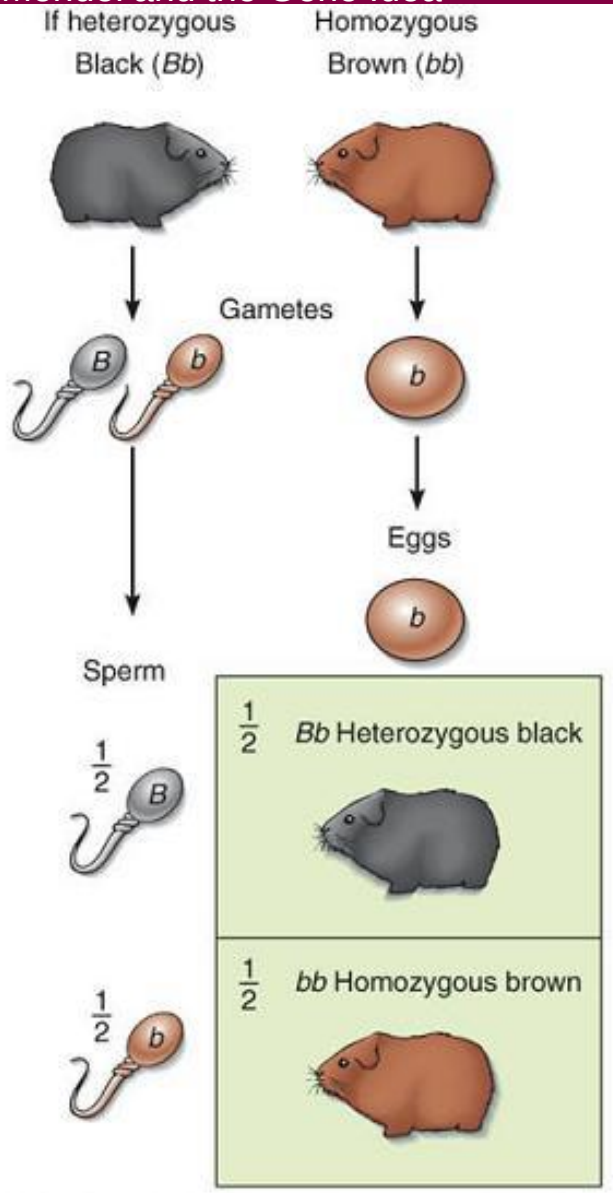
- A **testcross** is crossing an organism of unknown genotype with a homozygous recessive individual to determine the unknown genotype. The ratio of phenotypes in the offspring reveals the unknown genotype.

***Results*:** Matching the results to either prediction identifies the unknown parental genotype (either *PP* or *Pp* in this example). In this testcross, we transferred pollen from a white-flowered plant to the carpels of a purple-flowered plant; the opposite (reciprocal) cross would have led to the same results.

Figure 11-7. A test cross in guinea pigs.



(a) If a black guinea pig is mated with a brown guinea pig and all the offspring are black, the black parent probably has a homozygous genotype.



(b) If any of the offspring is brown, the black guinea pig must be heterozygous. The expected phenotypic ratio is 1 black to 1 brown.

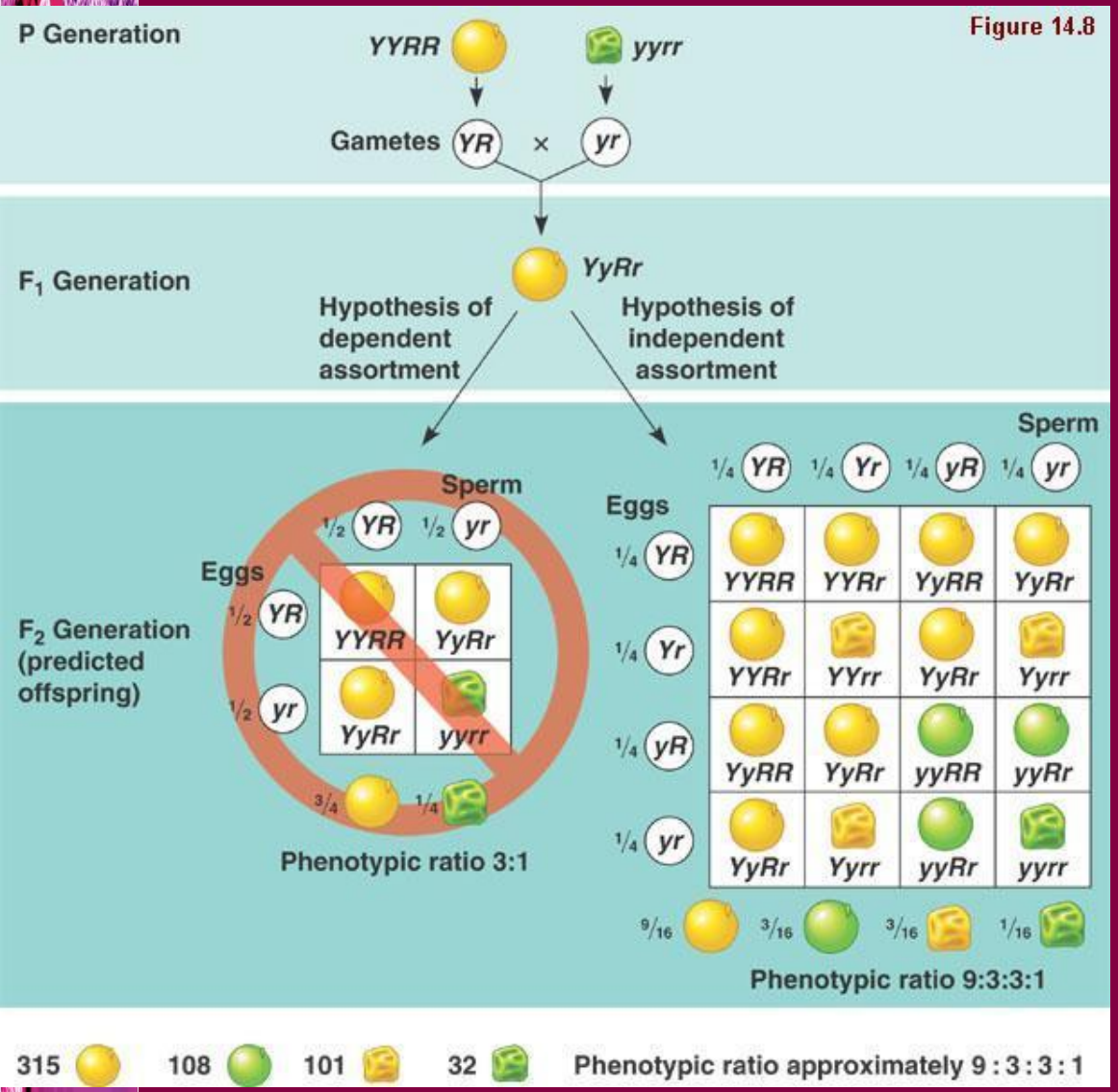
Monohybrid versus Dihybrid; and the Law of Independent Assortment

- A **monohybrid** organism is heterozygous with respect to a single gene of interest (in other words, one trait).
  - Example: ***Aa***. All the offspring from a cross between parents homozygous for different alleles are monohybrids, such as the F_1 progeny produced in Mendel's first set of crosses of true-breeding parents: ***AA × aa = Aa*.**
- A **dihybrid** organism is heterozygous with respect to two genes of interest (two traits).
  - Example: ***YyRr***. All the offspring from a cross between parents doubly homozygous for different alleles are dihybrids, for example: ***YYRR x yyrr = YyRr.***
  - *Are these two characters or traits transmitted from parents to offspring as a package? That is, will the **Y*** and ***R*** alleles always stay together, generation after generation?

Mendel's Model of Inheritance: The Law of Independent Assortment

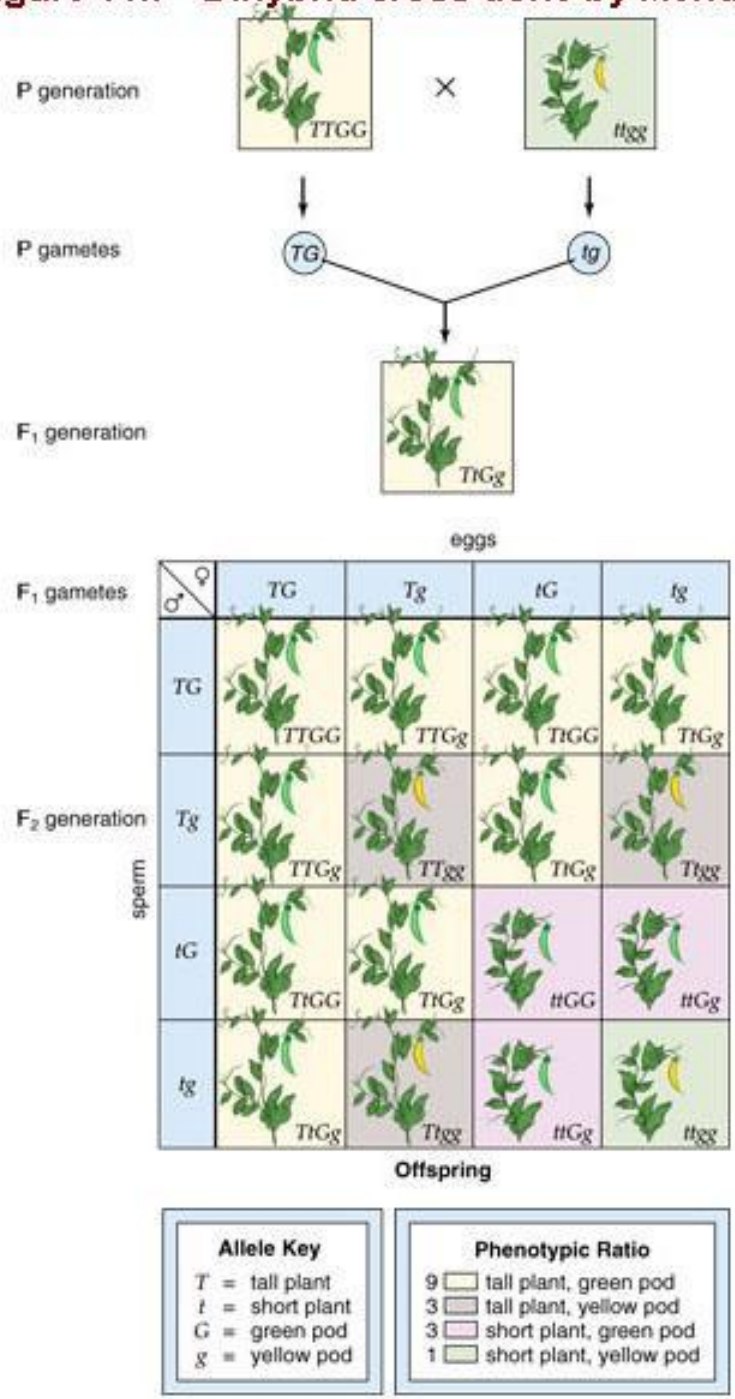
- Mendel identified his second law of inheritance by following two characters at the same time, such as seed color and seed shape.
- Law or Principle of Independent Assortment:
  - The *independent inheritance of two or more distinct traits*; it states that **the pair of alleles for one trait are distributed or assorted to the gametes independently (randomly) of the alleles for other traits (in meiosis)**.
    - In other words, **each pair** of genes (hereditary factors) **separates** (assorts, or distributes) **independently of the other pairs**.
    - New gene combinations not present in the parental (P) generation are produced.

Law of Independent Assortment



- Two true-breeding pea plants—one with yellow-round seeds and the other with green-wrinkled seeds-were crossed, producing **dihybrid F_1 plants**. Self-pollination of the F_1 dihybrids, which are **heterozygous** for both characters, produced the **F_2 generation**. Note that yellow color (***Y***) and round shape (***R***) are **dominant**.
- **Conclusion:** The results support the hypothesis of independent assortment. **The alleles for seed color and seed shape sort into gametes independently of each other**.

Figure 11.7 - Dihybrid cross done by Mendel.



Law of Independent Assortment in a Dihybrid Cross

- P generation plants differ in **two traits**—length of the stem and color of the pod. The **F_1** generation shows only the dominant traits, but all possible phenotypes appear among the **F_2** generation. The **9:3:3:1** ratio allowed Mendel to deduce that factors segregate into gametes independently of other factors.
- The law of independent assortment states that **each pair of factors segregate (assorts) independently of the other pairs**, and all possible combinations of factors can occur in the gametes.

Summary of Mendel's Laws or Principles of Heredity

Law of Segregation	*Law of Independent Assortment*
✓Each individual has **two factors (alleles)** for each trait. ✓The factors or **alleles segregate** (separate) during the formation of the gametes. ✓**Each gamete** (sperm or egg) contains only **one factor** from each pair of factors. ✓Fertilization gives each new individual two factors for each trait.	✓Each **pair of factors (alleles) distributes (assorts) into gametes independently of other pairs** for other traits. ✓All possible **combinations** of factors can occur in the gametes. *Note*: Strictly speaking, this law applies only to genes on different, nonhomolo-gous chromosomes. Genes located near each other on the same chromosome tend to be inherited together.

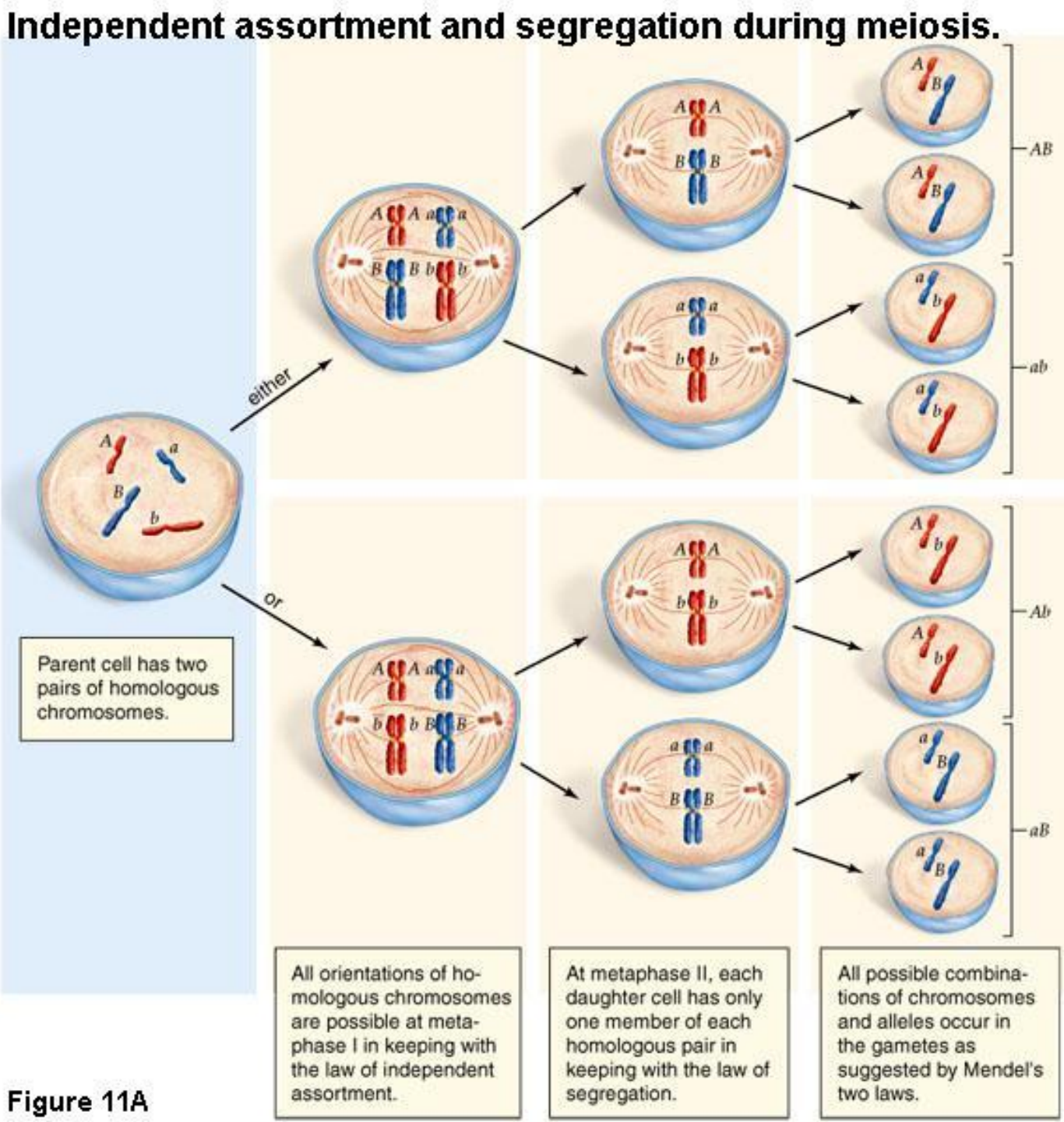


Figure 11A

Mendel's laws hold because of the events of meoisis. The homologous pairs of chromosomes line up randonmly at the metaphase plate during meioisis I. Therefore, the homologous chromosomes, and alleles they carry, segregate independently during gamete formation. All possible combinations of chromosomes and alleles occur in the gametes.

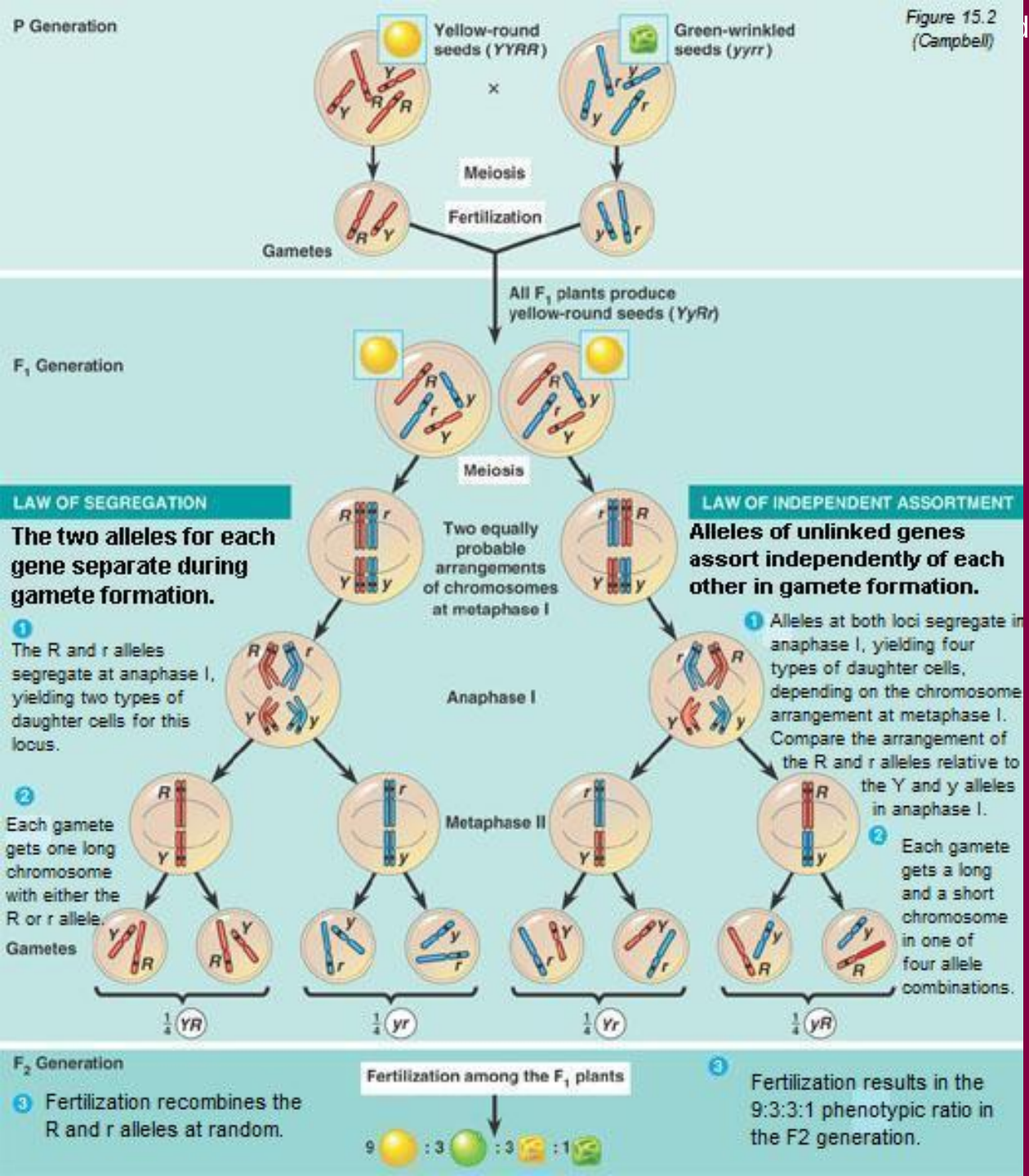


Figure 15.2 – The chromosomal basis of Mendel's laws. Correlation of the results of one of Mendel's dihybrid crosses with the behavior of chromosomes during meiosis.

Law of segregation: The two alleles for each gene separate during gamete formation. As an example, follow the fate of the long chromosomes (carrying *R* and *r*).

Law of independent assortment: Alleles of genes on nonhomologous chromosomes assort independently during gamete formation. As an example, follow both the long and short chromosomes along both paths.

Animations

- Monohybrid Cross:
  - ..\BIOLOGY-SOLOMON-Images\chapter11\Animations\monohybrid_v2.html
- Dihybrid Cross:
  - ..\BIOLOGY-SOLOMON-Images\chapter11\Animations\dihybrid_v2.html

Monohybrid Cross

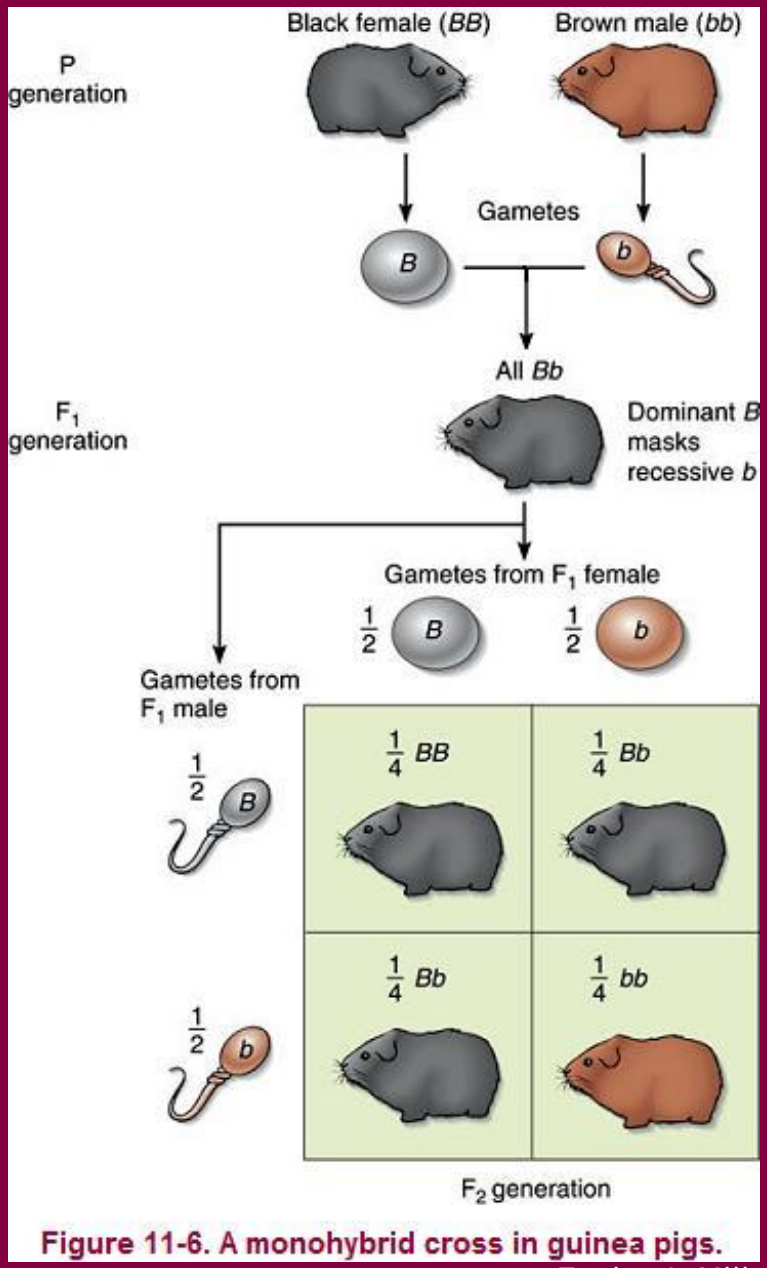


Figure 11-6. A monohybrid cross in guinea pigs.

Dihybrid Cross

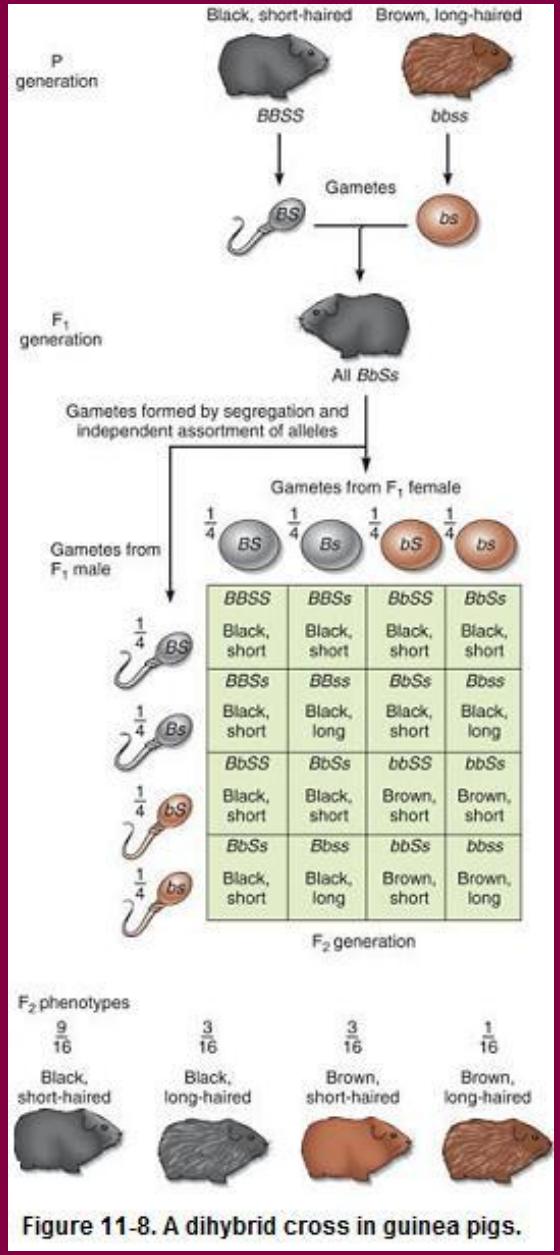


Figure 11-8. A dihybrid cross in guinea pigs.

TABLE 11-1

Mendel's Model of Inheritance

1. Alternative forms of a "factor" (what we now call a *gene*) account for variations in inherited traits.

Although Mendel only observed two forms (what we now call *alleles*) for each factor he studied, we now know that many genes have more than two alleles.

2. Inherited traits pass from parents to offspring as unmodified factors.

Mendel did not observe offspring of intermediate appearance, as a hypothesis of blending inheritance would have predicted. Exceptions to this concept are known today.

3. Each individual has two sets of factors, one of each pair inherited from the mother and one from the father.

It does not matter which parent contributes which set of factors.

4. The paired factors separate during the formation of reproductive cells (the principle of segregation).

Because of *meiosis*, which was discovered after Mendel's time, each parent passes one set of factors to each offspring.

5. Factors may be expressed or hidden in a given generation, but they are never lost.

For example, factors not expressed in the F_1 generation reappear in some F_2 individuals.

6. Each factor is passed to the next generation independently from all other factors (the principle of independent assortment).

Research since Mendel's time has revealed that there are exceptions to this principle.

Mendel's Model of Inheritance

Solving One-Trait Genetics Problems

1) **Decide on an appropriate allele key:** Assign letters to alleles to identify which allele is dominant and which is recessive. Use **uppercase letter** for ***dominant allele*** and **lowercase letter** for ***recessive allele***. For example:

   - If the **dominant** trait is *unattached earlobes* and the **recessive** trait is *attached earlobes*:
   - Allele key:
     - ***E*** = *unattached earlobes* (dominant trait)
     - ***e*** = *attached earlobes* (recessive trait)

Solving One-Trait Genetics Problems

2) Determine the **genotype of each parent** and indicate a cross (pairing).

- Example: Both parents that reproduce are ***heterozygous*** for unattached earlobes.
- * Remember that an individual is ***diploid*** (has two alleles for each trait).
- In the following diagram, P represents the parental generation. Their **genotypes** are:
  - **P: *Ee* × *Ee***

Solving One-Trait Genetics Problems

3) Determine the possible gametes that each parent can produce.

- * Remember that each gamete is ***haploid***, that is, *has only* ***one allele for each trait***.
- **P:** ***Ee*** × ***Ee***
- Gametes: *E, e* *E, e*

Solving One-Trait Genetics Problems: **The Punnett Square**

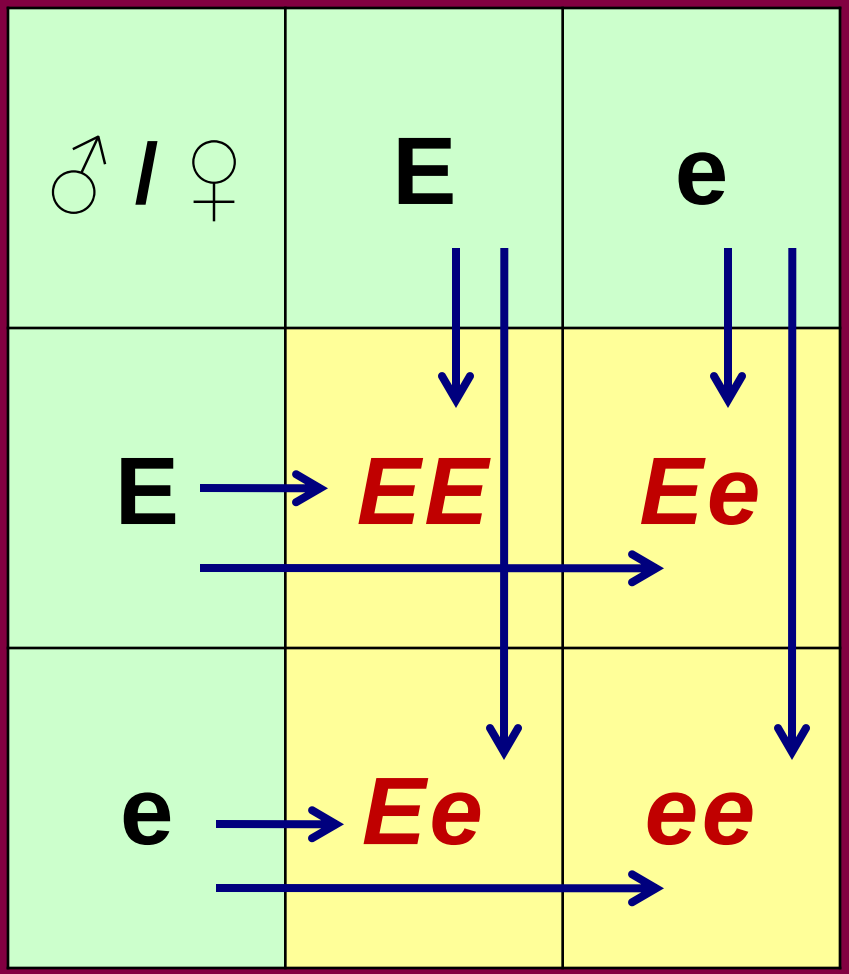
4) Determine the possible combinations of the alleles that can occur when gametes are paired by using the **Punnett Square**.

- The **Punnett Square** is a grid used to calculate the expected results of simple genetic crosses (in random fertilization). It allows us to determine the probability of genotypes and phenotypes of the offspring for a particular cross. It was introduced by the geneticist **R. C. Punnett** in the early 1900s.

Solving One-Trait Genetics Problems: **The Punnett Square**

4) All possible kinds of male gametes (sperm) are lined up vertically in the first column and all possible kinds of female gametes (eggs or ova) are lined up horizontally in the first row (or vice versa).

- Cross the gametes: Every possible combination of alleles is placed within the squares (alleles that can be donated by each progenitor are written inside the squares).
  - **P** (parents)**:** ***Ee*** × ***Ee***
  - Gametes: *E, e* *E, e*



Solving One-Trait Genetics Problems: **The Punnett Square**

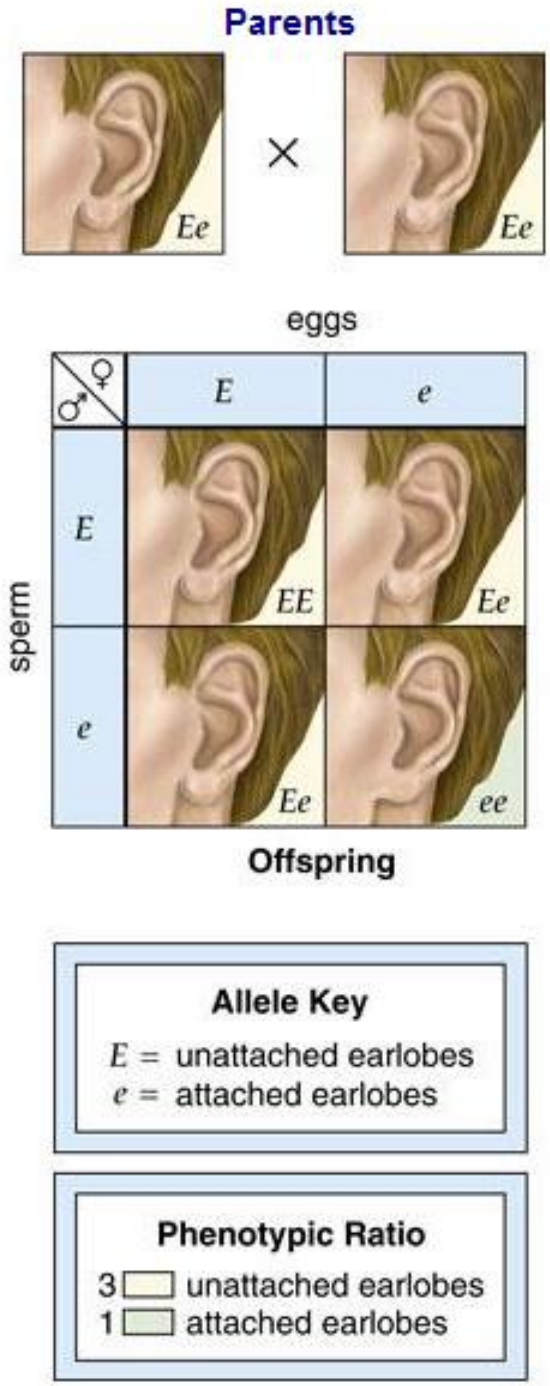
5) From the resulting genotypes, determine the phenotype of each combination of alleles. In other words, when the cross in this example occurs: What is the probability (chance) for each phenotype?

Ee × ***Ee***

♂/♀	**E**	**e**
E	***EE***	***Ee***
e	***Ee***	***ee***

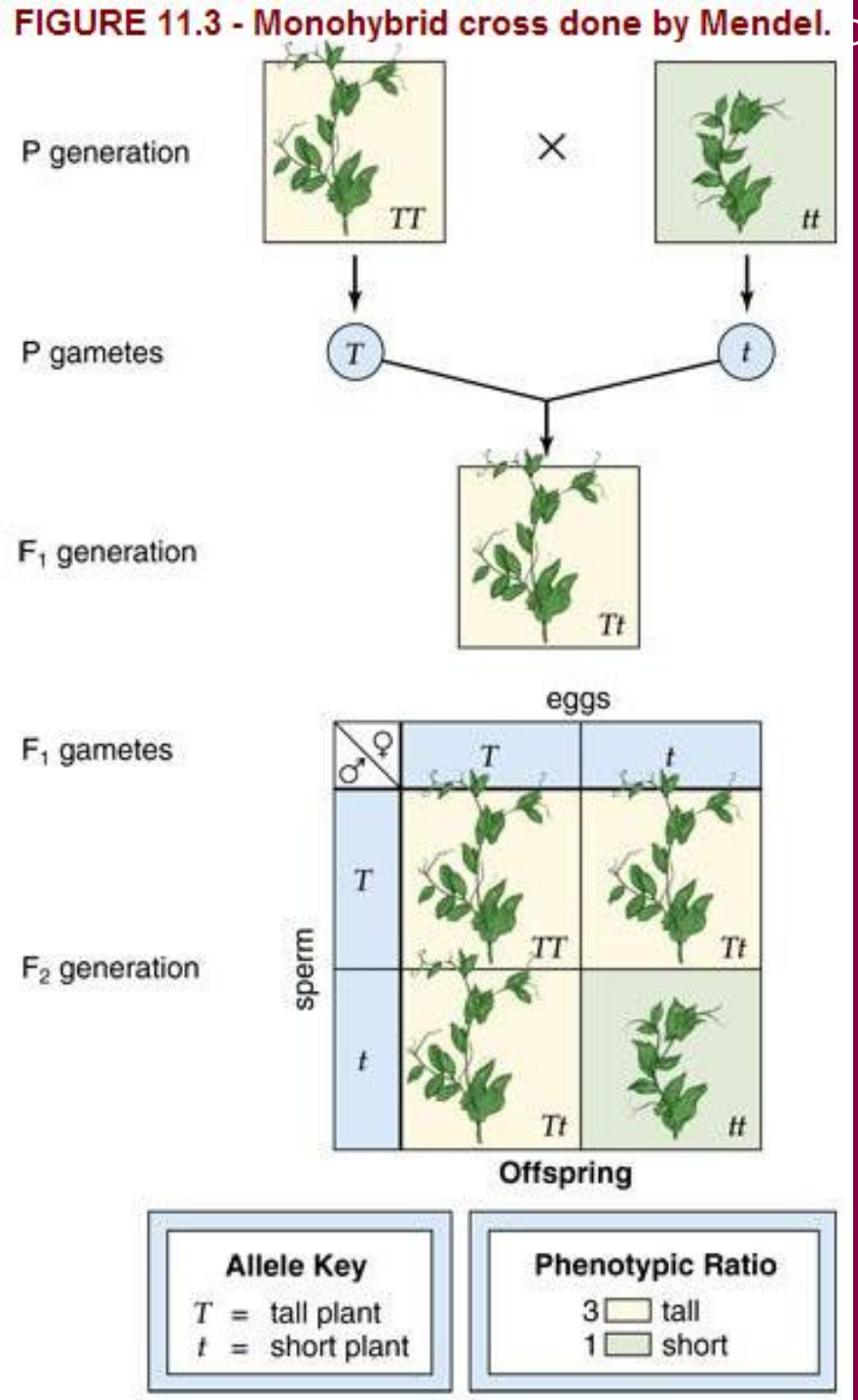
- * Remember the allele key:
  - ***E*** = *unattached earlobes*
  - ***e*** = *attached earlobes*
- Chance of <u>dominant phenotype</u> (unattached earlobes: ***EE, Ee, Ee)*** = ***¾ = 75%***
- Chance of <u>recessive phenotype</u> (attached earlobes: ***ee***) = **¼ = 25%**

FIGURE 11.5 - Genetic Inheritance in Humans.



Solving One-Trait Genetics Problems: **The Punnett Square**

- **FIGURE 11.5 – Genetic Inheritance in Humans.**
  - When the parents are heterozygous, each child has a 75% chance of having the dominant phenotype (unattached earlobes) and a 25% chance of having the recessive phenotype (attached earlobes).



Solving One-Trait Genetics Problems: **The Punnett Square**

- **FIGURE 11.3 – Monohybrid cross done by Mendel.**
- The P generation plants differ in one regard—length of the stem. The F_1 generation plants are all tall, but the factor for short has not disappeared because ¼ of the F_2 generation plants are short. The **3:1** ratio allowed Mendel to deduce that ***individuals have two discrete and separate genetic factors for each trait***.

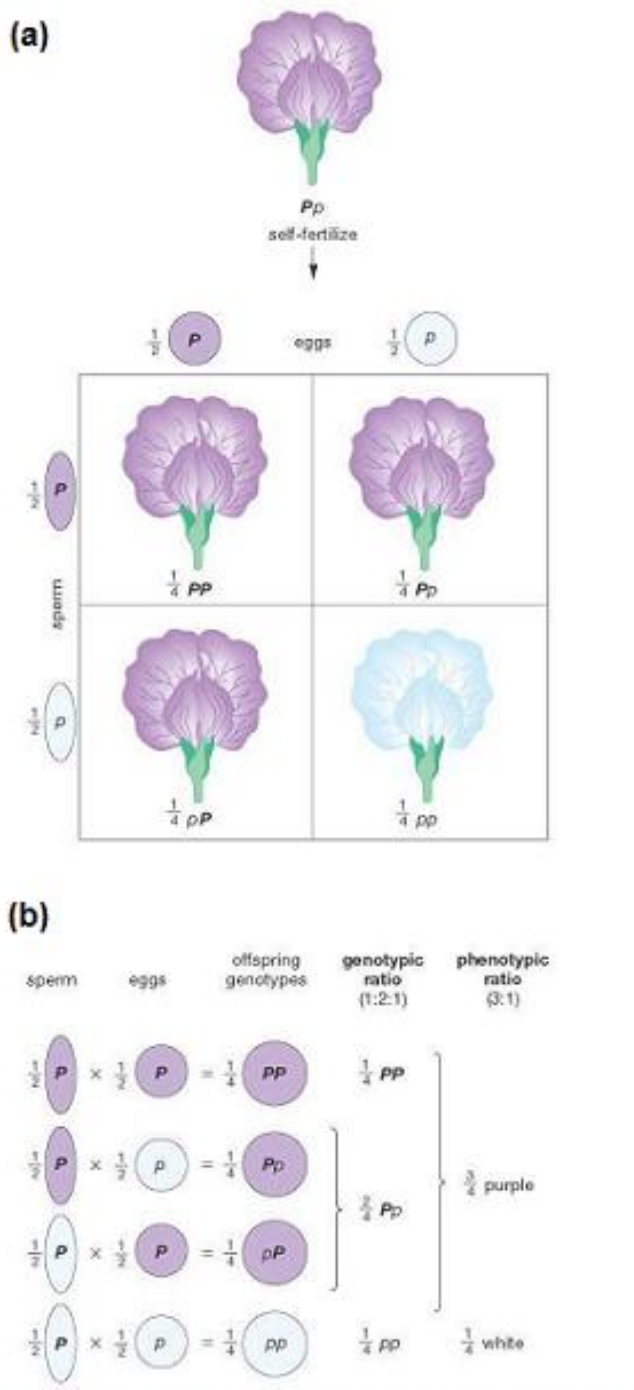


FIGURE 12-4 - Outcome of Single-Trait Cross

(a) Determining the Outcome of a Single-Trait Cross Using the Punnett Square

1) **Allele key**: Assign a letter to the different alleles; use the same letter for one trait, but uppercase for dominant and lowercase for recessive.
2) Write the **genotypes** of the parents and determine all the types of genetically different gametes that can be produced by the male and female parents.
3) Draw the **Punnett square**, with each row and column labeled with one of the possible genotypes of sperm and eggs, respectively.
4) Fill in the genotype of the offspring in each box by **combining** the genotype of sperm in its row with the genotype of the egg in its column.
5) **Count** the number of **offspring** with each genotype. (Note: Pp is the same as pP).
6) Convert the number of offspring of each genotype to a **fraction** of the total number of offspring. To determine phenotypic fractions, add the fractions of genotypes that would produce a given phenotype. It can also be expressed as a percentage.

Solving One-Trait Genetics Problems

- Make sure that you understand the difference between genotype and phenotype before trying to solve genetics problems.
- You also need to understand well homozygous vs. heterozygous, and dominant vs. recessive.

TABLE 11.1

Genotype Versus Phenotype

Genotype	*Genotype*	*Phenotype*
TT	Homozygous dominant	Tall plant
Tt	Heterozygous	Tall plant
tt	Homozygous recessive	Short plant

Solving One-Trait Genetics Problems

- Practice Problems 1.1

1. For each of the following genotypes, give all genetically different gametes, noting the proportion of each for the individual.
   a. *WW*
   b. *Ww*
   c. *Tt*
   d. *TT*
2. For each of the following, state whether a genotype (genetic makeup of an organism) or a type of gamete is represented.
   a. *D*
   b. *GG*
   c. *P*

Solving One-Trait Genetics Problems

- Solutions to Practice Problems 1.1

1. For each of the following genotypes, give all genetically different gametes, noting the proportion of each for the individual.

   a. *WW* → All ***W***

   b. *Ww* → ½ ***W***, ½ ***w***

   c. *Tt* → ½ ***T***, ½ ***t***

   d. *TT* → All ***T***

2. For each of the following, state whether a genotype (genetic makeup of an organism) or a type of gamete is represented.

   a. *D* → **Gamete**

   b. *GG* → **Genotype**

   c. *P* → **Gamete**

Solving One-Trait Genetics Problems: **Practice Problem 1.2**

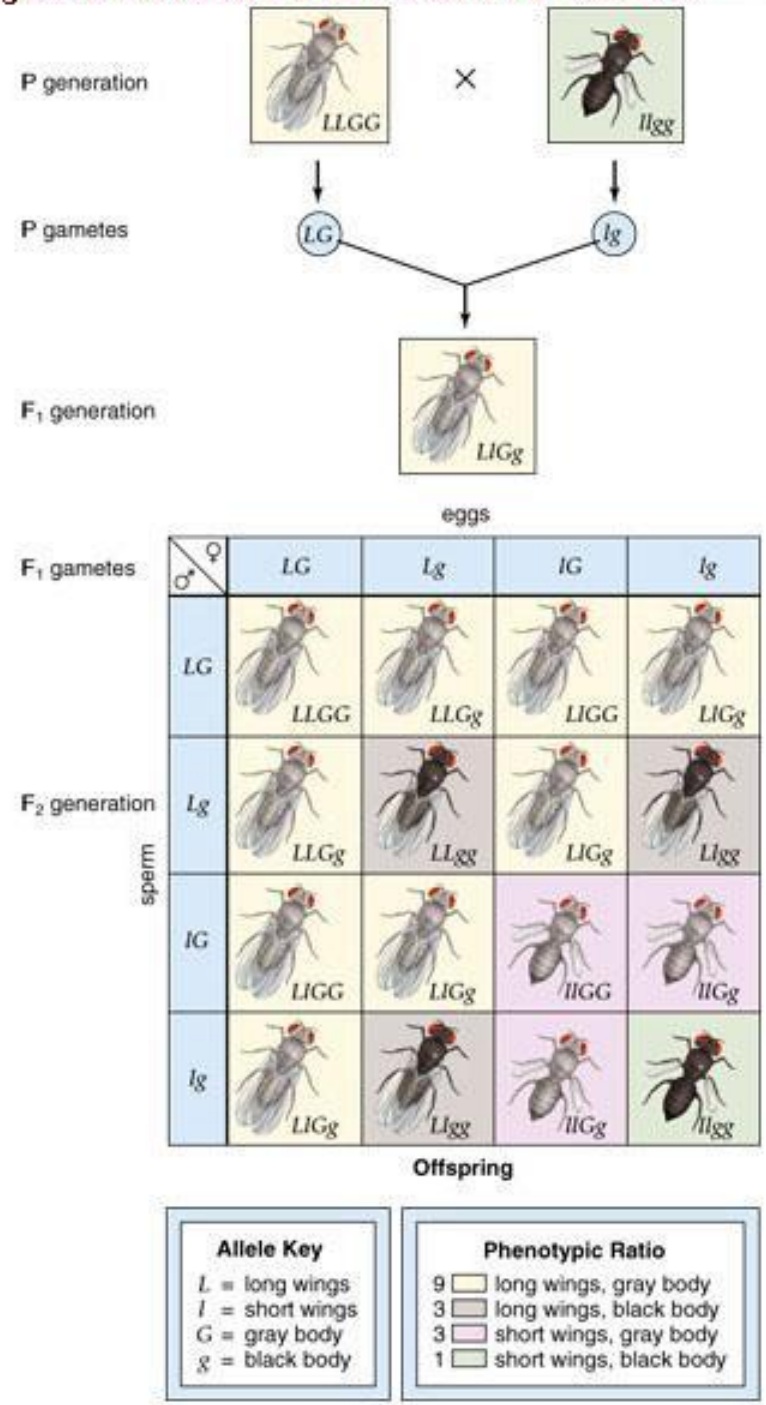
- In the garden pea, round seeds are dominant over wrinkled seeds. An investigator crosses a plant having round seeds with a plant having wrinkled seeds. He counts 400 offspring.
- **How many of the offspring have wrinkled seeds if the plant having round seeds is a heterozygote**?

Solving One-Trait Genetics Problems:
Practice Problem 1.2

- SOLUTION: * Remember that the problem tells us that round seeds are dominant over wrinkled seeds, and the plant with round seeds is heterozygous.
  - Allele key:
    - ***R*** = *round seeds* (dominant)
    - ***r*** = *wrinkled seeds* (recessive)
  - **P** (parents)**:** ***Rr*** × ***rr***
  - Gametes: *R, r* *r, r*
  - Chance of dominant phenotype (***Rr, Rr)*** ***= 2/4 = 50%***
  - Chance of recessive phenotype (***rr***) = ***2/4 = 50%***
  - After solving the problem by using the Punnett square the answer is:
    - **200 plants have wrinkled seeds** (50% of the 400 offspring).

♂ / ♀	***r***	***r***
R	***Rr***	***Rr***
r	***rr***	***rr***

Figure 11.8. Mendelian inheritance in fruit flies.



Two-Trait Genetics Problems

Mendelian Inheritance in Fruit Flies

- Each **P generation** fly has only one possible type of gamete because the P_1 fly is either homozygous dominant or homozygous recessive for both traits. All the **F_1** flies are heterozygous (*LlGg*) and have the same phenotype (long wings, gray body).
- The Punnett square shows the expected results when the F_1 flies are crossed. Each **F_1** fly (*LlGg*) produces four types of gametes because all possible combinations of chromosomes can occur in the gametes. Therefore, all possible phenotypes appear among the **F_2 generation**.
- Notice that **9/16** of the offspring have long wings and a gray body, **3/16** have long wings and a black body, **3/16** have short wings and a gray body, and **1/16** have short wings and a black body. This phenotypic ratio **(9:3:3:1)** is expected whenever **a heterozygote for two traits is crossed with another heterozygote for two traits** and simple dominance is present in both genes.

Practice Problem: **Two Traits**

- The steps to solve a two trait genetics problem are the same as for a one trait problem, but you need to keep in mind the law of independent assortment: ***each pair of alleles segregates independently of each other pair of alleles during gamete formation***. Also, remember that you will assign a different letter to each trait.
- Problem:
  - In humans, ***short fingers*** and ***widow's peak*** are **dominant** over ***long fingers*** and ***straight hairline***. A **heterozygote** in both regards reproduces with a similar **heterozygote**. ***What is the chance of any one child having the same phenotype as the parents?***

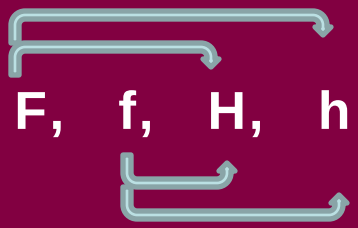
Solution to Practice Problem: **Two Traits**

- **Allele key:** *** You need to remember what is homozygous, heterozygous, dominant, recessive, etc.
  - ***F*** = short fingers (dominant)
  - ***f*** = long fingers (recessive)
  - ***H*** = widow's peak (dominant)
  - ***h*** = straight hairline (recessive)
- ***Genotype of parents*:** The problem indicates that both parents are **heterozygous for both traits** (Ff Hh); in other words, they both have **short fingers**, and **widow's peak**.
  - ***P*** (parents): ***FfHh*** × ***FfHh***
- **Determine gametes and possible combinations** of alleles that can occur when gametes are paired by using the **Punnett square** (see next slide).

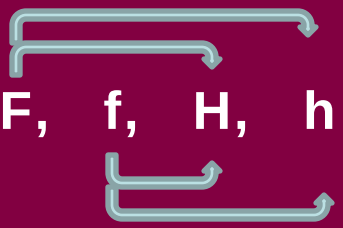
Solution to Practice Problem: **Two Traits**

- ***P*** (parents)**: *FfHh*** × ***FfHh***
- **Gametes: F, f, H, h** **x** **F, f, H, h**

- To pair the gametes in the Punnett square, combine them as follows and then write the combinations in the outer squares:



x



♂ / ♀				

Solution to Practice Problem: Two Traits

♂ / ♀	*FH*	*Fh*	*fH*	*fh*
FH	***FFHH***	***FFHh***	***FfHH***	***FfHh***
Fh	***FFHh***	***FFhh***	***FfHh***	***Ffhh***
fH	***FfHH***	***FfHh***	***ffHH***	***ffHh***
fh	***FfHh***	***Ffhh***	***ffHh***	***ffhh***

Interpretation of results and answer to question (remember allele key):

- *FFHH* = 1 = short fingers, widow's peak
- *FFHh* = 2 = short fingers, widow's peak
- *FfHH* = 2 = short fingers, widow's peak
- *FfHh* = 4 = short fingers, widow's peak

9 / 16 (This is the answer; why?)

- *FFhh* = 1 = short fingers, straight hairline
- *Ffhh* = 2 = short fingers, straight hairline

3 / 16

- *ffHH* = 1 = long fingers, widow's peak
- *ffHh* = 2 = long fingers, widow's peak

3 / 16

- *ffhh* = 1 = long fingers, straight hairline → 1 / 16

Solution to Practice Problem: Two Traits

♂ / ♀	*FH*	*Fh*	*fH*	*fh*
FH	***FFHH***	***FFHh***	***FfHH***	***FfHh***
Fh	***FFHh***	***FFhh***	***FfHh***	***Ffhh***
fH	***FfHH***	***FfHh***	***ffHH***	***ffHh***
fh	***FfHh***	***Ffhh***	***ffHh***	***ffhh***

Allele key
F = short fingers
f = long fingers
H = widow's peak
h = straight hairline

Phenotypic ratio
9 = short fingers, widow's peak
3 = short fingers, straight hairline
3 = long fingers, widow's peak
1 = long fingers, straight hairline

- The question was: **What is the chance of any one child having the same *phenotype* as the parents?** Remember: The parents are ***heterozygous*** for both traits, in other words, they both have **short fingers and widow's peak** (with genotype ***FfHh***).
- ***** Answer:** The chance of any one child having the same **phenotype** as the parents is **9/16** (Note that genotypes can be different).

Beyond Mendelian Genetics: Inheritance patterns are often more complex than predicted by simple Mendelian genetics.

- Mendel studied traits that were determined by one gene, for which there are only two alleles, one completely dominant to the other. However, alleles can show different ***degrees of dominance*** and recessiveness in relation to each other.
- **Complete dominance:** The situation in which the phenotypes of the heterozygote (Aa) and dominant homozygote (AA) are indistinguishable.
  - Example: The F_1 offspring of Mendel's classic pea crosses. The F_1 plants always looked like one of the two parental varieties because of the complete dominance of one allele over another.

Mendel's Experiment Shows Complete Dominance

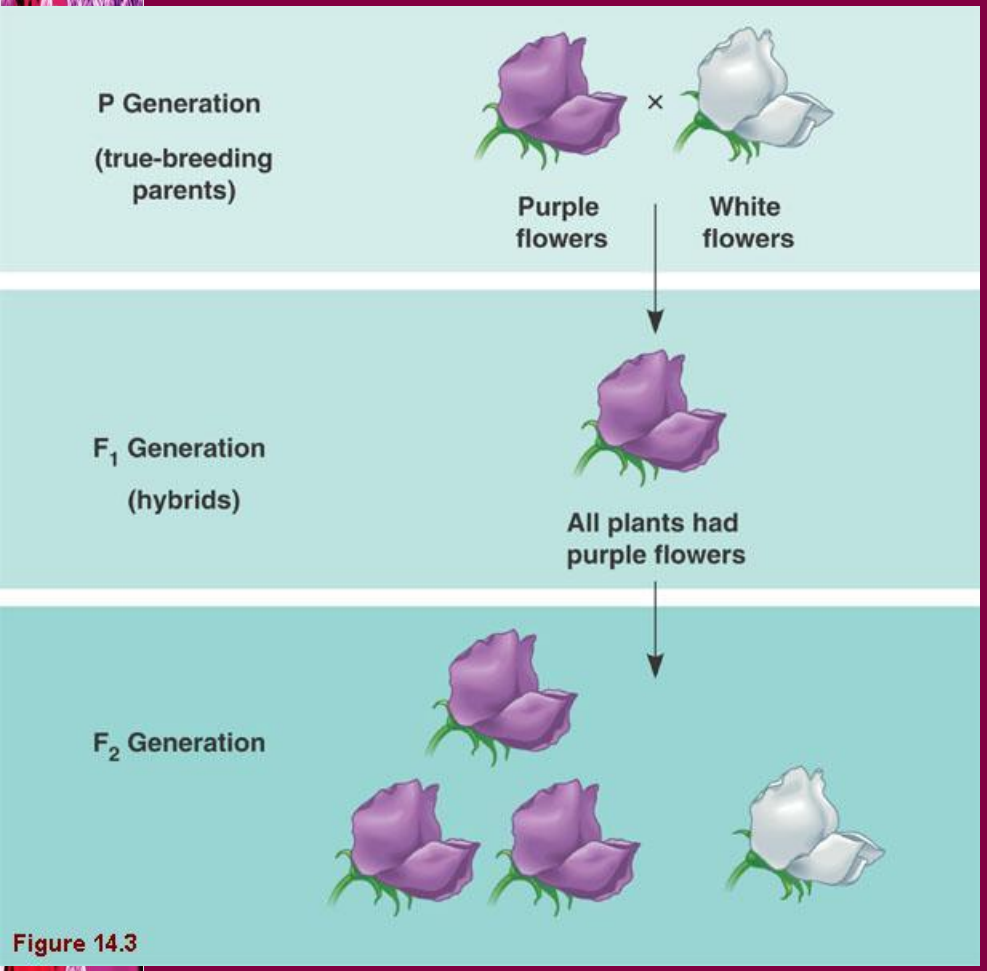


Figure 14.3

*** The white flower color did not disappear, but the *purple color dominated*.**

- Mendel crossed **true-breeding** purple-flowered pea plants and white-flowered pea plants. The resulting **F_1 hybrids** (*all purple*) were allowed to self-pollinate or were cross-pollinated with other F_1 hybrids. Flower color was then observed in the F_2 generation.
- ***Results*:** Both purple-flowered plants and white-flowered plants appeared in the **F_2** generation.
- In Mendel's experiment 705 plants had purple flowers, and 224 had white flowers, showing a ratio of approximately **3 purple: 1 white**.
- This showed that purple flower color is a ***dominant*** trait and white flower color is a ***recessive*** trait.

Beyond Mendelian Genetics

- **Incomplete dominance:** Inheritance pattern in which the offspring has an **intermediate phenotype** (the phenotype of heterozygotes is intermediate between the phenotypes of individuals homozygous for either allele).
  - Example: When a red-flowered plant and a white-flowered plant produce pink-flowered offspring. (See next slide.)

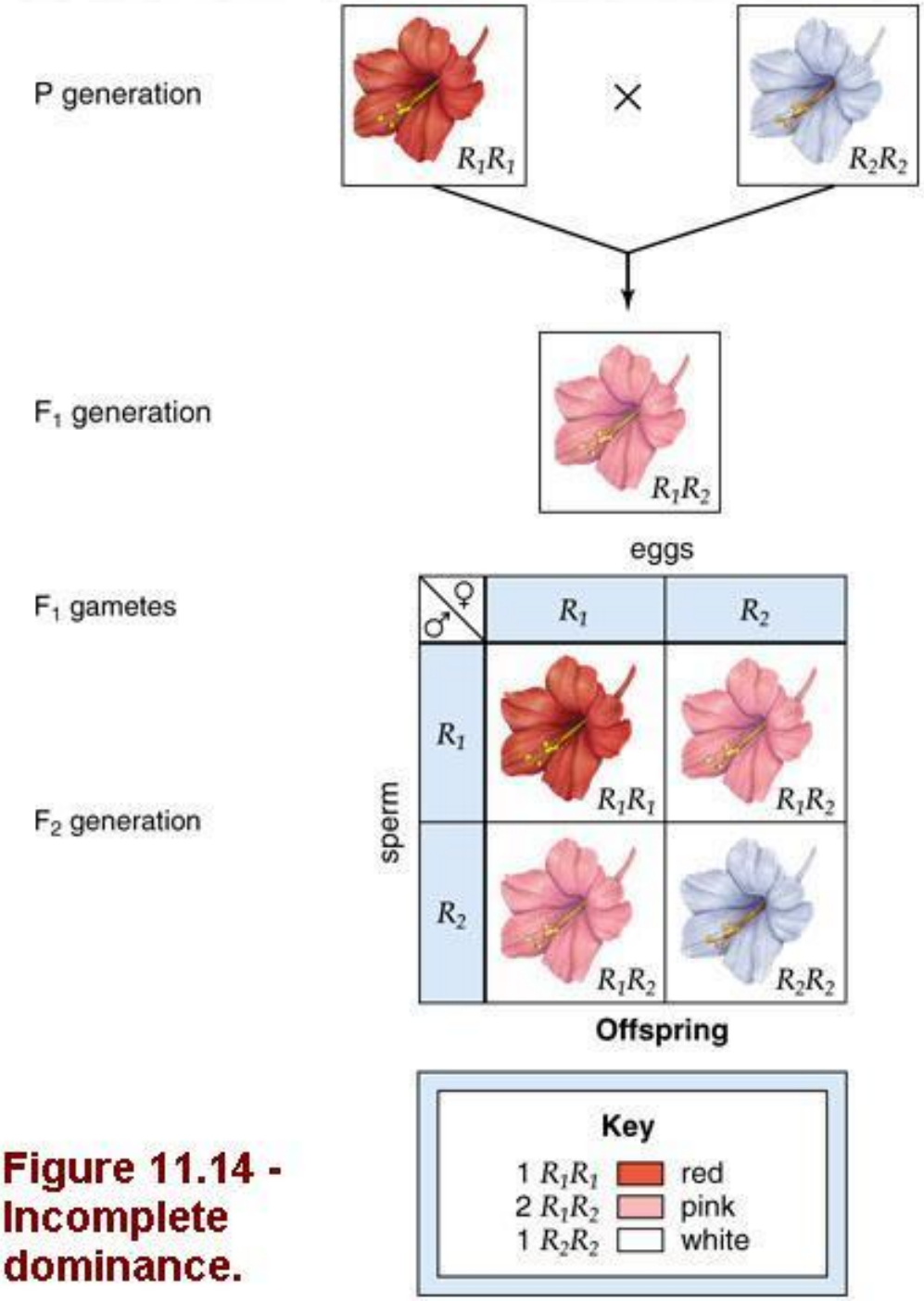


Figure 11.14 - Incomplete dominance.

Incomplete Dominance

- **Incomplete dominance** is illustrated by a cross between red- and white-flowered four-o'clocks. The F_1 plants are pink, a phenotype **intermediate** between those of the P generation; however the F_2 results show that neither the red nor the white allele has disappeared.

Animation: Incomplete Dominance

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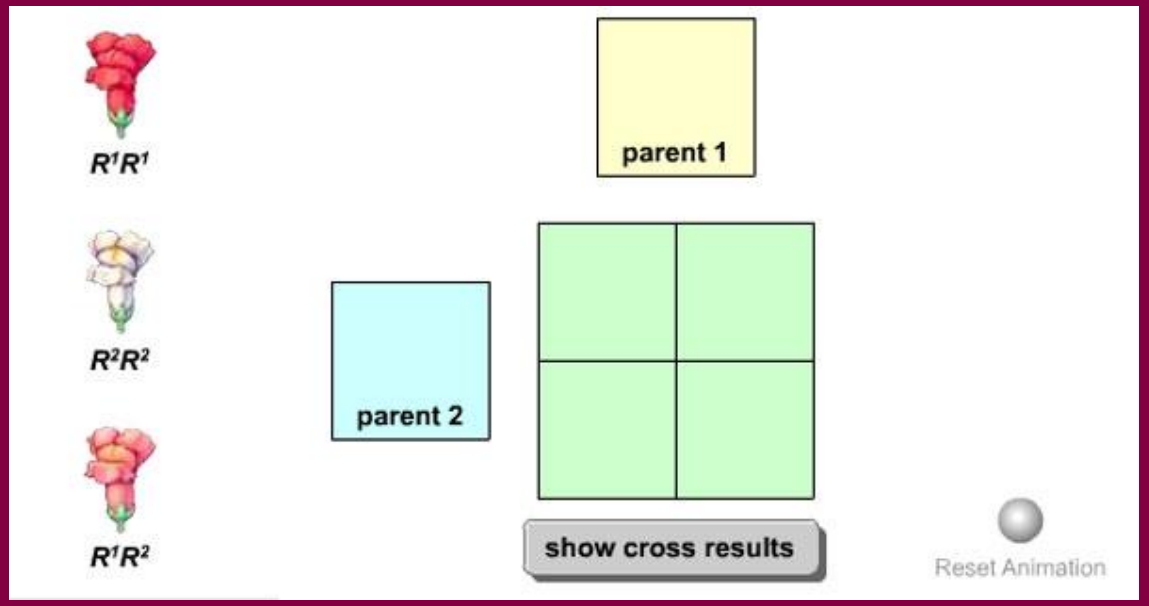


Beyond Mendelian Genetics:
Codominance and Multiple Alleles

- **Codominance:** Inheritance pattern in which both alleles of a gene are **equally expressed** (more than one dominant allele).
- **Multiple alleles:** Inheritance pattern in which there are more than two alleles for a particular trait; each individual has only two of all possible alleles.
  - Example: **ABO blood types** in humans. Three alleles for the same gene control the inheritance of ABO blood types. It is also an example of codominance because both I^A and I^B are dominant over ***i***.
    - I^A is dominant = Carbohydrate A on red blood cells.
    - I^B is dominant = Carbohydrate B on red blood cells.
    - ***i*** is recessive = Neither A nor B on red blood cells.

Codominance & Multiple Alleles

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

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

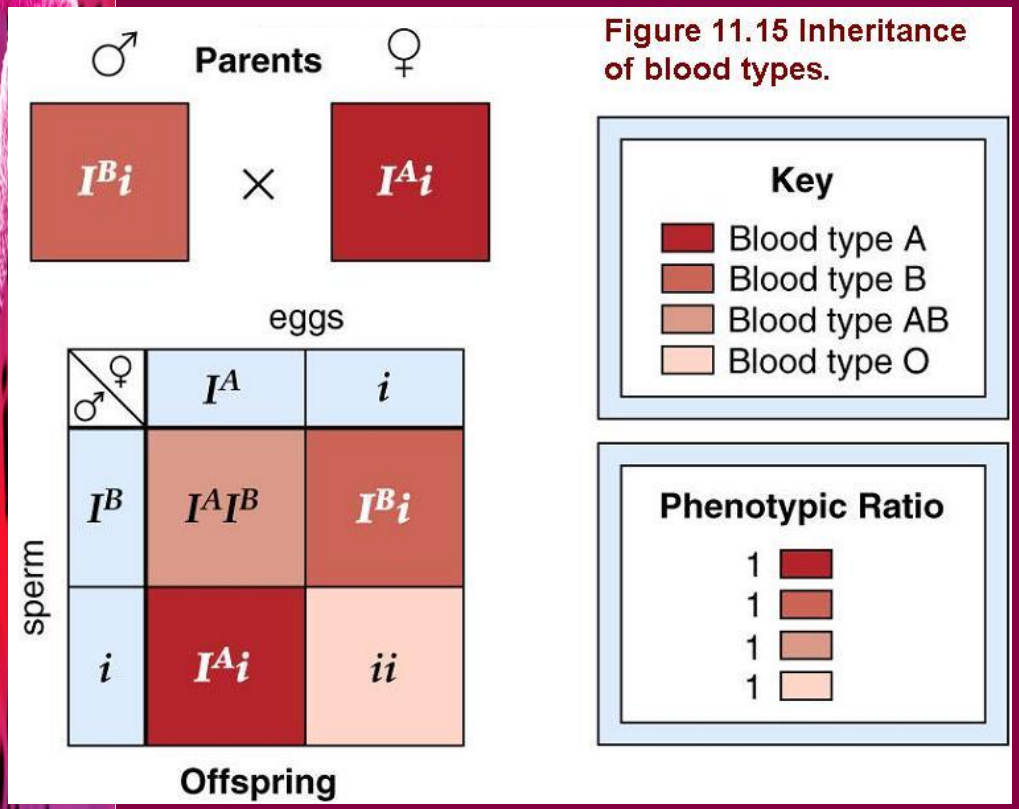


Figure 11.15 Inheritance of blood types.

Table 14.2 Determination of ABO Blood Group by Multiple Alleles

Genotype	Phenotype (Blood Group)	Red Blood Cells
I^AI^A or I^Ai	A	
I^BI^B or I^Bi	B	
I^AI^B	AB	
ii	O	

A mating between individuals with type A blood and type B blood can result in any one of the four blood types. Why? Because the parents are ***I*^{B}*i*** and ***I*^{A}*i***.

Codominance and Multiple Alleles

TABLE 19.6 Summary of ABO Blood Group Interactions

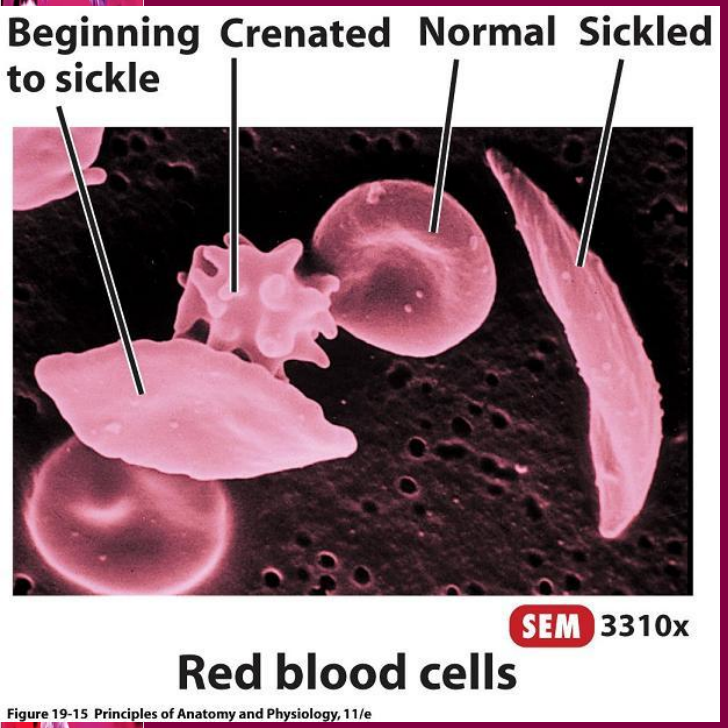
Characteristic	Blood Type A	Blood Type B	Blood Type AB	Blood Type O
Agglutinogen (antigen) on RBCs	A	B	Both A and B	Neither A nor B
Agglutinin (antibody) in plasma	anti-B	anti-A	Neither anti-A nor anti-B	Both anti-A and anti-B
Compatible donor blood types (no hemolysis)	A, O	B, O	A, B, AB, O	O
Incompatible donor blood types (hemolysis)	B, AB	A, AB	—	A, B, AB

Table 19-6 Principles of Anatomy and Physiology, 11/e

- People with type AB blood do not have anti-A or anti-B antibodies in their plasma. They are sometimes called *universal recipients* because theoretically they can receive blood from donors of all blood types. They have no antibodies to attack antigens on donated RBCs.
- People with type O blood have neither A nor B antigens on their RBCs and are sometimes called *universal donors* because theoretically they can donate blood to all four ABO blood types. Type O persons requiring blood may receive only type O blood.

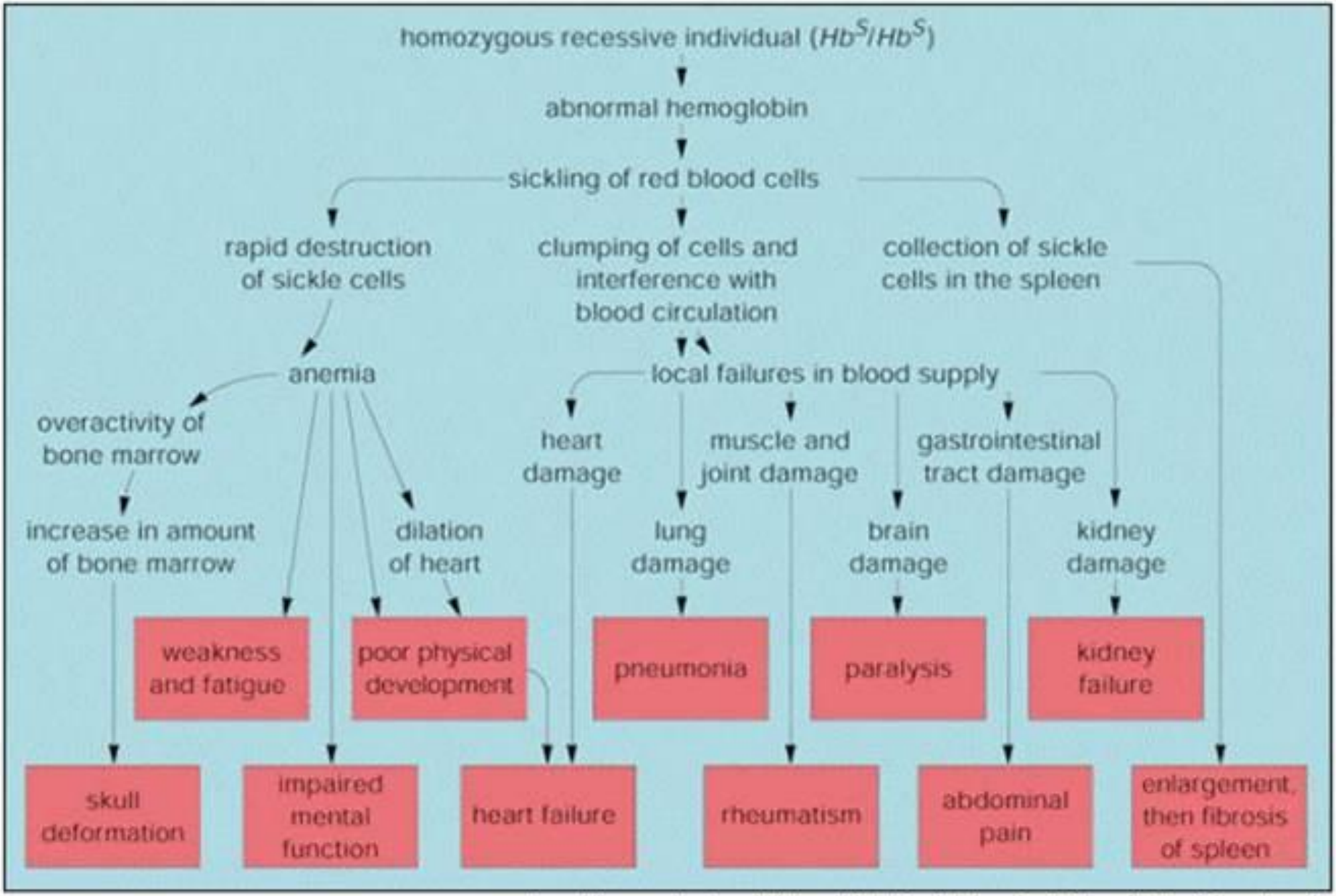
In practice, use of the terms universal recipient and universal donor is misleading and dangerous. Blood contains antigens and antibodies other than those associated with the ABO system that can cause transfusion problems.

Beyond Mendelian Genetics



- **Pleiotropy:** Inheritance pattern in which **one gene have multiple effects, or affects more than one phenotypic characteristic** of the individual.
- Example: sickle cell anemia.
  - Two alleles exist for the **hemoglobin** gene: Hb^A encodes normal (wild-type) hemoglobin; Hb^S (mutant allele) encodes defective hemoglobin. Hb^S homozygotes produce only the defective hemoglobin and suffer the disease. The altered hemoglobin affects the shape of the red blood cells, which clump together and impedes oxygen delivery and blood flow. Over time, this results in damage throughout the body tissues.

SICKLE CELL ANEMIA



http://trc.ucdavis.edu/biosci10v/bis10v/week4/06multipleeffects.html

Extending Mendelian Genetics for a Single Gene

Degree of dominance	Description	Example
Complete dominance of one allele	Heterozygous phenotype same as that of homozygous dominant	
Incomplete dominance of either allele	Heterozygous phenotype intermediate between the two homozygous phenotypes	
Codominance	Heterozygotes: Both phenotypes expressed	
Multiple alleles	In the whole population, some genes have more than two alleles	ABO blood group alleles I^A, I^B, i
Pleiotropy	One gene is able to affect multiple phenotypic characters	Sickle-cell disease

Extending Mendelian Genetics for a Single Gene

Extending Mendelian Genetics for Two or More Genes

- **Epistasis:** Inheritance pattern in which **one gene masks the expression of another gene that is at a different locus and is independently inherited.** In other words, one gene produces a phenotype and a second gene prevents that first gene from producing its phenotype.
- Example: In mice (and other mammals) black coat color **(*B*)** is dominant to brown **(*b*)**. A ***second gene*** determines whether or not pigment will be deposited in the hair.
  - The dominant allele **(*C*)** results in deposition of either black or brown pigment, depending on the genotype at the first locus. But if the mouse is homozygous recessive for the second locus **(*cc*)**, then the coat is white (albino), regardless of the genotype at the black/brown locus. The gene for pigment deposition is said to be **epistatic** to the gene that codes for black or brown pigment.

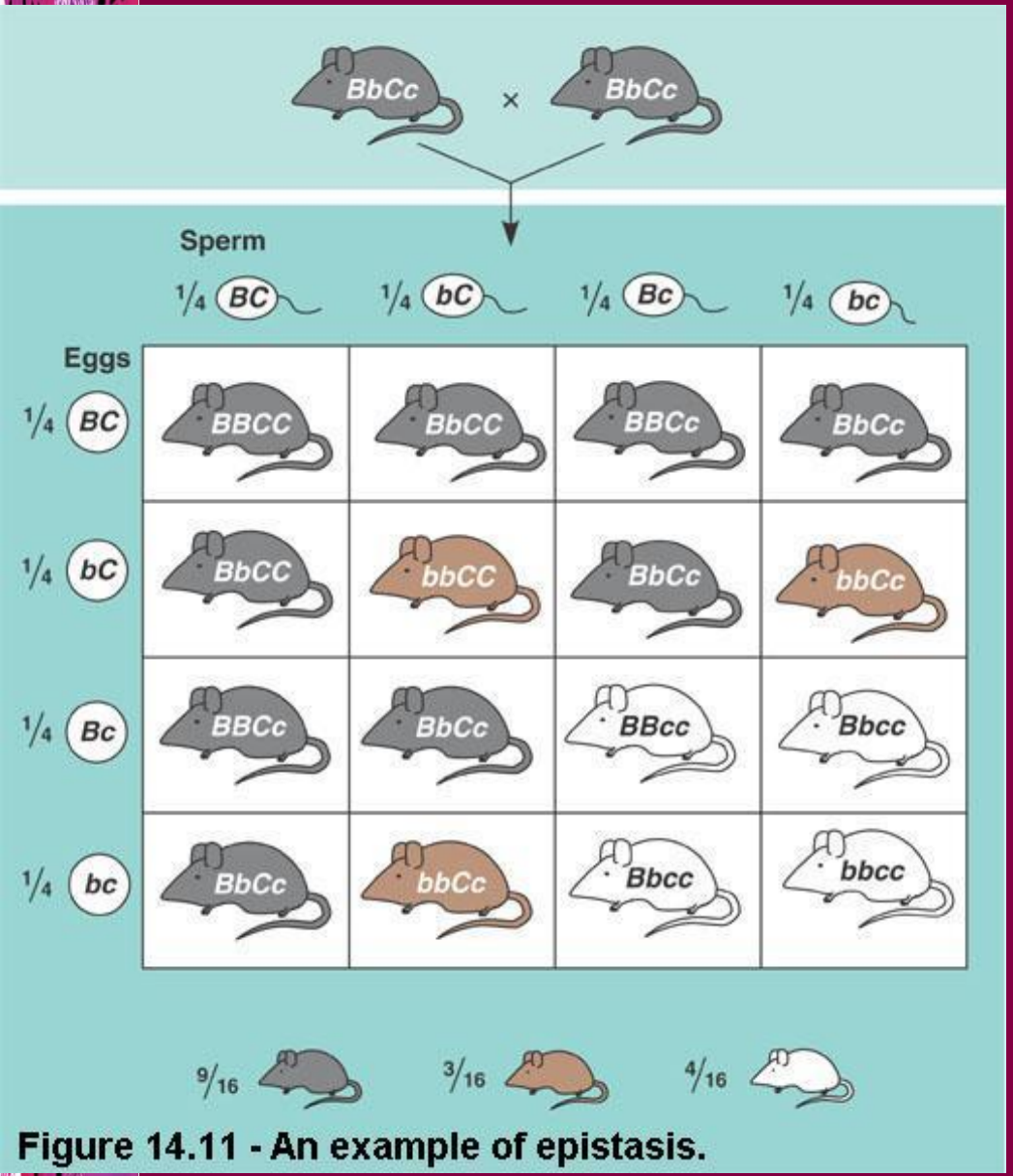


Figure 14.11 - An example of epistasis.

Epistasis

- This Punnett square illustrates the genotypes and phenotypes predicted for offspring of matings between two black mice of genotype BbCc. The C/c gene, which is epistatic to the B/b gene, controls whether or not pigment of any color will be deposited in the hair.

Epistasis

Figure 11-21. Epistasis in Labrador retrievers.

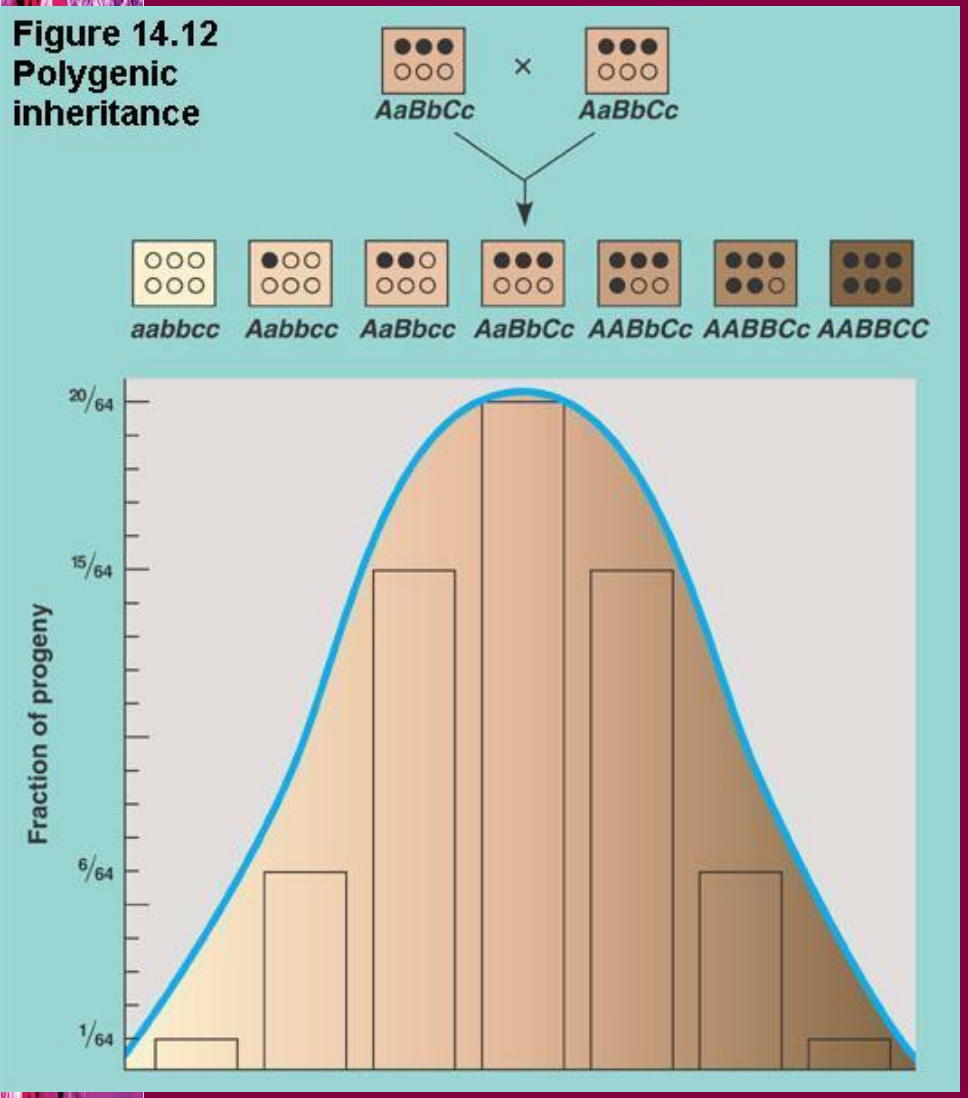
Black	Yellow	Chocolate
BBEE, BbEE, BBEe or *BbEe*	*BBee, Bbee* or *bbee*	*bbEE* or *bbEe*

- Two gene pairs interact to govern coat color in Labrador retrievers. Black Labs have the genotype *B_E_*; yellow Labs have the genotype *B_ee* or *bbee*; and chocolate Labs have the genotype *bbE_*. (The blanks represent either dominant or recessive alleles).

Extending Mendelian Genetics for Two or More Genes

- **Polygenic inheritance:** Pattern of inheritance in which ***a trait is controlled by two or more pairs of alleles***; each dominant allele contributes to the phenotype in an additive and like manner. (*It is the converse of pleitropy, where a single gene affects several phenotypic characters*.)
- **Examples: human height, eye color, skin color.**
- An **either-or** classification is impossible because the characters vary in the population along a continuum (in gradations). These are called **quantitative characters**.

A Simplified Model for Polygenic Inheritance



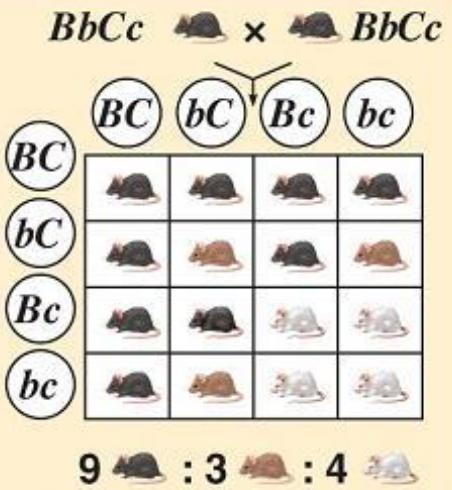
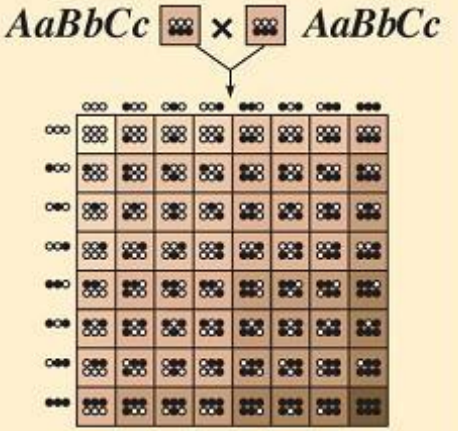
- Three separately inherited genes affect **skin pigmentation**. The heterozygous individuals (AaBbCc, top of figure), each carry three dark-skin alleles (black circles) and three light-skin alleles (open circles). The variations in genotype and skin color that can occur among offspring from a large number of hypothetical matings between these heterozygotes are shown above the graph. The *y*-axis represents the fraction of progeny with each skin color. The resulting histogram is smoothed into a bell-shaped curve by environmental factors that affect skin color (e.g. exposure to sun).

Polygenic Inheritance

Figure 11.17. Height in human beings.

- When you record the **heights** of a large group of people chosen at random, the values typically follow a bell-shaped curve. **Such a continuous distribution is due to control of a trait by several sets of alleles**. Environmental effects can also influence gene expression.

Extending Mendelian Genetics for Two or More Genes

Relationship among genes	Description	Example
Epistasis	One gene affects the expression of another	
Polygenic inheritance	A single phenotypic character is affected by two or more genes	

Extending Mendelian Genetics for Two or More Genes

Nature and Nurture: The Environmental Impact on Phenotype

- The expression of a phenotype for a character depends on environmental influences as well as on genotype.
  - For example, in human height, *nutrition* is a major factor.
  - Phenotypes of plants and animals are influenced by *temperature*, for example, plant leaves change color in fall.

Figure 11.18. Coat color in Himalayan rabbits.

The influence of the environment on the phenotype has been demonstrated by plucking out the fur from one area and applying an ice pack. The new fur that grew in was black instead of white, showing that **the enzyme that produces melanin (a dark pigment) in the rabbit is active only at low temperatures**.

Human Genetics and Pedigree Analysis

- A **pedigree** is a diagram, or a ***family tree***, showing the occurrence of heritable characters in parents and offspring over multiple generations. A pedigree is also useful to show the pattern of inheritance for a particular medical condition.
- Many genetic disorders (caused by alleles inherited from parents) can be explained on the basis of simple Mendelian inheritance.
  - **Dominantly Inherited Disorder** (autosomal dominant disorder)**:** ***Homozygous dominant*** individuals (with the alleles ***AA***) or ***heterozygous*** individuals (with the alleles ***Aa***) have the disorder.
  - **Recessively Inherited Disorder** (autosomal recessive disorder)**:** Only ***homozygous recessive*** individuals (with the alleles ***aa***) have the disorder.

Human Genetics Analysis: ***Pedigree***

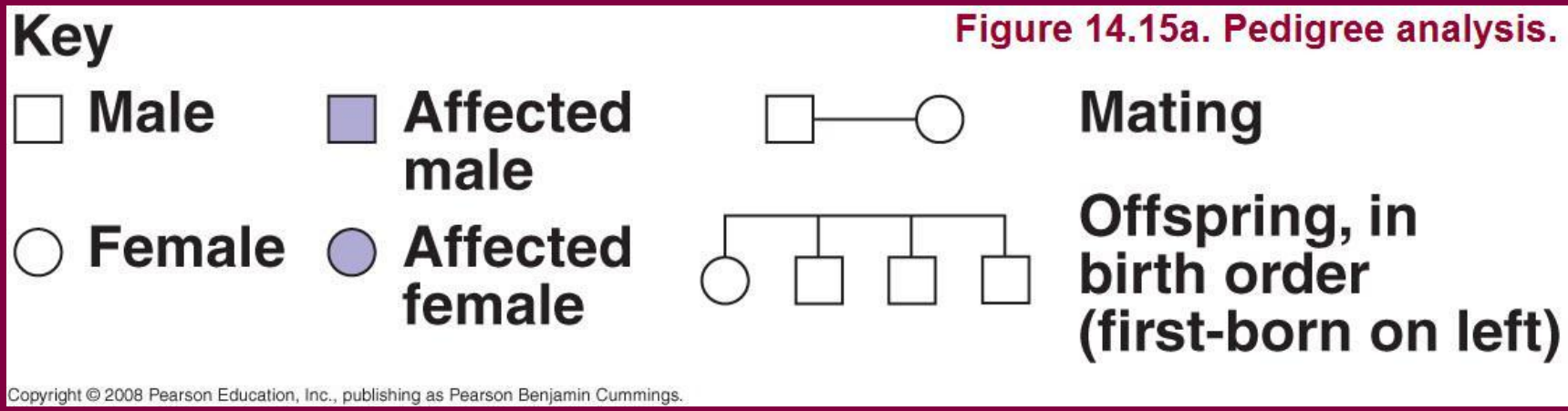
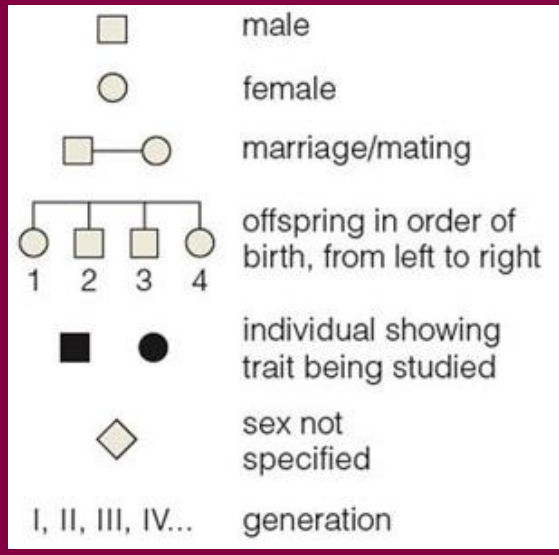


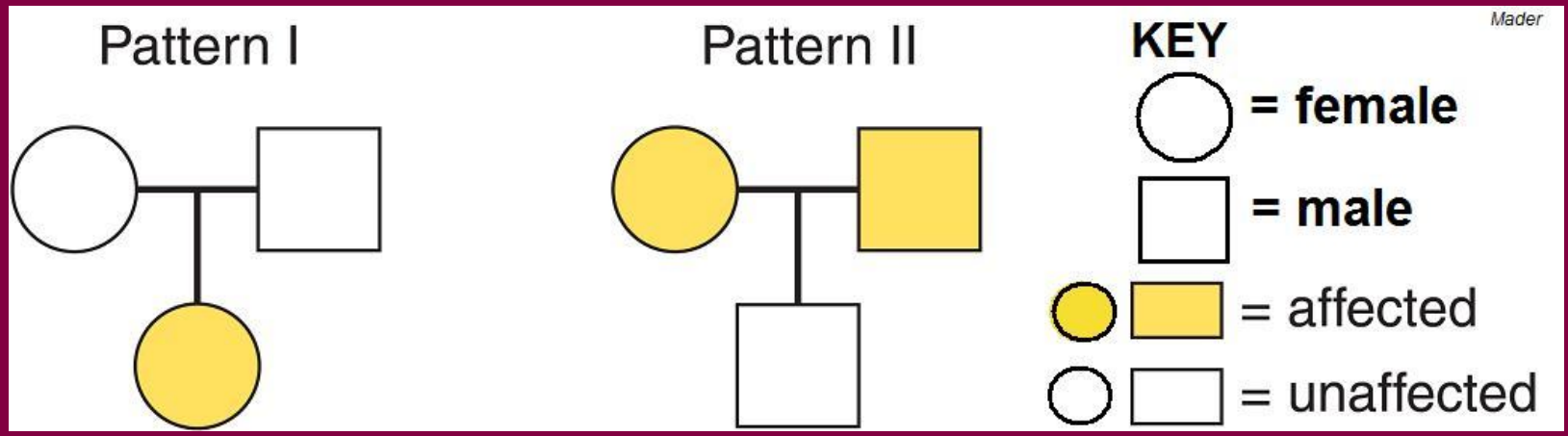
Figure 14.15a. Pedigree analysis.

- ***Male*** = square
- ***Female*** = circle
- ***Affected male*** = shaded square
- ***Affected female*** = shaded circle
- ***Horizontal line between square and circle*** = mating
- ***Horizontal line connecting vertical lines*** = offspring; in birth order (first-born on left)
- ***Roman numerals*** = generations



Human Genetics and Pedigree Analysis

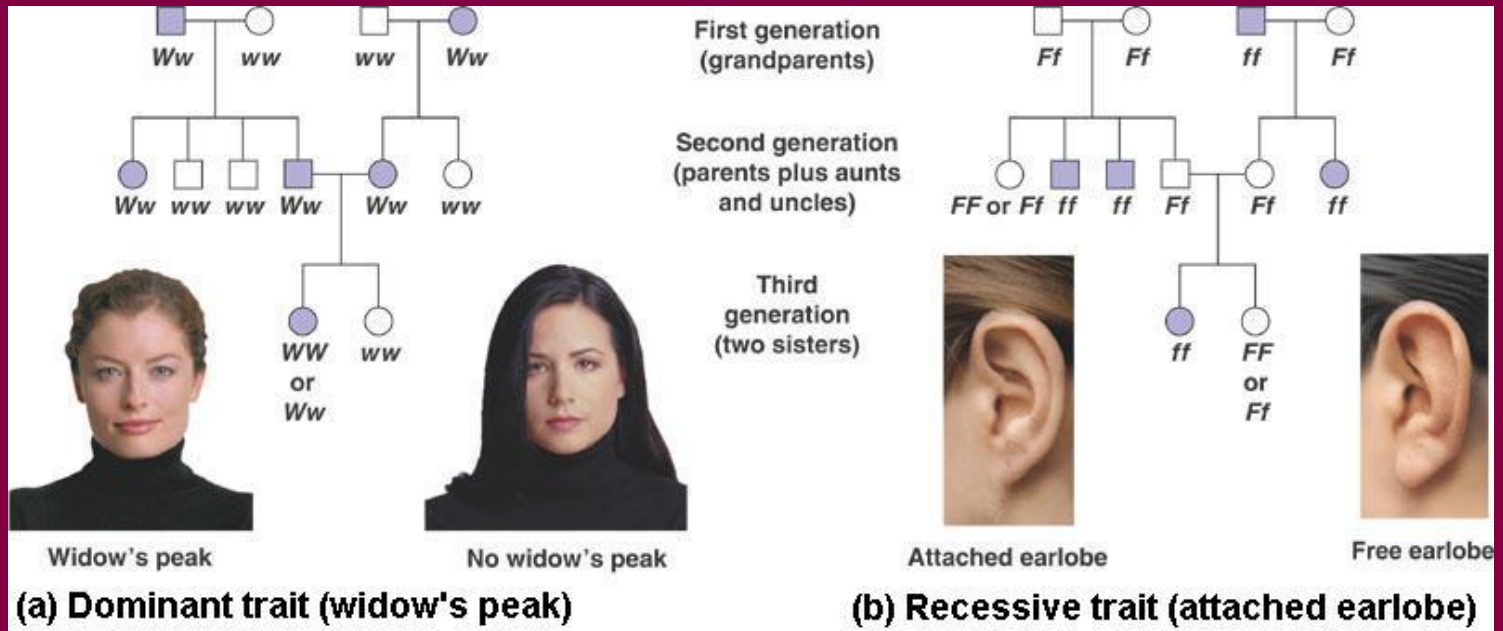
- In **pattern I**, the child is affected, but neither parent is; this can happen if the condition is **recessive** and the parents are ***Aa***. These parents are **carriers** because they appear normal but are capable of having a child with the genetic disorder.
- In **pattern II**, the child is unaffected, but the parents are affected. This can happen if the condition is **dominant** and the parents are ***Aa***.



Pedigree Analysis

a) In the third generation the second-born daughter lacks a **widow's peak**, although both of her parents had the trait. **The trait is due to a dominant allele**. If the trait were due to a ***recessive allele***, and both parents had the recessive phenotype, then *all* of their offspring would also have the recessive phenotype.

b) The first-born daughter in the third generation has **attached earlobes**, although both of her parents lack that trait (they have free earlobes). **The attached-lobe phenotype is due to a recessive allele**. If it were due to a dominant allele, then at least one parent would also have had the trait.



Human Genetics and Pedigree Analysis

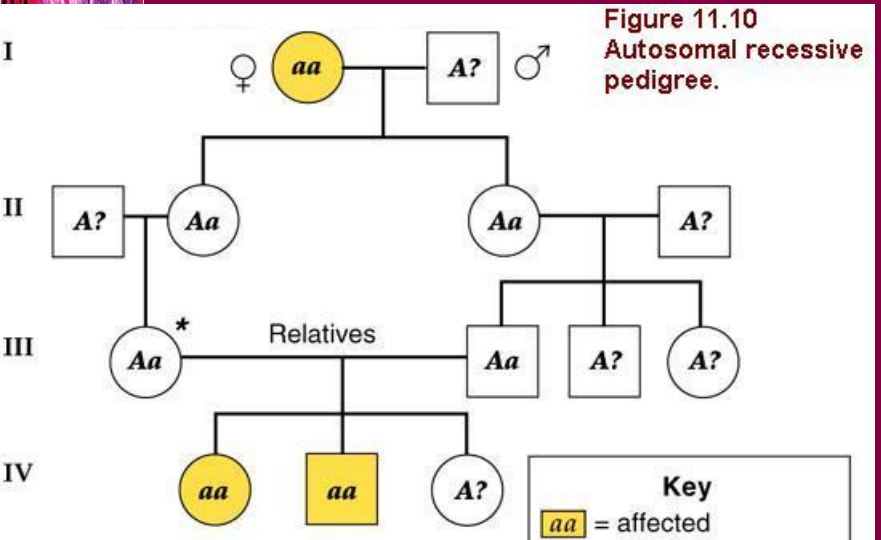


Figure 11.10 Autosomal recessive pedigree.

Key

aa = affected
Aa = carrier (unaffected)
AA = unaffected
A? = unaffected (one allele unknown)

Autosomal recessive disorders

- Most affected children have unaffected parents.
- Heterozygotes (*Aa*) have an unaffected phenotype.
- Two affected parents will always have affected children.
- Affected individuals with homozygous unaffected mates will have unaffected children.
- Close relatives who reproduce are more likely to have affected children.
- Both males and females are affected with equal frequency.

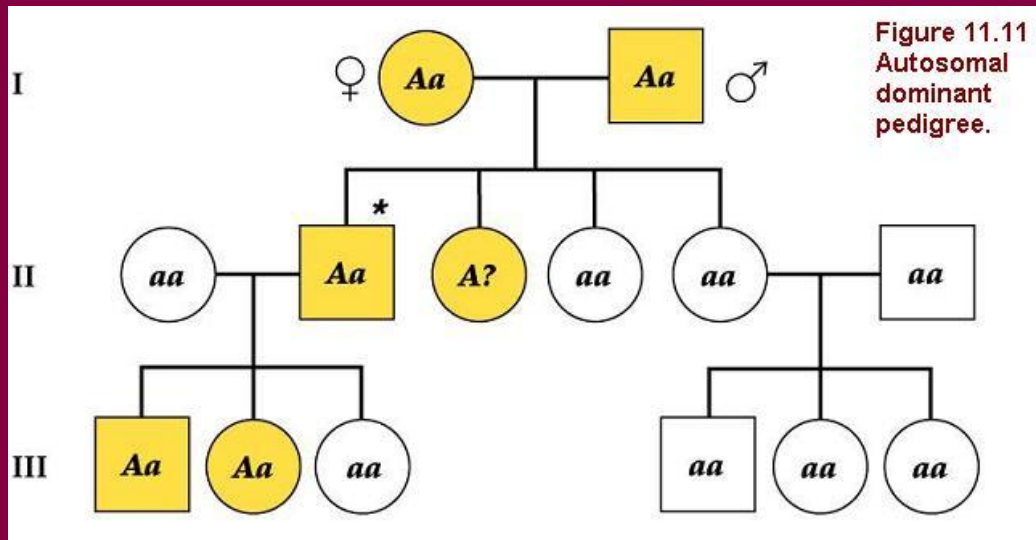


Figure 11.11 Autosomal dominant pedigree.

Key

AA = affected
Aa = affected
A? = affected (one allele unknown)
aa = unaffected

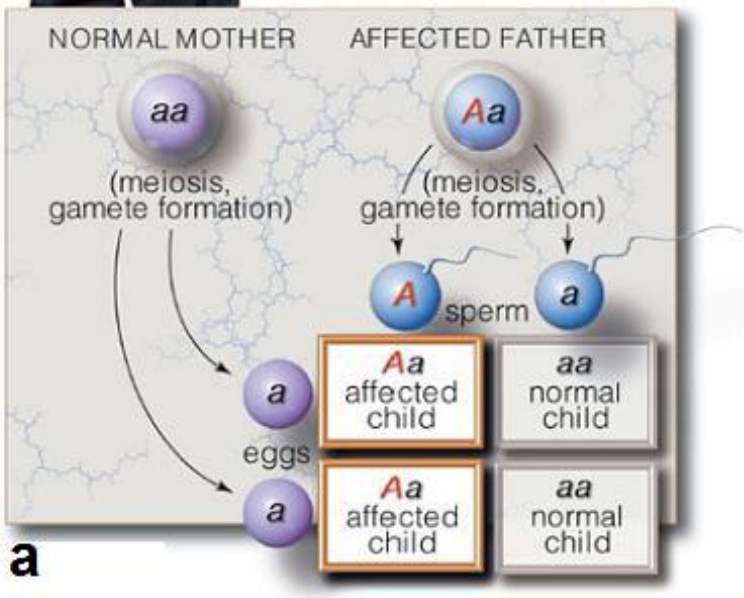
Autosomal dominant disorders

- Affected children will usually have an affected parent.
- Heterozygotes (*Aa*) are affected.
- Two affected parents can produce an unaffected unaffected child.
- Two unaffected parents will not have affected children.
- Both males and females are affected with equal frequency.

Human Inheritance Patterns

a. Autosomal dominant inheritance. A dominant allele (coded red) is fully expressed in carriers. The three males shown have achondroplasia, an autosomal dominant disorder.

b. Autosomal recessive inheritance. In this case, both parents are heterozygous carriers of the recessive allele (coded red).



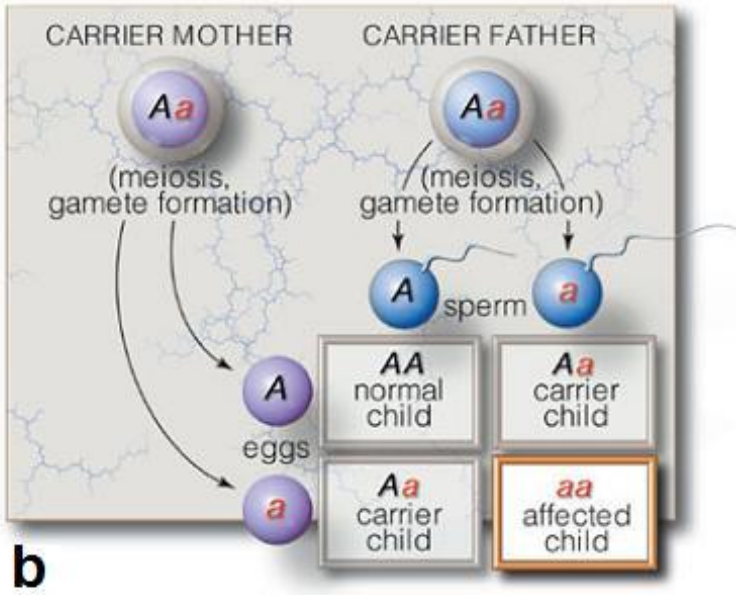
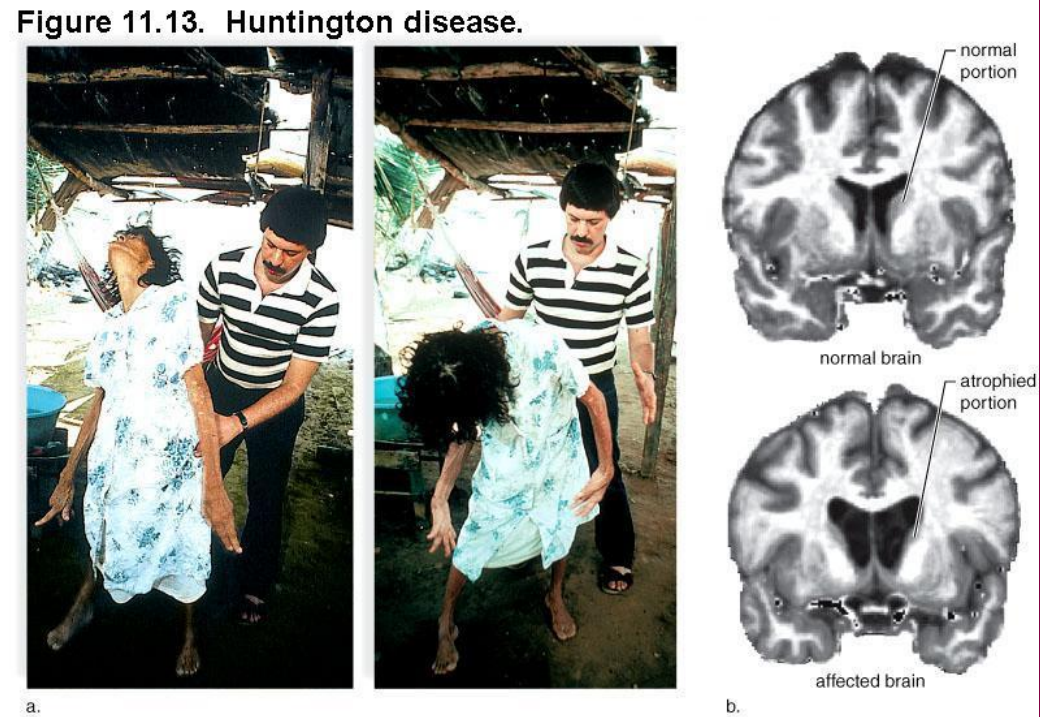


Figure 11.4

Dominantly Inherited Disorder: **Huntington's Disease**



a. Affected persons gradually lose psychomotor control of the body. The symptoms become worse over time.

b. Cross section of a normal brain and affected brain.

- It is a neurological disorder that leads to progressive degeneration of brain cells, which in turn causes severe muscle spasms and personality disorders.
- Occurs in about 1 in 20,000 persons and is caused by a single mutated copy of the gene for a protein called **huntingtin**.
- Most patients appear normal until they are of middle age.
- No effective treatment; death comes 10-15 years after the onset of symptoms.

Recessively Inherited Disorder: **Albinism**

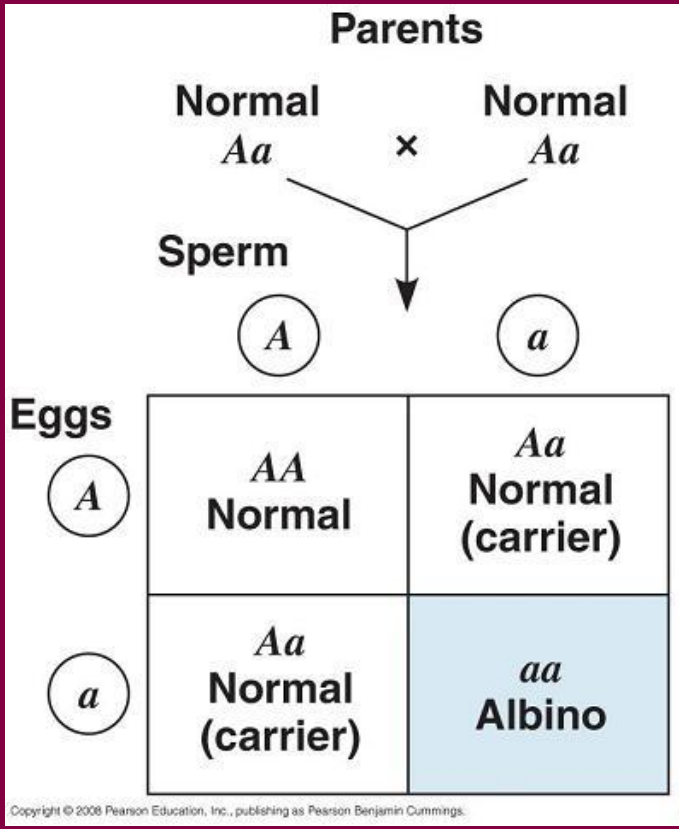


Figure 14.16. Albinism: a recessive trait.

- **Albinism: a recessive trait.** One of the two sisters shown here has normal coloration; the other is albino. Most recessive homozygotes are born to parents who are carriers of the disorder but themselves have a normal phenotype, the case shown in the Punnett square.

Recessively Inherited Disorder: **Cystic Fibrosis (CF)**

These brothers are undergoing antibiotic and percussion therapy. The antibiotic may be administered as an aerosol using a nebulizer. Mucus in the lungs can be loosened by a percussion vest or manual therapy.

- Related to *chromosome 7* and characterized by abnormal secretions related to the chloride ion (Cl^{--}) channel.
- Most common lethal genetic disease among Caucasians in the United States. One in 20 Caucasians & 1 in 2,000 newborns have the disorder.
- Symptoms: Extremely salty sweat, very thick and viscous mucus that interferes with the function of the lungs and pancreas.
- Life expectancy can be up to 35 years of age.

Testing a fetus for genetic disorders

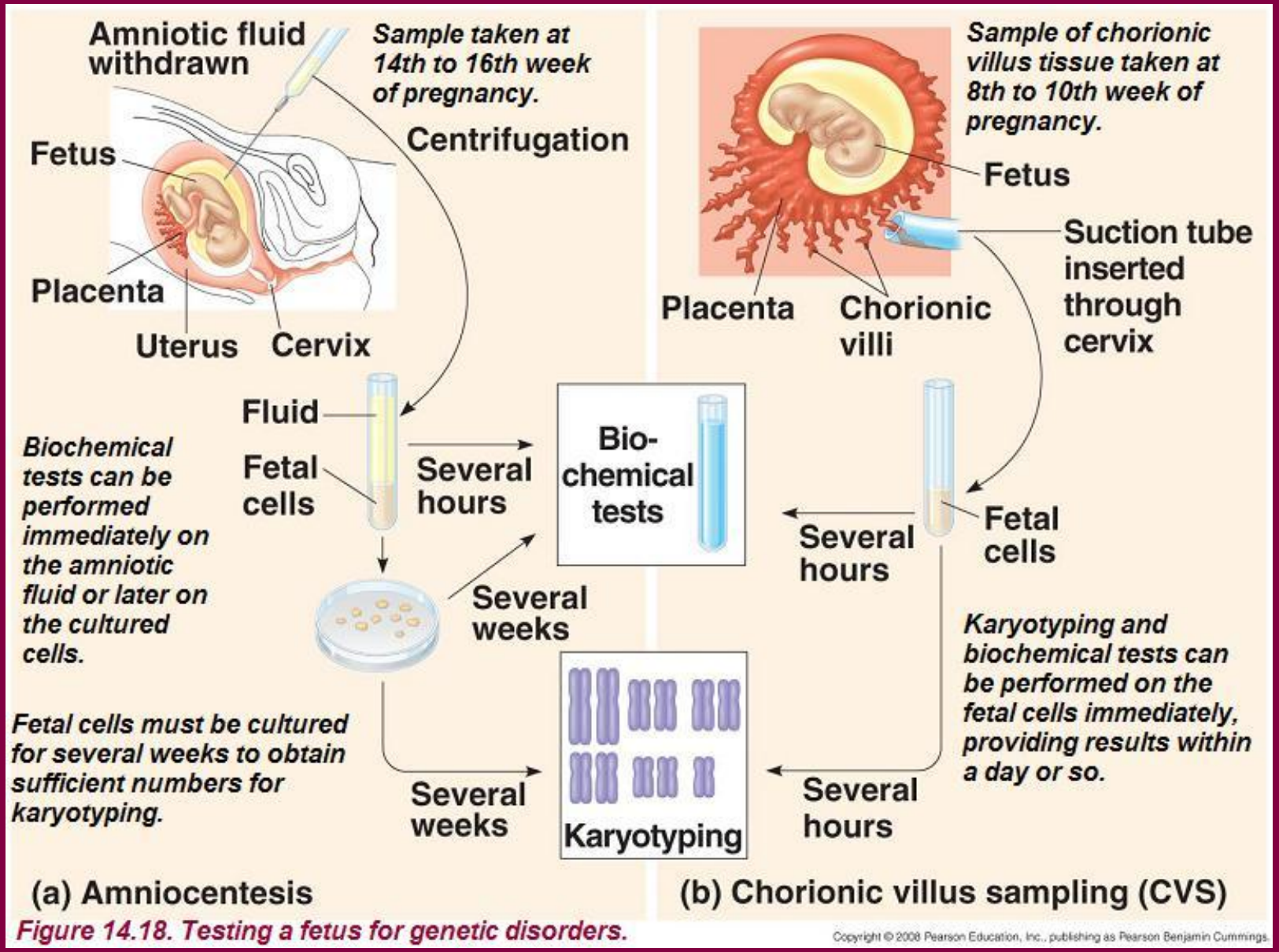


Figure 14.18. Testing a fetus for genetic disorders.

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