LECTURE PRESENTATIONS
For CAMPBELL BIOLOGY, NINTH EDITION
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Chapter 14

Mendel and the Gene Idea

Lectures by
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Overview: Drawing from the Deck of Genes

- What genetic principles account for the passing 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)
- This hypothesis can explain the reappearance of traits after several generations
- Mendel documented a particulate mechanism through his experiments with garden peas

Figure 14.1

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

- Advantages of pea plants for genetic study
  - There are many varieties with distinct heritable features, or **characters** (such as flower color); character variants (such as purple or white flowers) are called **traits**
  - Mating can be controlled
  - Each flower has sperm-producing organs (stamens) and egg-producing organ (carpel)
  - Cross-pollination (fertilization between different plants) involves dusting one plant with pollen from another

Figure 14.2

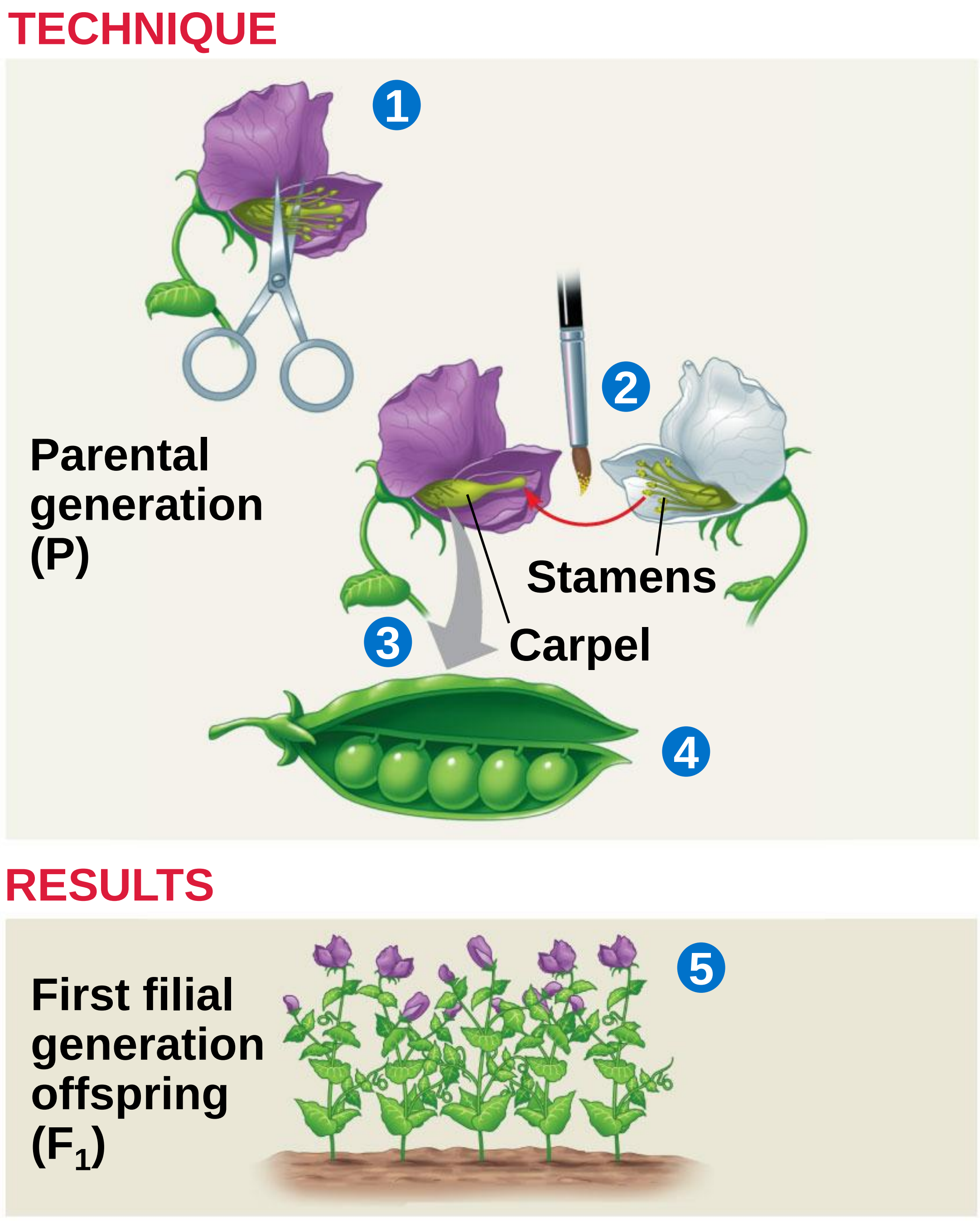

TECHNIQUE
1
2
Parental generation (P)
Stamens
3
Carpel
4
RESULTS
5
First filial generation offspring (F_1)

Figure 14.2a

TECHNIQUE

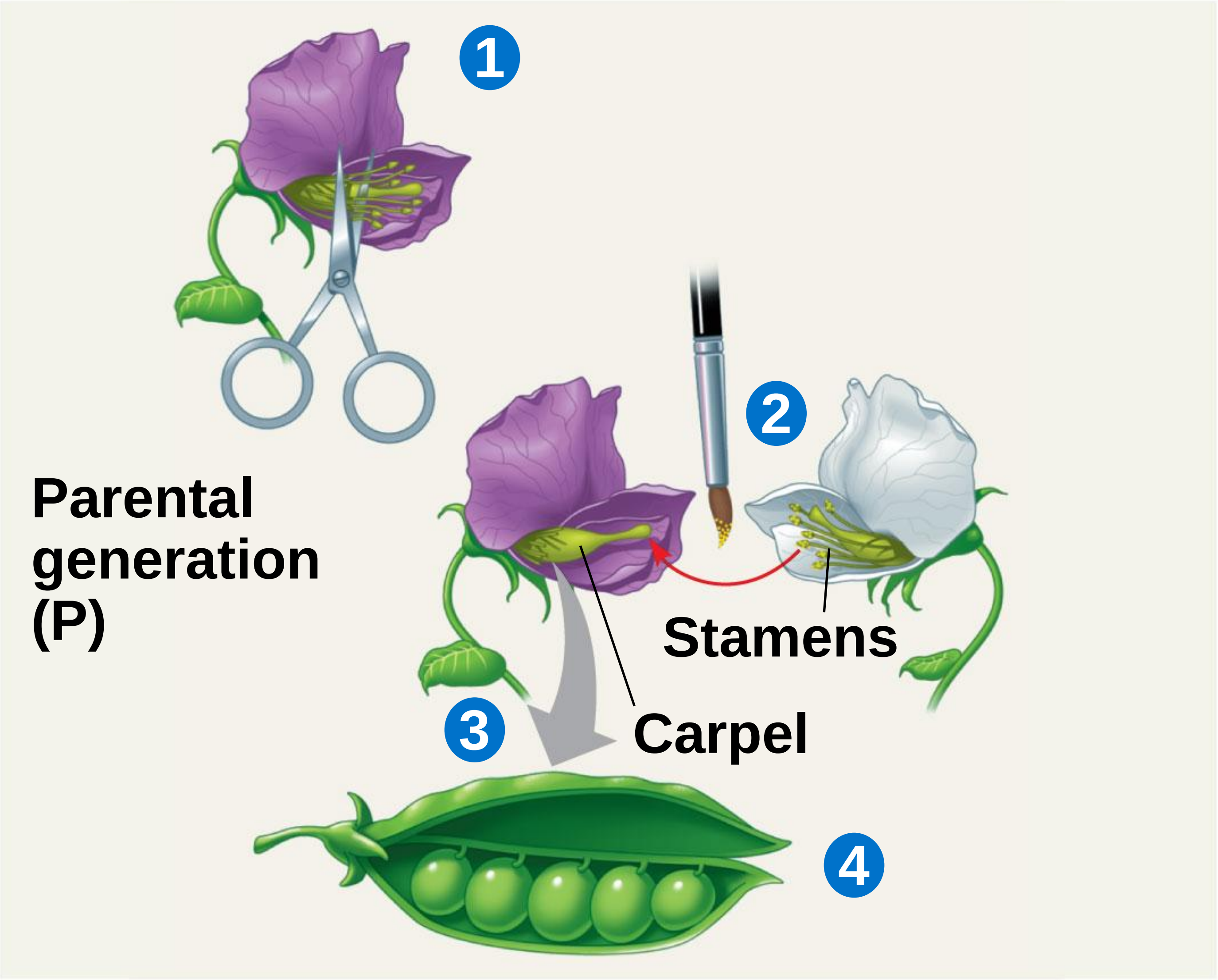


Figure 14.2b

RESULTS

- Mendel chose to track only those characters that occurred in two distinct alternative forms
- He also used 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 to one, purple to white flowers, in the F_2 generation

Figure 14.3-1

EXPERIMENT

P Generation
(true-breeding parents)

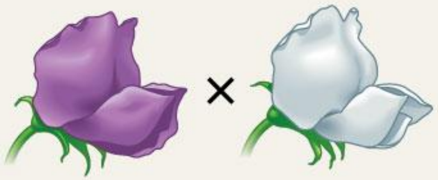


Figure 14.3-2

EXPERIMENT

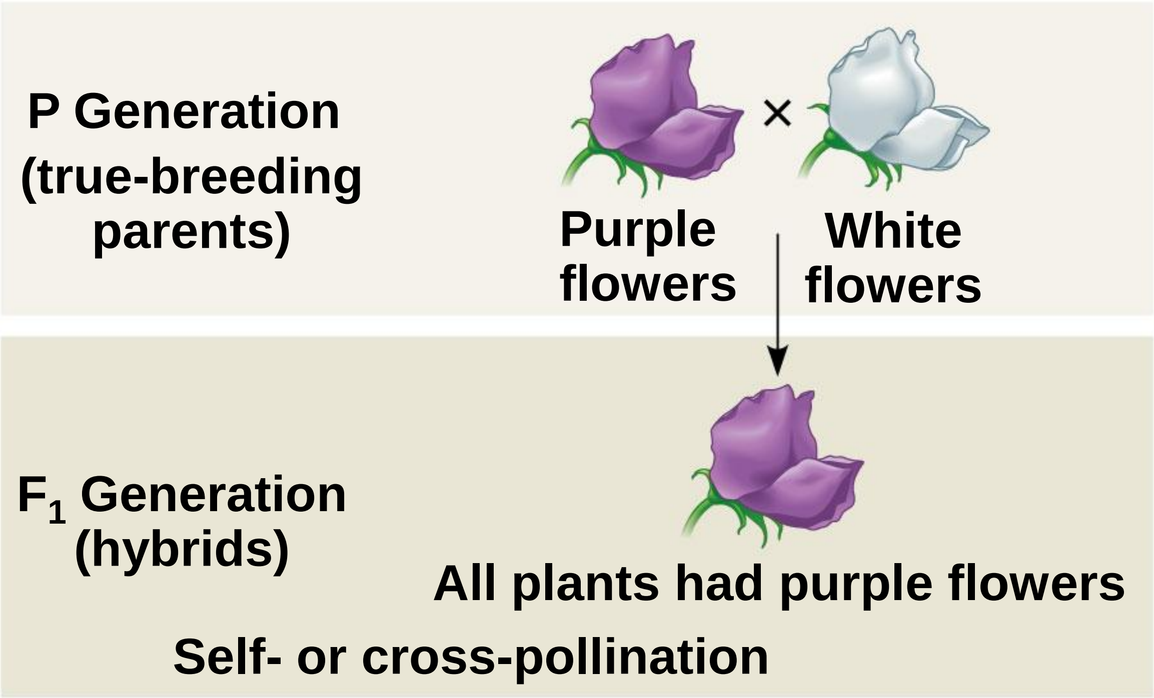
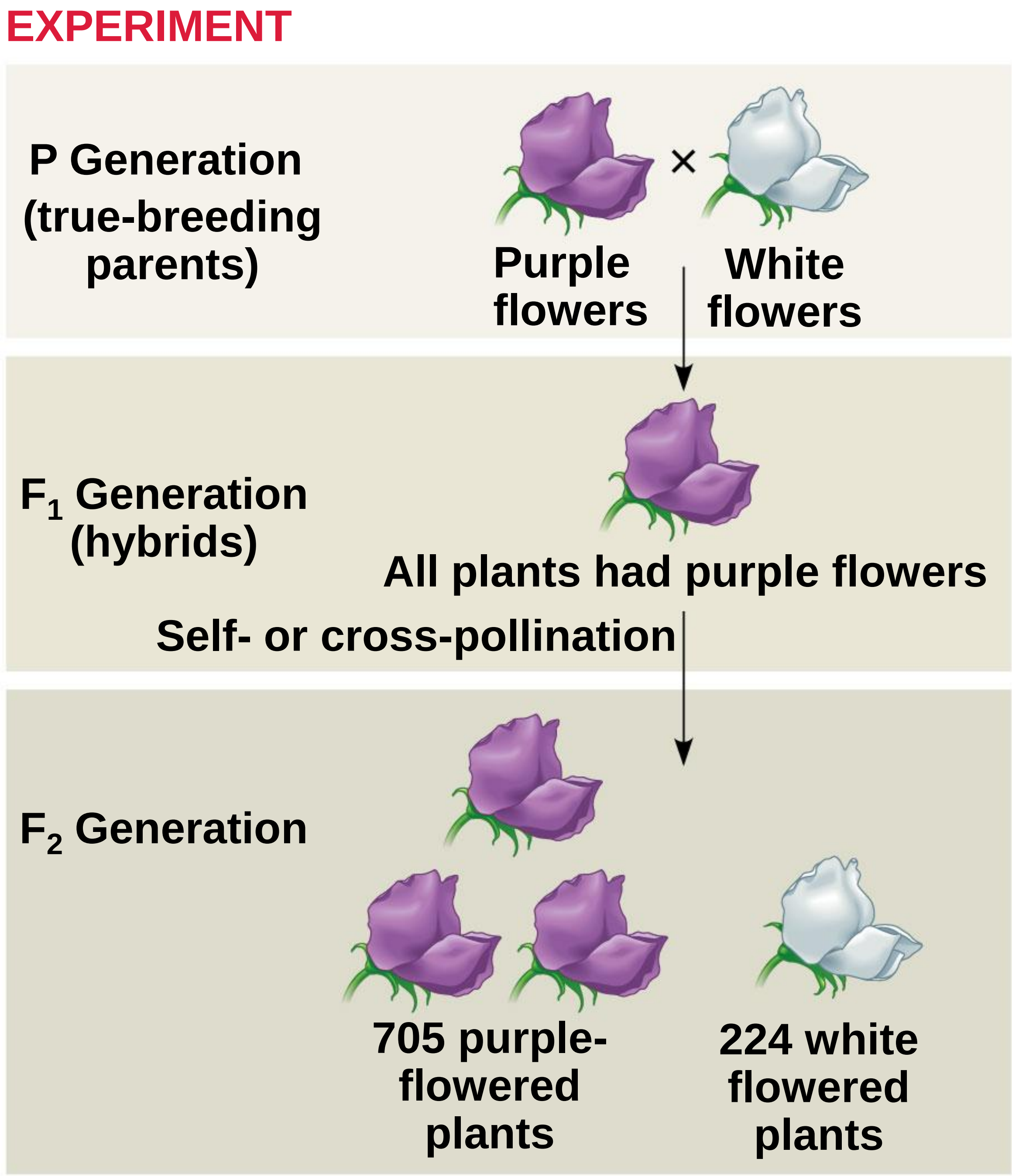


Figure 14.3-3


EXPERIMENT
P Generation
(true-breeding parents)
×
Purple flowers
White flowers
F1 Generation
(hybrids)
All plants had purple flowers
Self- or cross-pollination
F2 Generation
705 purple-flowered plants
224 white flowered plants

- 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 14.1

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

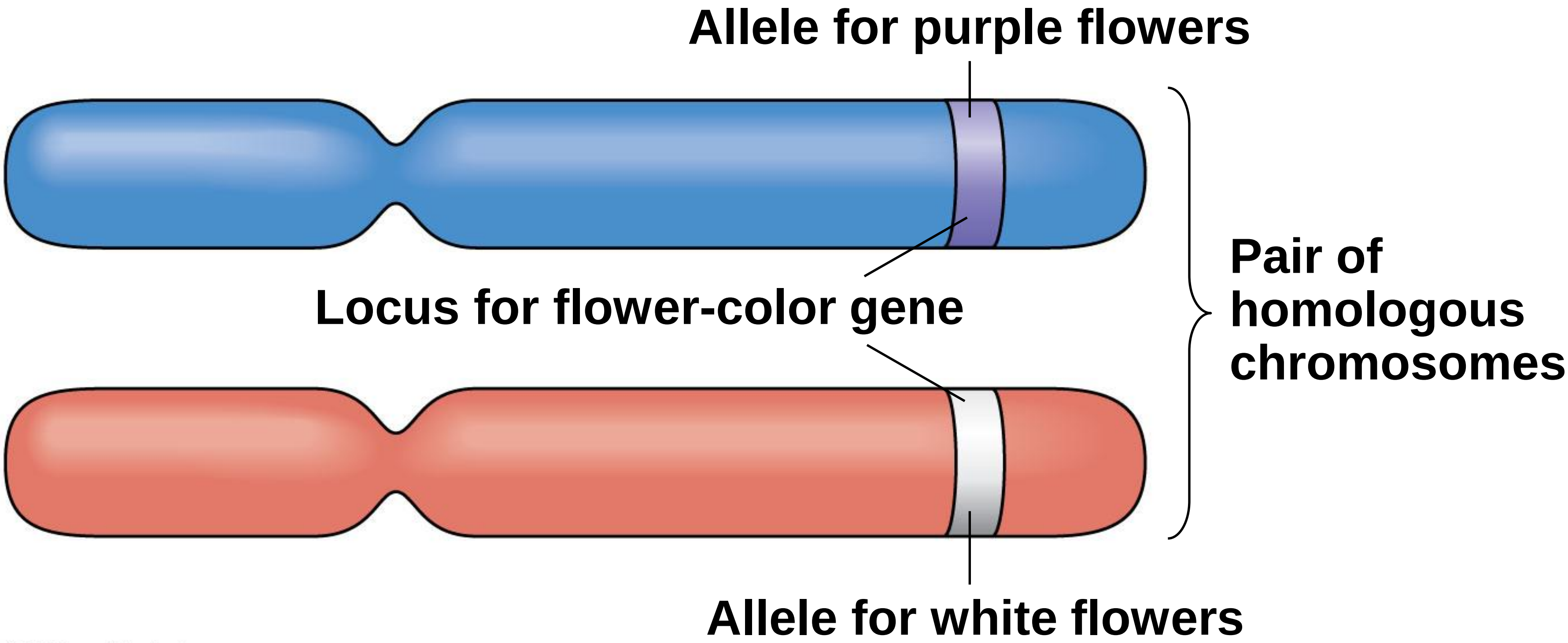
Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Seed color	Yellow 	×	Green 	6,022:2,001	3.01:1
Seed shape	Round 	×	Wrinkled 	5,474:1,850	2.96:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
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 now called **alleles**
- Each gene resides at a specific locus on a specific chromosome

Figure 14.4


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

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

- Mendel’s segregation model accounts for the 3:1 ratio he observed in the F_2 generation of his numerous crosses
- The possible combinations of sperm and egg can be shown using a **Punnett square**, a diagram for predicting the results of a genetic cross between individuals of known genetic makeup
- A capital letter represents a dominant allele, and a lowercase letter represents a recessive allele

Figure 14.5-1

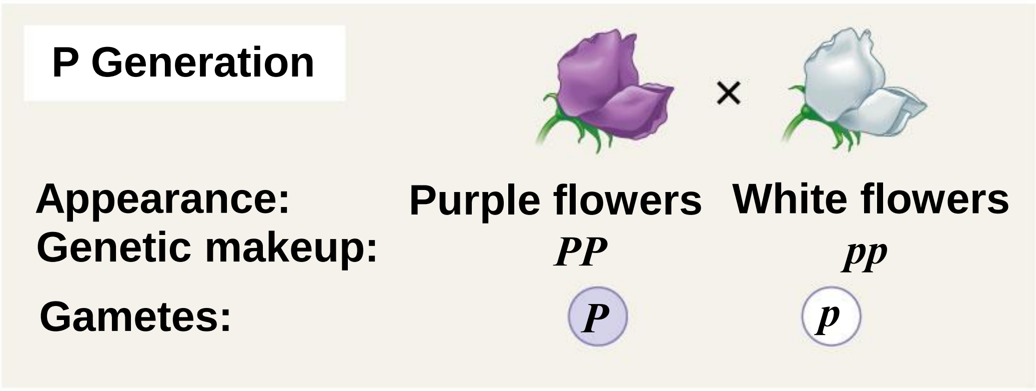
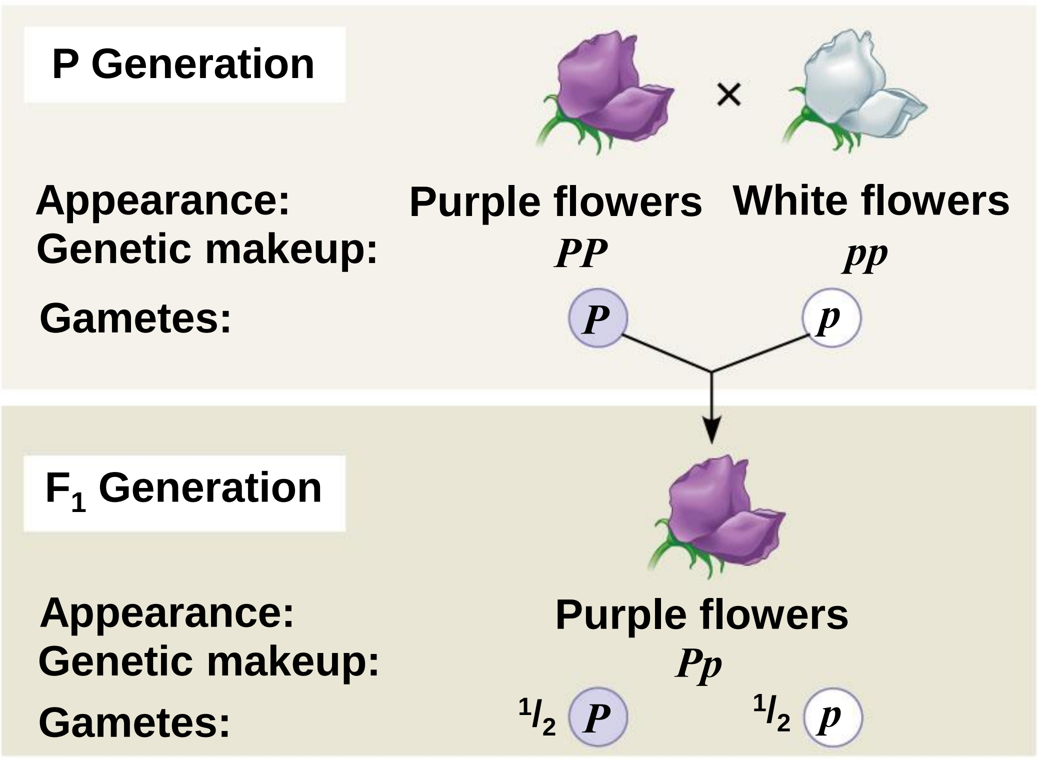
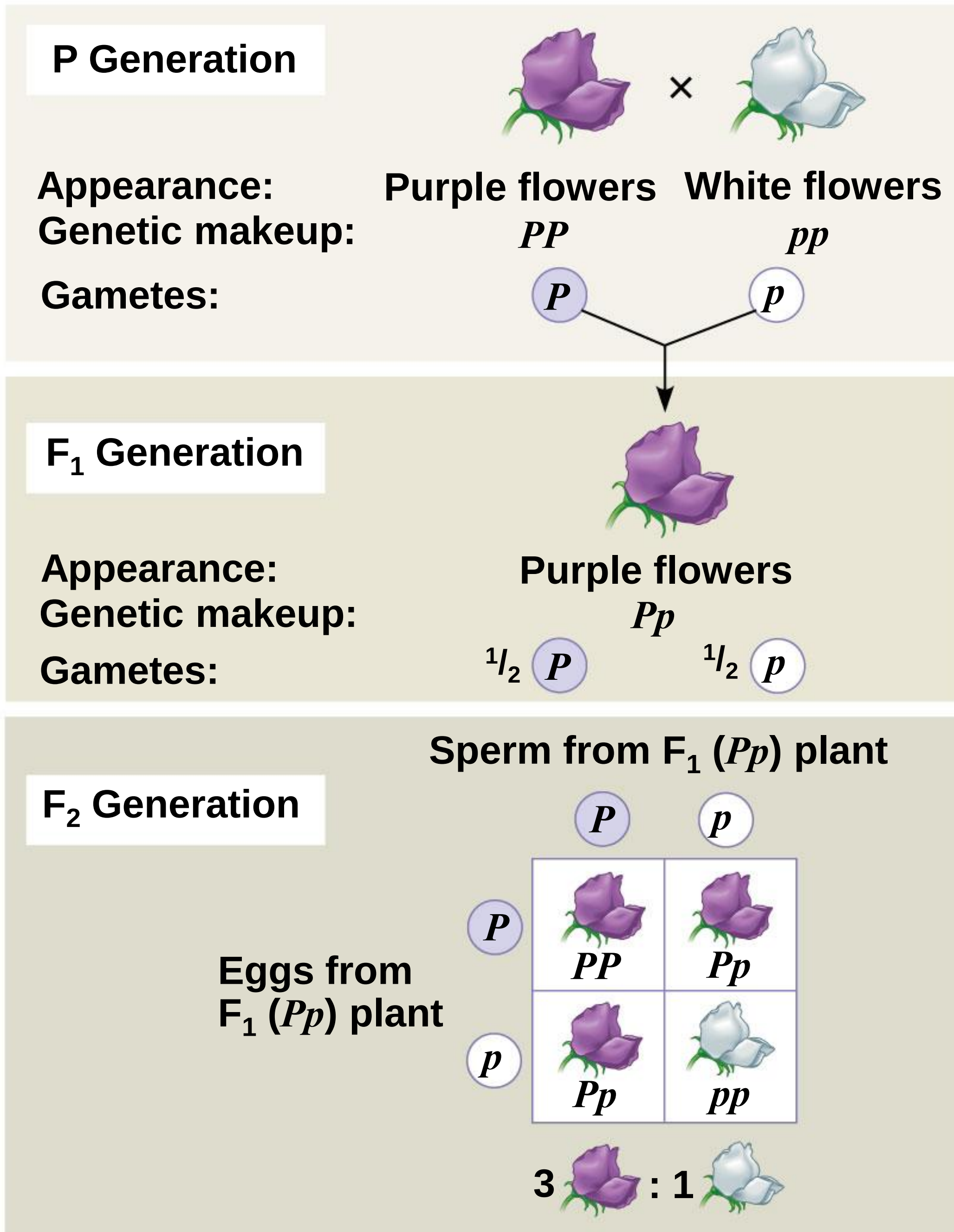

P Generation
×
Appearance:
Purple flowers
White flowers
Genetic makeup:
PP
pp
Gametes:
P
p

Figure 14.5-2



P Generation
×
Appearance:
Genetic makeup:
Gametes:
Purple flowers
PP
P
White flowers
pp
p
F1 Generation
Appearance:
Genetic makeup:
Gametes:
Purple flowers
Pp
1/2 P
1/2 p

Figure 14.5-3



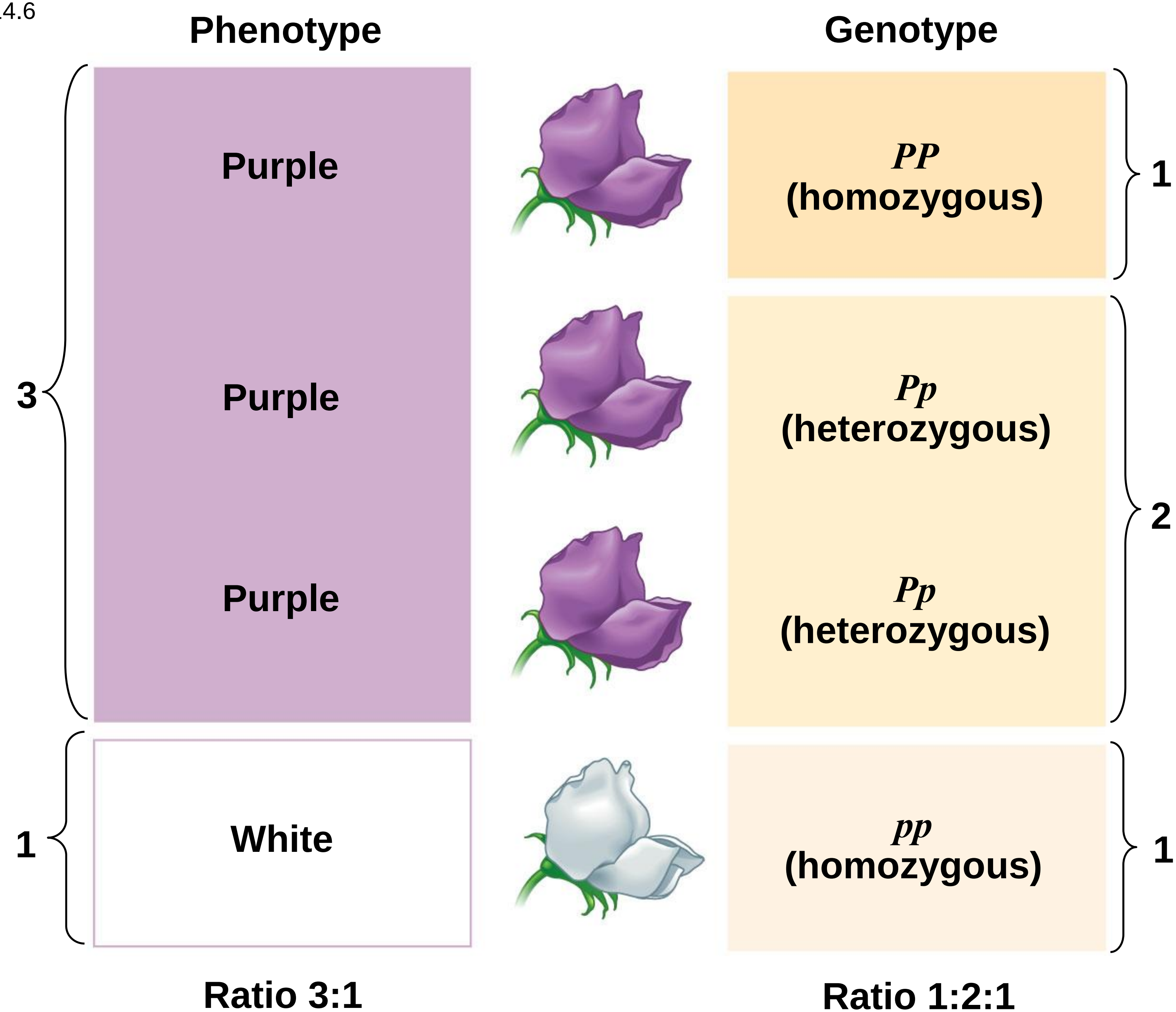
P Generation
×
Appearance:
Genetic makeup:
Gametes:
Purple flowers
PP
White flowers
pp
P
p
F1 Generation
Appearance:
Genetic makeup:
Gametes:
Purple flowers
Pp
1/2 P
1/2 p
F2 Generation
Sperm from F1 (Pp) plant
P
p
Eggs from
F1 (Pp) plant
P
p
PP
Pp
Pp
pp
3 : 1

Useful Genetic Vocabulary

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

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

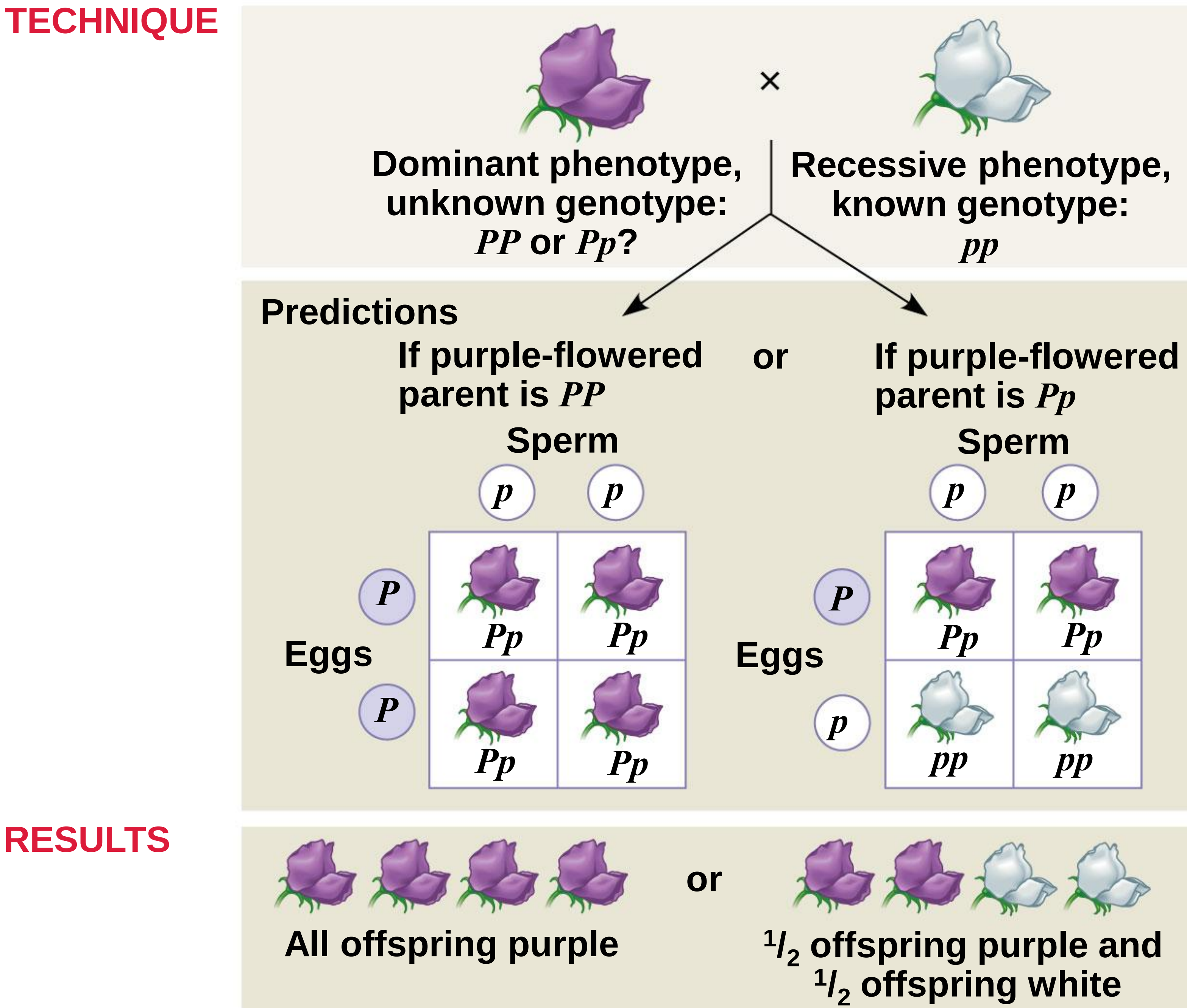
Figure 14.6


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

The Testcross

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

Figure 14.7

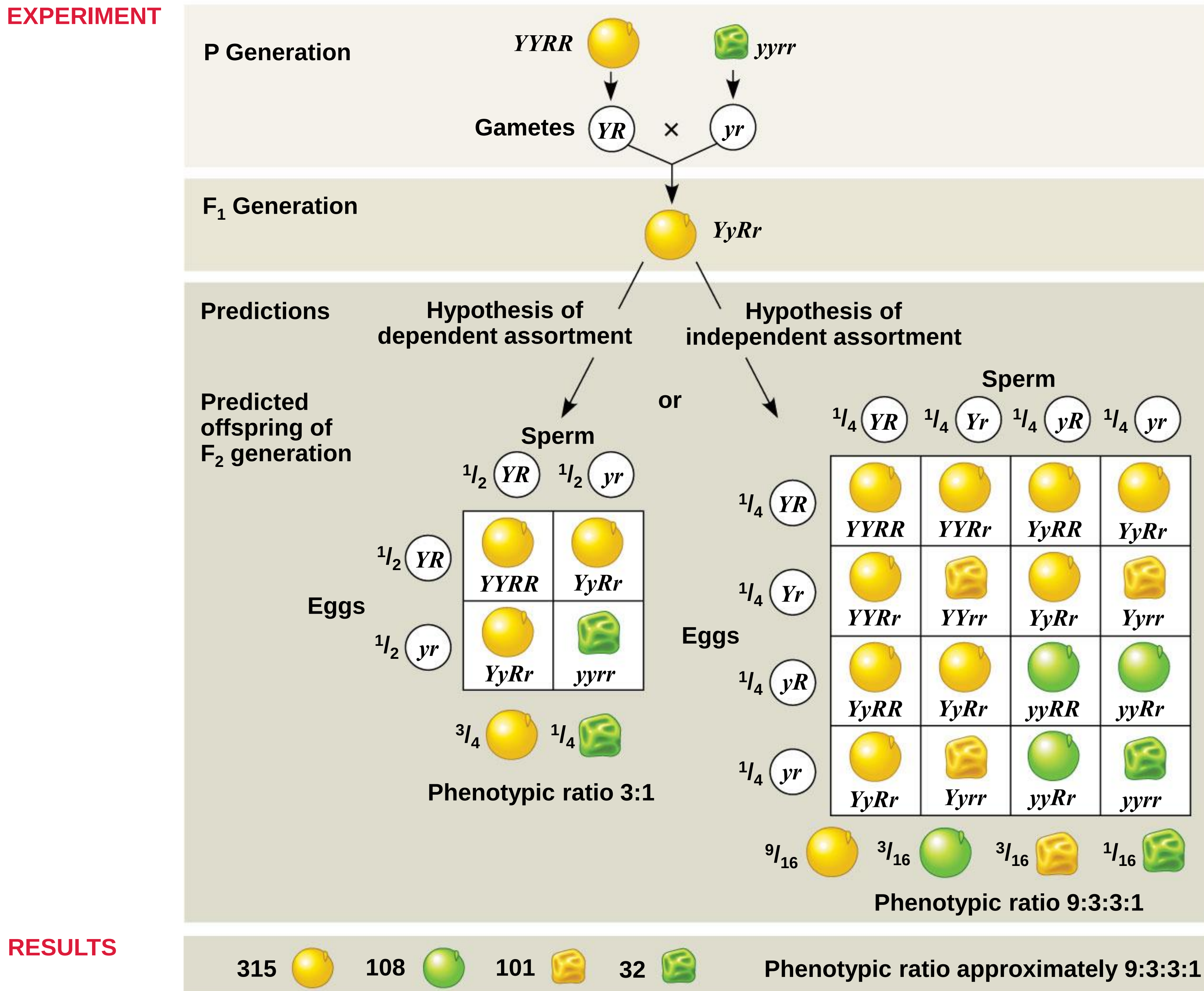

TECHNIQUE
×
Dominant phenotype, unknown genotype: PP or Pp?
Recessive phenotype, known genotype: pp
Predictions
If purple-flowered parent is PP
or
If purple-flowered parent is Pp
Sperm
p
p
Eggs
P
P
Pp
Pp
Pp
Pp
Sperm
p
p
Eggs
P
p
Pp
Pp
pp
pp
RESULTS
All offspring purple
or
1/2 offspring purple and 1/2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

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

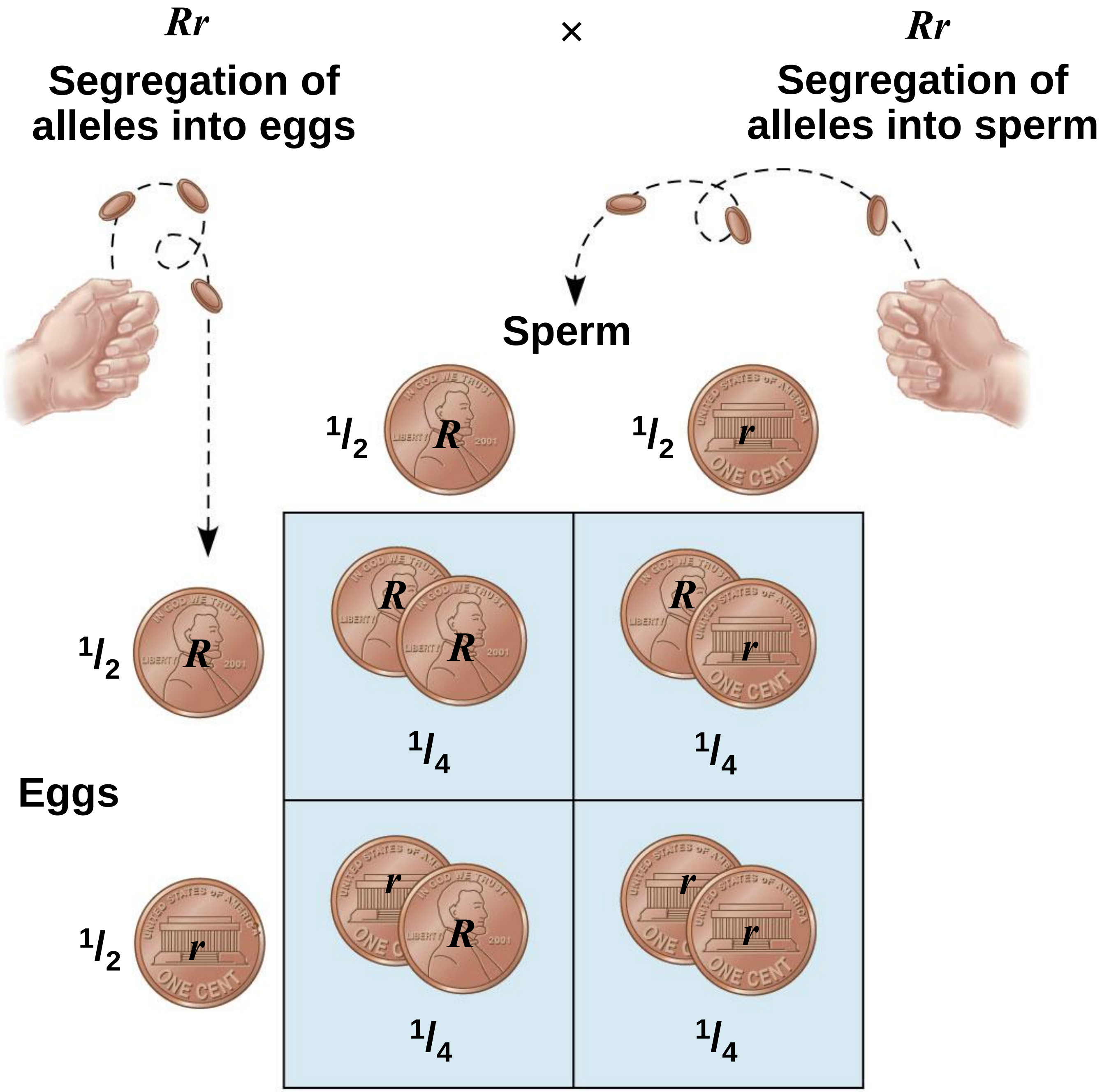
Concept 14.2: The laws of probability govern Mendelian inheritance

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

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
×
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

- 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

Solving Complex Genetics Problems with the Rules of Probability

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

Figure 14.UN01

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

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

Figure 14.UN02

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

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

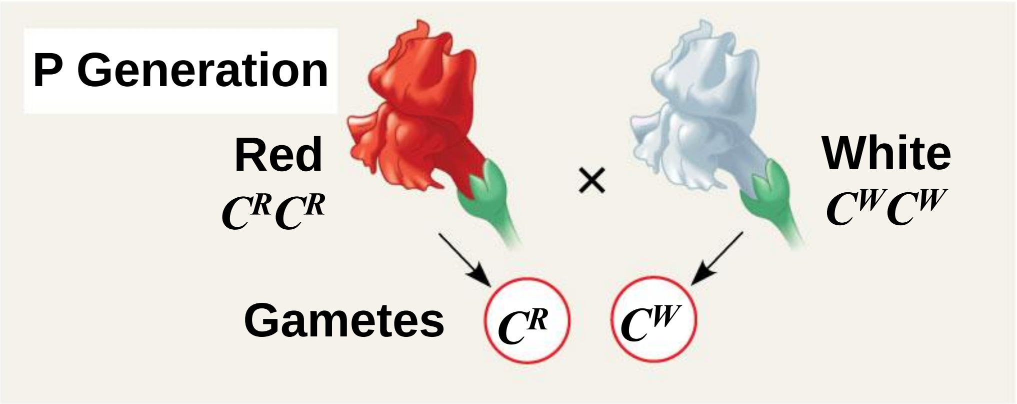


Figure 14.10-2

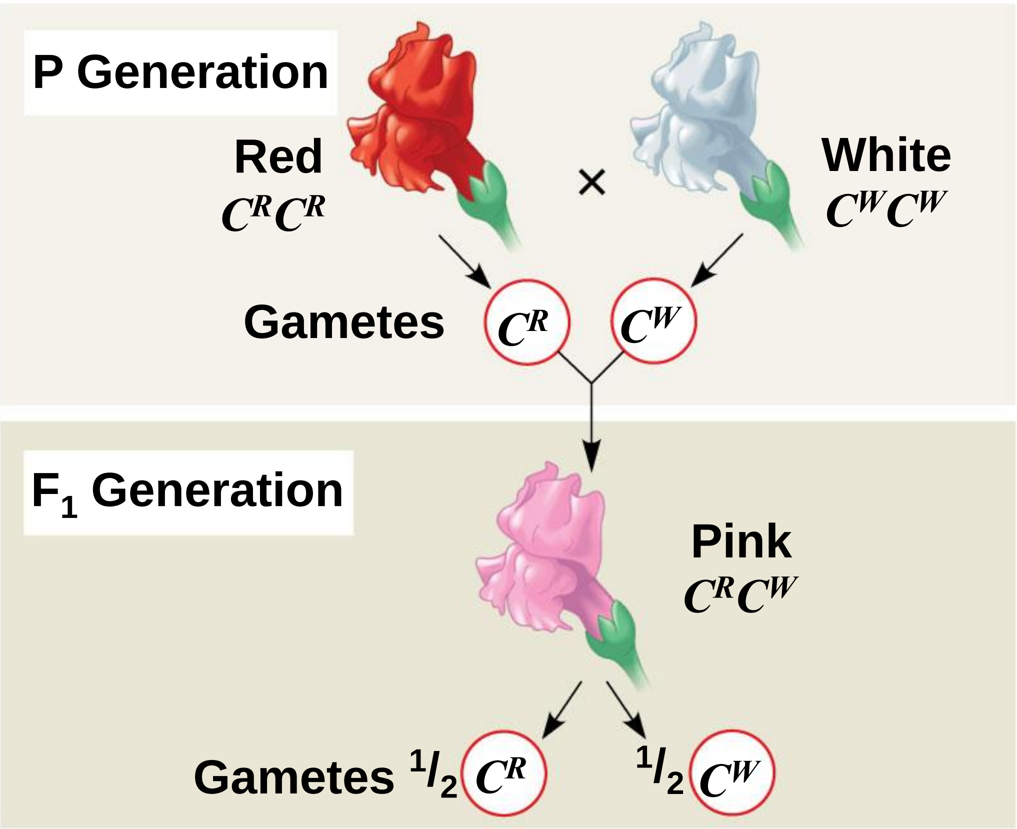
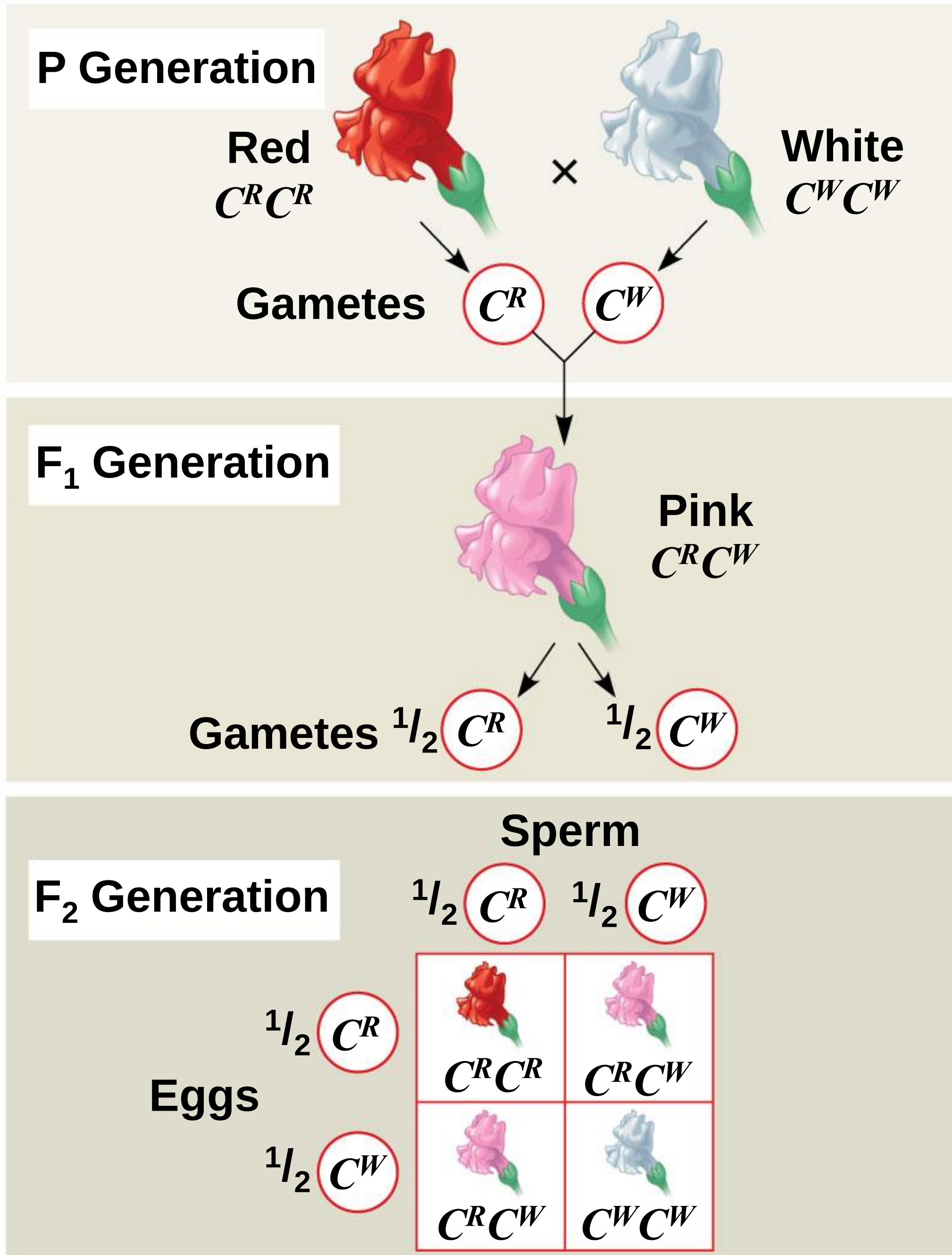


Figure 14.10-3



The Relation Between Dominance and Phenotype

- A dominant allele does not subdue a recessive allele; alleles don't interact that way
- Alleles are simply variations in a gene's nucleotide sequence
- For any character, dominance/recessiveness relationships of alleles depend on the level at which we examine the phenotype

- **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
- For example, one baby out of 400 in the United States is born with extra fingers or toes

- The allele for this unusual trait is dominant to the allele for the more common trait of five digits per appendage
- In this example, the recessive allele is far more prevalent than the population's dominant allele

Multiple Alleles

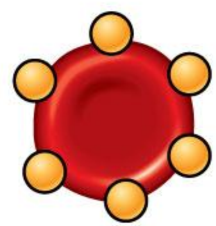
- Most genes exist in populations in more than two allelic forms
- For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme (I) that attaches A or B carbohydrates to red blood cells: I^A, I^B, and *i*.
- The enzyme encoded by the I^A allele adds 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^AI^A or I^Ai	I^BI^B or I^Bi	I^AI^B	ii
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

Pleiotropy

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

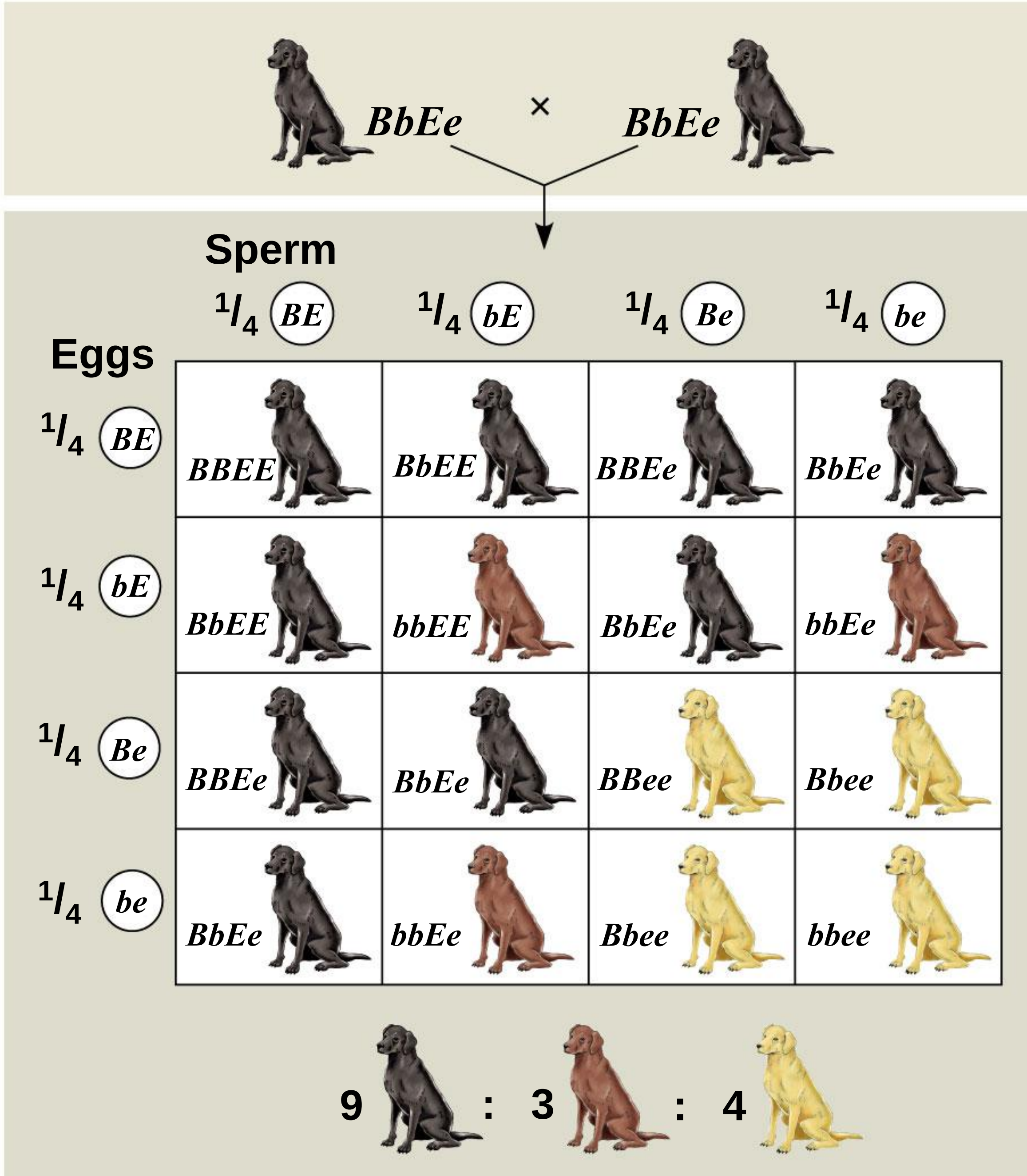
Extending Mendelian Genetics for Two or More Genes

- Some traits may be determined by two or more genes

Epistasis

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

Figure 14.12

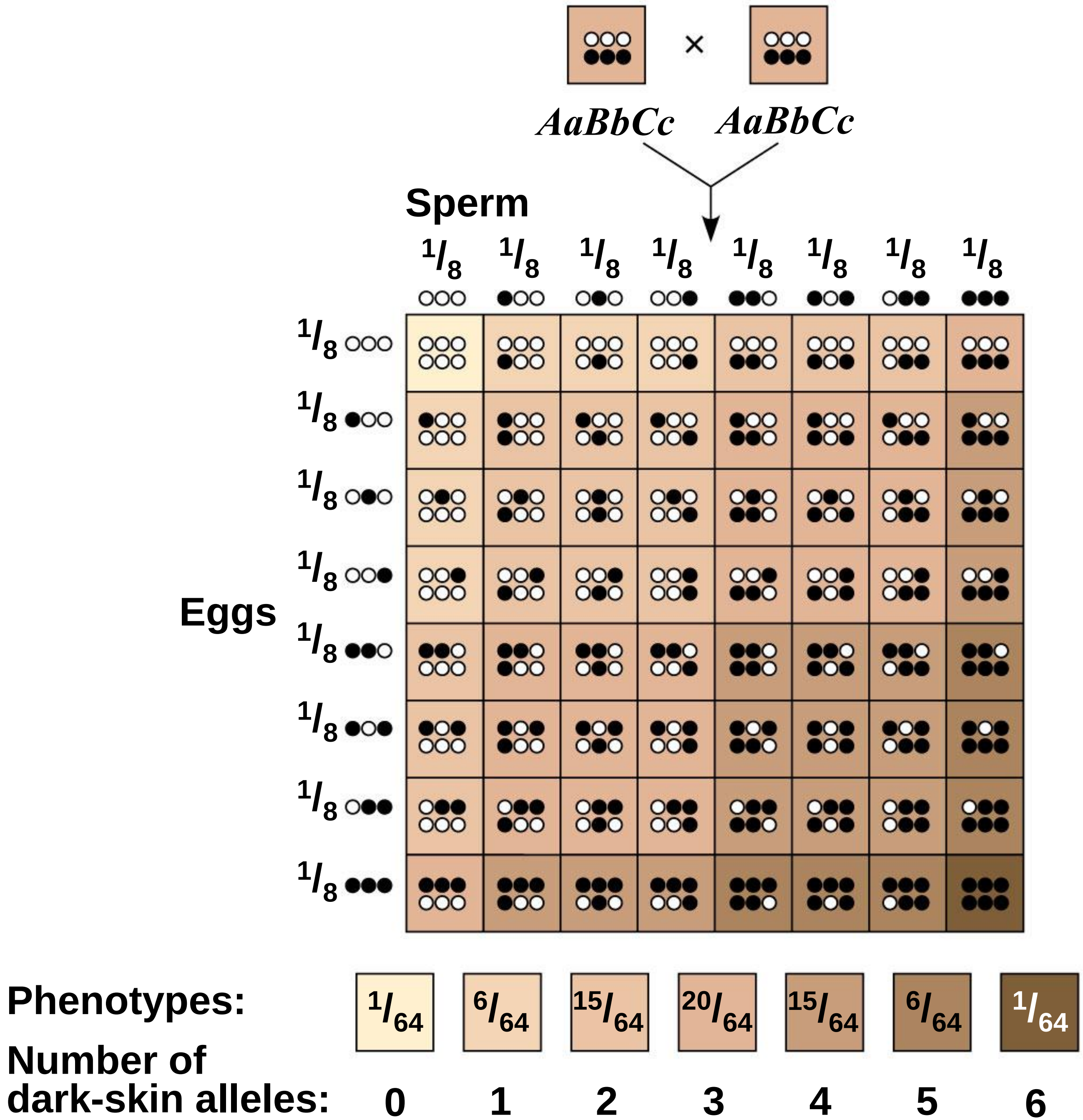


BbEe
×
BbEe
Sperm
1/4 BE
1/4 bE
1/4 Be
1/4 be
Eggs
1/4 BE
1/4 bE
1/4 Be
1/4 be
BBEE
BbEE
BBEe
BbEe
BbEE
bbEE
BbEe
bbEe
BBEe
BbEe
BBee
Bbee
BbEe
bbEe
Bbee
bbee
9 : 3 : 4

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
- Skin color in humans is an example of polygenic inheritance

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

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 **norm of reaction** is the phenotypic range of a genotype influenced by the environment
- For example, hydrangea flowers of the same genotype range from blue-violet to pink, depending on soil acidity

Figure 14.14

Figure 14.14a

Figure 14.14b

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

Integrating a Mendelian View of Heredity and Variation

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

Figure 14.UN03

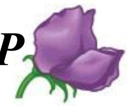
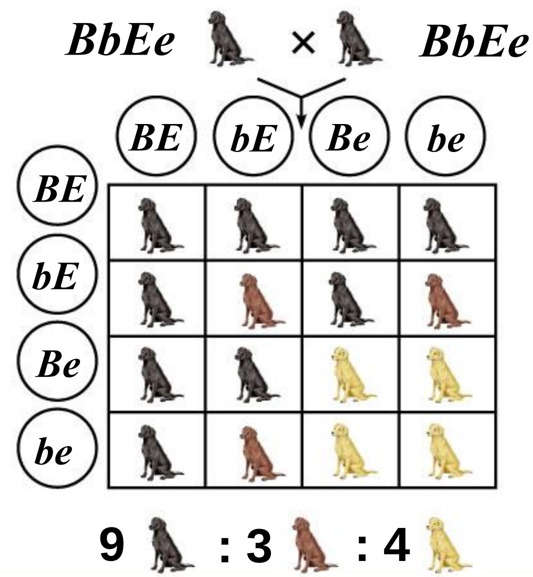
Relationship among alleles of a single gene	Description	Example
Complete dominance of one allele	**Heterozygous phenotype same as that of homo-zygous dominant**	PP  Pp 
Incomplete dominance of either allele	**Heterozygous phenotype intermediate between the two homozygous phenotypes**	 C^RC^R C^RC^W C^WC^W
Codominance	**Both phenotypes expressed in heterozygotes**	I^AI^B 
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**

Figure 14.UN04

Relationship among two or more genes	Description	Example
Epistasis	**The phenotypic expression of one gene affects that of another**	
Polygenic inheritance	**A single phenotypic character is affected by two or more genes**