PowerPoint® Lecture Presentation for

# Concepts of Genetics

Ninth Edition

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# Chapter 3
# Mendelian Genetics

Lectures by David Kass with contributions from
John C. Osterman.

# Chapter 3 Contents



*Continued*

# Chapter 3 Contents

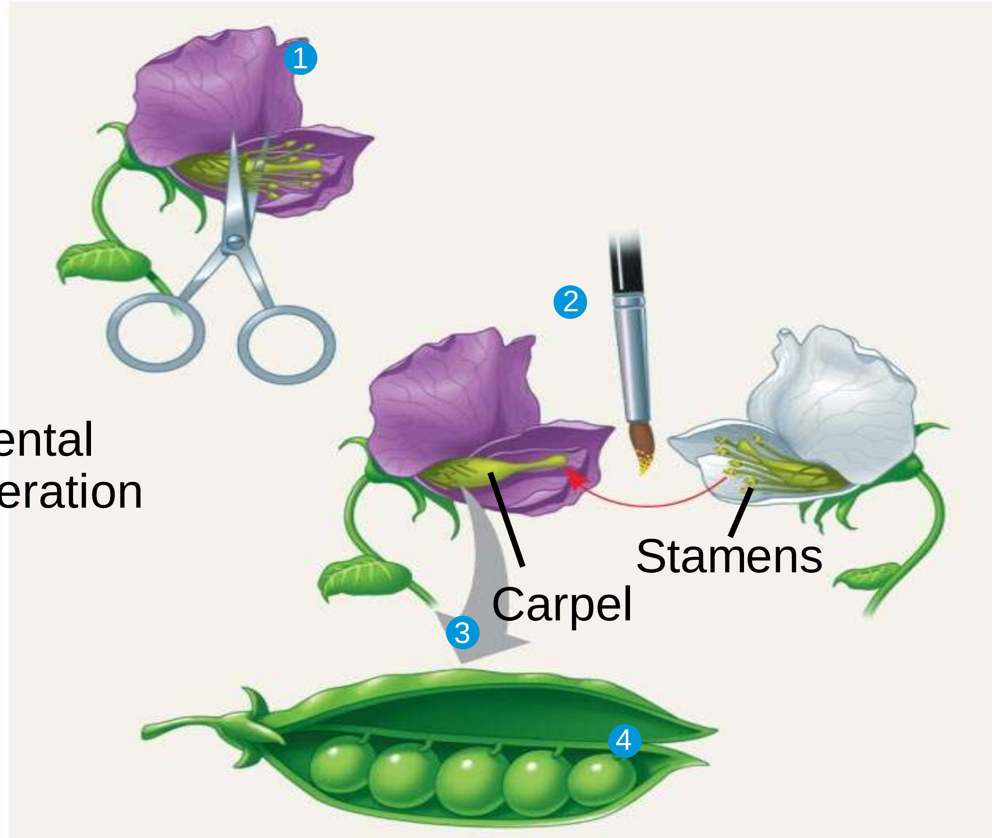
# 3.1 Mendel Used a Model Experimental Approach to Study Patterns of Inheritance

# Section 3.1

- Mendel chose the garden pea as his model system because it is easy to grow, can be crossbred artificially, and grows to maturity in one season.

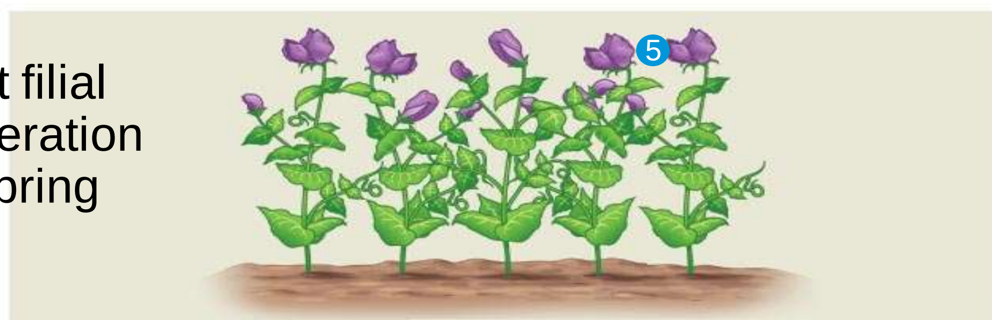
Fig. 14-2

## TECHNIQUE




Parental generation (P)

## RESULTS

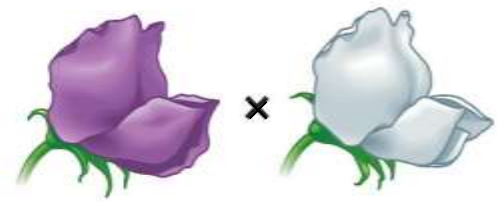



First filial generation offspring ($F_1$)

Fig. 14-3-1

EXPERIMENT

P Generation
(true-breeding
parents)




Purple flowers

White flowers

Fig. 14-3-2

EXPERIMENT

P Generation

(true-breeding parents)

×

Purple flowers

White flowers

F$_1$ Generation

(hybrids)

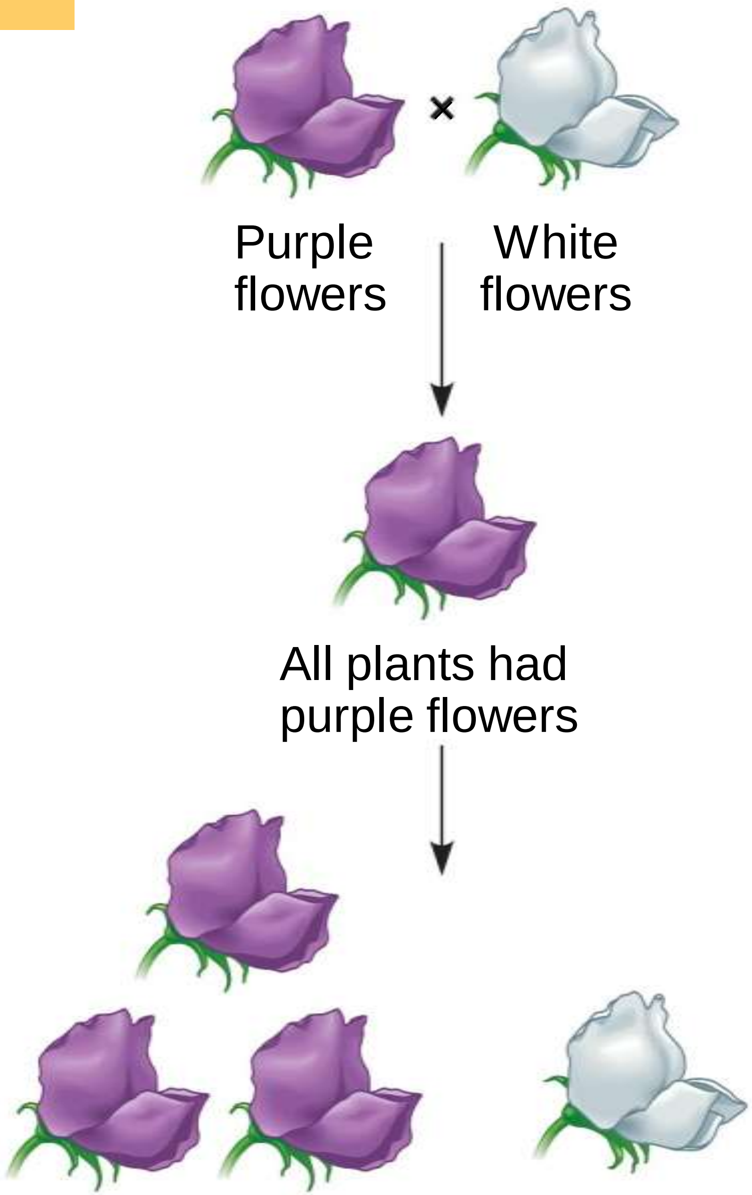
All plants had purple flowers

Fig. 14-3-3

## EXPERIMENT

P Generation
(true-breeding parents)

Purple flowers × White flowers



$F_1$ Generation
(hybrids)

All plants had purple flowers

$F_2$ Generation

705 purple-flowered plants

224 white-flowered plants

# Section 3.1

- Using seven visible features, each with two contrasting traits (**Figure 3.1**), and true-breeding strains, Mendel determined that discrete units of inheritance exist and predicted their behavior during the formation of gametes.

| Character | Contrasting traits | | $F_1$ results | $F_2$ results | $F_2$ ratio |
|---|---|---|---|---|---|
| **Seed shape** | round/wrinkled |  | all round | 5474 round<br>1850 wrinkled | 2.96:1 |
| **Seed color** | yellow/green |  | all yellow | 6022 yellow<br>2001 green | 3.01:1 |
| **Pod shape** | full/constricted |  | all full | 882 full<br>299 constricted | 2.95:1 |
| **Pod color** | green/yellow |  | all green | 428 green<br>152 yellow | 2.82:1 |
| **Flower color** | violet/white |  | all violet | 705 violet<br>224 white | 3.15:1 |
| **Flower position** | axial/terminal |  | all axial | 651 axial<br>207 terminal | 3.14:1 |
| **Stem height** | tall/dwarf |  | all tall | 787 tall<br>277 dwarf | 2.84:1 |



Figure 3.1

# Section 3.1

- Mendel’s postulates were eventually accepted as the basis for Mendelian, or transmission, genetics.

# 3.2 The Monohybrid Cross Reveals How One Trait Is Transmitted from Generation to Generation

# Section 3.2

- **Monohybrid crosses** involve a single pair of contrasting traits.
- The original parents are the $P_1$ generation, and their offspring are the $F_1$ generation.
- Offspring arising from selfing (self-fertilizing) the $F_1$ generation are the $F_2$ generation.

# Section 3.2

- In the $F_1$ generation of a monohybrid cross, all of the plants have just one of the two contrasting traits.
- In the $F_2$ generation, 3/4 of the plants exhibit the same trait as the $F_1$ generation, and 1/4 exhibit the contrasting trait that disappeared in the $F_1$ generation (**Figure 3.1**).

| Character | Contrasting traits | | $F_1$ results | $F_2$ results | $F_2$ ratio |
|---|---|---|---|---|---|
| **Seed shape** | round/wrinkled |  | all round | 5474 round<br>1850 wrinkled | 2.96:1 |
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| **Flower position** | axial/terminal |  | all axial | 651 axial<br>207 terminal | 3.14:1 |
| **Stem height** | tall/dwarf |  | all tall | 787 tall<br>277 dwarf | 2.84:1 |





Figure 3.1

# Table 14.1

**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  | 6,022:2,001 | 3.01:1 |
| Seed shape | Round  | × | Wrinkled  | 5,474:1,850 | 2.96:1 |
| Pod shape | Inflated  | × | Constricted  | 882:299 | 2.95:1 |
| Pod color | Green  | × | Yellow  | 428:152 | 2.82:1 |

# Section 3.2

- To explain these results, Mendel proposed the existence of "**particulate unit factors**" for each trait.
- He suggested that these factors (now called **genes**) and are passed unchanged from generation to generation, determining various traits expressed by each individual plant.

# Section 3.2

- Mendel’s monohybrid crosses were not sex dependent.
- For example, it did not matter whether a tall male plant pollinated a dwarf female plant, or vice versa. The results were the same either way.
- This is called a **reciprocal cross**.

# Section 3.2

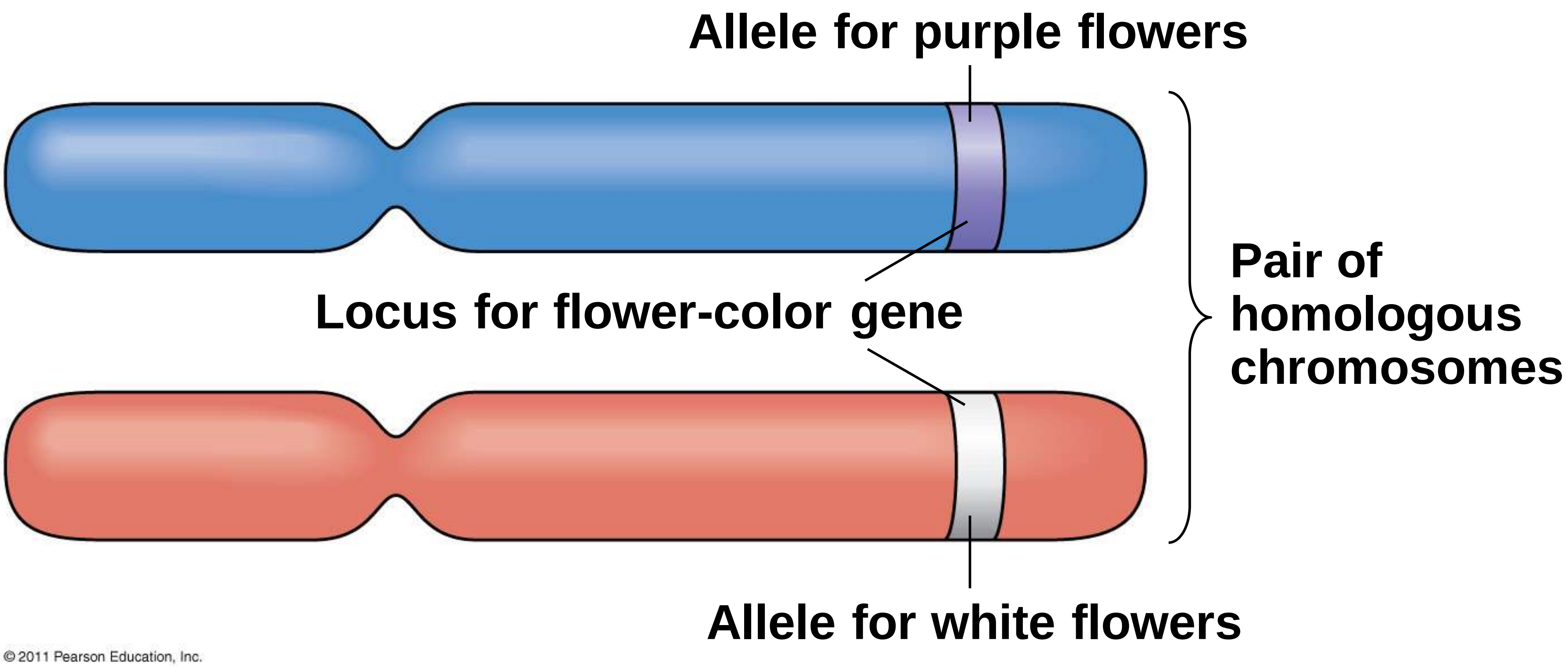
- Mendel proposed three postulates of inheritance:
  - Unit factors exist in pairs.
  - In the pair of unit factors for a single characteristic in an individual, one unit factor is dominant and the other is recessive.
  - The paired unit factors segregate (separate) independently during gamete formation.

# Section 3.2

- The **genotype** is the genetic makeup of an individual.
- The **phenotype** is the physical expression of the genetic makeup.

# Section 3.2

- When the alleles (alternative forms of a single gene) for a trait in an individual are the same, the individual is homozygous.
- If the alleles differ, the individual is heterozygous.


Allele for purple flowers
Locus for flower-color gene
Pair of homologous chromosomes
Allele for white flowers
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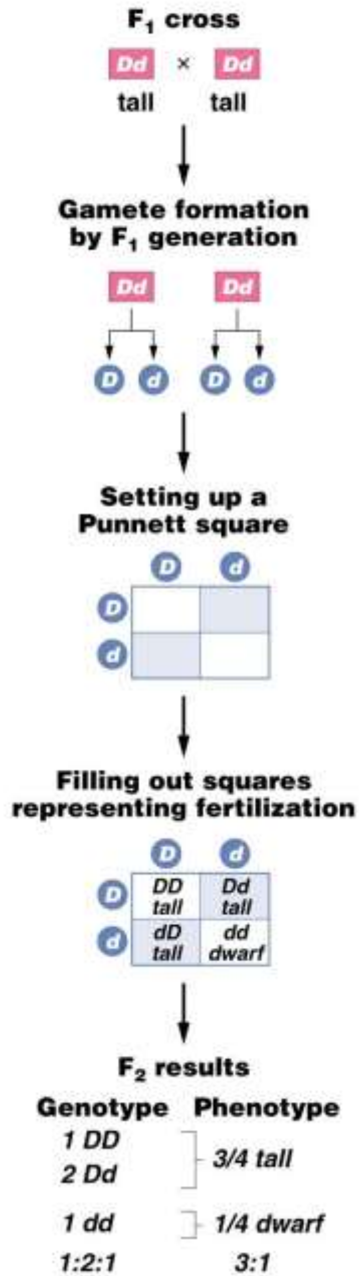
# Section 3.2

- A **Punnett square** allows the genotypes and phenotypes resulting from a cross to be visualized easily (**Figure 3.3**).



$F_1$ cross
Dd × Dd
tall tall
Gamete formation by $F_1$ generation
Dd Dd
D d D d
Setting up a Punnett square
D d
D
d
Filling out squares representing fertilization
D d
D DD tall Dd tall
d dD tall dd dwarf
$F_2$ results
Genotype Phenotype
1 DD
2 Dd
3/4 tall
1 dd
1/4 dwarf
1:2:1 3:1

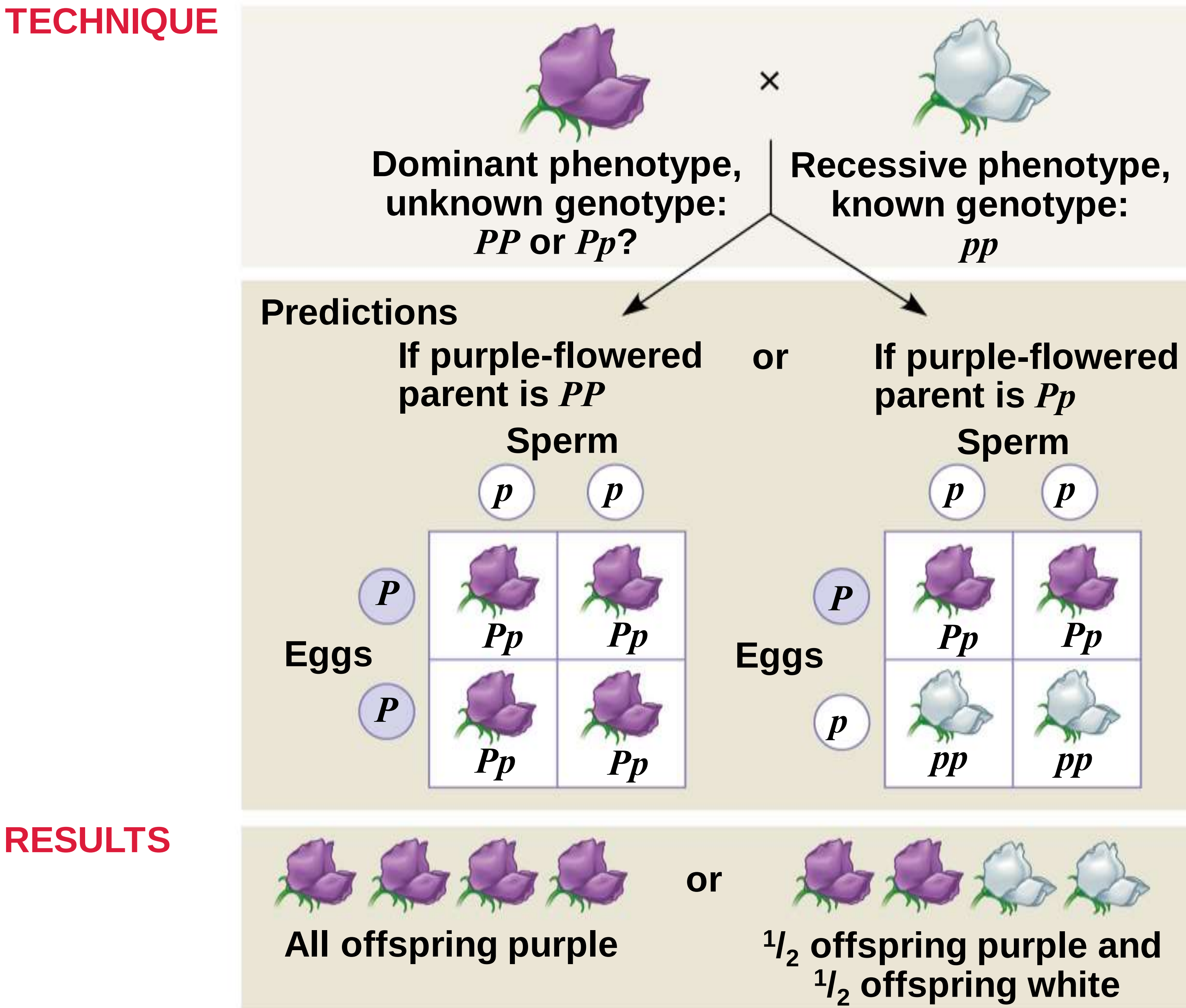



Figure 3.3

# Section 3.2

- A **testcross** is a way to determine whether an individual displaying the dominant phenotype is homozygous or heterozygous for that trait (**Figure 3.4**).

Figure 14.7


TECHNIQUE
×
Dominant phenotype, unknown genotype: PP or Pp?
Recessive phenotype, known genotype: pp
Predictions
If purple-flowered parent is PP
or
If purple-flowered parent is Pp
Sperm
p
p
Eggs
P
P
Pp
Pp
Pp
Pp
Sperm
p
p
Eggs
P
p
Pp
Pp
pp
pp
RESULTS
All offspring purple
or
1/2 offspring purple and 1/2 offspring white

# Testcross results

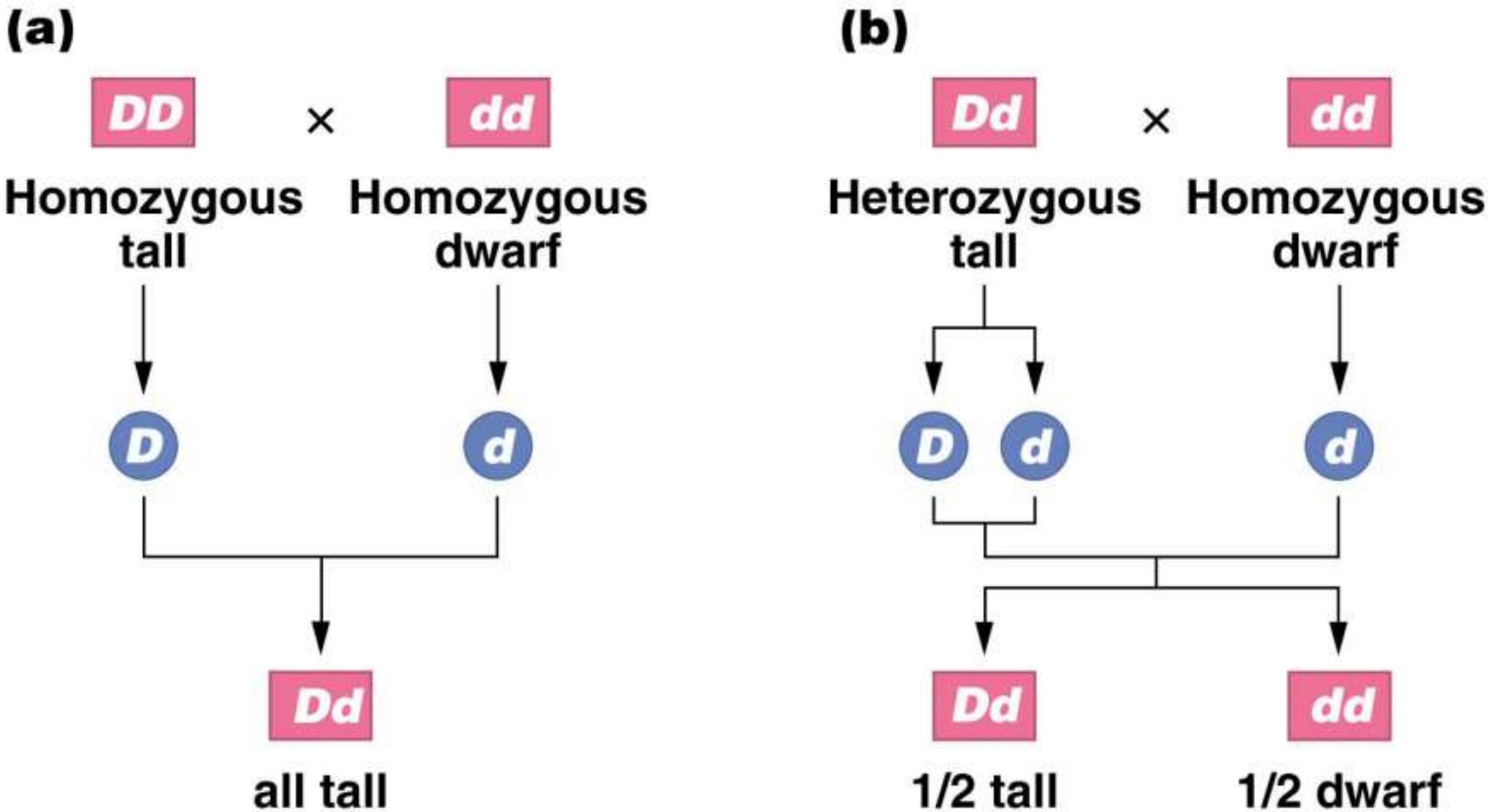


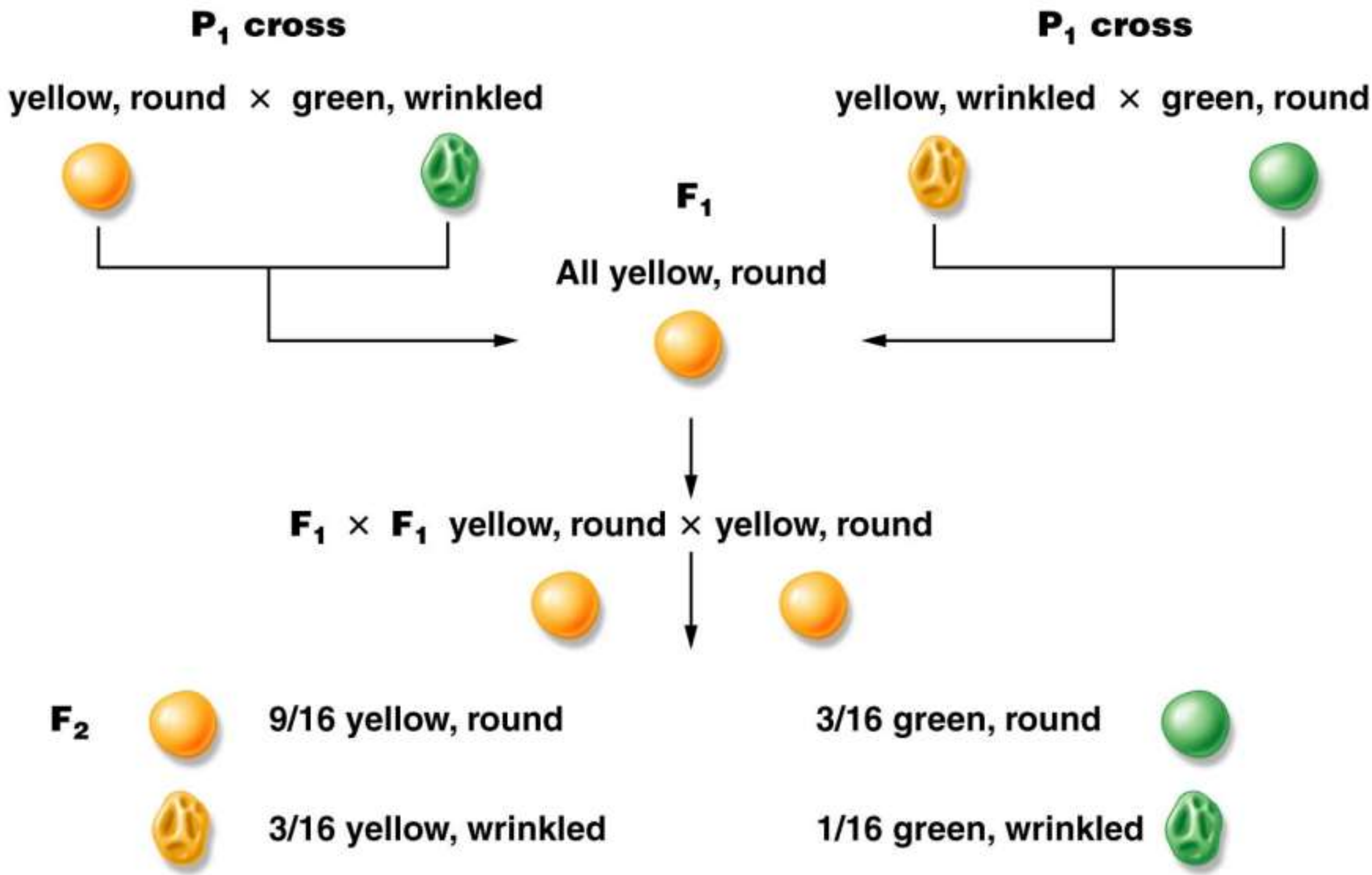



Figure 3.4

# 3.3 Mendel's Dihybrid Cross Generated a Unique $F_2$ Ratio

# Section 3.3

- A **dihybrid cross** involves two pairs of contrasting traits (**Figure 3.5**).



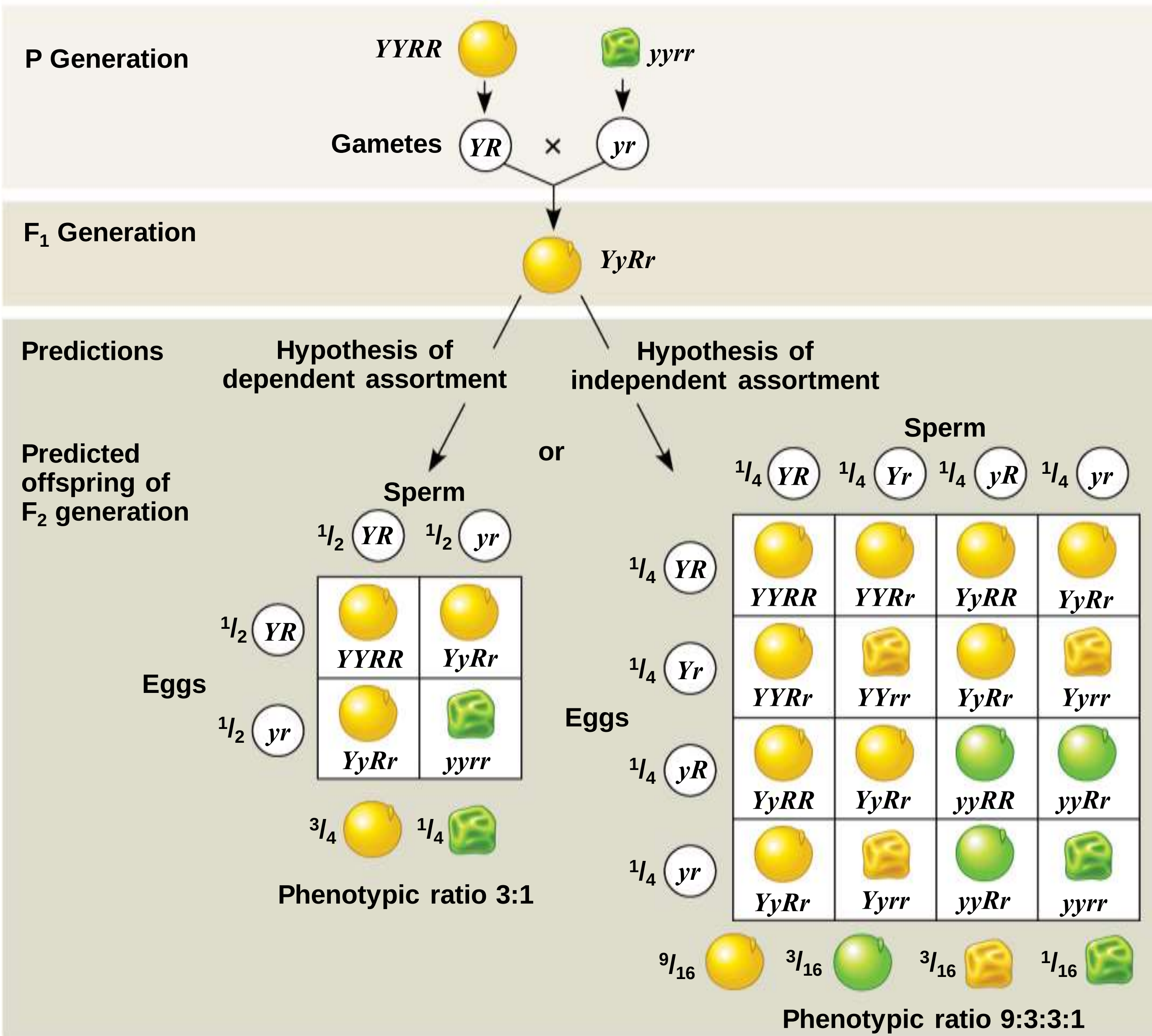
$P_1$ cross
yellow, round × green, wrinkled
$P_1$ cross
yellow, wrinkled × green, round
$F_1$
All yellow, round
$F_1$ × $F_1$ yellow, round × yellow, round
$F_2$
9/16 yellow, round
3/16 green, round
3/16 yellow, wrinkled
1/16 green, wrinkled




Figure 3.5

# Figure 14.8

**EXPERIMENT**



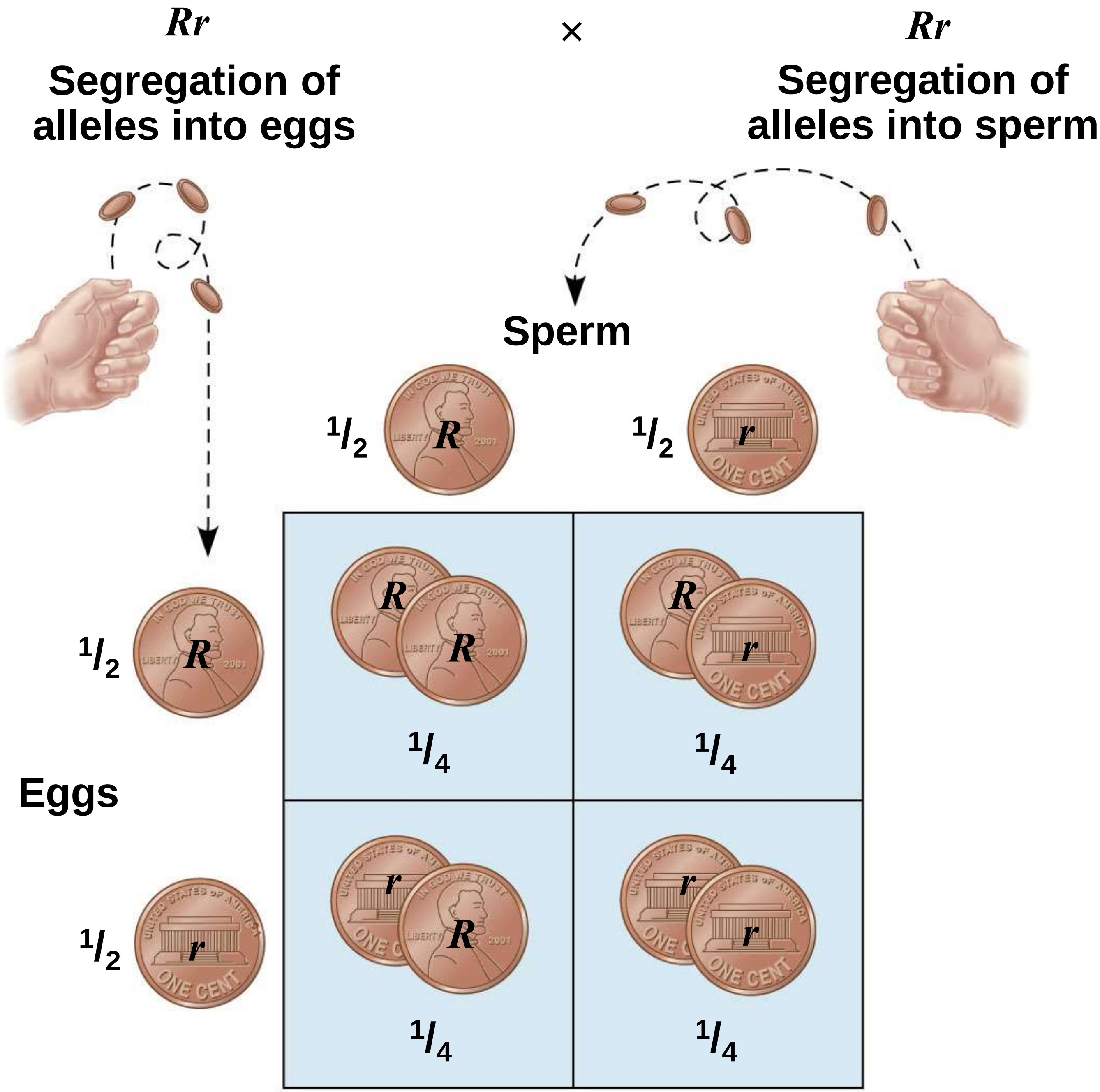

**RESULTS**

# Section 3.3

- The product law can be used to predict the frequency with which two independent events will occur simultaneously (**Figure 3.6**).

# The Multiplication and Addition Rules Applied to Crosses

- 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 $F_1$ 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



Rr
×
Rr
Segregation of alleles into eggs
Segregation of alleles into sperm
Sperm
1/2 R
1/2 r
Eggs
1/2 R
1/2 r
R R
1/4
R r
1/4
r R
1/4
r r
1/4
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**$F_1$ yellow, round × yellow, round**

| $F_2$ Of all offspring | Of all offspring | Combined probabilities |
|---|---|---|
| 3/4 are yellow | 3/4 are round | (3/4)(3/4) = 9/16 yellow, round |
| | and | |
| | 1/4 are wrinkled | (3/4)(1/4) = 3/16 yellow, wrinkled |
| 1/4 are green | 3/4 are round | (1/4)(3/4) = 3/16 green, round |
| | and | |
| | 1/4 are wrinkled | (1/4)(1/4) = 1/16 green, wrinkled |



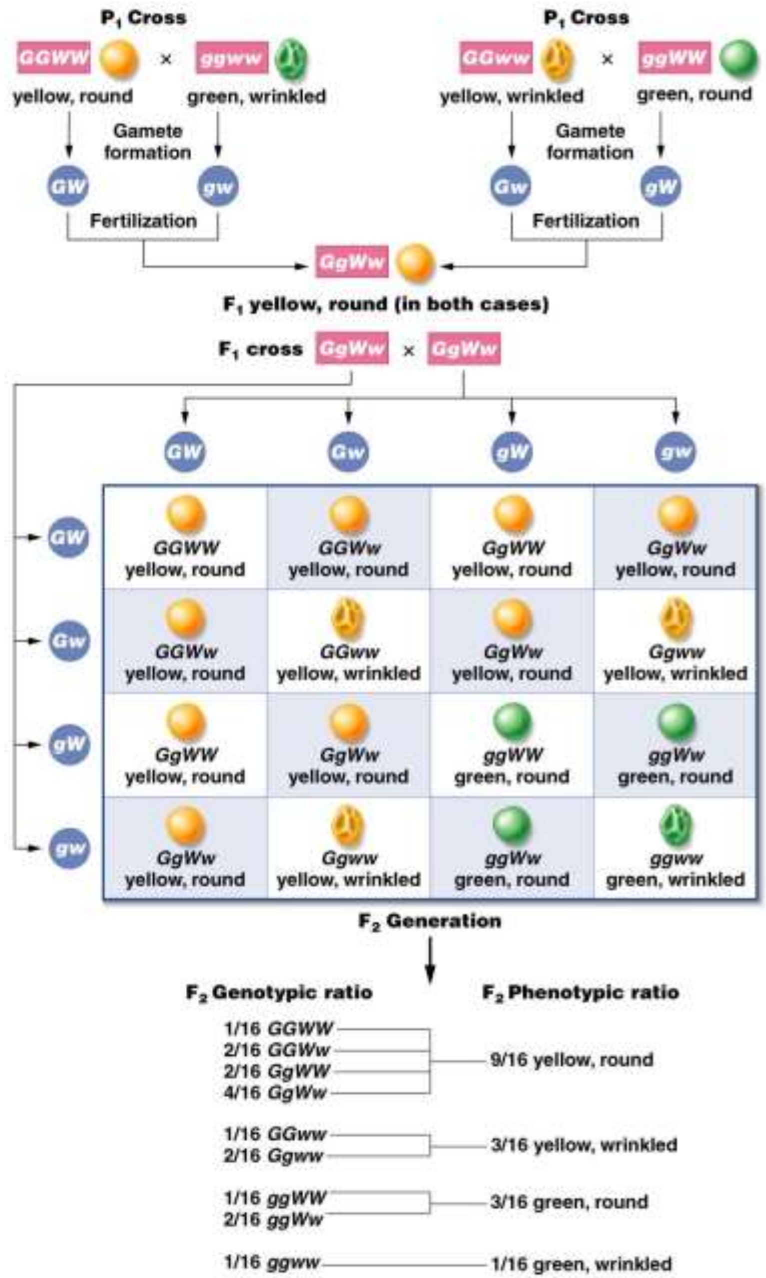
Figure 3.6

# Section 3.3

- Mendel's fourth postulate states that:
  - Traits assort independently during gamete formation
  - All possible combinations of gametes will form with equal frequency.

# Section 3.3

- A Punnett square of a dihybrid cross is shown in Figure 3.7.
- Note the 9:3:3:1 dihybrid ratio.



P1 Cross
GGWW
yellow, round
ggww
green, wrinkled
Gamete formation
GW
gw
Fertilization
P1 Cross
GGww
yellow, wrinkled
ggWW
green, round
Gamete formation
Gw
gW
Fertilization
GgWw
F1 yellow, round (in both cases)
F1 cross GgWw × GgWw
GW
Gw
gW
gw
GGWW
yellow, round
GGWw
yellow, round
GgWW
yellow, round
GgWw
yellow, round
GGWw
yellow, round
GGww
yellow, wrinkled
GgWw
yellow, round
Ggww
yellow, wrinkled
GgWW
yellow, round
GgWw
yellow, round
ggWW
green, round
ggWw
green, round
GgWw
yellow, round
Ggww
yellow, wrinkled
ggWw
green, round
ggww
green, wrinkled
F2 Generation
F2 Genotypic ratio
F2 Phenotypic ratio
1/16 GGWW
2/16 GGWw
2/16 GgWW
4/16 GgWw
9/16 yellow, round
1/16 GGww
2/16 Ggww
3/16 yellow, wrinkled
1/16 ggWW
2/16 ggWw
3/16 green, round
1/16 ggww
1/16 green, wrinkled

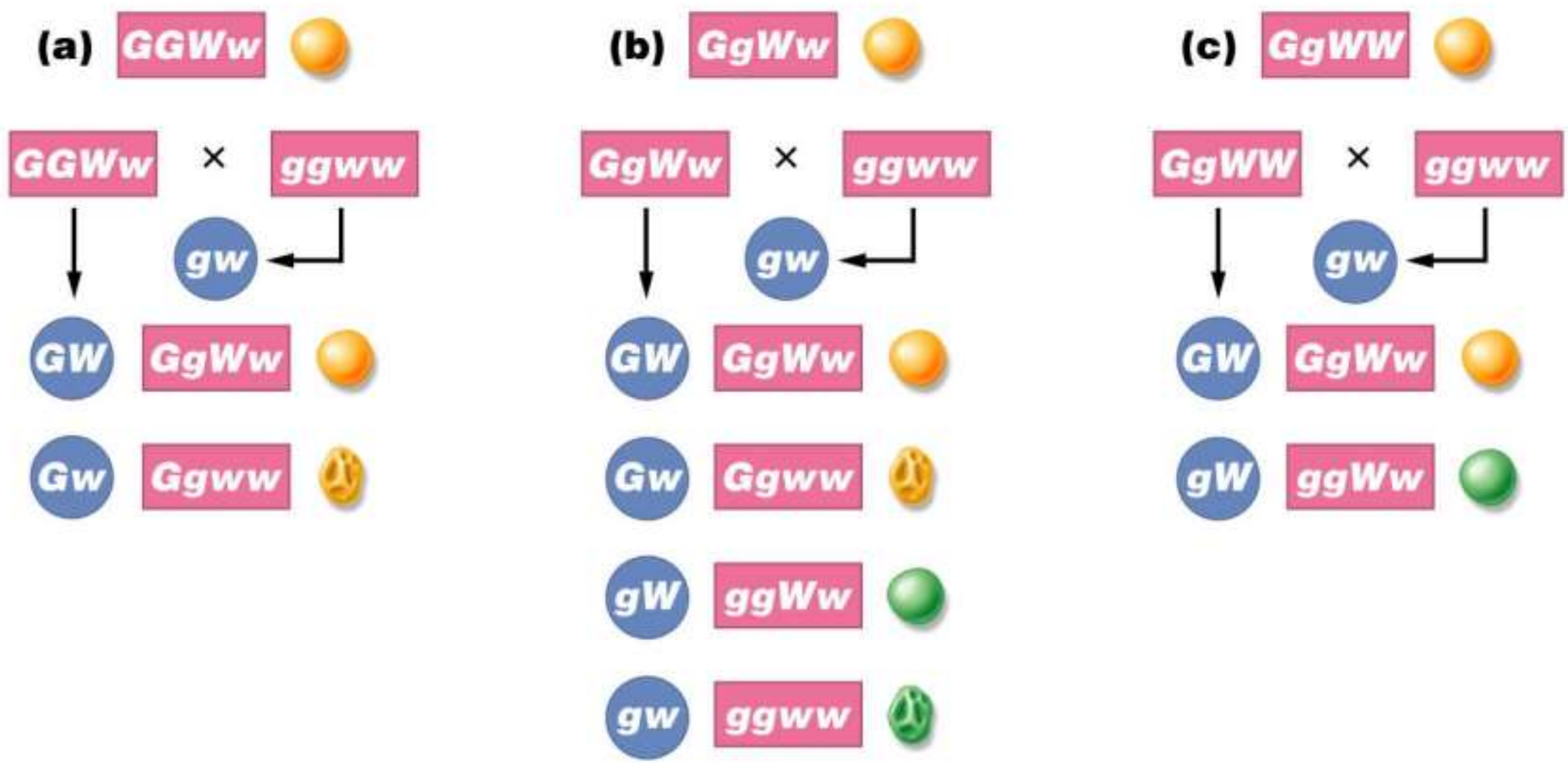



Figure 3.7

# Section 3.3

- **Testcrosses** can also be used with two independent traits (**Figure 3.8**).

## Testcross results of three yellow, round individuals




**(a) Phenotypic ratio**

1/2 yellow, round

1/2 yellow, wrinkled

**(b) Phenotypic ratio**

1/4 yellow, round

1/4 yellow, wrinkled

1/4 green, round

1/4 green, wrinkled

**(c) Phenotypic ratio**

1/2 yellow, round

1/2 green, round



Figure 3.8

# 3.4 The Trihybrid Cross Demonstrates That Mendel's Principles Apply to Inheritance of Multiple Traits

# Section 3.4

- **Trihybrid crosses** involving three independent traits show that Mendel's rules apply to any number of traits.

# Section 3.4

- The **forked-line** (branched diagram) method is easier to use than a Punnett square for analysis of inheritance of larger number of traits (**Figure 3.10**).

## Generation of $F_2$ trihybrid phenotypes

| *A* or *a* | *B* or *b* | *C* or *c* | Combined proportion |
|---|---|---|---|
| 3/4 *A* | 3/4 *B* | 3/4 *C* | (3/4)(3/4)(3/4) *ABC* = 27/64 *ABC* |
| | | 1/4 *c* | (3/4)(3/4)(1/4) *ABc* = 9/64 *ABc* |
| | 1/4 *b* | 3/4 *C* | (3/4)(1/4)(3/4) *AbC* = 9/64 *AbC* |
| | | 1/4 *c* | (3/4)(1/4)(1/4) *Abc* = 3/64 *Abc* |
| 1/4 *a* | 3/4 *B* | 3/4 *C* | (1/4)(3/4)(3/4) *aBC* = 9/64 *aBC* |
| | | 1/4 *c* | (1/4)(3/4)(1/4) *aBc* = 3/64 *aBc* |
| | 1/4 *b* | 3/4 *C* | (1/4)(1/4)(3/4) *abC* = 3/64 *abC* |
| | | 1/4 *c* | (1/4)(1/4)(1/4) *abc* = 1/64 *abc* |



Figure 3.10

# Section 3.4

- Some simple mathematical rules shown in **Table 3.1** apply in working genetics problems.

TABLE 3.1

## Simple Mathematical Rules Useful in Working Genetics Problems

| Crosses between Organisms Heterozygous for Genes Exhibiting Independent Assortment | | | |
|---|---|---|---|
| Number of Heterozygous Gene Pairs | Number of Different Types of Gametes Formed | Number of Different Genotypes Produced | Number of Different Phenotypes Produced* |
| $n$ | $2^n$ | $3^n$ | $2^n$ |
| 1 | 2 | 3 | 2 |
| 2 | 4 | 9 | 4 |
| 3 | 8 | 27 | 8 |
| 4 | 16 | 81 | 16 |

*The fourth column assumes a simple dominant–recessive relationship in each gene pair.



Table 3.1

# 3.5 Mendel's Work Was Rediscovered in the Early Twentieth Century

# Section 3.5

- Mendel suggested that heredity resulted in discontinuous variation, as opposed to the existing continuous variation hypothesis of his time—in which offspring were thought to be a blend of the parental phenotypes.

# 3.6 The Correlation of Mendel's Postulates with the Behavior of Chromosomes Provided the Foundation of Modern Transmission Genetics

# Section 3.6

- The **chromosomal theory of inheritance** proposed that the separation of chromosomes during meiosis could be the basis for Mendel’s principles of segregation and independent assortment.

# Section 3.6

- Three of Mendel's postulates and their basis in chromosomal segregation during meiosis are shown in **Figure 3.11**.

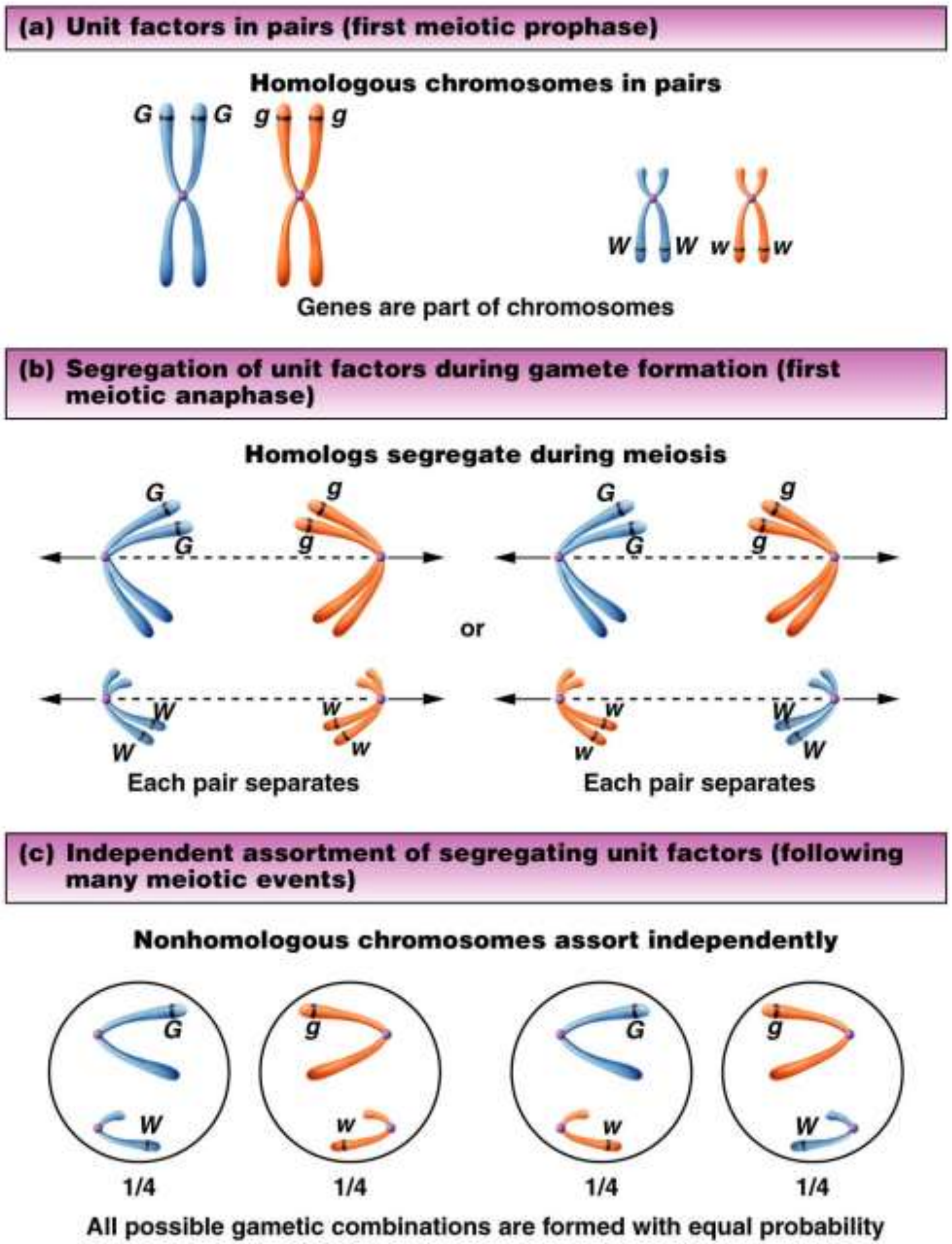

(a) Unit factors in pairs (first meiotic prophase)
Homologous chromosomes in pairs
G G g g
W W w w
Genes are part of chromosomes
(b) Segregation of unit factors during gamete formation (first meiotic anaphase)
Homologs segregate during meiosis
G G g g
or
G G g g
W W w w
Each pair separates
w w W W
Each pair separates
(c) Independent assortment of segregating unit factors (following many meiotic events)
Nonhomologous chromosomes assort independently
G W
g w
G w
g W
1/4
1/4
1/4
1/4
All possible gametic combinations are formed with equal probability

Figure 3.11

# 3.7 Independent Assortment Leads to Extensive Genetic Variation

# Section 3.7

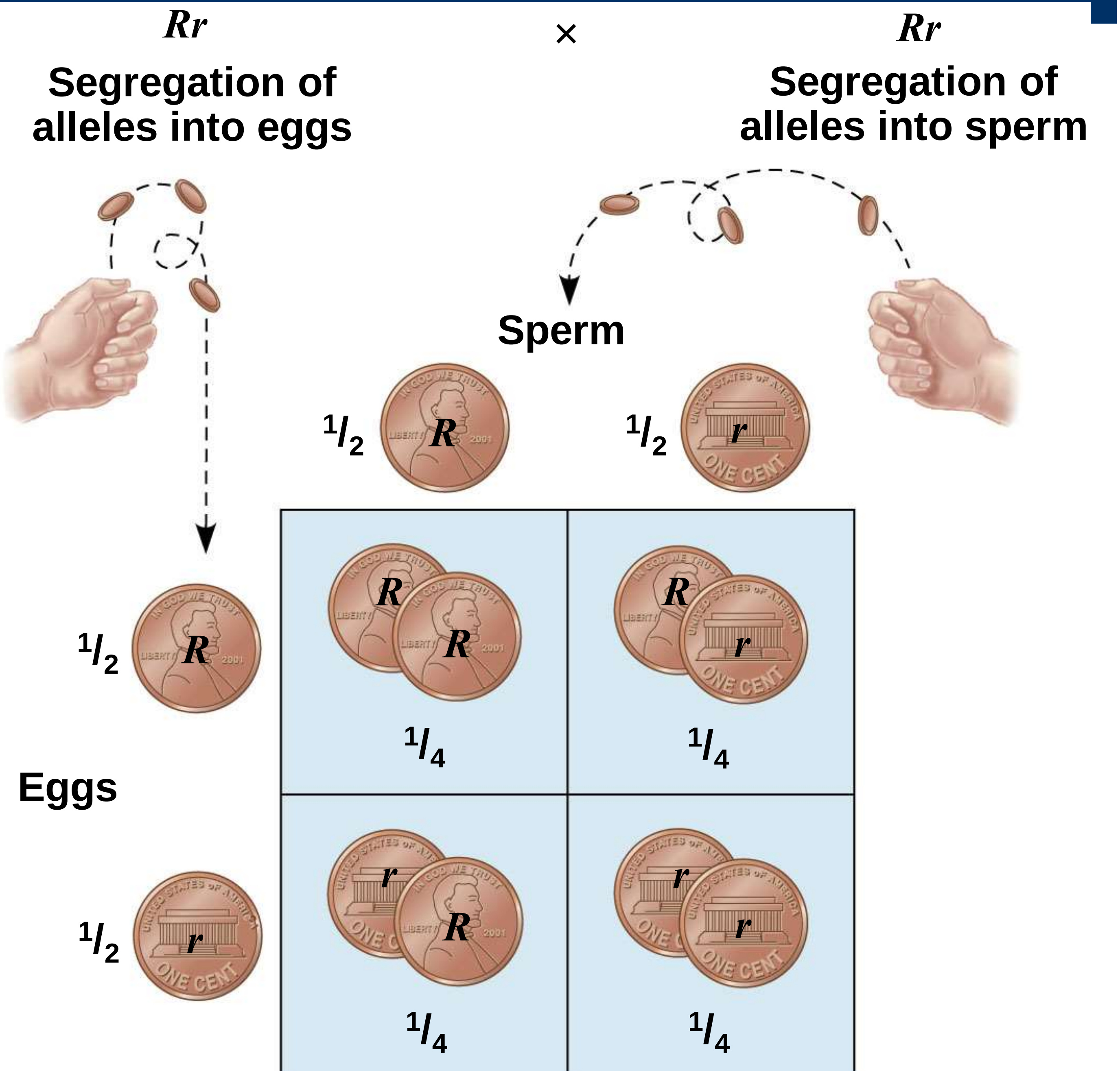
- A major consequence of independent assortment is the production of genetically dissimilar gametes.
- Genetic variation results from independent assortment and is very important to the process of evolution.

# 3.8 Laws of Probability Help to Explain Genetic Events

# Section 3.8

- The probability of two independent events occurring at the same time can be calculated using the **product law**.
- The probability of both events occurring is the product of the probability of each individual event.

Figure 14.9



Rr
×
Rr
Segregation of alleles into eggs
Segregation of alleles into sperm
Sperm
Eggs
1/2 R
1/2 r
1/2 R
1/2 r
R R
1/4
R r
1/4
r R
1/4
r r
1/4

# Section 3.8

- The **sum law** is used to calculate the probability of a generalized outcome that can be accomplished in more than one way.
- The sum law states that the probability of obtaining any single outcome, where that outcome can be achieved in two or more events, is equal to the sum of the individual probabilities of all such events.

- The **addition rule** states that the probability that any one of two or more exclusive events will occur is calculated by adding together their individual probabilities
- The rule of addition can be used to figure out the probability that an $F_2$ plant from a monohybrid cross will be heterozygous rather than homozygous

# Section 3.8

- When one event depends on another, the likelihood of the desired outcome is the **conditional probability**.

# Section 3.8

- The **binomial theorem** can be used to calculate the probability of any specific set of outcomes among a large number of potential events.

# 3.9 Chi-Square Analysis Evaluates the Influence of Chance on Genetic Data

# Section 3.9

- Chance deviation from an expected outcome is diminished by larger sample size.

# Section 3.9

- When we assume that data will fit a given ratio, we establish what is called the **null hypothesis**—so named because it assumes that there is no real difference between the measured values (or ratio) and the predicted values (or ratio).
- The apparent difference can be attributed purely to chance.

# Section 3.9

- **Chi-square ($\chi^2$)** analysis is used to test how well the data fit the null hypothesis.
- **Table 3.3** shows the steps in $\chi^2$ calculations for the $F_2$ generation of a monohybrid cross.

TABLE 3.3

## Chi-Square Analysis

**(a) Monohybrid Cross**

| Expected Ratio | Observed ($o$) | Expected ($e$) | Deviation ($o - e$) | Deviation ($d^2$) | $d^2/e$ |
|---|---|---|---|---|---|
| 3/4 | 740 | 3/4(1000) = 750 | 740 − 750 = −10 | $(-10^2) = 100$ | 100/750 = 0.13 |
| 1/4 | 260 | 1/4(1000) = 250 | 260 − 250 = +10 | $(+10)^2 = 100$ | 100/250 = 0.40 |
| | Total = 1000 | | | | $\chi^2 = 0.53$ |
| | | | | | $p = 0.48$ |

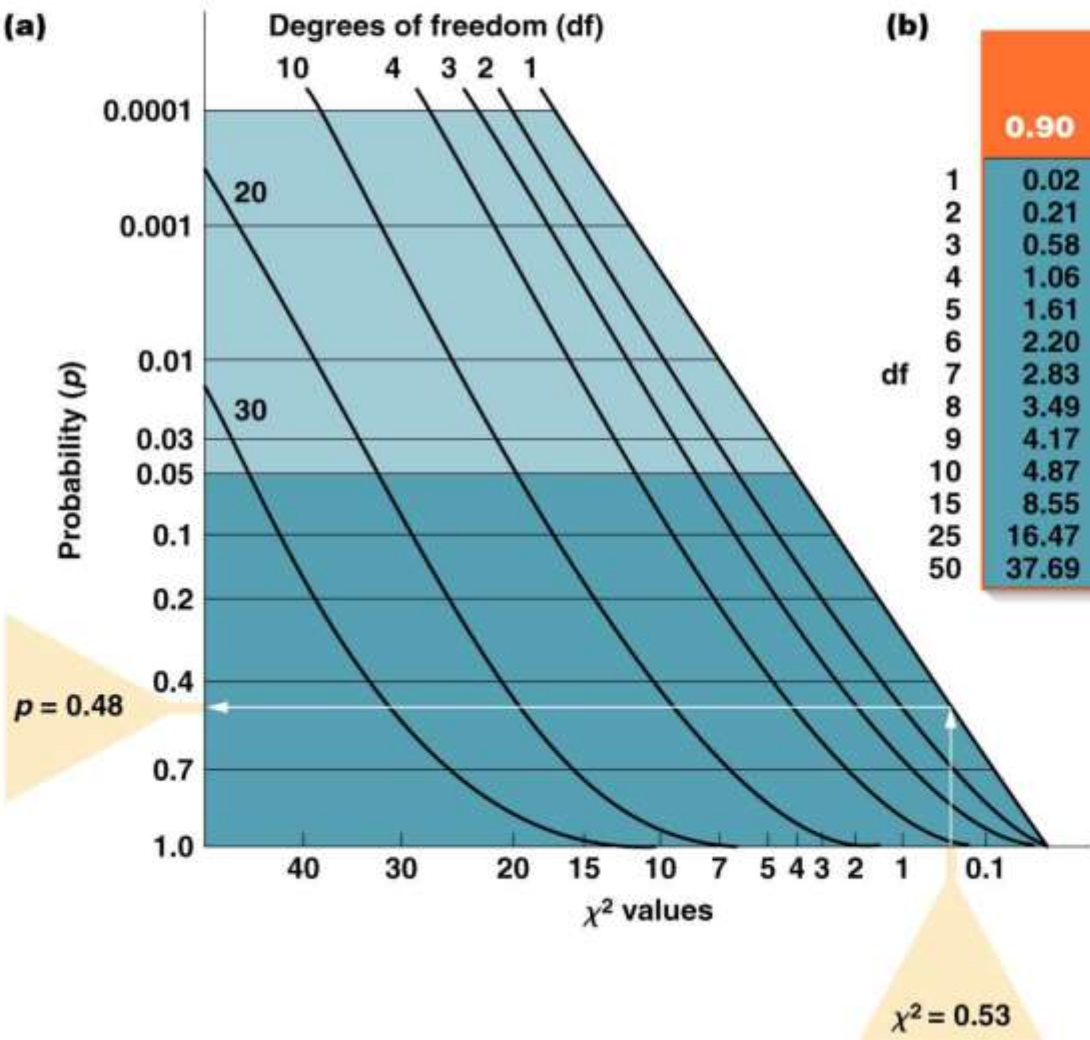
**(b) Dihybrid Cross**

| Expected Ratio | $o$ | $e$ | $(o - e)$ | $d^2$ | $d^2/e$ |
|---|---|---|---|---|---|
| 9/16 | 587 | 567 | +20 | 400 | 0.71 |
| 3/16 | 197 | 189 | +8 | 64 | 0.34 |
| 3/16 | 168 | 189 | −21 | 441 | 2.33 |
| 1/16 | 56 | 63 | −7 | 49 | 0.78 |
| | Total = 1008 | | | | $\chi^2 = 4.16$ |
| | | | | | $p = 0.26$ |

# Section 3.9

- Chi-square analysis requires that the degree of freedom (df) be taken into account, since more deviation is expected with a higher degree of freedom (**Figure 3.12**).
- The degree of freedom is equal to $n$ – 1 where $n$ is the number of different categories into which each datum point may fall.

(a)




(b)

| df | 0.90 | 0.50 | 0.20 | 0.05 | 0.01 | 0.001 |
|---|---|---|---|---|---|---|
| | **Probability (p)** | | | | | |
| 1 | 0.02 | 0.46 | 1.64 | 3.84 | 6.64 | 10.83 |
| 2 | 0.21 | 1.39 | 3.22 | 5.99 | 9.21 | 13.82 |
| 3 | 0.58 | 2.37 | 4.64 | 7.82 | 11.35 | 16.27 |
| 4 | 1.06 | 3.36 | 5.99 | 9.49 | 13.28 | 18.47 |
| 5 | 1.61 | 4.35 | 7.29 | 11.07 | 15.09 | 20.52 |
| 6 | 2.20 | 5.35 | 8.56 | 12.59 | 16.81 | 22.46 |
| 7 | 2.83 | 6.35 | 9.80 | 14.07 | 18.48 | 24.32 |
| 8 | 3.49 | 7.34 | 11.03 | 15.51 | 20.09 | 26.13 |
| 9 | 4.17 | 8.34 | 12.24 | 16.92 | 21.67 | 27.88 |
| 10 | 4.87 | 9.34 | 13.44 | 18.31 | 23.21 | 29.59 |
| 15 | 8.55 | 14.34 | 19.31 | 25.00 | 30.58 | 37.30 |
| 25 | 16.47 | 24.34 | 30.68 | 37.65 | 44.31 | 52.62 |
| 50 | 37.69 | 49.34 | 58.16 | 67.51 | 76.15 | 86.60 |

$\chi^2$ values

Fails to reject the null hypothesis

Rejects the null hypothesis



Figure 3.12

# Section 3.9

- Once the number of degrees of freedom is determined, the $\chi^2$ value can be interpreted in terms of a corresponding probability value ($p$).

# 3.10 Pedigrees Reveal Patterns of Inheritance of Human Traits

# Section 3.10

- A **pedigree** shows a family tree with respect to a given trait. Pedigree analysis reveals patterns of inheritance.

# Section 3.10

- Standard pedigree conventions are shown in **Figure 3.13** and representative pedigrees are shown in **Figure 3.14**.

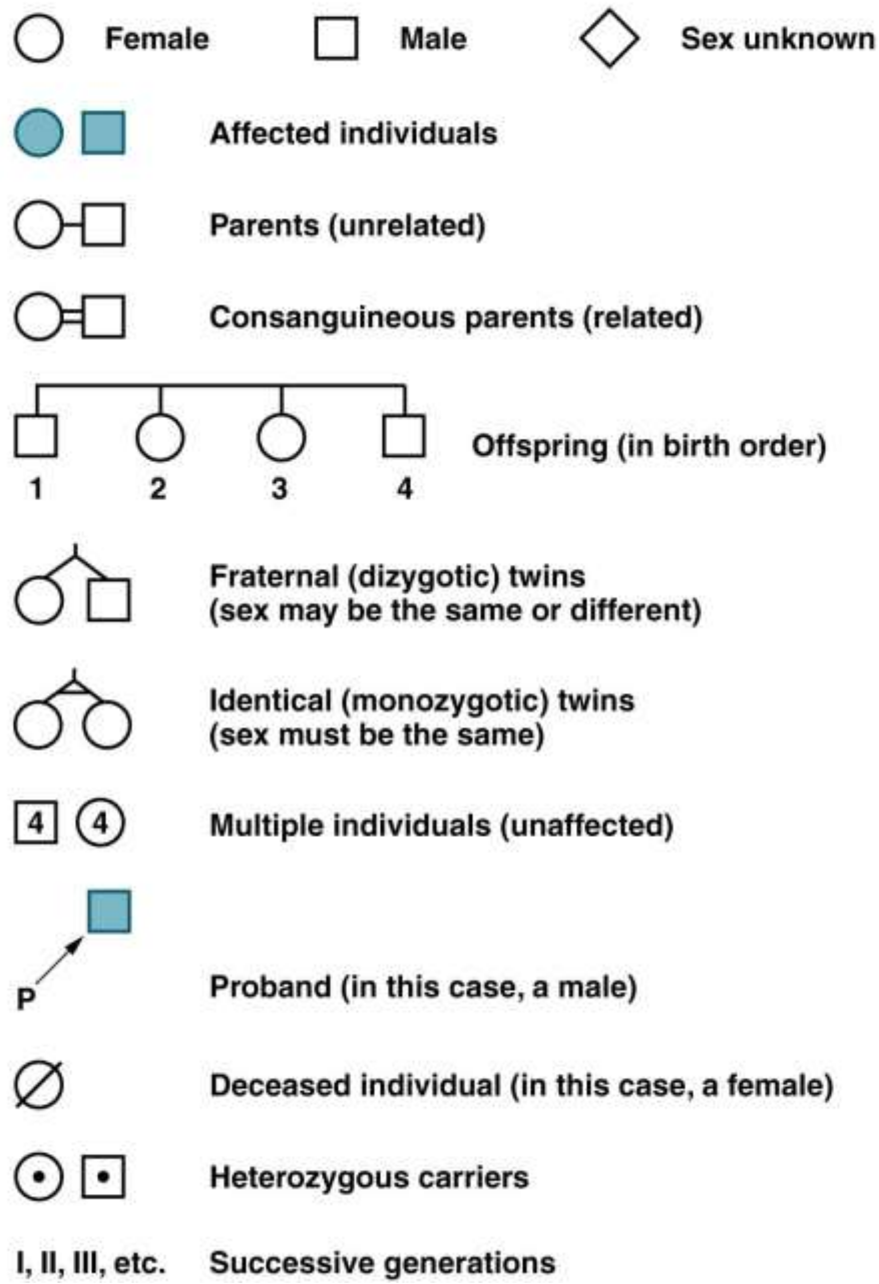
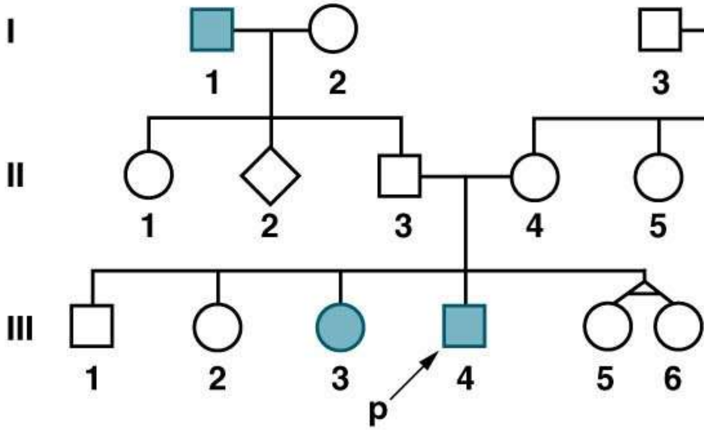

Female
Male
Sex unknown
Affected individuals
Parents (unrelated)
Consanguineous parents (related)
1
2
3
4
Offspring (in birth order)
Fraternal (dizygotic) twins
(sex may be the same or different)
Identical (monozygotic) twins
(sex must be the same)
4
4
Multiple individuals (unaffected)
P
Proband (in this case, a male)
Deceased individual (in this case, a female)
Heterozygous carriers
I, II, III, etc.
Successive generations



Figure 3.13

## (a) Autosomal Recessive Trait

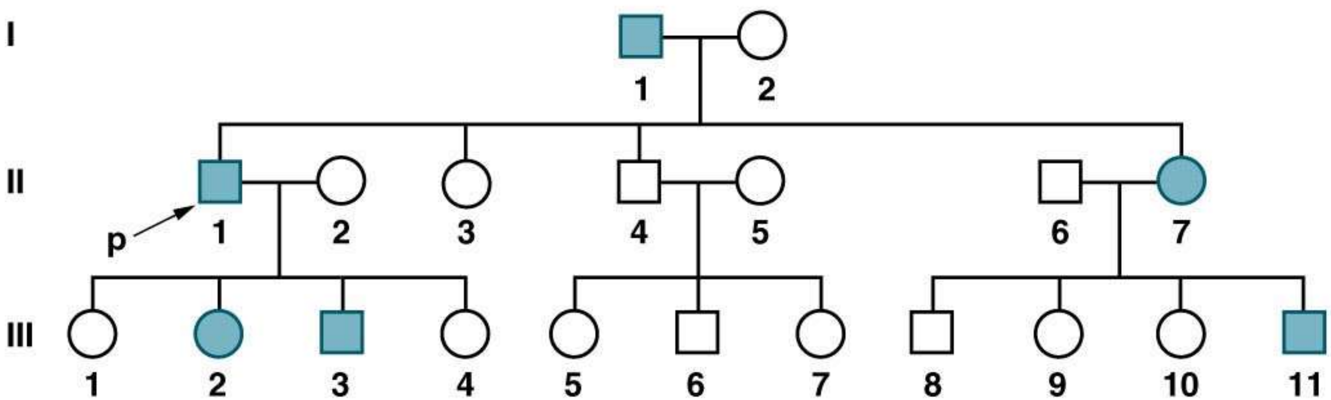



Either I-3 or I-4 must be heterozygous

Recessive traits typically skip generations

Recessive autosomal traits appear equally in both sexes

## (b) Autosomal Dominant Trait




I-1 is heterozygous for a dominant allele

Dominant traits almost always appear in each generation

Affected individuals all have an affected parent. Dominant autosomal traits appear equally in both sexes



Figure 3.14

# Section 3.10

- Parents are connected by a single horizontal line, and vertical lines lead to their offspring.
- If the parents are related (consanguineous), such as first cousins, they are connected by a double line.
- Offspring are called sibs (siblings) and are connected by a horizontal sibship line.

# Section 3.10

- Twins are indicated by diagonal lines stemming from a vertical line connected to the sibship line.
- For identical (monozygotic) twins, the diagonal lines are linked by a horizontal line.
- Fraternal (dizygotic) twins lack this connecting line.

# Section 3.10

- Pedigree analysis of human traits has been an extremely valuable tool in human genetic studies (see **Table 3.4** for some examples of human traits for which pedigree analysis is very useful).

**TABLE 3.4**

## Representative Recessive and Dominant Human Traits

| Recessive Traits | Dominant Traits |
|---|---|
| Albinism | Achondroplasia |
| Alkaptonuria | Brachydactyly |
| Ataxia telangiectasia | Congenital stationary night blindness |
| Color blindness | Ehler–Danlos syndrome |
| Cystic fibrosis | Hypotrichosis |
| Duchenne muscular dystrophy | Huntington disease |
| Galactosemia | Hypercholesterolemia |
| Hemophilia | Marfan syndrome |
| Lesch–Nyhan syndrome | Neurofibromatosis |
| Phenylketonuria | Phenylthiocarbamide tasting |
| Sickle-cell anemia | Porphyria (some forms) |
| Tay–Sachs disease | Widow's peak |



Table 3.4