CAMPBELL
BIOLOGY TENTH EDITION
Reece • Urry • Cain • Wasserman • Minorsky • Jackson

14

Mendel and the Gene Idea

Lecture Presentation by Nicole Tunbridge and Kathleen Fitzpatrick

Drawing from the Deck of Genes

- What 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)
- 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

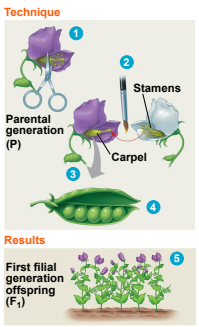
- Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments

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



- 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

Figure 14.3-2

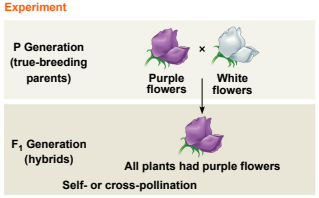
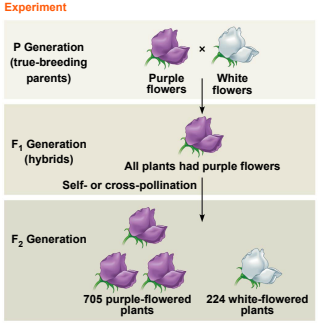


Figure 14.3-3



- 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
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
Flower position	Axial	×	Terminal	651:207	3.14:1
Stem length	Tall	×	Dwarf	787:277	2.84:1

Table 14.1a

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Table 14.1b

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Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
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Flower position	Axial	×	Terminal	651:207	3.14:1
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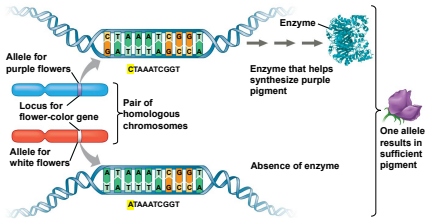
Mendel's Model

- 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

- 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



- 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
- 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 (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

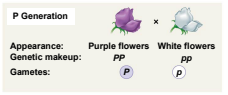


Figure 14.5-2

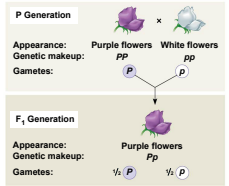
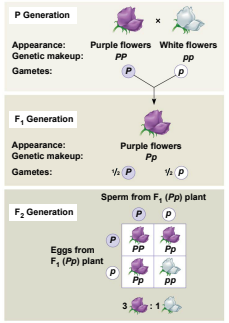


Figure 14.5-3

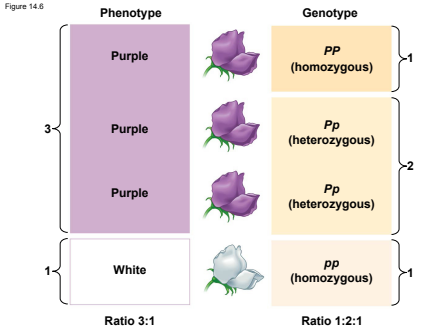


Useful Genetic Vocabulary

- An organism with two identical alleles for a character is **homozygous** for the gene controlling that character
- An organism that has two different alleles for a gene is **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

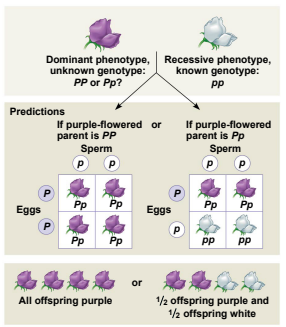
Figure 14.6



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



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

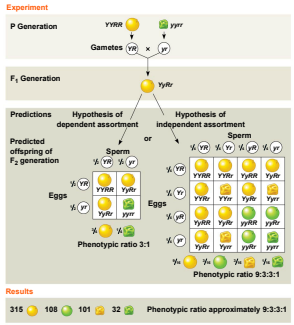


Figure 14.8a

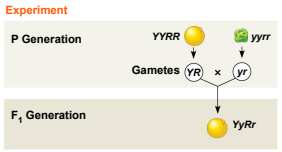
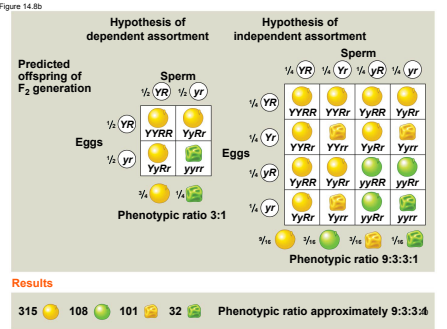


Figure 14.8b



- Using a dihybrid cross, Mendel developed the **law of independent assortment**
- It states that each pair of alleles segregates independently of each 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

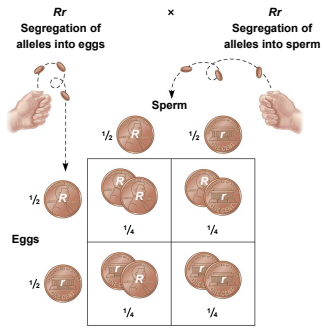
Concept 14.2: Probability laws govern Mendelian inheritance

- 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



- 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 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) = $\frac{1}{16}$

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

Figure 14.UN02

$ppyyRr$	$\frac{1}{4}$ (probability of pp) × $\frac{1}{2}$(yy) × $\frac{1}{2}$(Rr)	= $\frac{1}{16}$
$ppYyrr$	$\frac{1}{4}$ × $\frac{1}{2}$ × $\frac{1}{2}$	= $\frac{1}{16}$
$Ppyyrr$	$\frac{1}{2}$ × $\frac{1}{2}$ × $\frac{1}{2}$	= $\frac{2}{16}$
$PPyyrr$	$\frac{1}{4}$ × $\frac{1}{2}$ × $\frac{1}{2}$	= $\frac{1}{16}$
$ppyyrr$	$\frac{1}{4}$ × $\frac{1}{2}$ × $\frac{1}{2}$	= $\frac{1}{16}$
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

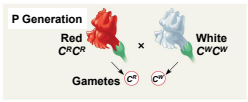


Figure 14.10-2

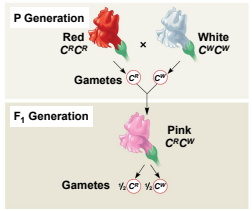
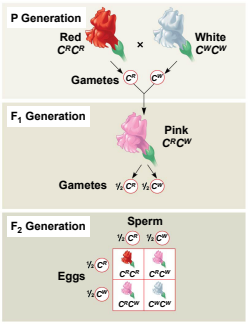


Figure 14.10-3



The 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

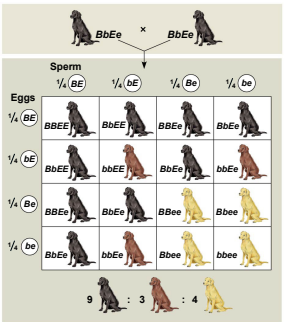
Extending Mendelian Genetics for Two or More Genes

- 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 *E* for color and *e* for no color) determines whether the pigment will be deposited in the hair

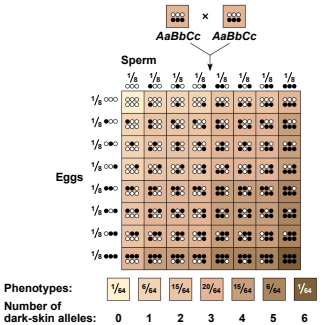
Figure 14.12



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



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

Figure 14.14a

Figure 14.14b

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

Concept 14.4: Many human traits follow Mendelian patterns of inheritance

- Humans are not good subjects for genetic research
  - Generation time is too long
  - Parents produce relatively few offspring
  - Breeding experiments are unacceptable
- However, basic Mendelian genetics endures as the foundation of human genetics

Pedigree Analysis

- A **pedigree** is a family tree that describes the interrelationships of parents and children across generations
- Inheritance patterns of particular traits can be traced and described using pedigrees

Figure 14.15

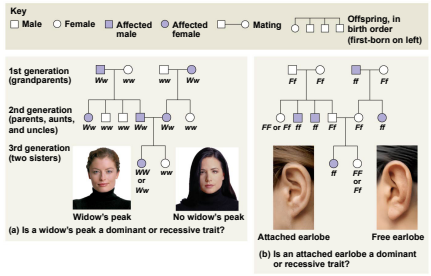


Figure 14.15a

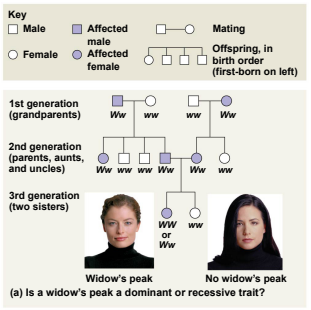


Figure 14.15aa

Widow's peak

Figure 14.15ab

No widow's peak

Figure 14.15b

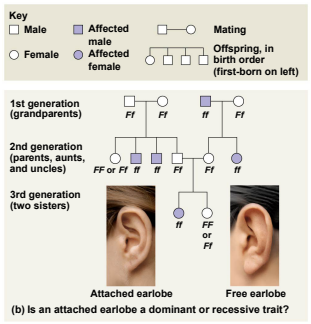
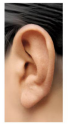


Figure 14.15ba

Attached earlobe

Figure 14.15bb



Free earlobe

- Pedigrees can also be used to make predictions about future offspring
- We can use the multiplication and addition rules to predict the probability of specific phenotypes

Recessively Inherited Disorders

- Many genetic disorders are inherited in a recessive manner
- These range from relatively mild to life-threatening

The Behavior of Recessive Alleles

- Recessively inherited disorders show up only in individuals homozygous for the allele
- **Carriers** are heterozygous individuals who carry the recessive allele but are phenotypically normal; most individuals with recessive disorders are born to carrier parents
- Albinism is a recessive condition characterized by a lack of pigmentation in skin and hair

Figure 14.16

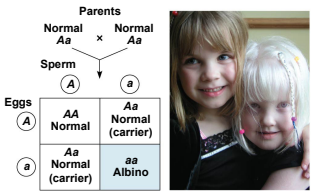


Figure 14.16a

- If a recessive allele that causes a disease is rare, then the chance of two carriers meeting and mating is low
- Consanguineous matings (i.e., matings between close relatives) increase the chance of mating between two carriers of the same rare allele
- Most societies and cultures have laws or taboos against marriages between close relatives

Cystic Fibrosis

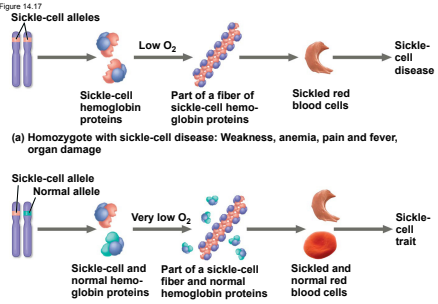
- **Cystic fibrosis** is the most common lethal genetic disease in the United States, striking one out of every 2,500 people of European descent
- The cystic fibrosis allele results in defective or absent chloride transport channels in plasma membranes leading to a buildup of chloride ions outside the cell
- Symptoms include mucus buildup in some internal organs and abnormal absorption of nutrients in the small intestine

Sickle-Cell Disease: A Genetic Disorder with Evolutionary Implications

- **Sickle-cell disease** affects one out of 400 African-Americans
- The disease is caused by the substitution of a single amino acid in the hemoglobin protein in red blood cells
- In homozygous individuals, all hemoglobin is abnormal (sickle-cell)
- Symptoms include physical weakness, pain, organ damage, and even paralysis

- Heterozygotes (said to have sickle-cell trait) are usually healthy but may suffer some symptoms
- About one out of ten African Americans has sickle-cell trait, an unusually high frequency
- Heterozygotes are less susceptible to the malaria parasite, so there is an advantage to being heterozygous in regions where malaria is common

Figure 14.17



Dominantly Inherited Disorders

- Some human disorders are caused by dominant alleles
- Dominant alleles that cause a lethal disease are rare and arise by mutation
- *Achondroplasia* is a form of dwarfism caused by a rare dominant allele

Figure 14.18

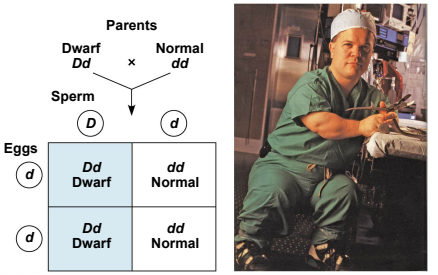
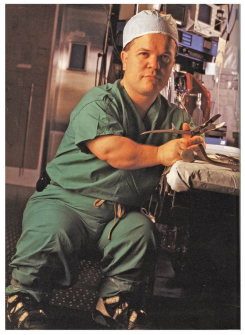


Figure 14.18a



- The timing of onset of a disease significantly affects its inheritance
- **Huntington's disease** is a degenerative disease of the nervous system
- The disease has no obvious phenotypic effects until the individual is about 35 to 40 years of age
- Once the deterioration of the nervous system begins the condition is irreversible and fatal

Multifactorial Disorders

- Many diseases, such as heart disease, diabetes, alcoholism, mental illnesses, and cancer have both genetic and environmental components
- No matter what our genotype, our lifestyle has a tremendous effect on phenotype

Genetic Testing and Counseling

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease

Counseling Based on Mendelian Genetics and Probability Rules

- Using family histories, genetic counselors help couples determine the odds that their children will have genetic disorders
- It is important to remember that each child represents an independent event in the sense that its genotype is unaffected by the genotypes of older siblings

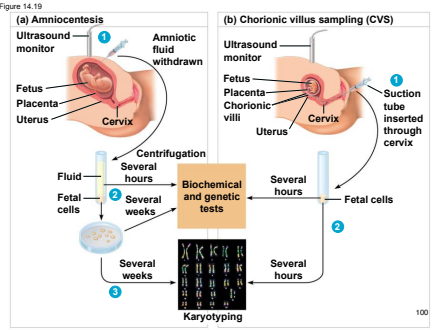
Tests for Identifying Carriers

- For a growing number of diseases, tests are available that identify carriers and help define the odds more accurately
- The tests enable people to make more informed decisions about having children
- However, they raise other issues, such as whether affected individuals fully understand their genetic test results

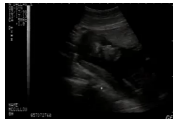
Fetal Testing

- In **amniocentesis**, the liquid that bathes the fetus is removed and tested
- In **chorionic villus sampling** (**CVS**), a sample of the placenta is removed and tested
- Other techniques, such as *ultrasound* and *fetoscopy*, allow fetal health to be assessed visually in utero

Figure 14.19



Video: Ultrasound of Human Fetus



Newborn Screening

- Some genetic disorders can be detected at birth by simple tests that are now routinely performed in most hospitals in the United States
- One common test is for phenylketonuria (PKU), a recessively inherited disorder that occurs in one of every 10,000–15,000 births in the United States

Figure 14.UN03a

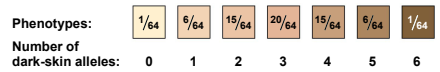


Figure 14.UN03b

Figure 14.UN04

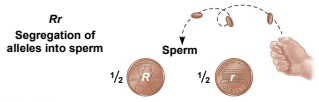


Figure 14.UN05

Relationship among alleles of a single gene	Description	Example
Complete dominance of one allele	Heterozygous phenotype same as that of homozygous 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 population some genes have more than two alleles	ABO blood group alleles I^A, I^B, i
Pleiotropy	One gene affects multiple phenotypic characters	Sickle-cell disease

Figure 14.UN06

Relationship among two or more genes	Description	Example
Epistasis	The phenotypic expression of one gene affects the expression of another gene	*BbEe* × *BbEe*; 9 : 3 : 4
Polygenic inheritance	A single phenotypic character is affected by two or more genes	*AaBbCc* × *AaBbCc*

Figure 14.UN07

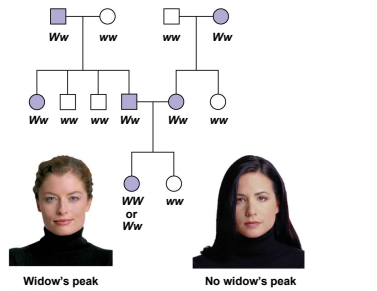


Figure 14.UN08

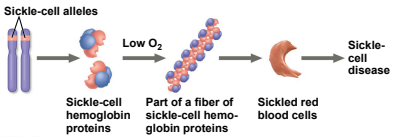


Figure 14.UN09

Figure 14.UN10

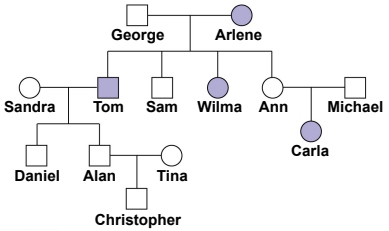


Figure 14.UN11