Chapter 14

Mendel and the Gene Idea

Overview: Drawing from the Deck of Genes

- **What genetic principles account for the passing of traits from parents to offspring?**
- **The _____is the idea that genetic material from the two parents blends together (like blue and yellow paint blend to make green)**

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

Mendel used the scientific approach to identify two laws of inheritance

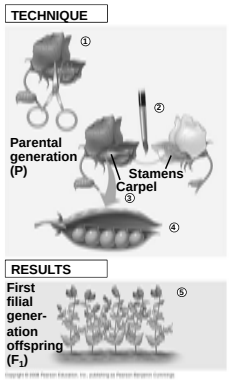
- **Mendel discovered the basic principles of heredity by breeding _____in carefully planned experiments**

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 _____**
- **Mating of plants can be _____**
- **Each pea plant has sperm-producing organs (_____) and egg-producing organs (_____)**
- **Cross-pollination (fertilization between different plants) can be achieved by dusting one plant with pollen from another**

Fig. 14-2



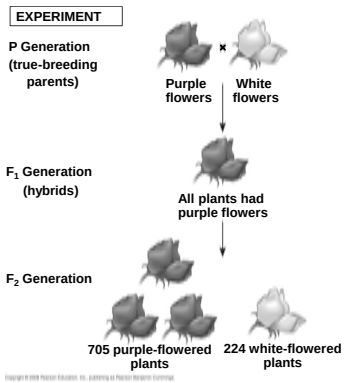
- **Mendel chose to track only those characters that varied in an either-or manner**
- **He also used varieties that were _____ (plants that produce offspring of the same variety when they self-pollinate)**

- **In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called _____**
- **The true-breeding parents are the P generation**
- **The hybrid offspring of the P generation are called the F_1 generation**
- **When ____individuals self-pollinate, the ____ 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 _____**
- **When Mendel crossed the F_1 hybrids, many of the F_2 plants had purple flowers, but some had _____**
- **Mendel discovered a ratio of about three to one, purple to white flowers, in the F_2 generation**

Fig. 14-3-3



- **Mendel reasoned that only the purple flower factor was affecting flower color in the ____hybrids**
- **Mendel called the purple flower color a _____ trait and the white flower color a _____trait**
- **Mendel observed the same pattern of inheritance in six other pea plant characters, each represented by two traits**
- **What Mendel called a _____is what we now call a gene**

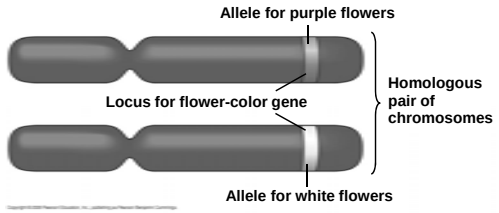
Table 14-1

Mendel's Model

- Mendel developed a hypothesis to explain the _____ 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 _____

- The first concept is that alternative versions of _____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 _____ on a specific chromosome

Fig. 14-4



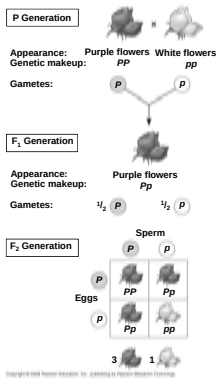
- The second concept is that for each character an organism inherits two _____, one from each parent
- Mendel made this deduction without knowing about the role of chromosomes
- The two alleles at a locus on a chromosome may be identical, as in the true-breeding plants of Mendel's P generation
- Alternatively, the two alleles at a locus may differ, as in the _____hybrids

- The third concept is that if the two alleles at a locus differ, then one (the _____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 _____

- The fourth concept, now known as the _____, states that 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 somatic cells of an organism
- This _____ of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis

- Mendel's segregation model accounts for the _____ratio he observed in the F_2 generation of his numerous crosses
- The possible combinations of sperm and egg can be shown using a _____, 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 _____ allele

Fig. 14-5-3

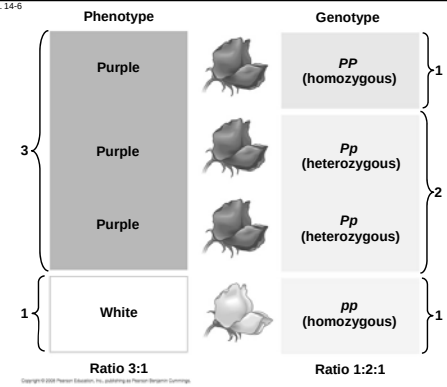


Useful Genetic Vocabulary

- An organism with two identical alleles for a character is said to be _____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, _____are not true-breeding

- Because of the different effects of dominant and recessive alleles, an organism's traits do not always reveal its _____
- 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, ___ and ___ plants have the same phenotype (purple) but different genotypes

Fig. 14-6



The Testcross

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

Fig. 14-7a

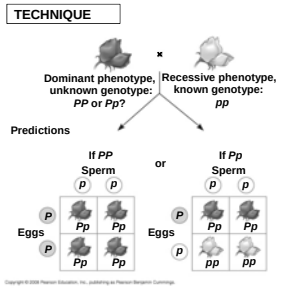
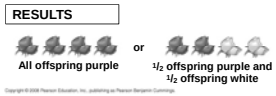


Fig. 14-7b

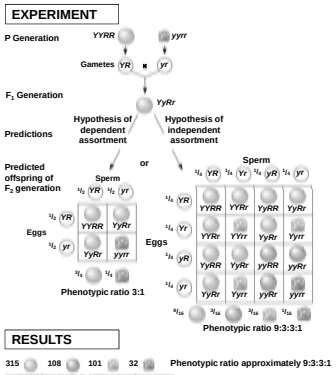


The Law of Independent Assortment

- **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 _____**

- **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 _____ 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**

Fig. 14-8



- **Using a dihybrid cross, Mendel developed the _____**
- **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**
- **Genes located near each other on the same chromosome tend to be inherited _____**

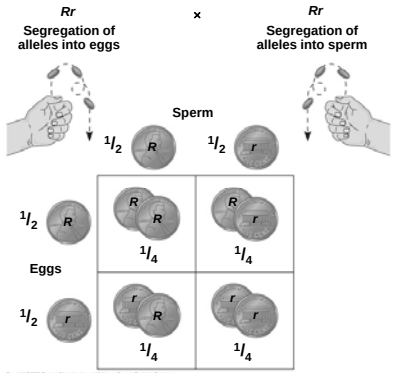
The laws of probability govern Mendelian inheritance

- Mendel's laws of segregation and independent assortment reflect the rules of_____
- When tossing a coin, the outcome of one toss has no impact on the outcome of the _____
- 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 _____allele

Fig. 14-9



- The _____states that the probability that any one of two or more exclusive events will occur is calculated by adding together their individual probabilities
- The _____ 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 _____
- 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 together

Fig. 14-UN1

ppyyRr	¼ (probability of *pp*) × ½ (*yy*) × ½ (*Rr*)	= 1/16
ppYyrr	¼ × ½ × ½	= 1/16
Ppyyrr	½ × ½ × ½	= 2/16
PPyyrr	¼ × ½ × ½	= 1/16
ppyyrr	¼ × ½ × ½	= 1/16
Chance of *at least two* recessive traits		= 5/16 or 3/8

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 _____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

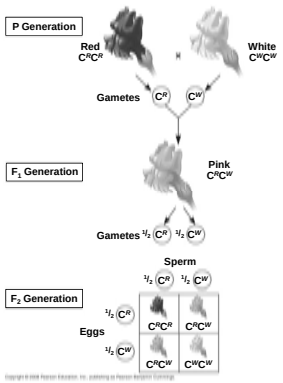
Extending Mendelian Genetics for a Single Gene

- Inheritance of characters by a single gene may deviate from simple _____ 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_____
- In _____ dominance, the phenotype of F_1 hybrids is somewhere between the phenotypes of the two parental varieties
- In _____, two dominant alleles affect the phenotype in separate, distinguishable ways

Fig. 14-10-3



The Relation Between Dominance and Phenotype

- A dominant allele does not subdue a recessive allele; alleles don't interact
- 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 _____

- _____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 _____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 _____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 _____ 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 _____

Fig. 14-11

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

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

Genotype	Red blood cell appearance	Phenotype (blood group)
I^AI^A or I^Ai		A
I^BI^B or I^Bi		B
I^AI^B		AB
ii		O

(b) Blood group genotypes and phenotypes

Pleiotropy

- Most genes have multiple _____ 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

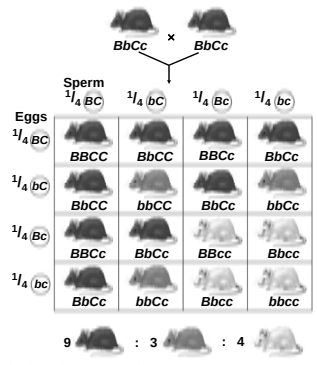
Extending Mendelian Genetics for Two or More Genes

- Some traits may be determined by two or more _____

Epistasis

- In epistasis, a gene at one locus alters the phenotypic expression of a gene at a _____
- For example, in mice 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

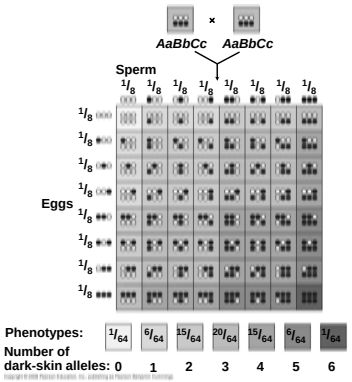
Fig. 14-12



Polygenic Inheritance

- Quantitative characters are those that vary in the population along a continuum
- Quantitative variation usually indicates polygenic inheritance, an _____effect of two or more genes on a single phenotype
- Skin color in humans is an example of polygenic inheritance

Fig. 14-13



Nature and Nurture: The Environmental Impact on Phenotype

- Another departure from Mendelian genetics arises when the phenotype for a character depends on _____ 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 _____

- Norms of reaction are generally broadest for polygenic characters
- Such characters are called _____ 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 _____

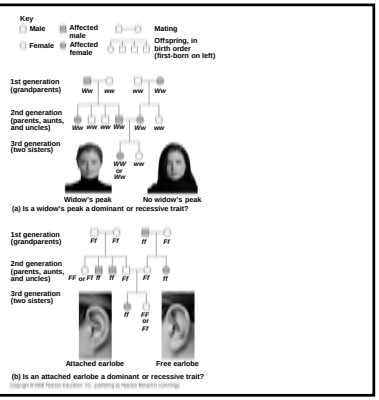
Many human traits follow Mendelian patterns of inheritance

- Humans are not good subjects for genetic research
  - _____ time is too long
  - Parents produce relatively _____
  - _____ 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 _____
- Inheritance patterns of particular traits can be traced and described using pedigrees

Fig. 14-15



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

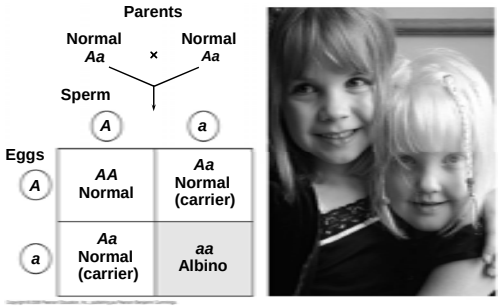
Recessively Inherited Disorders

- Many genetic disorders are inherited in a _____ manner

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 _____(i.e., pigmented)**
- **Albinism is a recessive condition characterized by a lack of pigmentation in skin and hair**

Fig. 14-16



- **If a recessive allele that causes a disease is rare, then the chance of two carriers meeting and mating is low**
- **_____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 _____in the United States,striking one out of every 2,500 people of European descent**
- **The cystic fibrosis allele results in defective or absent _____ transport channels in plasma membranes**
- **Symptoms include mucus buildup in some internal organs and abnormal absorption of nutrients in the small intestine**

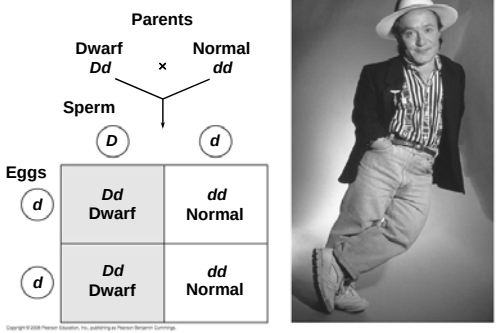
Sickle-Cell Disease

- **Sickle-cell disease affects one out of _____ African-Americans**
- **The disease is caused by the substitution of a single amino acid in the hemoglobin protein in red blood cells**
- **Symptoms include physical weakness, pain, organ damage, and even paralysis**

Dominantly Inherited Disorders

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

Fig. 14-17



Huntington's Disease

- Huntington's disease is a _____disease of the nervous system
- The disease has no obvious phenotypic effects until the individual is about 35 to 40 years of age

Multifactorial Disorders

- Many diseases, such as heart disease and cancer, have both genetic and _____
- Little is understood about the genetic contribution to most multifactorial diseases

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_____that their children will have genetic disorders

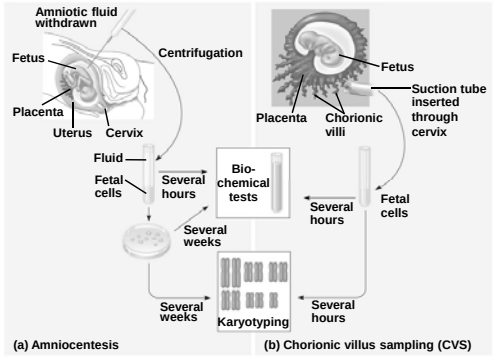
Tests for Identifying Carriers

- For a growing number of diseases, tests are available that identify _____and help define the odds more accurately

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 _____ and *fetoscopy*, allow fetal health to be assessed visually in utero**

Fig. 14-18



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**

Fig. 14-UN2

Degree of dominance	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	Heterozygotes: Both phenotypes expressed	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

Fig. 14-UN3

Relationship among genes	Description	Example
Epistasis	One gene affects the expression of another	*BbCc* × *BbCc*; *BC bC Bc bc*; 9 : 3 : 4
Polygenic inheritance	A single phenotypic character is affected by two or more genes	*AaBbCc* × *AaBbCc*

Fig. 14-UN4

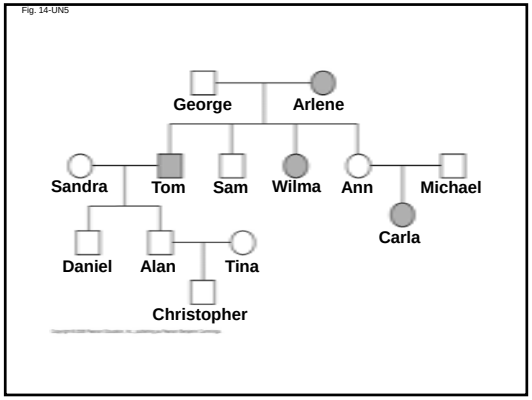


Fig. 14-UN5
George
Arlene
Sandra
Tom
Sam
Wilma
Ann
Michael
Carla
Daniel
Alan
Tina
Christopher