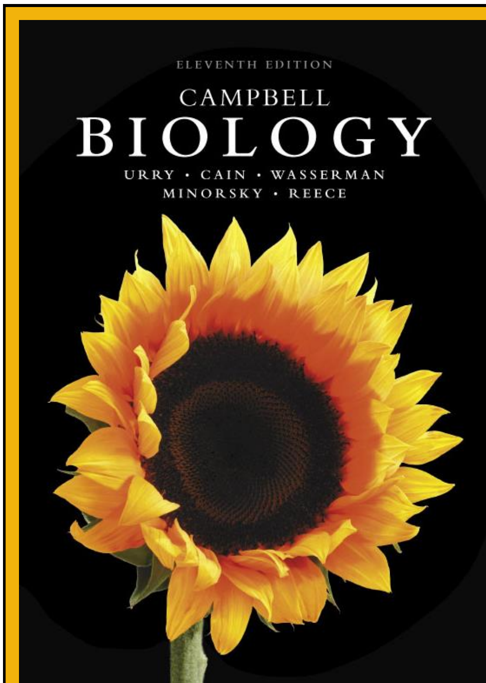

Chapter 14

Mendel and the Gene Idea

Lecture Presentations by
Nicole Tunbridge and
Kathleen Fitzpatrick

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Figure 14.1a

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

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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

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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

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- 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

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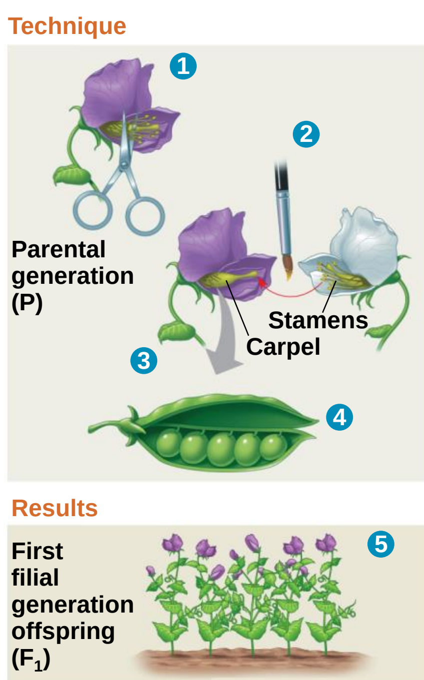
Figure 14.2



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- 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)

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- 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

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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

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Figure 14.3_1

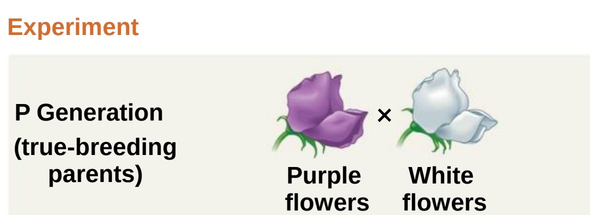

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Figure 14.3_2

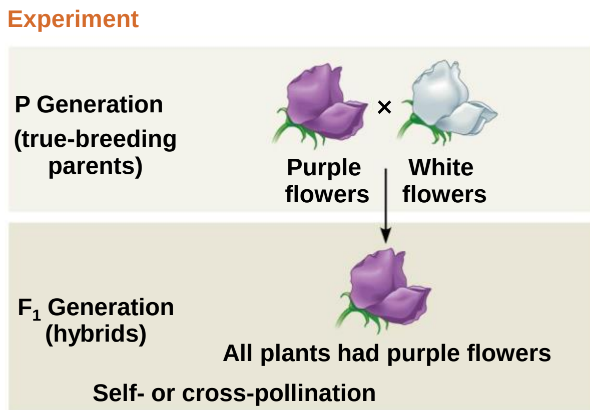

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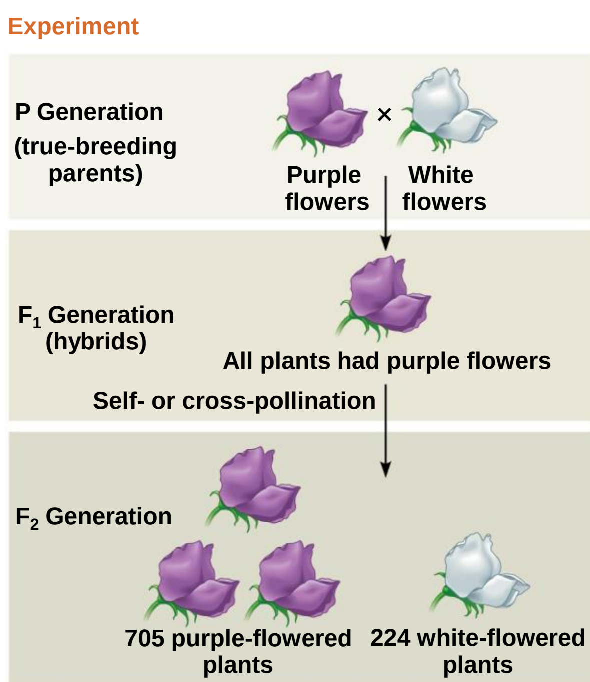
Figure 14.3_3



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- 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

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- 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

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

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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

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- 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

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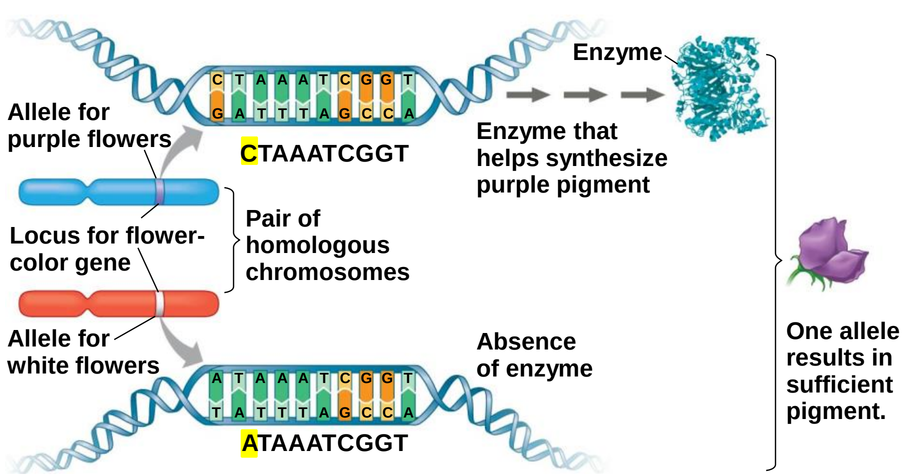
Figure 14.4



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- 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

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- 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

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- 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

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- 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

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Figure 14.5_1

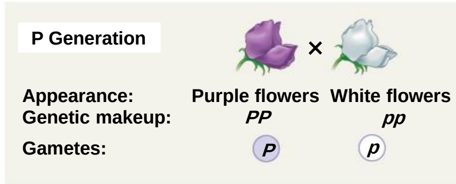

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Figure 14.5_2

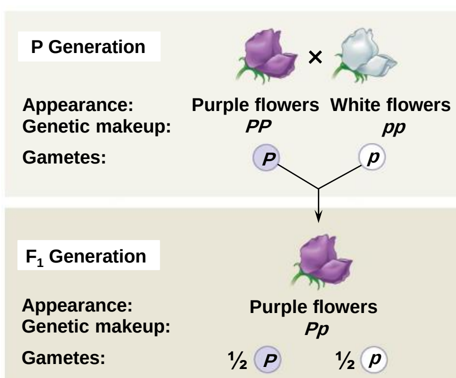

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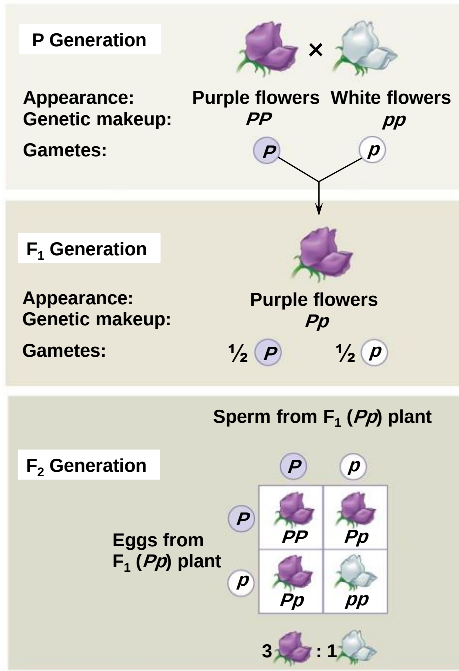
Figure 14.5_3



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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

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- 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

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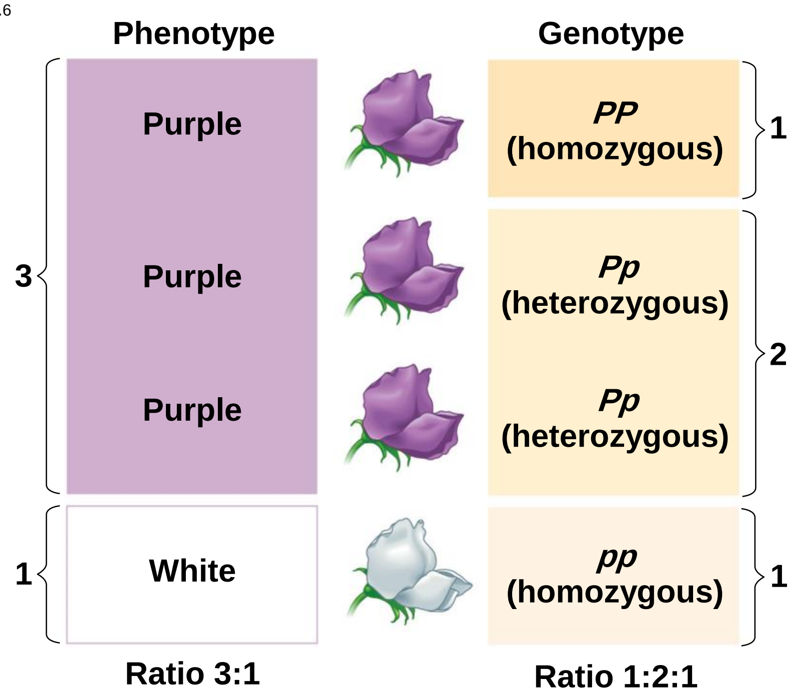
Figure 14.6



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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

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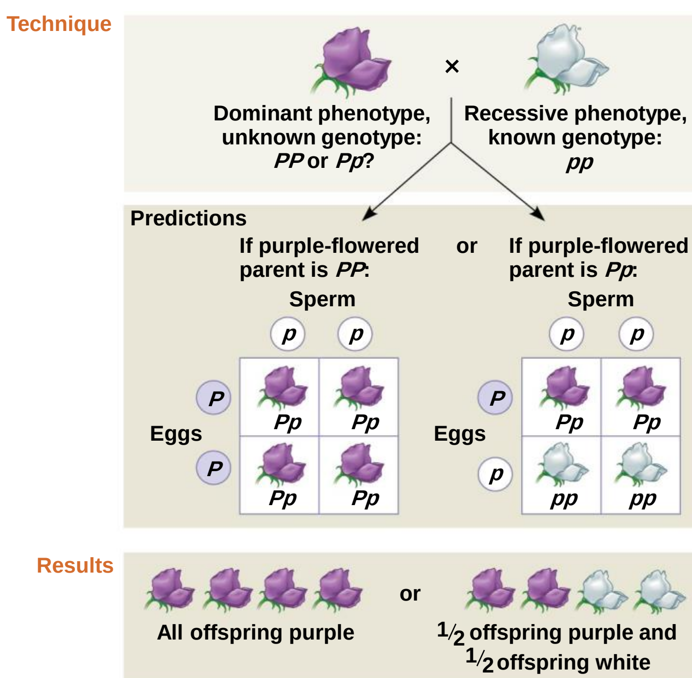
Figure 14.7



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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**

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- 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

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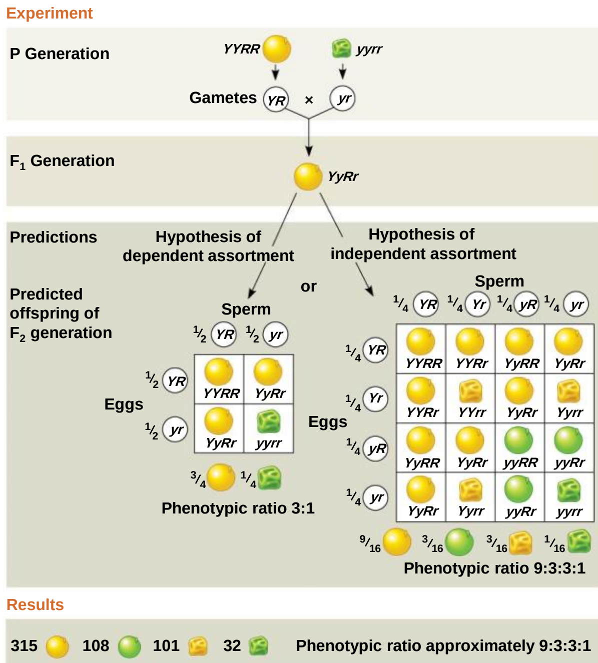
Figure 14.8



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- 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

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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

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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

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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

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Figure 14.10_1

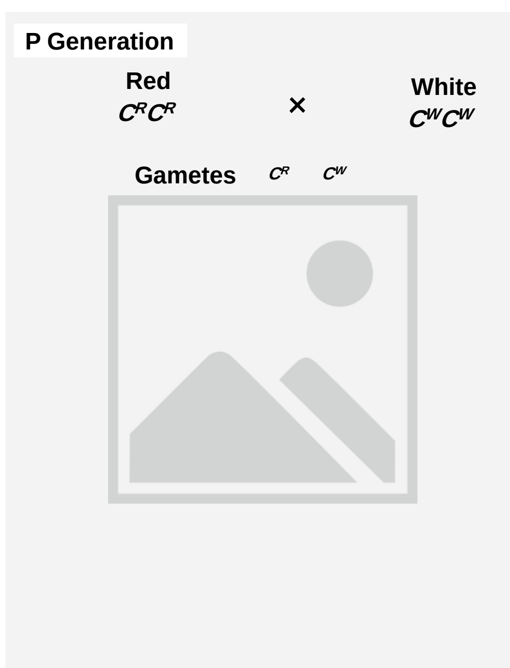

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Figure 14.10_2

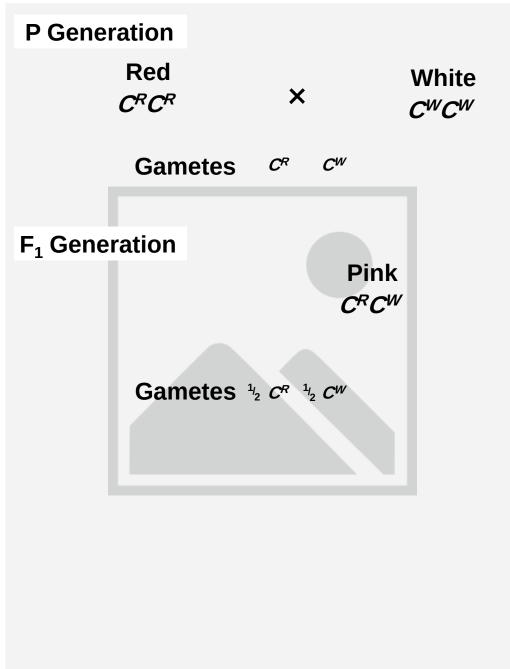

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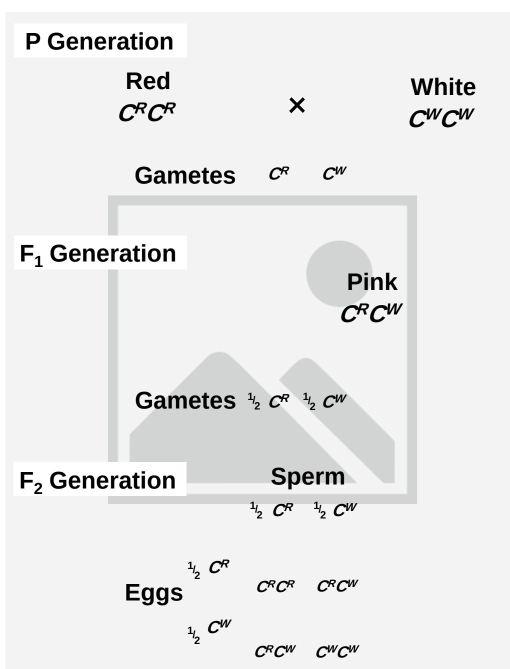
Figure 14.10_3



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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

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- **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

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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

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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

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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 with surface carbohydrates				
Phenotype (blood group)	A	B	AB	O

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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

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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

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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

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- 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

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Figure 14.12

BbEe × *BbEe*

Eggs \ Sperm	1/4 *BE*	1/4 *bE*	1/4 *Be*	1/4 *be*
1/4 *BE*	*BBEE*	*BbEE*	*BBEe*	*BbEe*
1/4 *bE*	*BbEE*	*bbEE*	*BbEe*	*bbEe*
1/4 *Be*	*BBEe*	*BbEe*	*BBee*	*Bbee*
1/4 *be*	*BbEe*	*bbEe*	*Bbee*	*bbee*

9 : 3 : 4

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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

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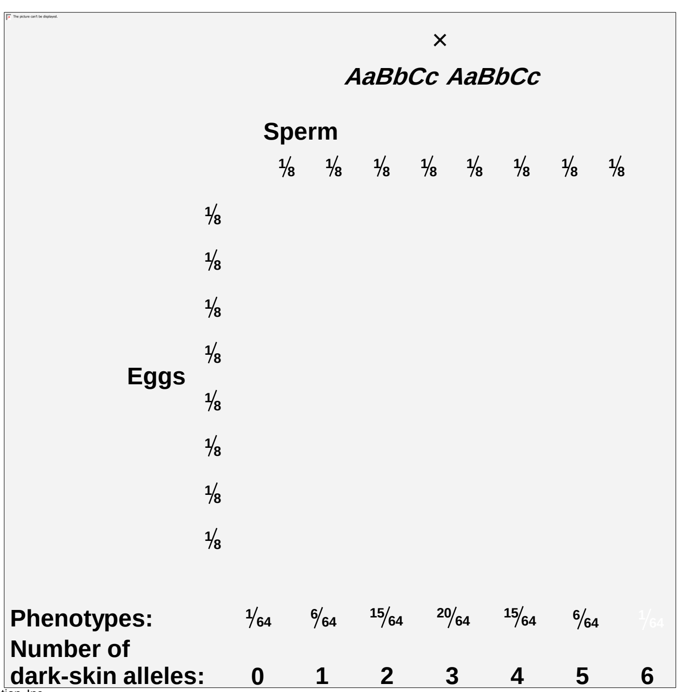
Figure 14.13



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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

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