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Chapter 14

Mendelian Genetics

PowerPoint® Lecture Presentations for

Biology

Eighth Edition

Neil Campbell and Jane Reece

Lectures by Chris Romero, updated by Erin Barley with contributions from Joan Sharp

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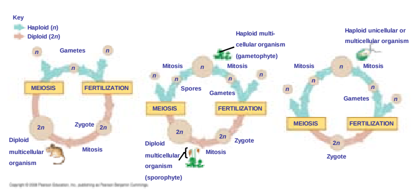
Fig. 13-6

Sexual Life Cycles



Animals   Plants and some algae   Most fungi and some protists

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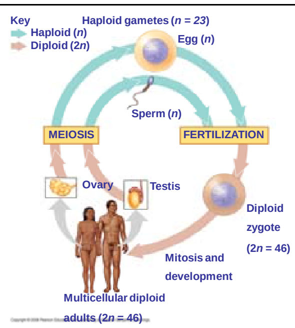
Fig. 13-5



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The BIG PICTURE of Meiosis

- One Interphase just like Mitosis
  - G1, S-Phase, G2
  - diploid 46 chromosomes replicate to form 92 sisters joined in pairs
- Followed by Two Cell Divisions instead of One
  - In meiosis I, homologous chromosomes separate resulting in two haploid daughters with 46 sisters joined in 23 pairs
    - it is called the ***reductional division***
  - In meiosis II, sister chromatids separate resulting in four haploid daughters with 23 unjoined chromosomes (much like mitosis)
    - it is called the ***equational division***

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Genetics is the study of heredity and variation....

- **Genetic inheritance in sexually reproducing organisms**
  - One gamete from mom, one from dad
  - One set of autosomes from mom, one from dad
  - One allele at each autosomal locus from mom, one from dad
  - One X from mom, one X or one Y from dad
- **Genetic variation results from mutation and mixing**
  - Everyday mutation produces altered allele sequences
  - Crossover mutation mixes DNA from mom and dad chromatids
  - Independent assortment of grandparent chromosomes in meiosis
  - Random fertilization between gametes gives 70 trillion options

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Key Terms and Definitions I

- Genome ____________________________________
- Chromosome _________________________________
- Gene ______________________________________
- Locus _____________________________________
- Allele _____________________________________
- Genotype ___________________________________
- Phenotype __________________________________

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Key Terms and Definitions II

- Genotype vs. Phenotype
  - gene level ___________________________________
  - visual level _________________________________
  - protein level ________________________________
  - physiological level ___________________________
  - organismal level ______________________________

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How might knowing about the transmission of genes (and their resultant proteins) be useful?

- Explain the observed phenotypes around us
- Understand the basic causes of genetic disorders
- Predict disease and, thus, potentially prevent it
- Make genetic changes to eradicate disease?
- Genetically modify other organisms for our use?
- Make changes to strengthen our species?

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Gregor Mendel started the study of genetics in a world without any understanding of genes!

- He was trying to determine the principles that account for the passing of traits from parents to offspring.
- There were two competing ideas at the time:
  - 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
- He was a University of Vienna trained scientist working in one of the few places where science was practiced at the time (roughly the time of the American Civil War).
  - Mendel was a really smart, well-trained scientist

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Key Terms and Definitions III

- Character ________________________________
- Trait ____________________________________
- True-breeding ____________________________
- Crossing _________________________________
- Hybridization _____________________________
- P Generation ______________________________
- F1 Generation _____________________________
- F2 Generation _____________________________

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Key Terms and Definitions IV

- Homozygous _______________________________
- Heterozygous ______________________________
- Dominant allele ____________________________
- Recessive allele ____________________________

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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. Why not?

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Mendel was both really smart and a little lucky!

- He picked the simplest genetic experiment possible
  - Plant characters with only two traits
  - Single genes produce those traits
  - Thus, only two alleles for each of those genes
  - One allele was perfectly dominant, the other perfectly recessive
- He took it out to two generations
  - He crossed the parental generation
  - Then self-pollinated the off-spring to look at what genes they got
  - Only the second generation of off-spring shows the key principles

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Table 14-1

All of his targeted characters were either/or

(--*KISS*--)

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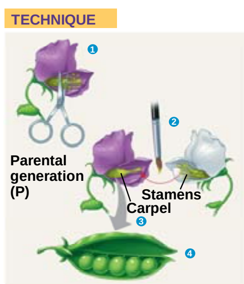
Fig. 14-2a



Purple and white plants both were true-breeding

1. Removed stamens from purple flowers
2. Transferred pollen from stamens of white flower to carpel of purple flower
3. Pollinated carpel matured into pod
4. Planted seeds (plant embryo) from pod

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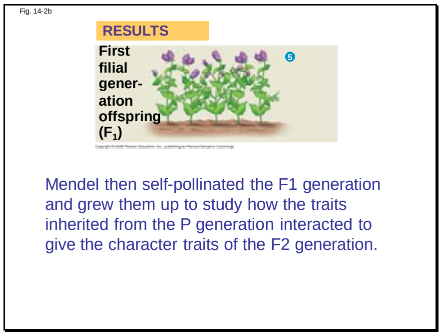

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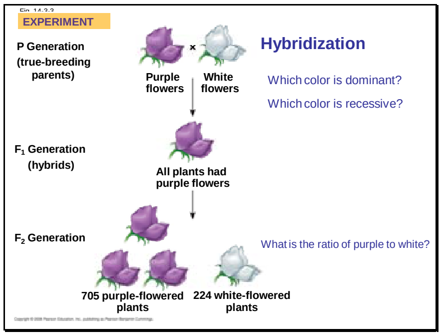

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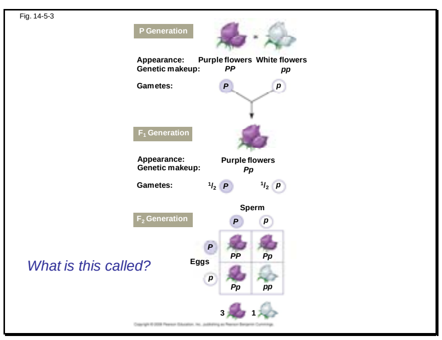

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Mendel's Model in the Terms of Modern Genetics

1. Alternative versions of **genes** account for variations in inherited characters
2. For each character an organism inherits **two alleles**, one from each parent
3. 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
4. the **law of segregation**, states that the two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes

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Fig. 13-7-3

Mendel didn't know about chromosomes but he showed that what you got from mom and dad segregated in the gametes

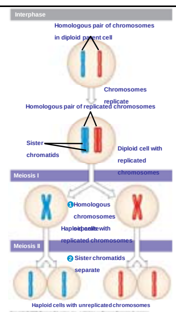

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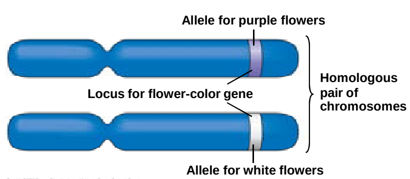
Fig. 14-4



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- 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

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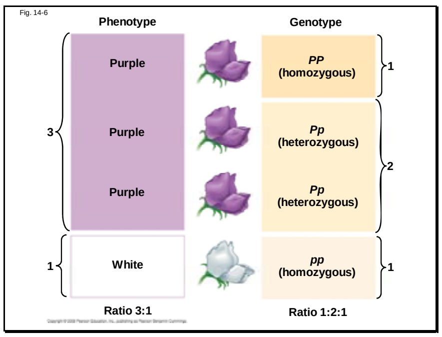

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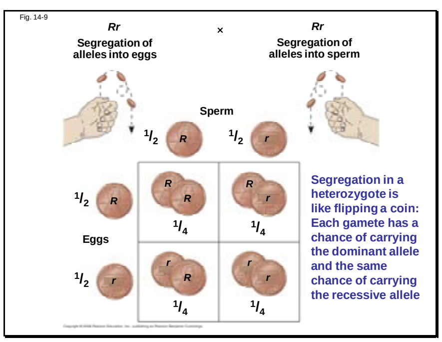

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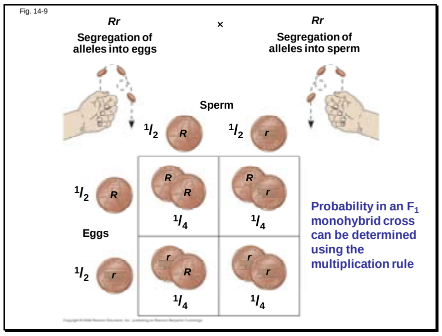

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The Testcross

- How can we tell the genotype of an individual with the dominant 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 **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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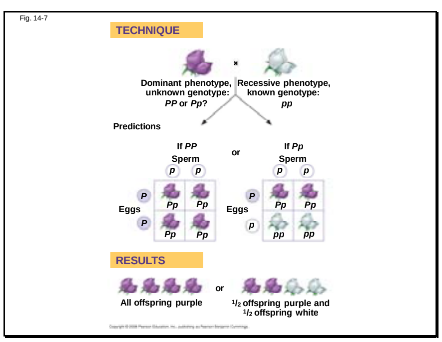

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The Law of Segregation

- Stated Simply: Gametes are haploid
- Mendel derived the law of segregation by following a single character
- All of 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*

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The Law of Independent Assortment

- 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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Table 14-1

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

Character	Dominant Trait	x	Recessive Trait	F_2 Generation Dominant:Recessive	Ratio
Flower color	Purple	x	White	705:224	3.15:1
Flower position	Axial	x	Terminal	651:207	3.14:1
Seed color	Yellow	x	Green	6,022:2,001	3.01:1
Seed shape	Round	x	Wrinkled	5,474:1,850	2.96:1
Pod shape	Inflated	x	Constricted	882:299	2.95:1
Pod color	Green	x	Yellow	428:152	2.82:1
Stem length	Tall	x	Dwarf	787:277	2.84:1

Remember:

He had lots

to choose

from.....

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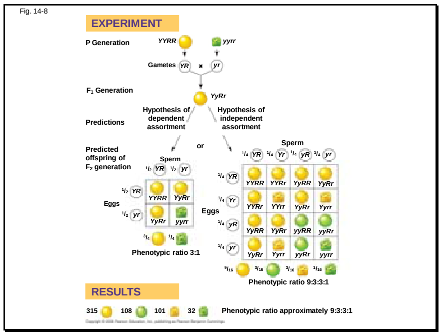

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The Law of Independent Assortment

- Simply Stated: Mom's and dad's allele loci end up in gametes independently of their other loci
- 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 together
- Mendel was lucky his targets were all on different chromosomes!

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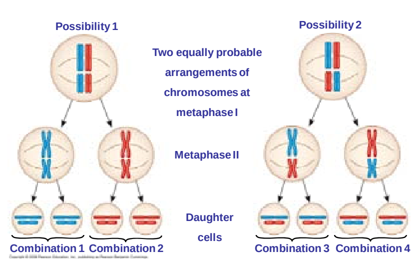
Independent assortment of chromosomes



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Review Questions

Which of the following about the law of segregation is false?

A) It states that each of two alleles for a given trait segregate into different gametes.

B) It can be explained by the segregation of homologous chromosomes during meiosis.

C) It can account for the 3:1 ratio seen in the F2 generation of Mendel's crosses.

D) It can be used to predict the likelihood of transmission of certain genetic diseases within families.

E) It is a method that can be used to determine the number of chromosomes in a plant.

Mendel's second law of independent assortment has its basis in which of the following events of meiosis I?

A) Synapsis of homologous chromosomes

B) Crossing over

C) Alignment of tetrads at the equator

D) Separation of homologs at anaphase

E) Separation of cells at telophase

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Extending Mendelian Genetics

- Mendel went for the simplest relationship between genotype and phenotype he could find
- 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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Mendelian Inheritance for a Single Gene

- When alleles are not completely dominant or recessive = incomplete dominance or co-dominance
- When a gene has more than two alleles = multiple alleles
- When a gene produces multiple phenotypes = pleiotropy

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Degrees of Dominance

- **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are identical.
  - Example: PP and Pp pea plant flowers are purple.
- In **incomplete dominance**, the phenotype of F_1 hybrids is somewhere between the phenotypes of the two parental varieties
  - Example: snapdragon flowers

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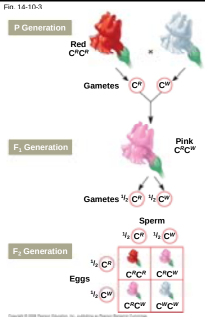
Fig. 14-10-3



Incomplete Dominance

Compare the genotype and phenotype of heterozygotes with homozygotes.

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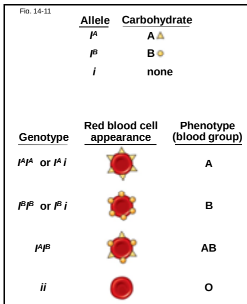
Fig. 14-11



Multiple Alleles: Most genes exist in populations in more than two allelic forms

Example: Three alleles for the ABO blood group carbohydrates

Codominance: Two dominant alleles affect the phenotype in separate, distinguishable ways

Both alleles are dominant and both phenotypes are expressed.

Example: AB blood type

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The Dominance-Phenotype Relationship

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

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Example 1: Albinism in humans occurs when both alleles at a locus produce defective enzymes in the biochemical pathway leading to melanin.

Given that heterozygotes are normally pigmented, which of the following statements is/are correct?

1. One normal allele produces as much melanin as two normal alleles.
2. Each defective allele produces a little bit of melanin.
3. Two normal alleles are needed for normal melanin production.
4. The two alleles are codominant.
5. The amount of sunlight will not affect skin color of heterozygotes.

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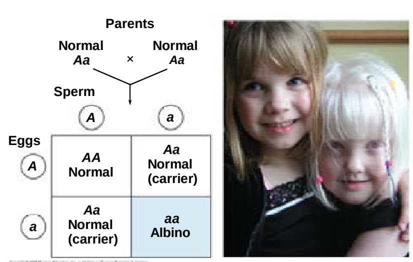
Fig. 14-16



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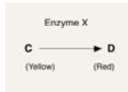
Example 2: Assume the last step in the synthesis of the red pigment of apples is catalyzed by enzyme X, which changes compound C to D.

Thinking about dominance and enzyme action, what can you accurately say about a heterozygote with one allele for an effective enzyme X and one allele for an ineffective enzyme X?

1. The phenotype will probably be yellow but cannot be red.
2. The phenotype will probably be red but cannot be yellow.
3. The phenotype will be a yellowish red.
4. The phenotype will be either yellow or red.
5. The phenotype will be either yellowish red or red.



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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

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Multiple Alleles

Imagine a locus with four different alleles for fur color in an animal. The alleles are named D^a, D^b, D^c, and D^d. If you crossed two heterozygotes, D^aD^b and D^cD^d, what genotype proportions would you expect in the offspring?

1. 25% D^aD^c, 25% D^aD^d, 25% D^bD^c, 25% D^bD^d
2. 50% D^aD^b, 50% D^cD^d
3. 25% D^aD^a, 25% D^bD^b, 25% D^cD^c, 25% $D^dD^dD^cD^d$
4. 50% D^aD^c, 50% D^bD^d
5. 25% D^aD^b, 25% D^cD^d, 25% D^cD^c, 25% D^dD^d

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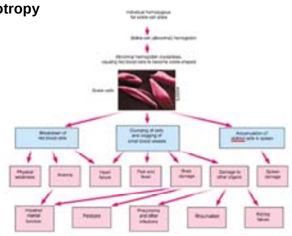
Pleiotropy

Most genes have multiple phenotypic effects, a property called **pleiotropy**



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Mendelian Genetics for Two or More Genes

Some traits may be determined by two or more genes

- Epistasis
- Polygenic
- Environmental Impact

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Epistasis

- In **epistasis**, a gene at one locus alters the phenotypic expression of a gene at a second locus
- 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

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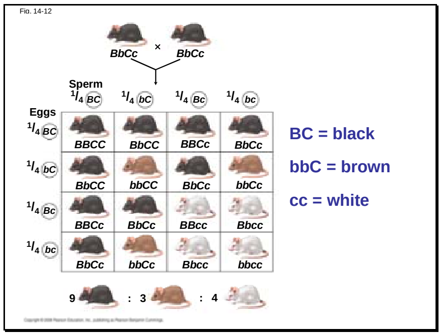

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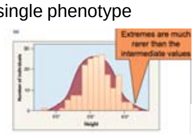
Polygenic Inheritance

- **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
- Skin color and height in humans is an example of polygenic inheritance



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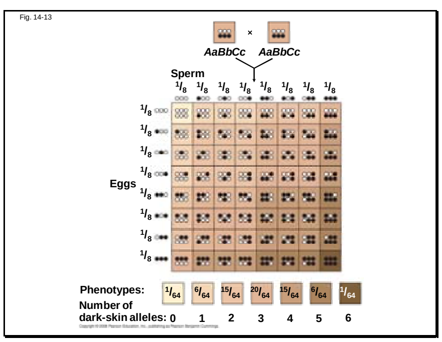

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Nature and Nurture: The Environmental Impact on Phenotype

- 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

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Fig. 14-14

Some alleles are heat-sensitive

ex. Arctic foxes make fur pigment only when the weather is warm

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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
- 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

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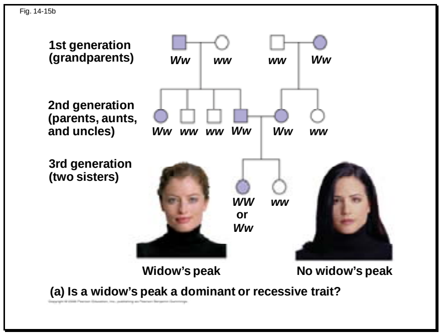

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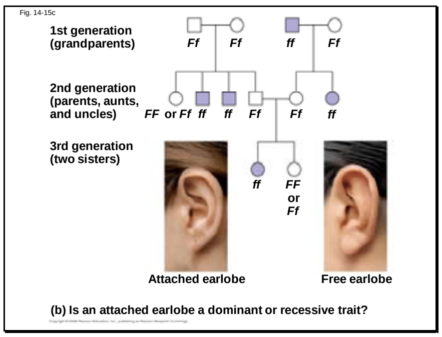

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Many genetic disorders are inherited recessively

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

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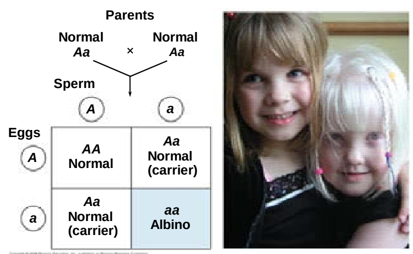
Fig. 14-16



What does the gene code for?

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Many genetic disorders are inherited recessively

- 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

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Recessive Disease I: 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 (1 in 25 are carriers)
- The cystic fibrosis allele results in defective or absent chloride transport channels in plasma membranes
- Symptoms include mucus buildup in some internal organs and abnormal absorption of nutrients in the small intestine

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Recessive Disease II: Sickle-Cell Disease

- **Sickle-cell disease** affects one out of 400 African-Americans
- 1 in 10 African-Americans have sickle cell trait
- 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

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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

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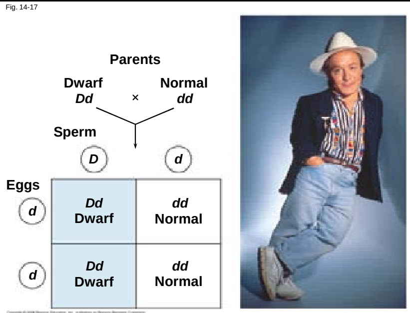
Fig. 14-17



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Huntington's Disease

- **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

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Multifactorial Disorders

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

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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