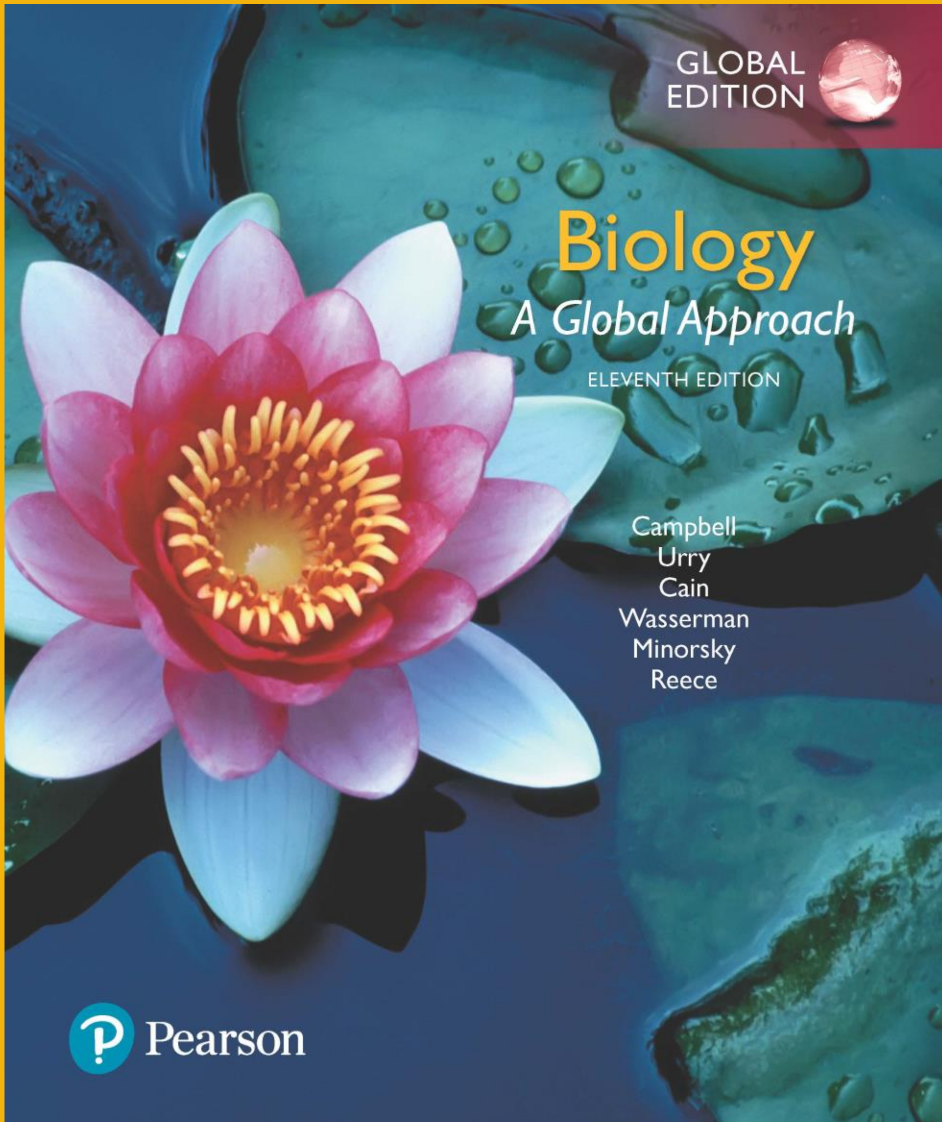


Chapter 14

Mendelian Genetics

Lecture Presentations by
Nicole Tunbridge and
Kathleen Fitzpatrick

Drawing from the Deck of Genes

- What principles account for the transmission of traits from parents to offspring?
- The “blending” hypothesis is the idea that genetic material from the two parents blends together (like blue and yellow paint blend to make green)

- The “particulate” hypothesis is the idea that parents pass on discrete heritable units (genes)
- Mendel documented a particulate mechanism through his experiments with garden peas

Figure 14.1

Figure 14.1a

Mendel (third from right, holding a sprig of fuchsia) with his fellow monks

Concept 14.1: Mendel used the scientific approach to identify two laws of inheritance

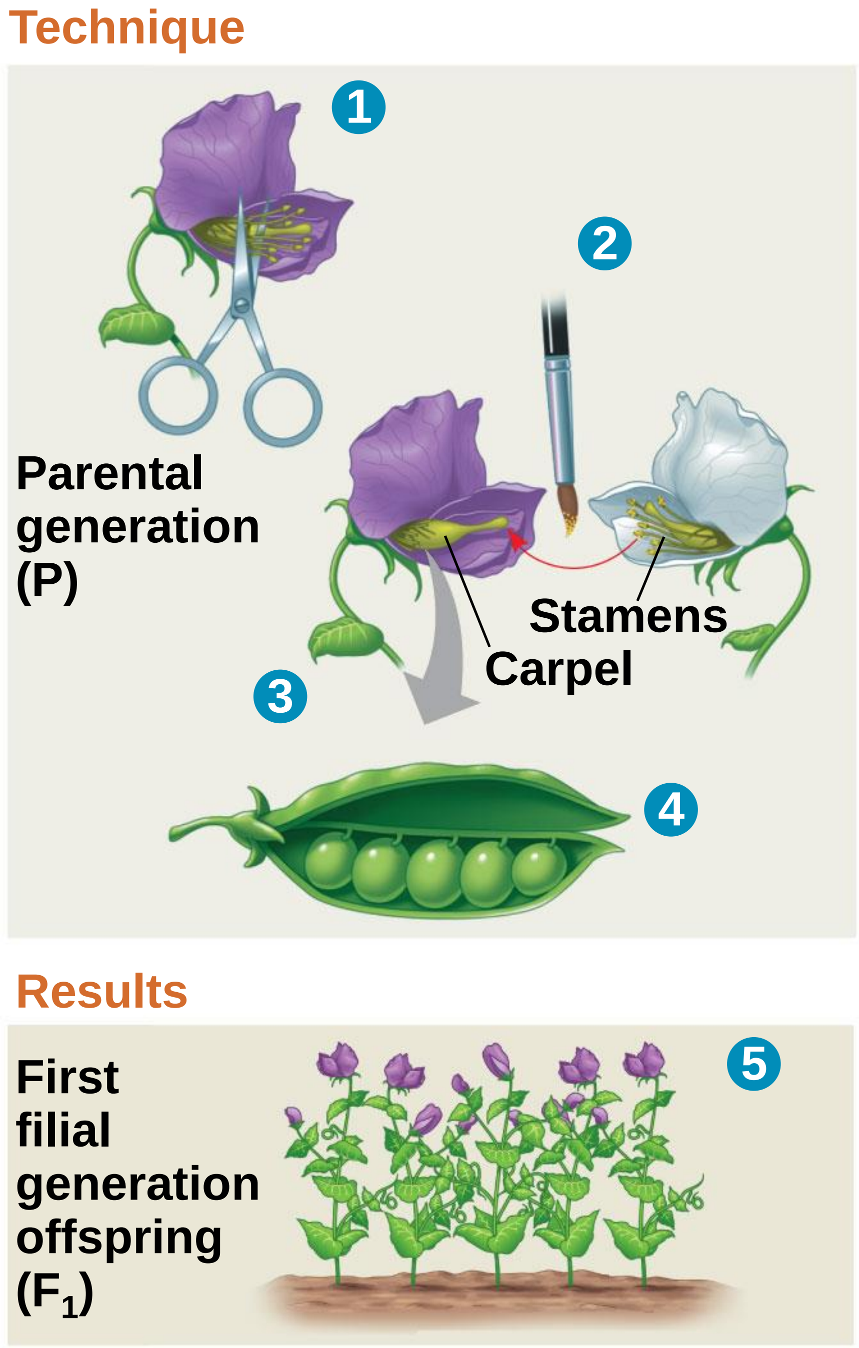
- Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments

Mendel's Experimental, Quantitative Approach

- Mendel's approach allowed him to deduce principles that had remained elusive to others
- A heritable feature that varies among individuals (such as flower color) is called a **character**
- Each variant for a character, such as purple or white color for flowers, is called a **trait**
- Peas were available to Mendel in many different varieties

- Other advantages of using peas
  - Short generation time
  - Large numbers of offspring
  - Mating could be controlled; plants could be allowed to self-pollinate or could be cross-pollinated

Figure 14.2



Technique
1
2
Parental
generation
(P)
Stamens
Carpel
3
4
Results
5
First
filial
generation
offspring
(F1)

- Mendel chose to track only those characters that occurred in two distinct alternative forms
- He also started with varieties that were **true-breeding** (plants that produce offspring of the same variety when they self-pollinate)

- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called **hybridization**
- The true-breeding parents are the **P generation**
- The hybrid offspring of the P generation are called the **F_1 generation**
- When F_1 individuals self-pollinate or cross-pollinate with other F_1 hybrids, the **F_2 generation** is produced

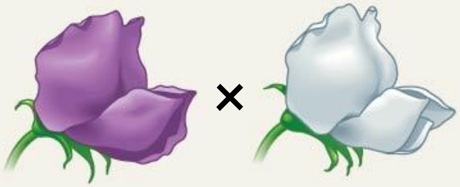
The Law of Segregation

- When Mendel crossed contrasting, true-breeding white- and purple-flowered pea plants, all of the F_1 hybrids were purple
- When Mendel crossed the F_1 hybrids, many of the F_2 plants had purple flowers, but some had white
- Mendel discovered a ratio of about three purple flowers to one white flower in the F_2 generation

Figure 14.3_1

Experiment

P Generation (true-breeding parents)



Purple flowers × **White flowers**

Figure 14.3_2

Experiment

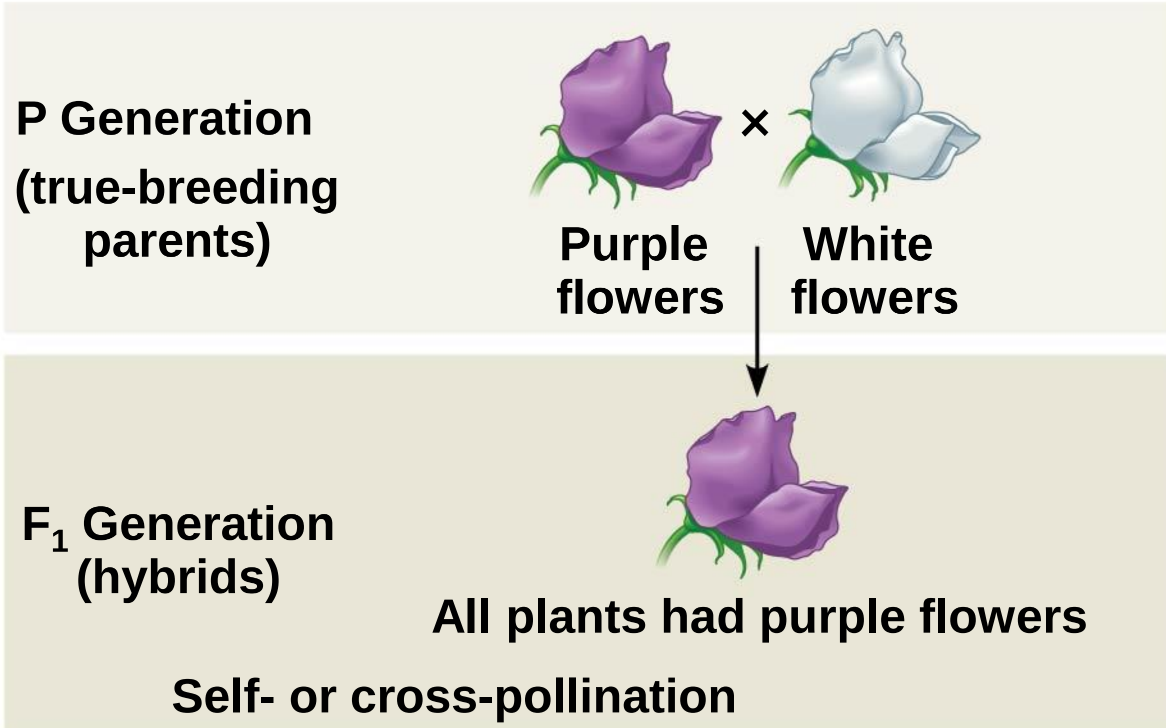
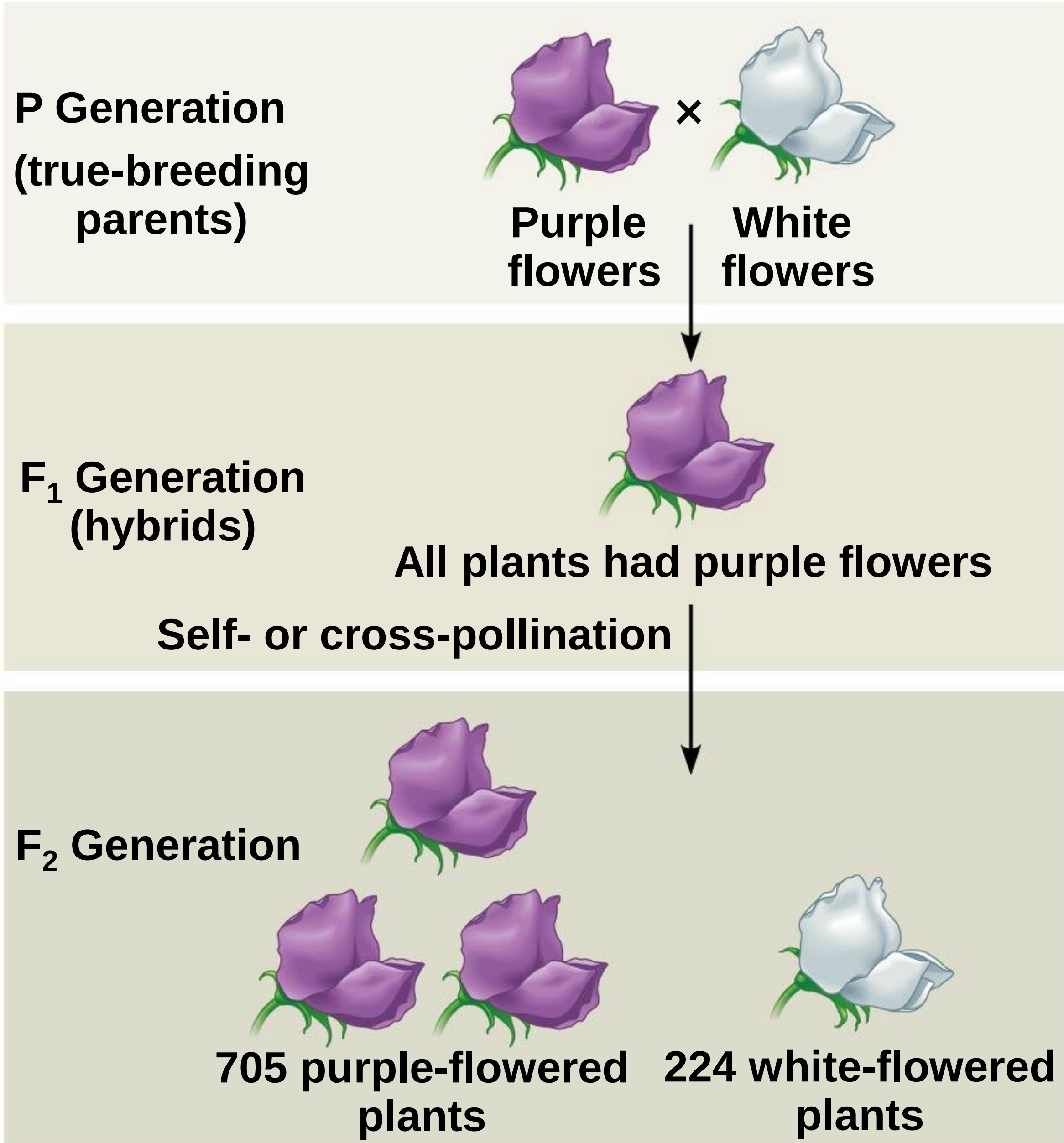


Figure 14.3_3

Experiment



- Mendel reasoned that only the purple flower factor was affecting flower color in the F_1 hybrids
- Mendel called the purple flower color a dominant trait and the white flower color a recessive trait
- The factor for white flowers was not diluted or destroyed because it reappeared in the F_2 generation

- Mendel observed the same pattern of inheritance in six other pea plant characters, each represented by two traits
- What Mendel called a “heritable factor” is what we now call a gene

Table 14.1a

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
Seed color	Yellow 	×	Green 	6,022:2,001	3.01:1
Seed shape	Round 	×	Wrinkled 	5,474:1,850	2.96:1

Table 14.1b

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

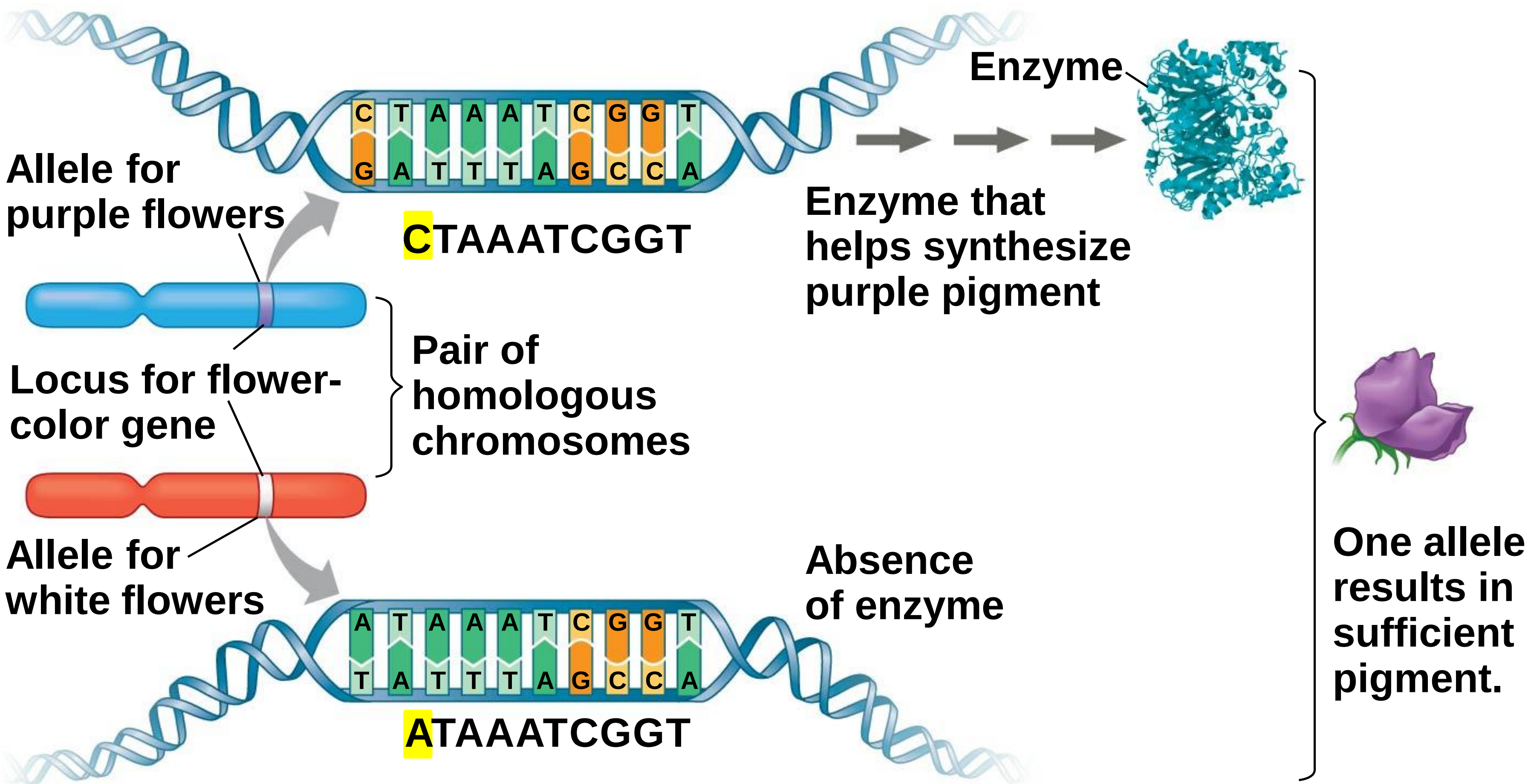
Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Pod color	Green 	×	Yellow 	428:152	2.82:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

Mendel's Model

- Mendel developed a hypothesis to explain the 3:1 inheritance pattern he observed in F_2 offspring
- Four related concepts make up this model
- These concepts can be related to what we now know about genes and chromosomes

- First: alternative versions of genes account for variations in inherited characters
- For example, the gene for flower color in pea plants exists in two versions, one for purple flowers and the other for white flowers
- These alternative versions of a gene are called **alleles**
- Each gene resides at a specific locus on a specific chromosome

Figure 14.4



Enzyme
C T A A A T C G G T
G A T T T A G C C A
Allele for purple flowers
CTAAATCGGT
Enzyme that helps synthesize purple pigment
Pair of homologous chromosomes
Locus for flower-color gene
Allele for white flowers
Absence of enzyme
One allele results in sufficient pigment.
A T A A A T C G G T
T A T T T A G C C A
ATAAATCGGT

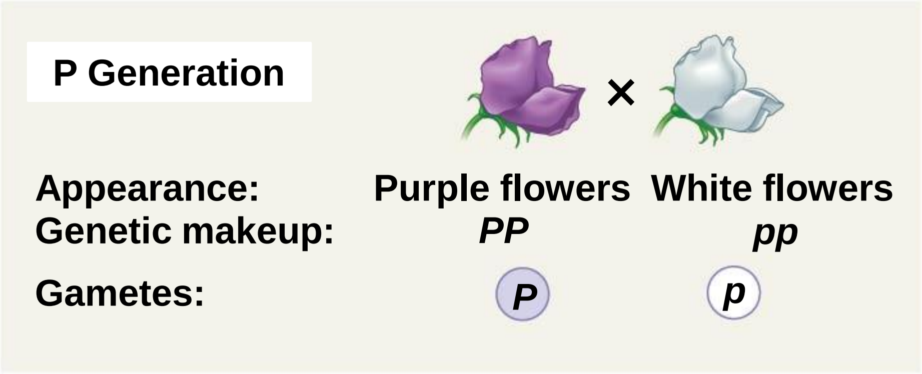
- Second: for each character, an organism inherits two alleles, one from each parent
- Mendel made this deduction without knowing about chromosomes
- The two alleles at a particular locus may be identical, as in the true-breeding plants of Mendel's P generation
- Or the two alleles at a locus may differ, as in the F_1 hybrids

- Third: if the two alleles at a locus differ, then one (the **dominant allele**) determines the organism's appearance, and the other (the **recessive allele**) has no noticeable effect on appearance
- In the flower-color example, the F_1 plants had purple flowers because the allele for that trait is dominant

- Fourth (the **law of segregation**): the two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes
- Thus, an egg or a sperm gets only one of the two alleles that are present in the organism
- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis

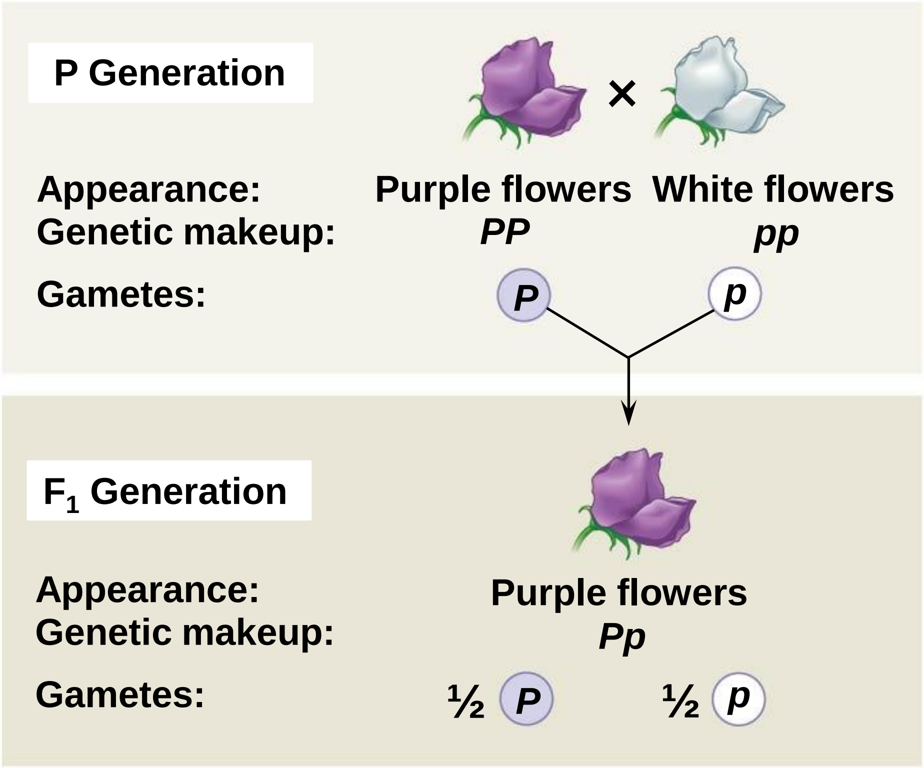
- The model accounts for the 3:1 ratio observed in the F_2 generation of Mendel's crosses
- Possible combinations of sperm and egg can be shown using a **Punnett square**
- A capital letter represents a dominant allele, and a lowercase letter represents a recessive allele

Figure 14.5_1



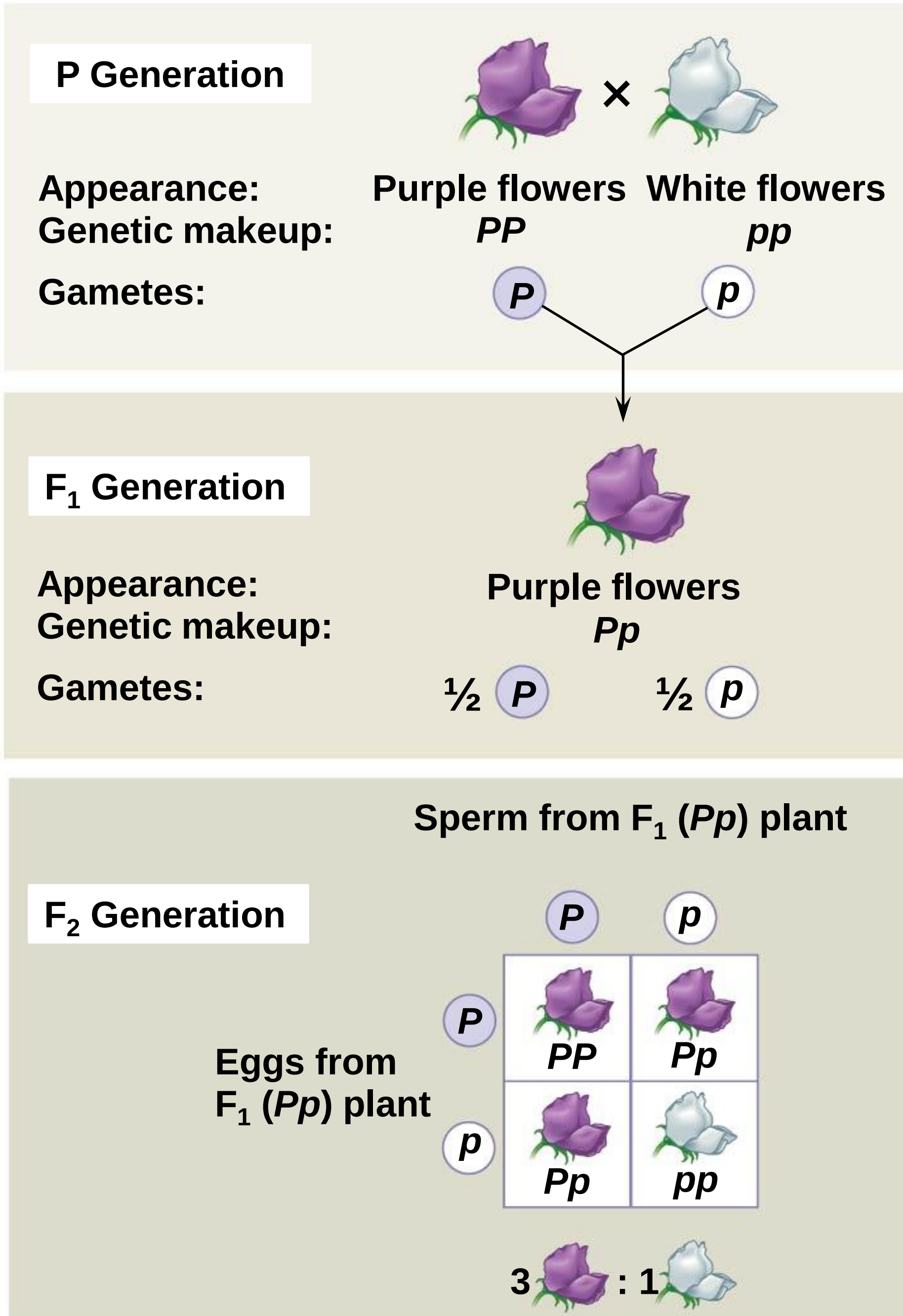
P Generation
×
Appearance:
Genetic makeup:
Gametes:
Purple flowers
PP
P
White flowers
pp
p

Figure 14.5_2



P Generation
×
Appearance:
Genetic makeup:
Gametes:
Purple flowers
PP
White flowers
pp
P
p
F1 Generation
Appearance:
Genetic makeup:
Gametes:
Purple flowers
Pp
½ P
½ p

Figure 14.5_3



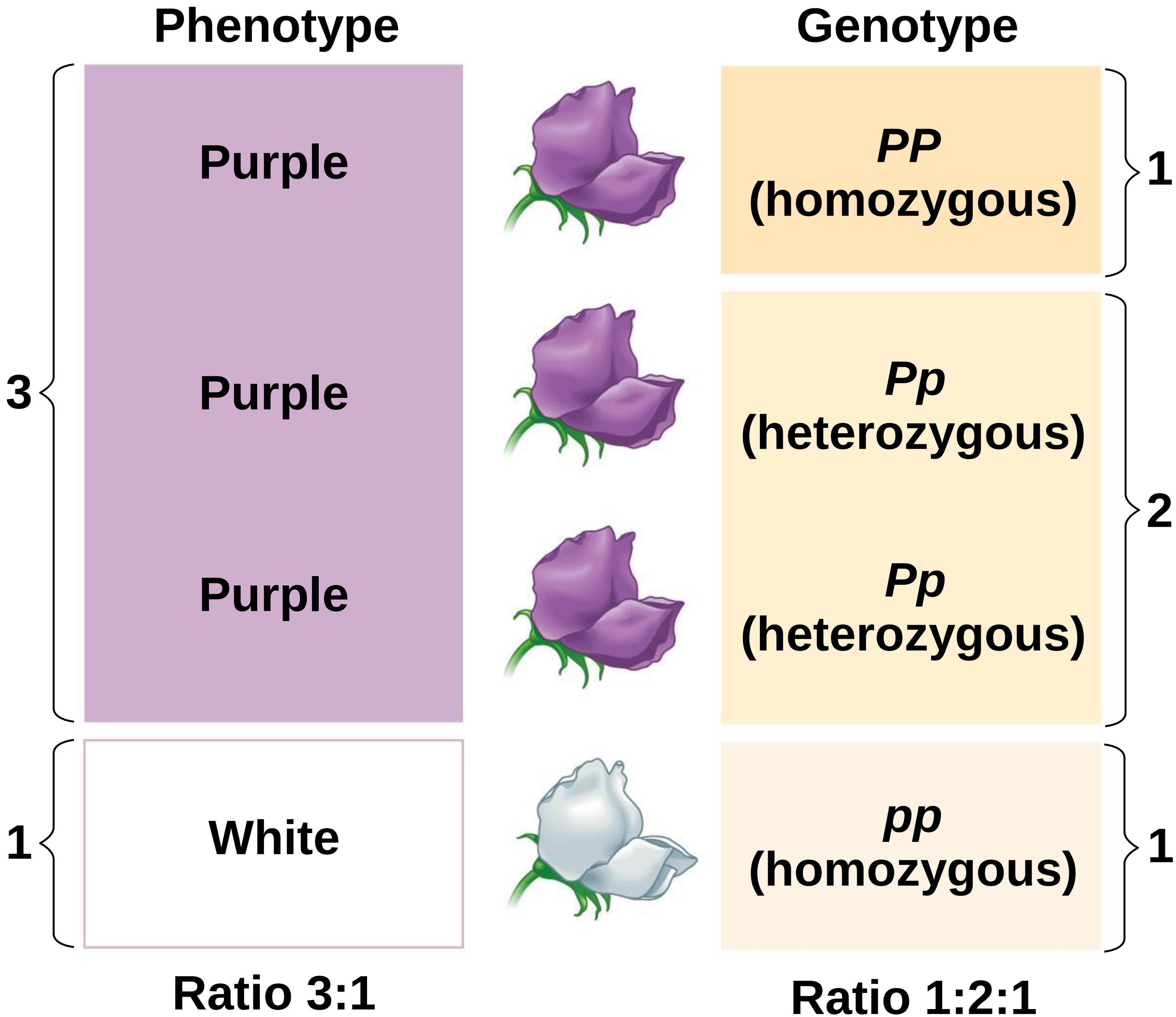
P Generation
Appearance:
Genetic makeup:
Gametes:
Purple flowers
PP
White flowers
pp
P
p
F1 Generation
Appearance:
Genetic makeup:
Gametes:
Purple flowers
Pp
½ P
½ p
Sperm from F1 (Pp) plant
F2 Generation
P
p
Eggs from
F1 (Pp) plant
P
p
PP
Pp
Pp
pp
3 : 1

Useful Genetic Vocabulary

- An organism with two identical alleles for a character is called a **homozygote**
- It is said to be **homozygous** for the gene controlling that character
- An organism with two different alleles for a gene is a **heterozygote** and is said to be **heterozygous** for the gene controlling that character
- Unlike homozygotes, heterozygotes are not true-breeding

- An organism’s traits do not always reveal its genetic composition
- Therefore, we distinguish between an organism’s **phenotype**, or physical appearance, and its **genotype**, or genetic makeup
- In the example of flower color in pea plants, *PP* and *Pp* plants have the same phenotype (purple) but different genotypes

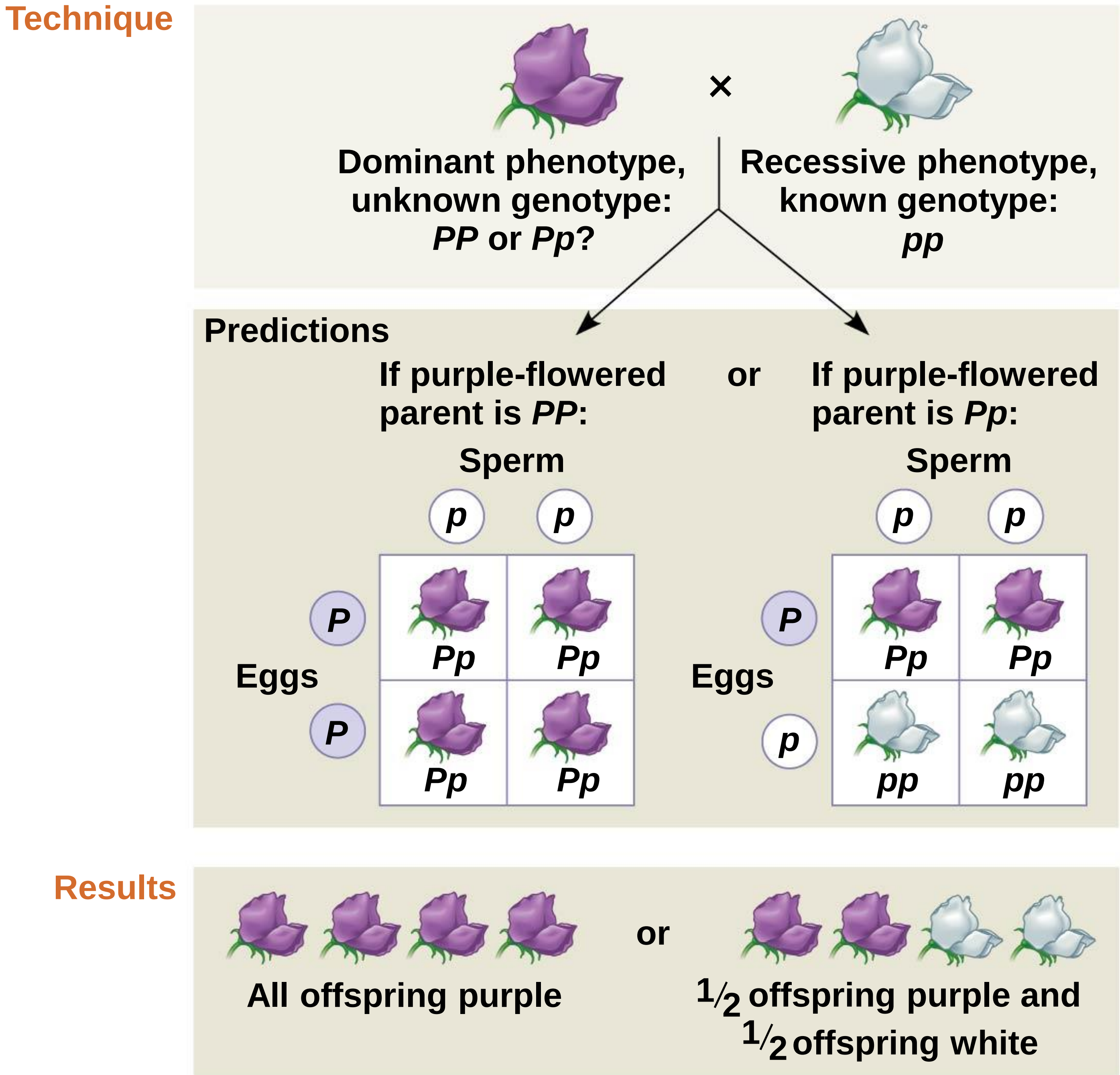
Figure 14.6


Phenotype
Genotype
Purple
Purple
Purple
White
3
1
PP
(homozygous)
Pp
(heterozygous)
Pp
(heterozygous)
pp
(homozygous)
1
2
1
Ratio 3:1
Ratio 1:2:1

The Testcross

- An individual with the dominant phenotype could be either homozygous dominant or heterozygous
- To determine the genotype we can carry out a **testcross**: breeding the mystery individual with a homozygous recessive individual
- If any offspring display the recessive phenotype, the mystery parent must be heterozygous

Figure 14.7

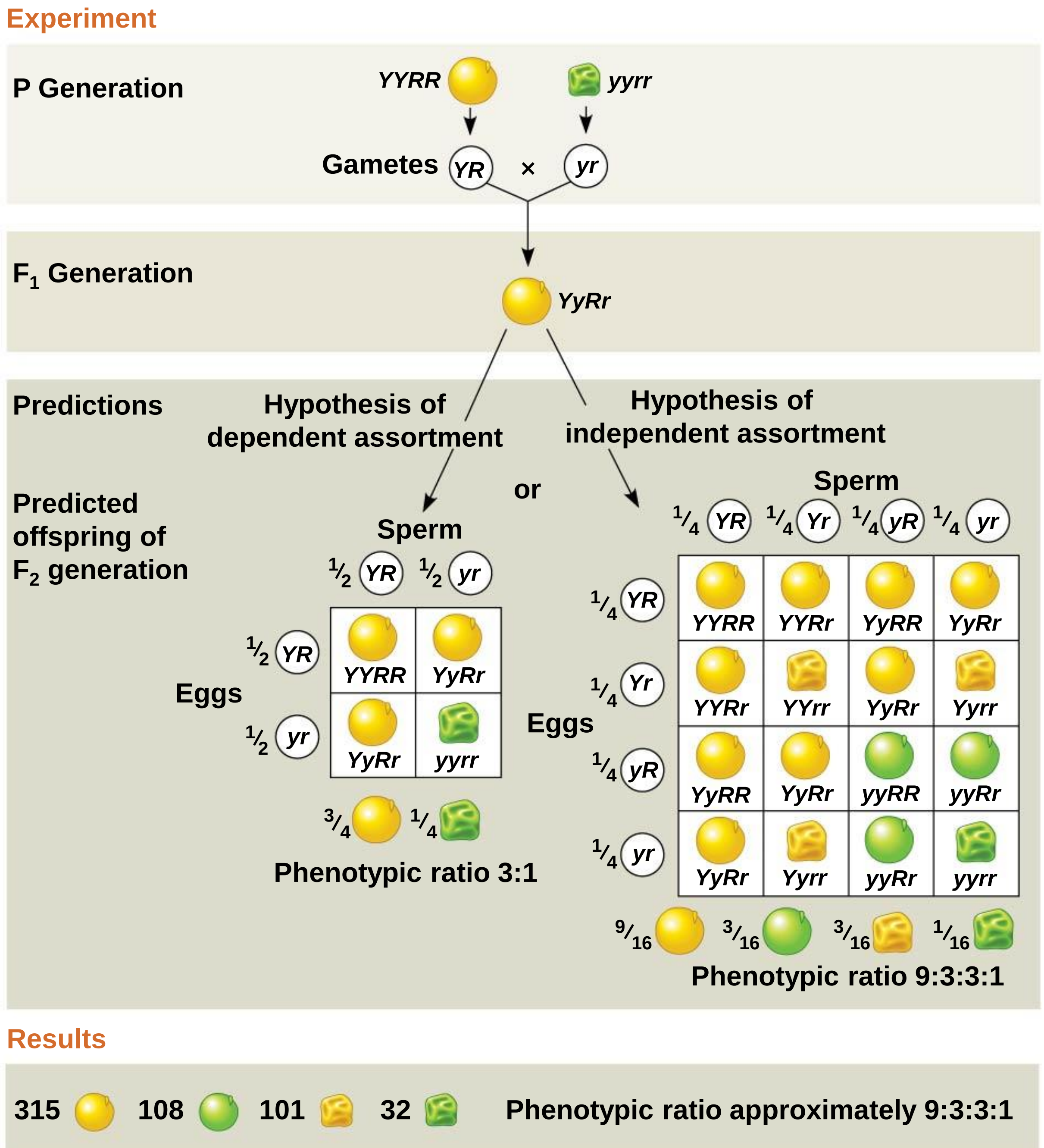

Technique
×
Dominant phenotype, unknown genotype: PP or Pp?
Recessive phenotype, known genotype: pp
Predictions
If purple-flowered parent is PP:
or
If purple-flowered parent is Pp:
Sperm
p
p
Eggs
P
P
Pp
Pp
Pp
Pp
Sperm
p
p
Eggs
P
p
Pp
Pp
pp
pp
Results
All offspring purple
or
1/2 offspring purple and 1/2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

The Law of Independent Assortment

- Mendel derived the law of segregation by following a single character
- The F_1 offspring produced in this cross were **monohybrids**, heterozygous for one character
- A cross between such heterozygotes is called a **monohybrid cross**

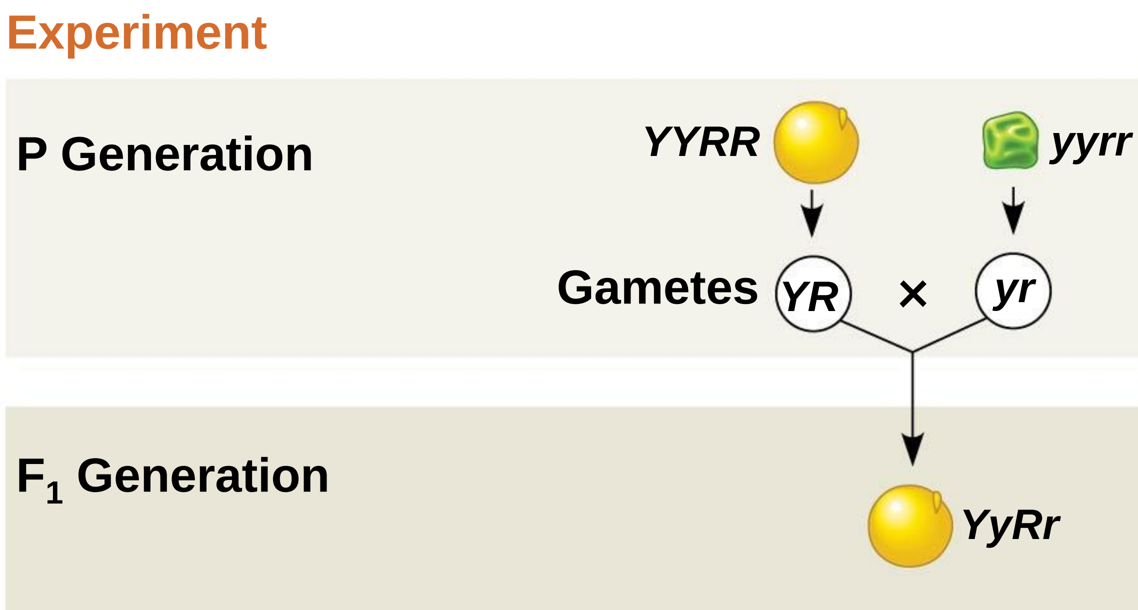
- Mendel identified his second law of inheritance by following two characters at the same time
- Crossing two true-breeding parents differing in two characters produces **dihybrids** in the F_1 generation, heterozygous for both characters
- A **dihybrid cross**, a cross between F_1 dihybrids, can determine whether two characters are transmitted to offspring as a package or independently

Figure 14.8



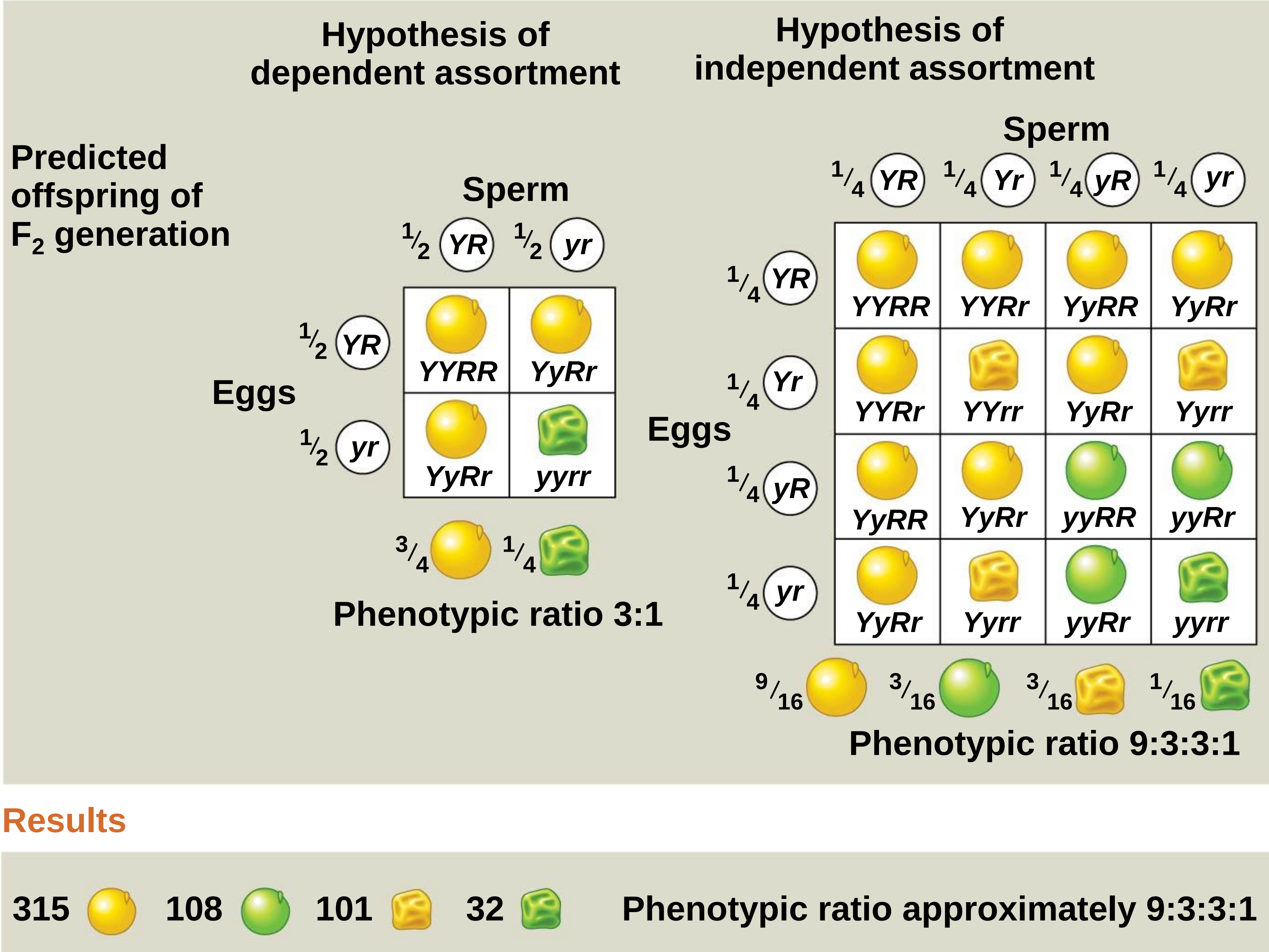
Experiment
P Generation
YYRR
yyrr
Gametes
YR
×
yr
F1 Generation
YyRr
Predictions
Hypothesis of dependent assortment
Hypothesis of independent assortment
or
Predicted offspring of F2 generation
Sperm
1/2 YR
1/2 yr
Eggs
1/2 YR
1/2 yr
YYRR
YyRr
YyRr
yyrr
3/4
1/4
Phenotypic ratio 3:1
Sperm
1/4 YR
1/4 Yr
1/4 yR
1/4 yr
Eggs
1/4 YR
1/4 Yr
1/4 yR
1/4 yr
YYRR
YYRr
YyRR
YyRr
YYRr
YYrr
YyRr
Yyrr
YyRR
YyRr
yyRR
yyRr
YyRr
Yyrr
yyRr
yyrr
9/16
3/16
3/16
1/16
Phenotypic ratio 9:3:3:1
Results
315
108
101
32
Phenotypic ratio approximately 9:3:3:1

Figure 14.8a



Experiment
P Generation
YYRR
yyrr
Gametes
YR
×
yr
F1 Generation
YyRr

Figure 14.8b



Hypothesis of dependent assortment
Hypothesis of independent assortment
Predicted offspring of F2 generation
Sperm
1/2 YR 1/2 yr
Eggs
1/2 YR
1/2 yr
YYRR YyRr
YyRr yyrr
3/4 1/4
Phenotypic ratio 3:1
Sperm
1/4 YR 1/4 Yr 1/4 yR 1/4 yr
Eggs
1/4 YR
1/4 Yr
1/4 yR
1/4 yr
YYRR YYRr YyRR YyRr
YYRr YYrr YyRr Yyrr
YyRR YyRr yyRR yyRr
YyRr Yyrr yyRr yyrr
9/16 3/16 3/16 1/16
Phenotypic ratio 9:3:3:1
Results
315 108 101 32
Phenotypic ratio approximately 9:3:3:1

Figure 14.8c

Figure 14.8d

- Using a dihybrid cross, Mendel developed the **law of independent assortment**
- It states that each pair of alleles segregates independently of any other pair of alleles during gamete formation
- This law applies only to genes on different, nonhomologous chromosomes or those far apart on the same chromosome
- Genes located near each other on the same chromosome tend to be inherited together

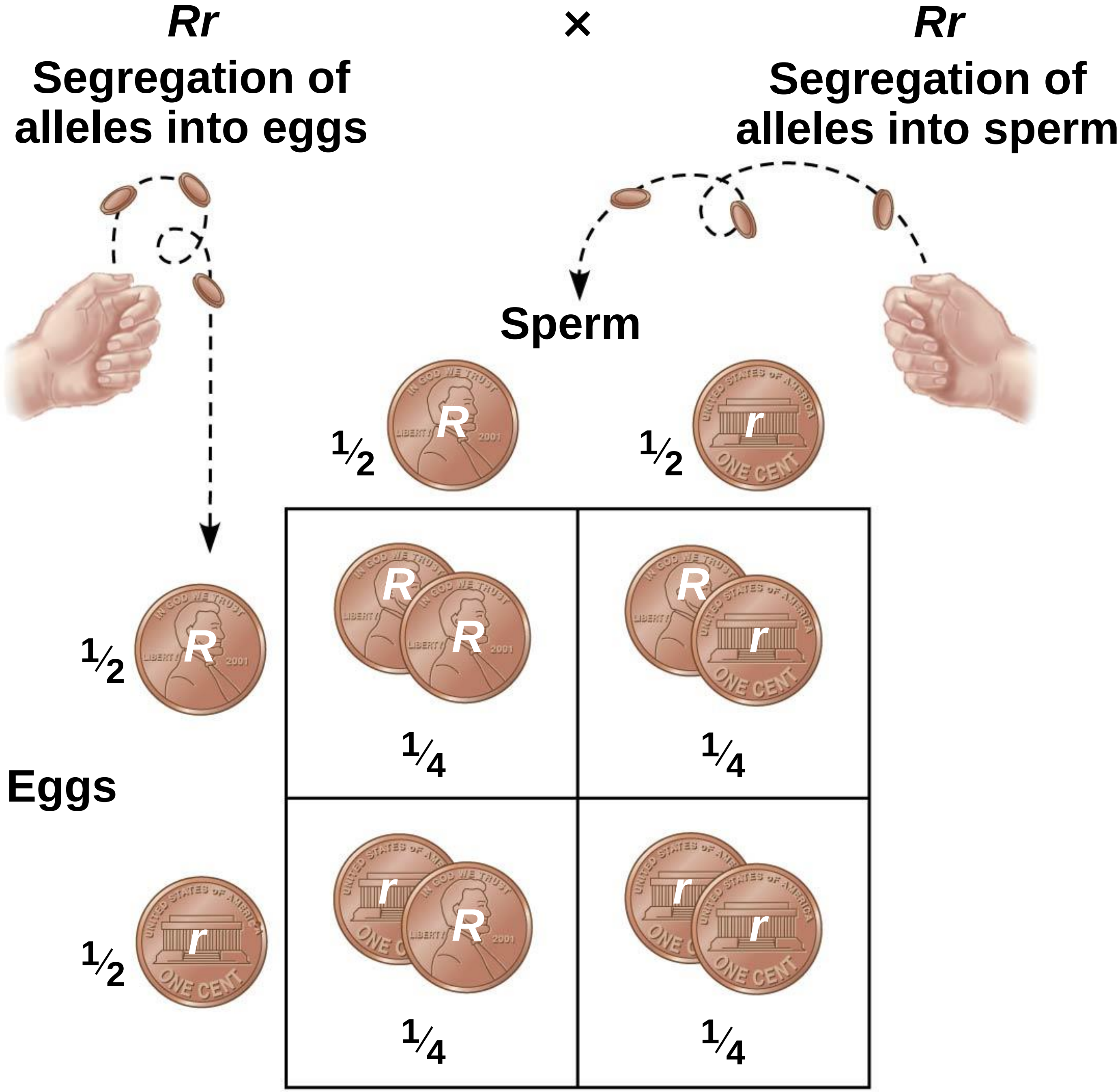
Concept 14.2: Probability laws govern Mendelian inheritance

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

The Multiplication and Addition Rules Applied to Monohybrid 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 ½ chance of carrying the dominant allele and a ½ chance of carrying the recessive allele

Figure 14.9


Rr
Segregation of alleles into eggs
×
Rr
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

- The **addition rule** states that the probability that any one of two or more mutually 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

Solving Complex Genetics Problems with the Rules of Probability

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

Figure 14.UN01

Probability of *YYRR* = $\frac{1}{4}$ (probability of *YY*) × $\frac{1}{4}$ (*RR*) = $\frac{1}{6}$

Probability of *YyRR* = $\frac{1}{2}$ (*Yy*) × $\frac{1}{4}$ (*RR*) = $\frac{1}{8}$

PpYyRr X Ppyyrr

ppyyRr	$\frac{1}{4}$ (probability of *pp*) $\times \frac{1}{2}$ (*yy*) $\times \frac{1}{2}$ (*Rr*)	$= \frac{1}{16}$
ppYyrr	$\frac{1}{4} \times \frac{1}{2} \times \frac{1}{2}$	$= \frac{1}{16}$
Ppyyrr	$\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2}$	$= \frac{2}{16}$
PPyyrr	$\frac{1}{4} \times \frac{1}{2} \times \frac{1}{2}$	$= \frac{1}{16}$
ppyyrr	$\frac{1}{4} \times \frac{1}{2} \times \frac{1}{2}$	$= \frac{1}{16}$
Chance of *at least two* recessive traits		$= \frac{6}{16}$ or $\frac{3}{8}$

Concept 14.3: Inheritance patterns are often more complex than predicted by simple Mendelian genetics

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

Extending Mendelian Genetics for a Single Gene

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

Degrees of Dominance

- **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are identical
- In **incomplete dominance**, the phenotype of F_1 hybrids is somewhere between the phenotypes of the two parental varieties
- In **codominance**, two dominant alleles affect the phenotype in separate, distinguishable ways

Figure 14.10_1

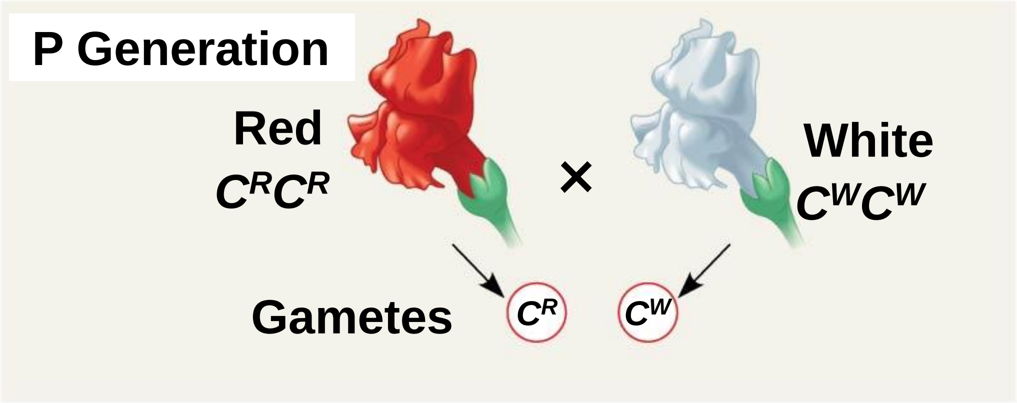


Figure 14.10_2

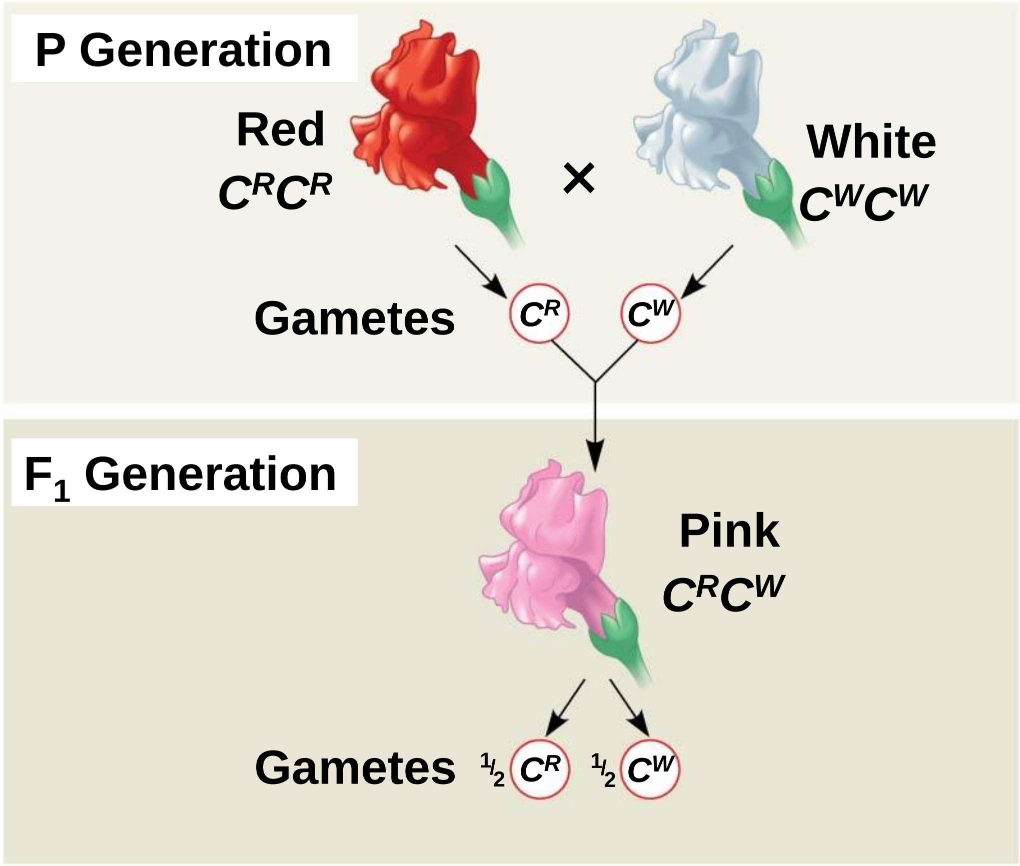
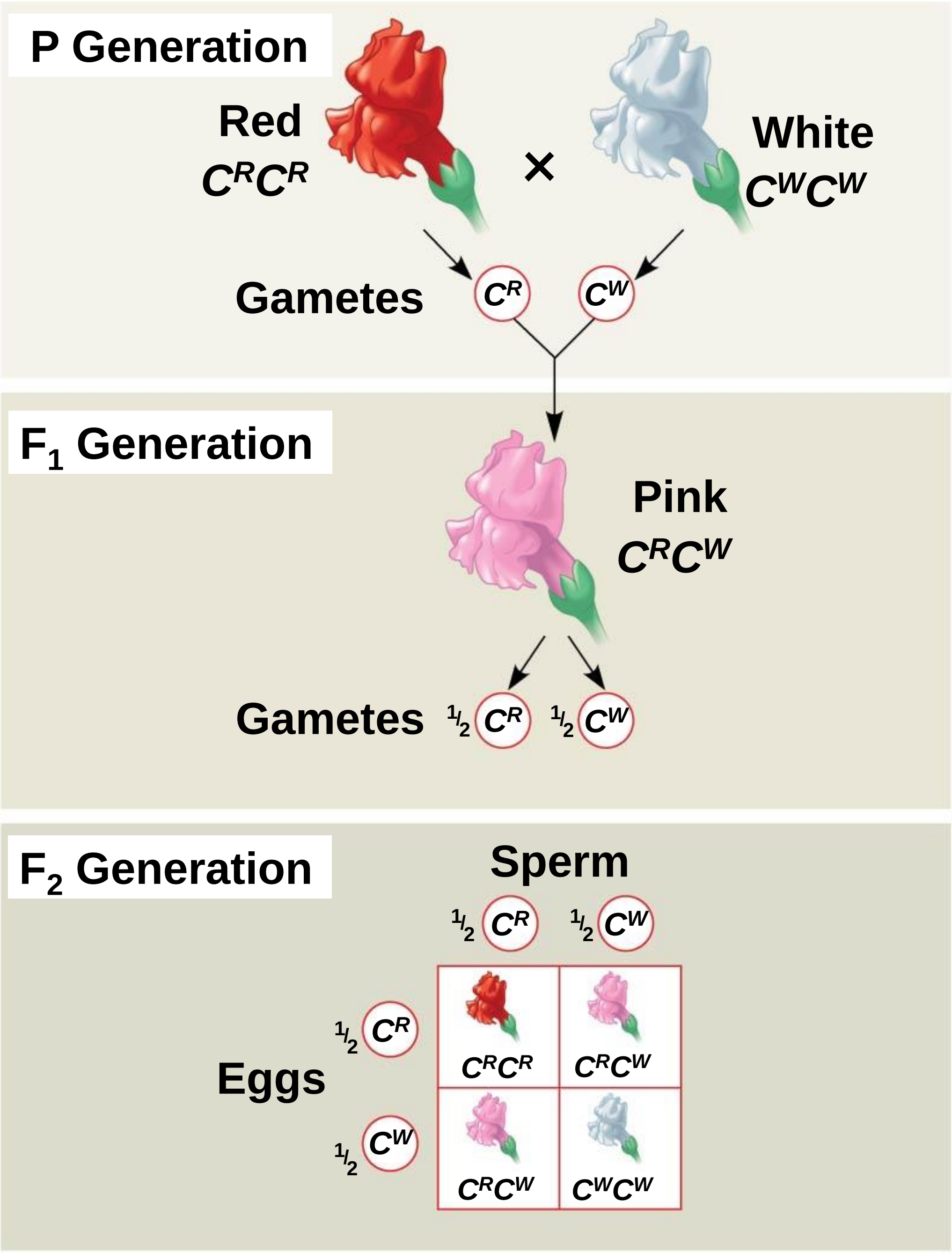


Figure 14.10_3



The Relationship Between Dominance and Phenotype

- In the case of pea shape, the dominant allele codes for an enzyme that converts an unbranched form of starch in the seed to a branched form
- The recessive allele codes for a defective form of the enzyme, which leads to an accumulation of unbranched starch
- This causes water to enter the seed, which then wrinkles as it dries

- **Tay-Sachs disease** is fatal; a dysfunctional enzyme causes an accumulation of lipids in the brain
  - At the organismal level, the allele is recessive
  - At the biochemical level, the phenotype (i.e., the enzyme activity level) is incompletely dominant
  - At the molecular level, the alleles are codominant

Frequency of Dominant Alleles

- Dominant alleles are not necessarily more common in populations than recessive alleles
- One baby out of 400 in the United States is born with extra fingers or toes
- This condition, polydactyly, is caused by a dominant allele, found much less frequently in the population than the recessive allele

Multiple Alleles

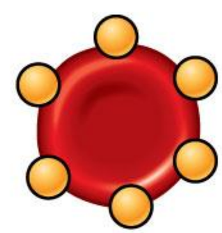
- Most genes exist in populations in more than two allelic forms
- For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme that attaches A or B carbohydrates to red blood cells: I^A, I^B, and *i*
- The enzyme encoded by the I^A allele adds the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

Figure 14.11

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

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

(b) Blood group genotypes and phenotypes

Genotype	$I^A I^A$ or $I^A i$	$I^B I^B$ or $I^B i$	$I^A I^B$	ii
Red blood cell with surface carbohydrates				
Phenotype (blood group)	A	B	AB	O

Pleiotropy

- Most genes have multiple phenotypic effects, a property called **pleiotropy**
- For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as cystic fibrosis (higher than normal level of salt in their sweat, A persistent cough that produces thick mucus (sputum), Wheezing, Breathlessness, Exercise intolerance, Repeated lung infections)
- and sickle-cell disease (Anemia, Episodes of pain, Painful swelling of hands and feet, Frequent infectionsm, Delayed growth, Vision problems)

Extending Mendelian Genetics for Two or More Genes

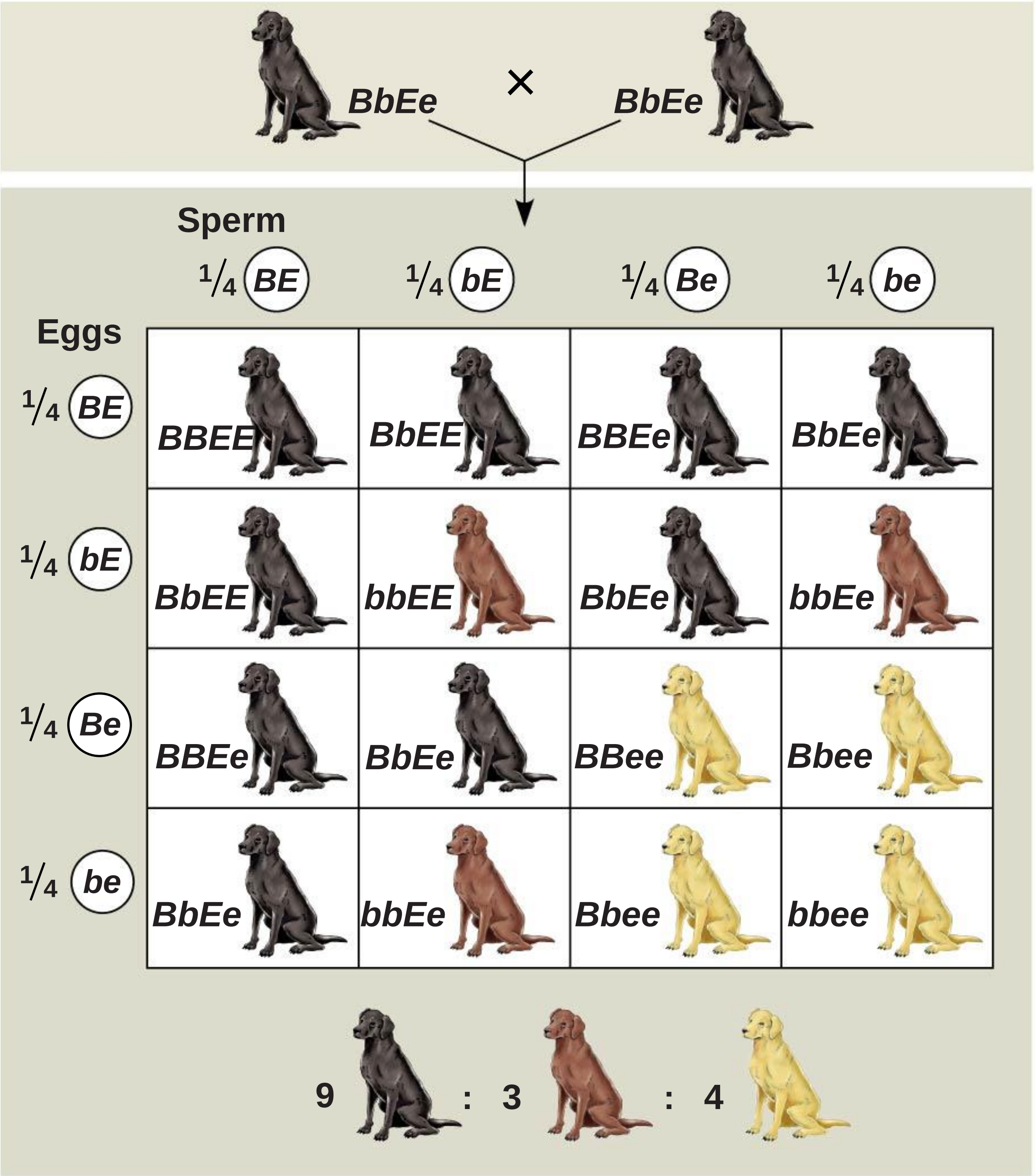
- Some traits may be determined by two or more genes
- In epistasis, one gene affects the phenotype of another due to interaction of their gene products
- In polygenic inheritance, multiple genes independently affect a single trait

Epistasis

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

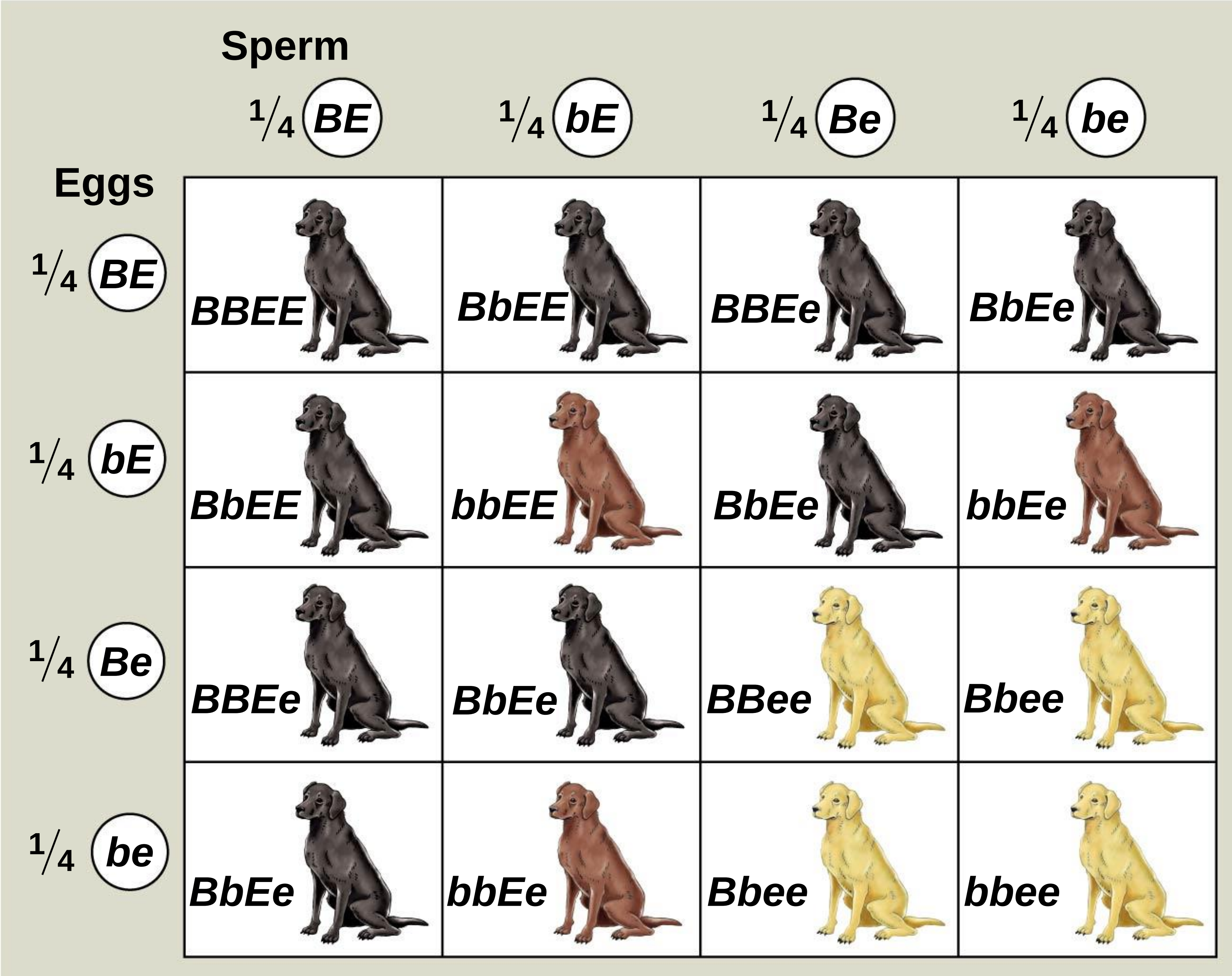
- If heterozygous black labs (genotype *BbEe*) are mated, we might expect the dihybrid F_2 ratio of 9:3:3:1
- However, a Punnett square shows that the phenotypic ratio will be 9 black to 3 chocolate to 4 yellow labs
- Epistatic interactions produce a variety of ratios, all of which are modified versions of 9:3:3:1

Figure 14.12



BbEe
×
BbEe
Sperm
1/4 BE
1/4 bE
1/4 Be
1/4 be
Eggs
1/4 BE
1/4 bE
1/4 Be
1/4 be
BBEE
BbEE
BBEe
BbEe
BbEE
bbEE
BbEe
bbEe
BBEe
BbEe
BBee
Bbee
BbEe
bbEe
Bbee
bbee
9 : 3 : 4

Figure 14.12a



Polygenic Inheritance

- **Quantitative characters** are those that vary in the population along a continuum
- Quantitative variation usually indicates **polygenic inheritance**, an additive effect of two or more genes on a single phenotype
- Height is a good example of polygenic inheritance: Over 180 genes affect height
- Skin color in humans is also controlled by many separately inherited genes

Figure 14.13

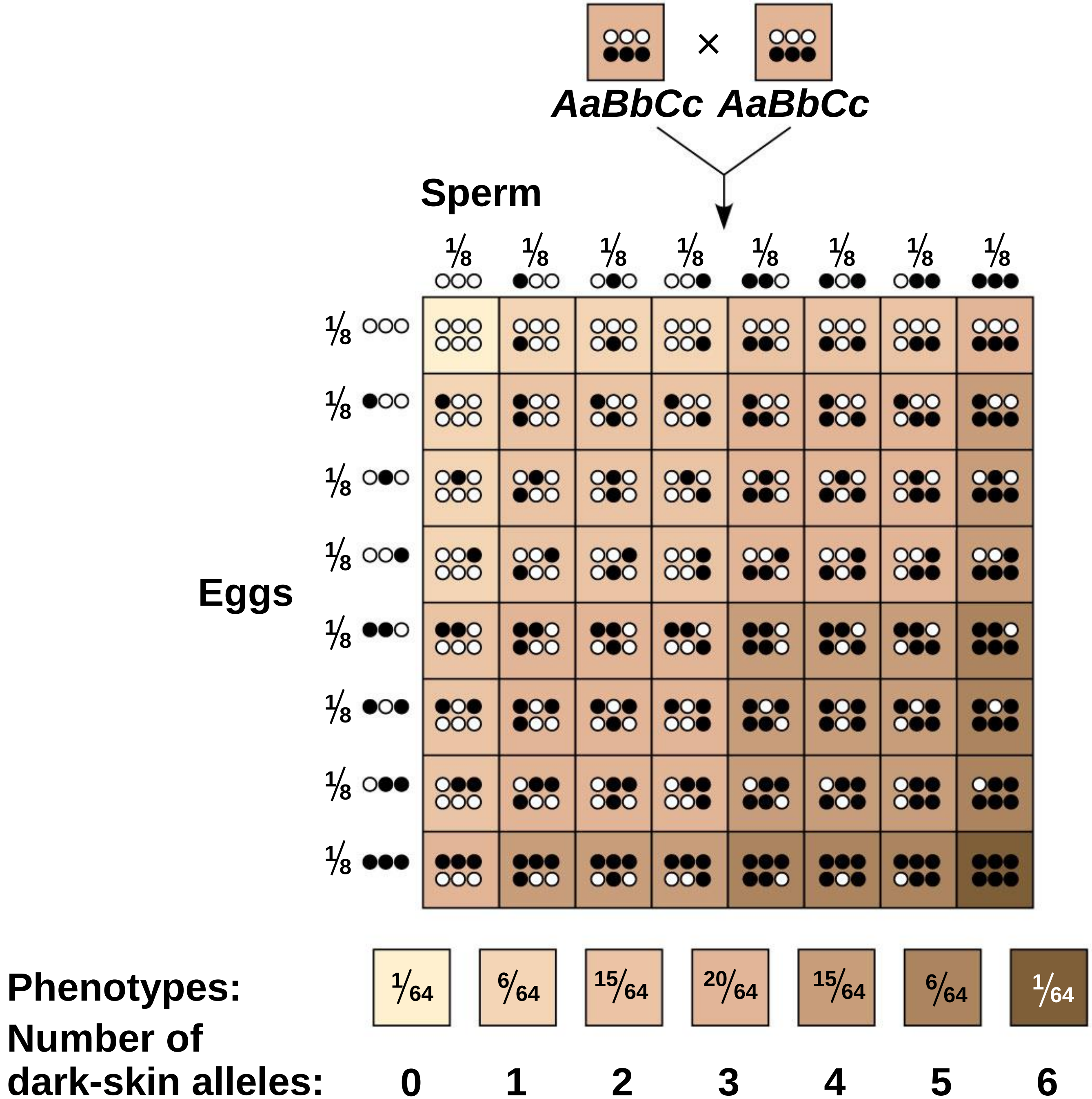
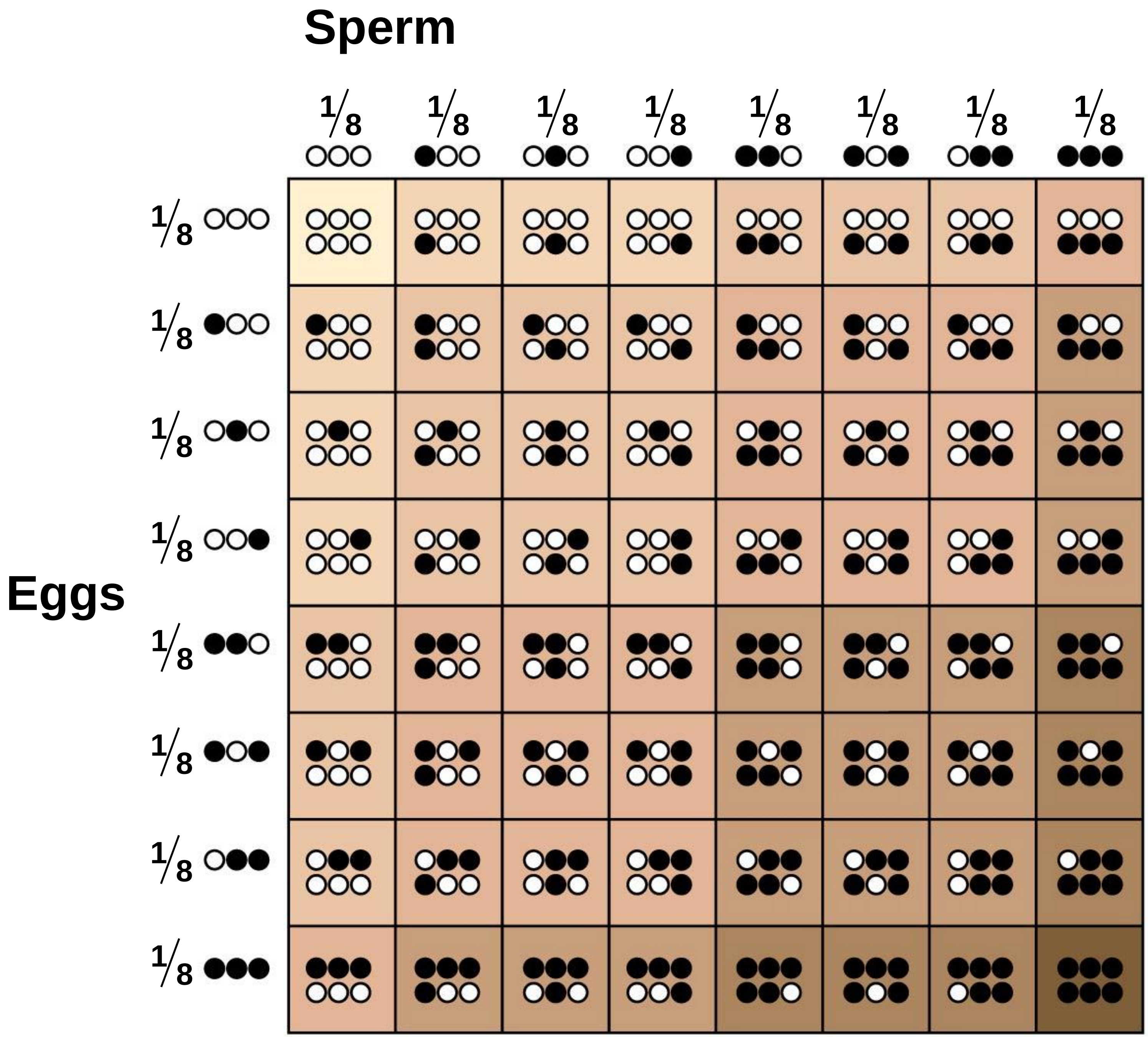

AaBbCc
AaBbCc
Sperm
Eggs
1/8
Phenotypes:
1/64
6/64
15/64
20/64
15/64
6/64
1/64
Number of dark-skin alleles:
0
1
2
3
4
5
6

Figure 14.13a



Nature and Nurture: The Environmental Impact on Phenotype

- Another departure from Mendelian genetics arises when the phenotype for a character depends on environment as well as genotype
- The phenotypic range is broadest for polygenic characters
- Traits that depend on multiple genes combined with environmental influences are called **multifactorial**

Figure 14.14

(a) Hydrangeas grown in basic soil

(b) Hydrangeas of the same genetic variety grown in acidic soil with free aluminum

A Mendelian View of Heredity and Variation

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