

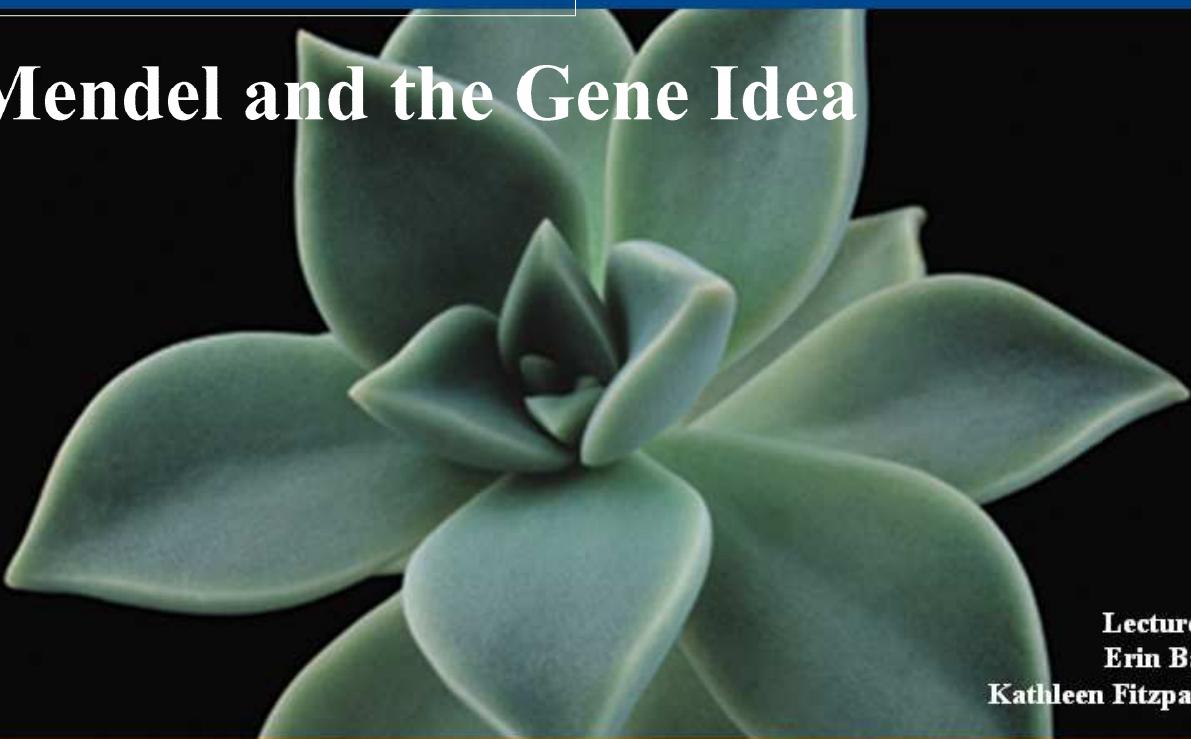
**LECTURE PRESENTATIONS**

**For CAMPBELL BIOLOGY, NINTH EDITION**

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# Chapter 14

## Mendel and the Gene Idea



Lectures by  
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## **Overview: Drawing from the Deck of Genes**

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

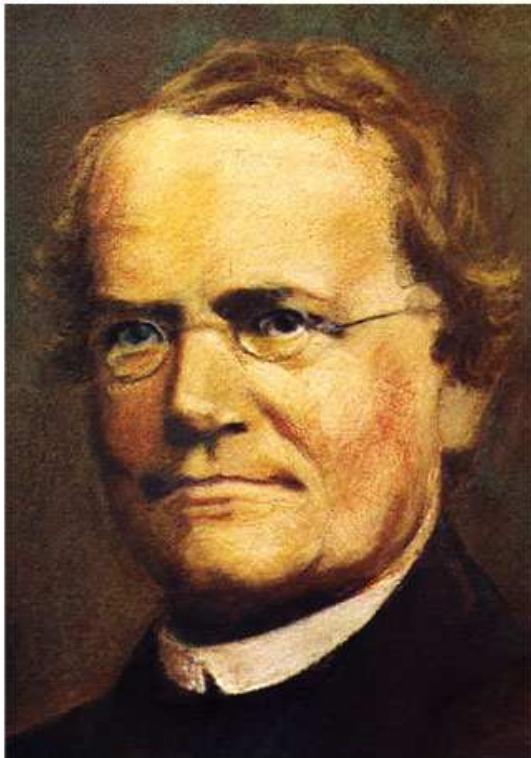
- The “particulate” hypothesis is the idea that parents pass on discrete heritable units (genes)
  - This hypothesis can explain the reappearance of traits after several generations
  - Mendel documented a particulate mechanism through his experiments with garden peas

Figure 14.1



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## **Concept 14.1: Mendel used the scientific approach to identify two laws of inheritance**

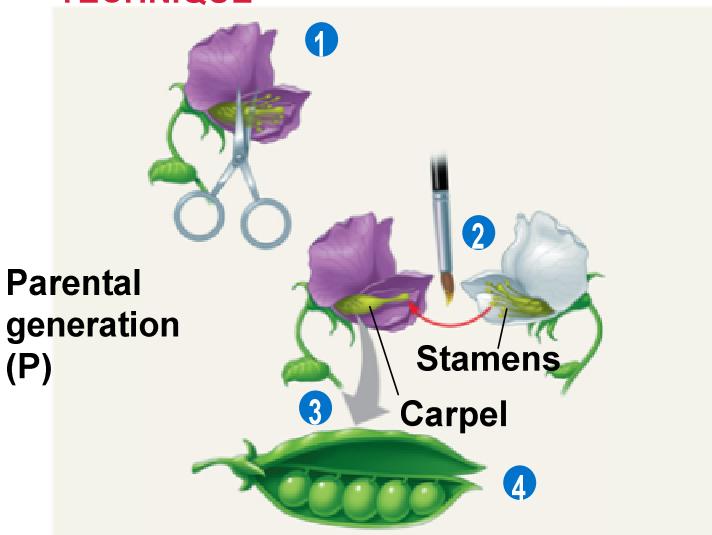
- Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments

## Mendel's Experimental, Quantitative Approach

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

Figure 14.2

### TECHNIQUE



### RESULTS

First filial  
generation  
offspring  
(F<sub>1</sub>)



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- 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)

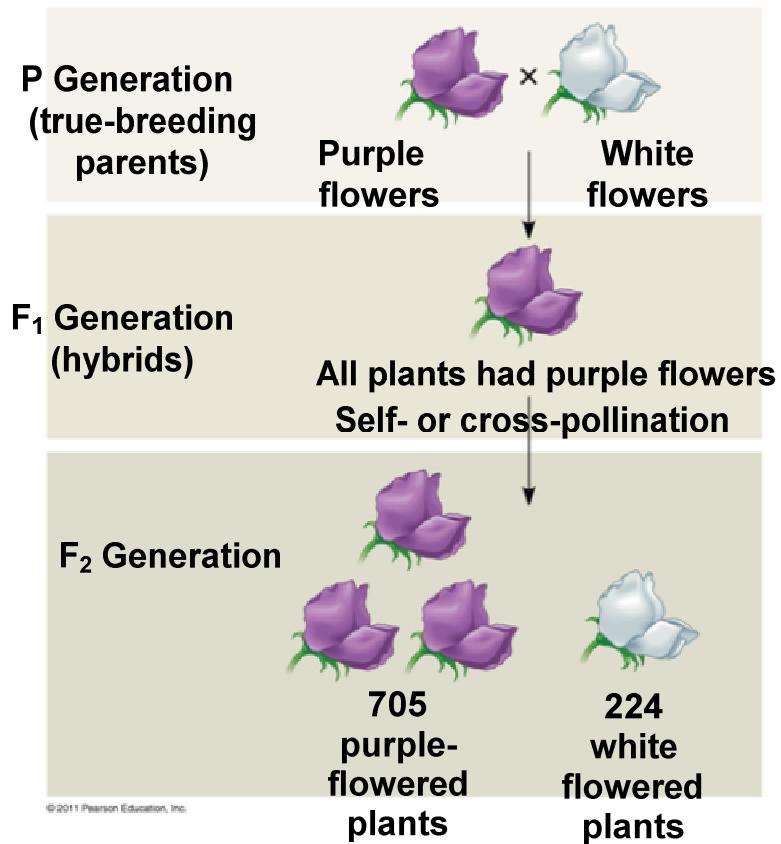
- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called **hybridization**
  - The true-breeding parents are the **P generation**
  - The hybrid offspring of the P generation are called the **F<sub>1</sub> generation**
  - When F<sub>1</sub> individuals self-pollinate or cross-pollinate with other F<sub>1</sub> hybrids, the **F<sub>2</sub> generation** is produced

## The Law of Segregation

- When Mendel crossed contrasting, true-breeding white- and purple-flowered pea plants, all of the F<sub>1</sub> hybrids were purple
- When Mendel crossed the F<sub>1</sub> hybrids, many of the F<sub>2</sub> plants had purple flowers, but some had white
- Mendel discovered a ratio of about three to one, purple to white flowers, in the F<sub>2</sub> generation

Figure 14.3-3

## EXPERIMENT



- Mendel reasoned that only the purple flower factor was affecting flower color in the F<sub>1</sub> 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<sub>2</sub> 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

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

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**Table 13.1** Seven Characters Mendel Studied and His Experimental Results

Character			F <sub>2</sub> Generation	
DOMINANT FORM	×	RECESSIVE FORM	DOMINANT:RECESSIVE	RATIO
	Purple flowers	×		705:224 3.15:1
	Yellow seeds	×		6022:2001 3.01:1
	Round seeds	×		5474:1850 2.96:1
	Green pods	×		428:152 2.82:1
	Inflated pods	×		882:299 2.95:1
	Axial flowers	×		651:207 3.14:1
	Tall plants	×		787:277 2.84:1

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$V_1 = 37$        $\frac{V_1 + gV_1}{2} = 112$  -  $\frac{V_1}{2} = 56$

$g = 37 \frac{3}{5}$        $V_1 W + gW = 300$   $\text{Reih. Vierl. } 27 - 37$

$gV_1 = 75 \frac{3}{5}$        $W = 150$   $\text{Hälfte } 165 + 15$

$V_1 W = 150$        $gV_1 = 75 \frac{3}{5} \text{ R. } 65 - 10$

$gW = 150$        $g = 37 \text{ dR. } 27 - 10$

$W = 150$        $V = 37 \text{ Vierl. } 93 + 56$

343 6V&V 951 7/2 <sup>Nat</sup> Lorraine t  
92 B 100 16 7/2 ~~traum~~  
166 W 150 14 2 <sup>etwas</sup>  
Nehre l

$x: 385 = 59 : 296$  von annehmen Wild will nicht  
 $\frac{39}{152} : \frac{95}{125} = 340$  sich hindern <sup>bleiben</sup>

$179 \frac{95}{125} : 296 = 65$  75

$W 150 \quad Y_4 \quad N$   $\frac{1}{2} \text{ längere } O. K. O.$

$6B \quad 75 \quad Y_8 \quad gV_1$

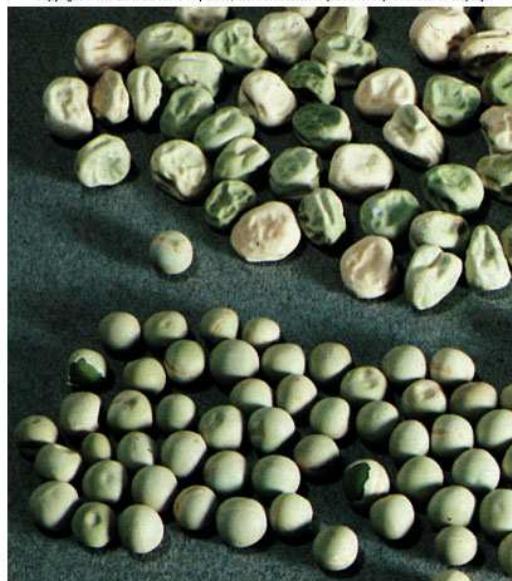
$dB \quad 37 \quad Y_{16} \quad g$

$6V \quad 300 \quad Y_2 \quad gW + V_1 W$

$V \quad 37 \quad Y_{16} \quad V$

## Page from Mendel's notebook

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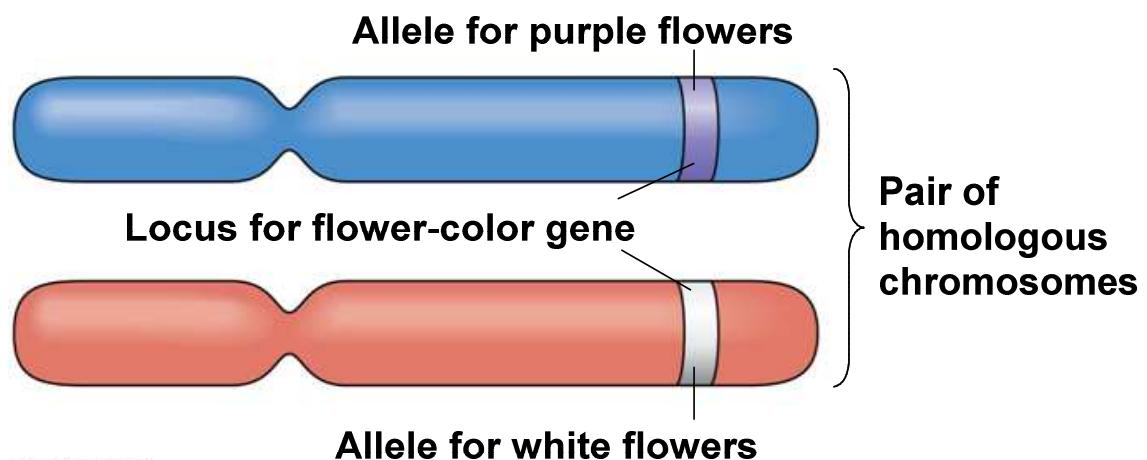


## ***Mendel's Model***

- 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 now called **alleles**
  - Each gene resides at a specific locus on a specific chromosome

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 the role of chromosomes
- The two alleles at a particular locus may be identical, as in the true-breeding plants of Mendel's P generation
- Alternatively, the two alleles at a locus may differ, as in the F<sub>1</sub> hybrids

= (Law of Independent Assortment)  
- Genes that are located on different chromosomes assort independently of one another.

## Mendelian Inheritance

- Mendel's Second Law of Heredity
  - (Law of Independent Assortment)
    - Genes that are located on different chromosomes assort independently of one another.

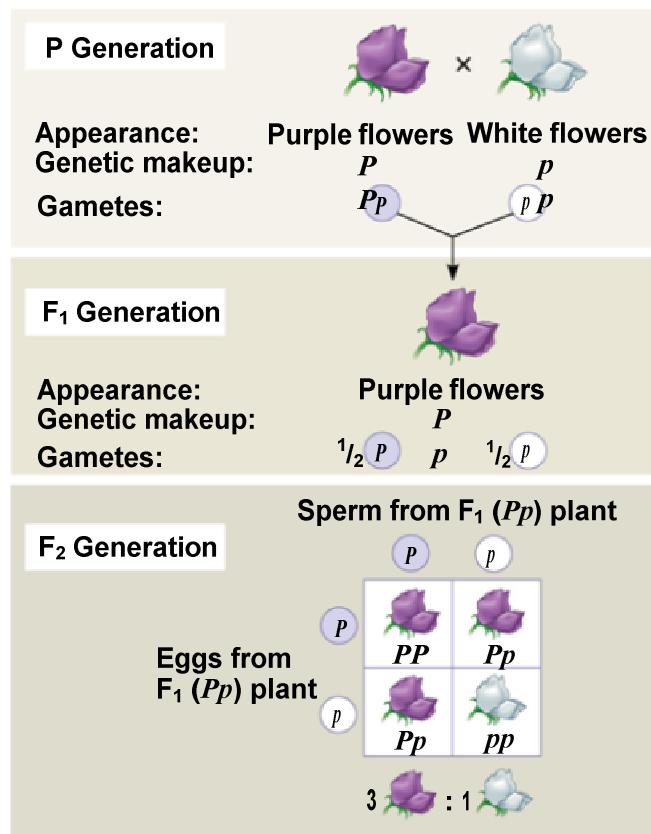
- Third: if the two alleles at a locus differ, then one (the **dominant allele**) determines the organism's appearance, and the other (the **recessive allele**) has no noticeable effect on appearance
- In the flower-color example, the  $F_1$  plants had purple flowers because the allele for that trait is dominant

= Law of Dominance

- Fourth: (now known as the **law of segregation**): the two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes
- Thus, an egg or a sperm gets only one of the two alleles that are present in the organism
- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis

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

Figure 14.5-3

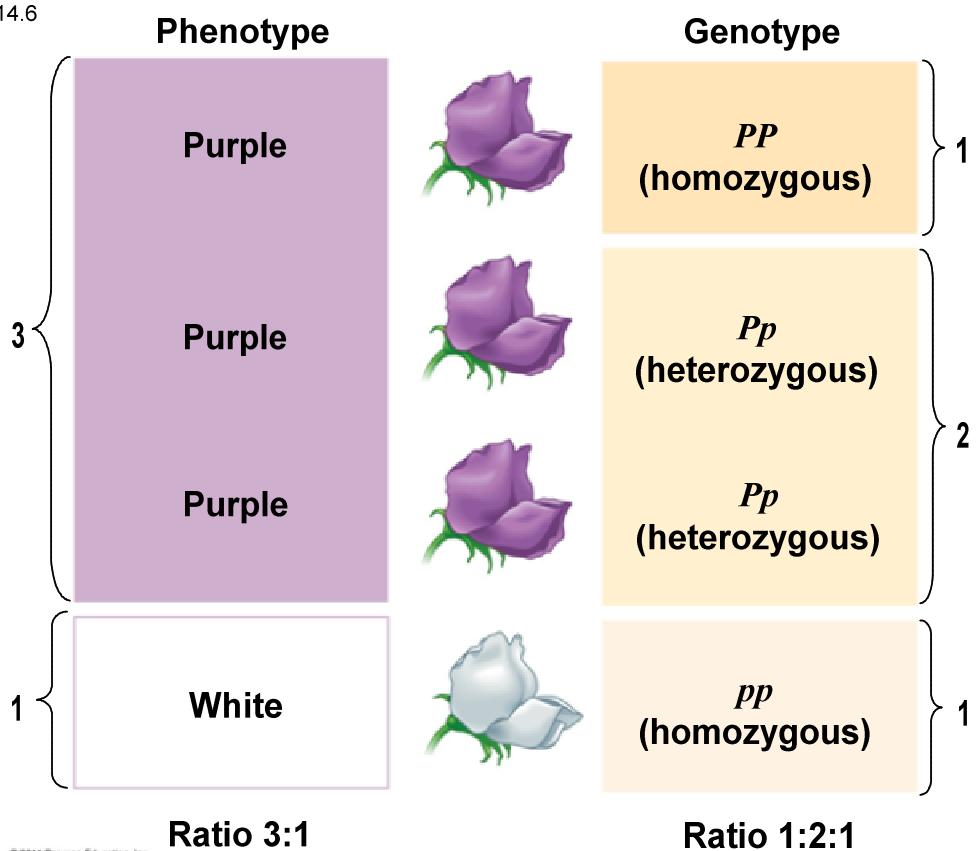


## *Useful Genetic Vocabulary*

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

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

Figure 14.6



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Cross a pure Flying (F) gorilla with a homozygous recessive gorilla. Provide the possible phenotypes and genotypes of the offspring.

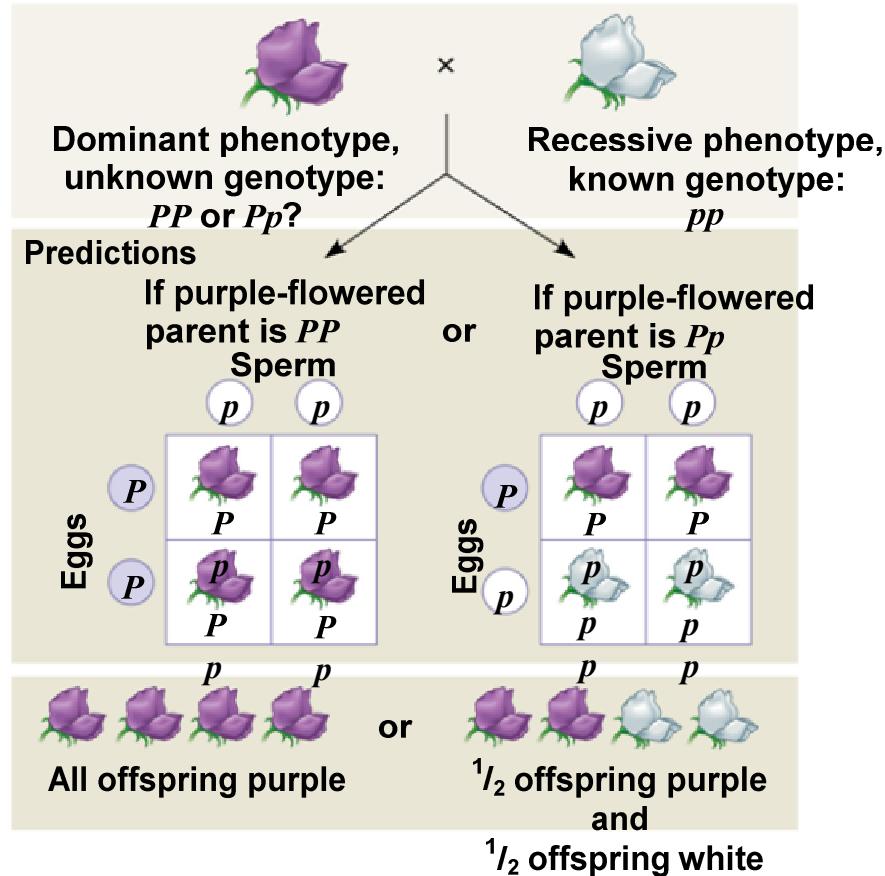
Randomly select any 2 of the F1 offspring and cross them. Provide the possible phenotypes and genotypes of the offspring.

## *The Testcross*

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

## TESTCROSS

Figure 14.7  
**TECHNIQUE**



## RESULTS

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## Testcross

What is it? A cross to determine if an unknown genotype that shows a dominant trait is either homozygous dominant or heterozygous

Method: Cross the unknown parent that shows the dominant trait with a homozygous recessive parent

Show the 2 possible crosses and results:

Rationale:

If you do the testcross and all of the offspring show the dominant trait then it is likely (but not a guarantee) that the unknown parent that shows the dominant trait is homozygous dominant.

If you do the testcross and any of the offspring show the recessive trait then it is a guarantee that the unknown parent that shows the dominant trait is heterozygous.

Why would this testcross be beneficial??

## Testcross

Practice Problem: You are a dragon breeder. You see a dragon advertised as homozygous dominant (fire breathing) for sale for \$2000. You know a true pure homozygous dominant dragon is worth \$100,000 and that a heterozygous dragon (fire breathing) is worth \$2. (F = fire breathing ability, f = no fire)

What do you do before you buy the dragon?

Show the possible crosses that could be made:

You do the cross and have all but one fire breathing baby dragons. Should you buy the dragon? Why or why not?

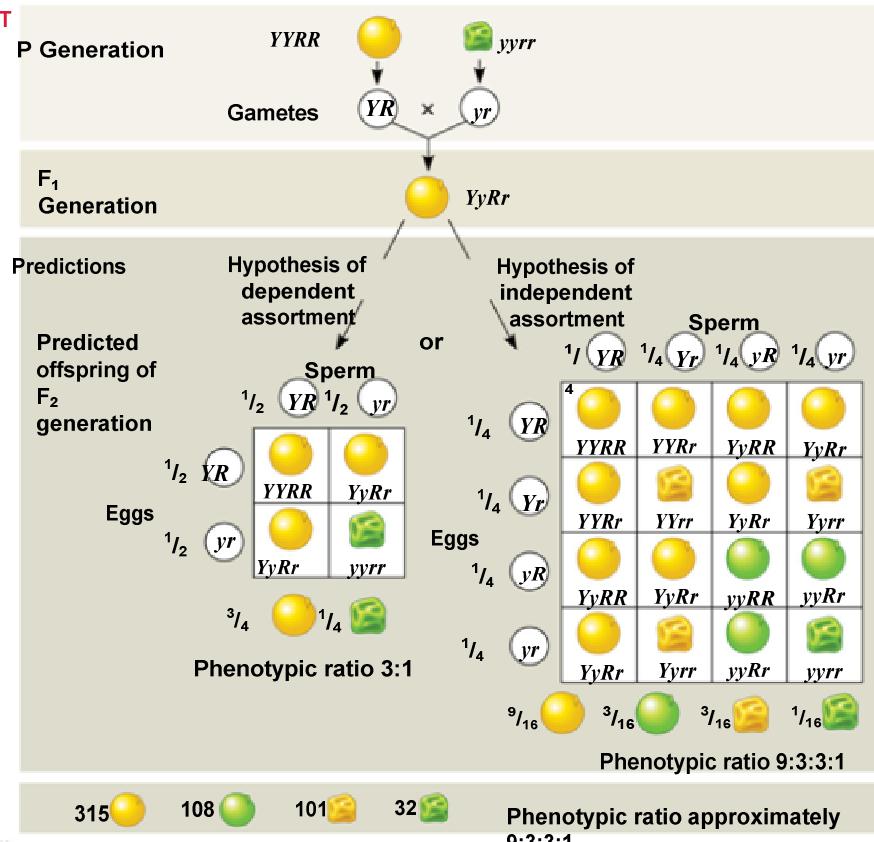
## The Law of Independent Assortment

- Mendel derived the law of segregation by following a single character
- The F<sub>1</sub> offspring produced in this cross were **monohybrids**, individuals that are heterozygous for one character
- A cross between such heterozygotes is called a **monohybrid cross**

- Mendel identified his second law of inheritance by following two characters at the same time
- Crossing two true-breeding parents differing in two characters produces **dihybrids** in the F<sub>1</sub> generation, heterozygous for both characters
- A **dihybrid cross**, a cross between F<sub>1</sub> dihybrids, can determine whether two characters are transmitted to offspring as a package or independently

Figure 14.8

**EXPERIMENT**



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$AaPp \times AaPp$

	AP	Ap	aP	ap
AP				

\* Practice problems!

1. R - running      B - black  
r - waltzing      b - brown

P<sub>1</sub> - RrBb x RRBB

possible gametes: parent 1: RB, Rb, rB, rb  
parent 2: RB, RB, RB, RB

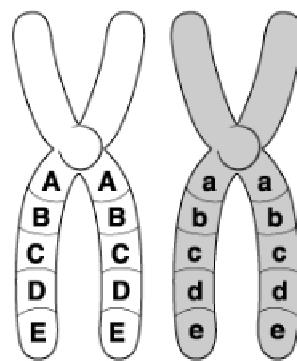
Phenotype

Genotype

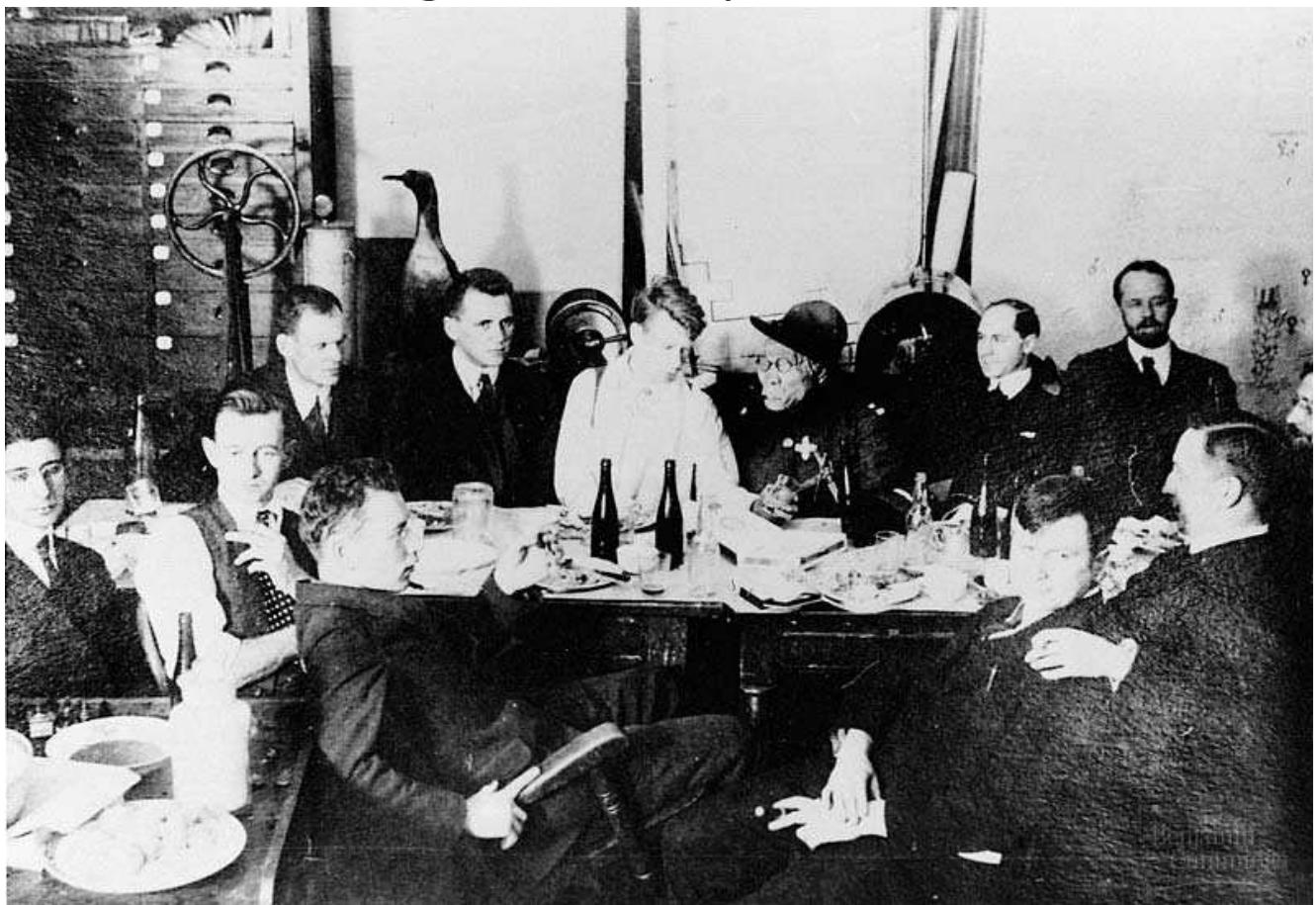
- Using a dihybrid cross, Mendel developed the **law of independent assortment**
- The law of independent assortment states that each pair of alleles segregates independently of each other pair of alleles during gamete formation
- Strictly speaking, this law applies only to genes on different, nonhomologous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

## **Forever Linked?**

Some genes appear to be inherited together, or "linked." If two genes are found on the same chromosome, does it mean they are linked forever? Study the diagram, which shows four genes labeled A–E and a–e, and then answer the questions on the next slide.

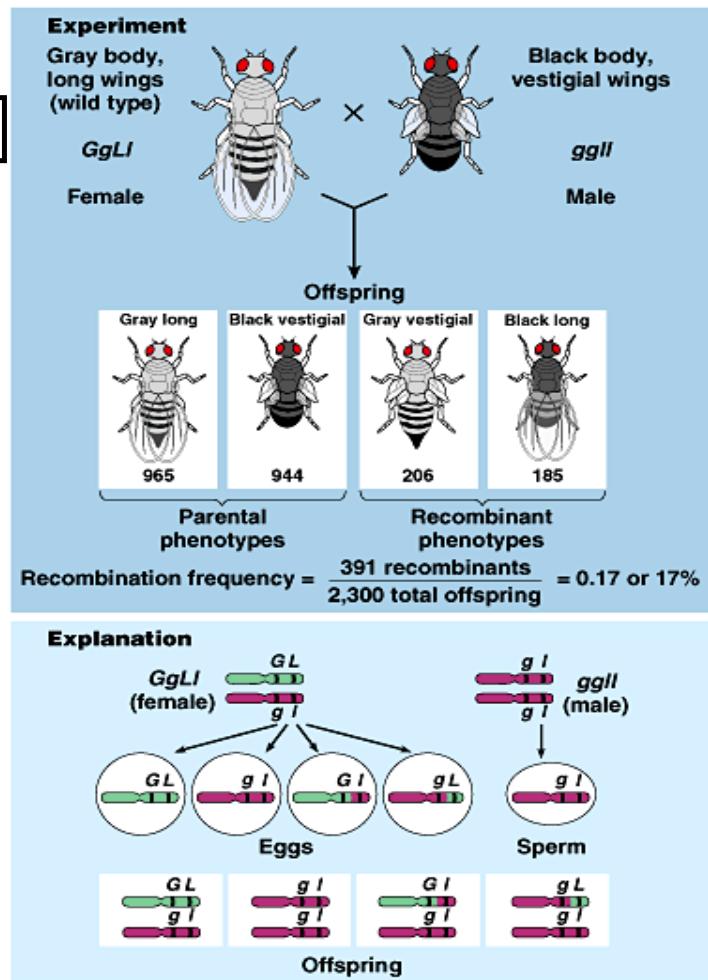


## Morgan Fruit Fly Room



- T.H. Morgan was looking at fruit flies and came up with gene linkage
- gene linkage - genes which are located on the same chromosome. Genes are inherited together and there is no independent assortment
- Morgan crossed fruit flies  $GgWw \times ggww$ 
  - expected ratios: 25% / 25% / 25% / 25%
  - actual ratios:  
41.5% dom/dom : 41.5% rec/rec : 8.5% dom/rec :  
8.5% rec/dom
    - = these genes are on the same linked chromosomes and dependently assort
- linkage groups (humans=23) are homologous chromosomes
- fruit flies have 4 linkage groups (= 4 pairs of chrom.)
- conclusions:
  1. each chromosome is a group of linked genes
  2. the chromosomes assort independently, not the genes

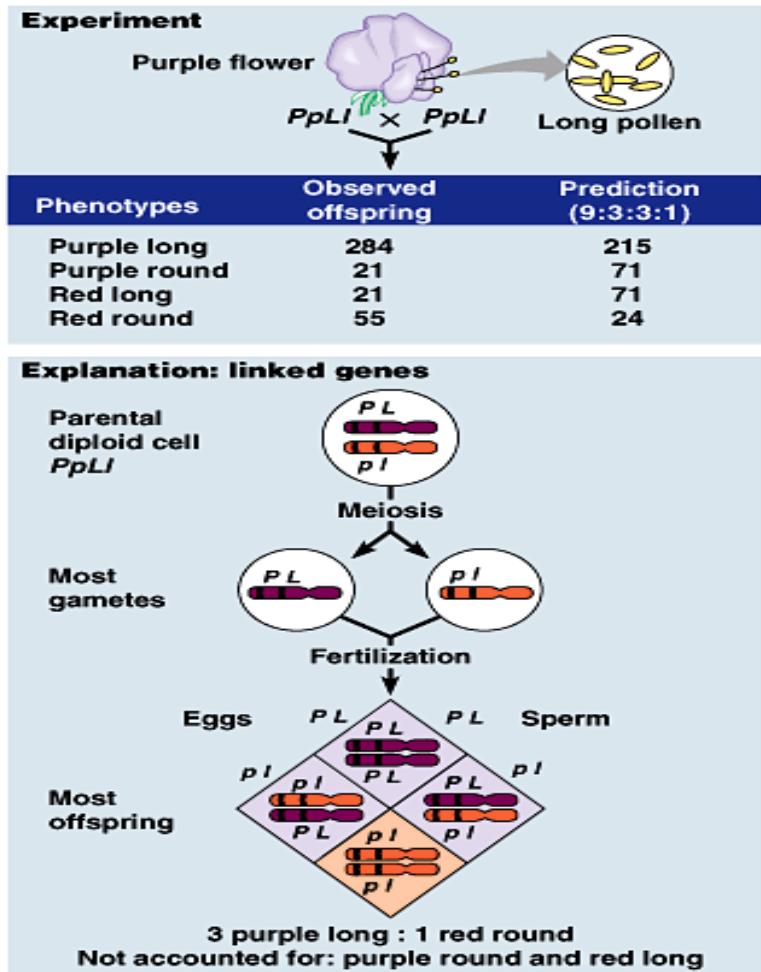
## *Drosophila melanogaster*



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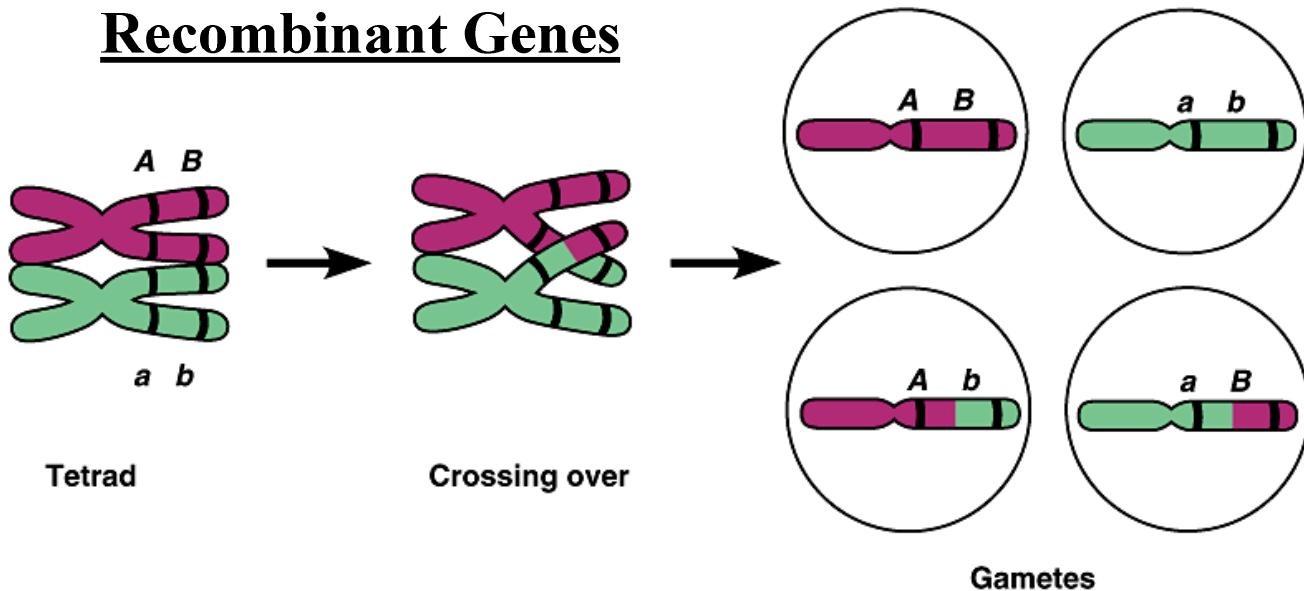
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## Linked Genes vs non-Linked Genes



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## Recombinant Genes



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Crossing Over - increases genetic diversity

- 1st studies of crossing over were done by Morgan using fruit flies *Drosophila melanogaster*
- crossed GgLI x ggII - should get 1:1:1:1 ratio if not linked
- b/c of recombination frequencies = 965:944:206:185 = linked genes

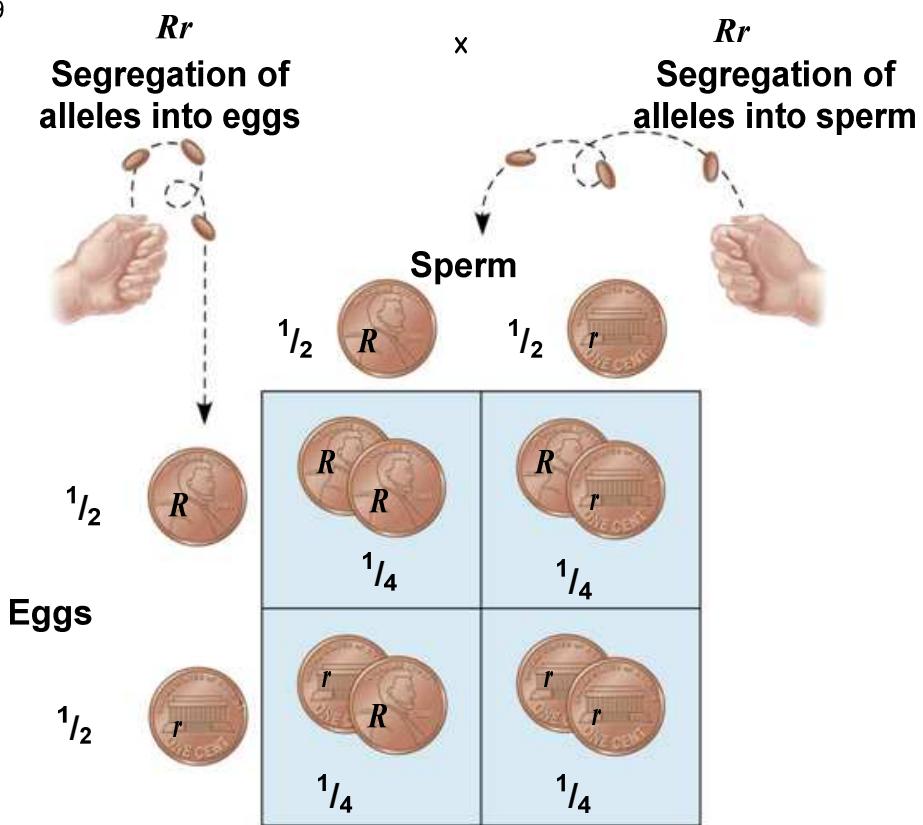
## **Concept 14.2: The laws of probability govern Mendelian inheritance**

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

## The Multiplication and Addition Rules Applied to Monohybrid Crosses

- The **multiplication rule** states that the probability that two or more independent events will occur together is the product of their individual probabilities
  - Probability in an  $F_1$  monohybrid cross can be determined using the multiplication rule
  - Segregation in a heterozygous plant is like flipping a coin: Each gamete has a  $\frac{1}{2}$  chance of carrying the dominant allele and a  $\frac{1}{2}$  chance of carrying the recessive allele
- e.g. the odds of a head and a head together is  $1/2 \times 1/2 = 1/4$

Figure 14.9



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- 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
  - e.g.  $Rr(25\%) + rR(25\%) = 50\%$  chance of heterozygous

## **Solving Complex Genetics Problems with the Rules of Probability**

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

Figure 14.UN01

**Probability of YYRR =  $1/4$  (probability of YY)  $\times$   $1/4$  (RR) =  $1/16$**

**Probability of YyRR =  $1/2$  (Yy)  $\times$   $1/4$  (RR) =  $1/8$**

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YY and RR are like heads + heads

Yy is like heads + tails

Figure 14.UN02

## Read the book

$ppyyRr$	$1/4$	(probability of $pp$ ) $\times$ $1/2$ ( $yy$ ) $\times$ $1/2$ ( $Rr$ )	$= 1/16$
$ppYyrr$	$1/4 \times 1/2 \times 1/2$		$= 1/16$
$Ppyyrr$	$1/2 \times 1/2 \times 1/2$		$= 2/16$
$PPyyrr$	$1/4 \times 1/2 \times 1/2$		$= 1/16$
$ppyyrr$	$1/4 \times 1/2 \times 1/2$		$= 1/16$
<hr/> <b>Chance of <i>at least two recessive traits</i></b>			$= 6/16$ or $3/8$

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**Table 13.2** Some Dominant and Recessive Traits in Humans

Recessive Traits	Phenotypes	Dominant Traits	Phenotypes
Albinism	Lack of melanin pigmentation	Mid-digital hair	Presence of hair on middle segment of fingers
Alkaptonuria	Inability to metabolize homogentisic acid	Brachydactyly	Short fingers
Red-green color blindness	Inability to distinguish red or green wavelengths of light	Huntington disease	Degeneration of nervous system, starting in middle age
Cystic fibrosis	Abnormal gland secretion, leading to liver degeneration and lung failure	Phenylthiocarbamide (PTC) sensitivity	Ability to taste PTC as bitter
Duchenne muscular dystrophy	Wasting away of muscles during childhood	Camptodactyly	Inability to straighten the little finger
Hemophilia	Inability to form blood clots	Hypercholesterolemia (the most common human Mendelian disorder—1 in 500)	Elevated levels of blood cholesterol and risk of heart attack
Sickle cell anemia	Defective hemoglobin that causes red blood cells to curve and stick together	Polydactyly	Extra fingers and toes

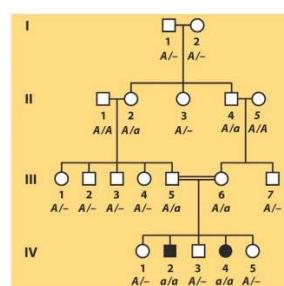
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**Table 13.3** Some Important Genetic Disorders

Disorder	Symptom	Defect	Dominant/ Recessive	Frequency Among Human Births
Cystic fibrosis	Mucus clogs lungs, liver, and pancreas	Failure of chloride ion transport mechanism	Recessive	1/2500 (Caucasians)
Sickle cell anemia	Blood circulation is poor	Abnormal hemoglobin molecules	Recessive	1/625 (African Americans)
Tay-Sachs disease	Central nervous system deteriorates in infancy	Defective enzyme (hexosaminidase A)	Recessive	1/3500 (Ashkenazi Jews)
Phenylketonuria	Brain fails to develop in infancy	Defective enzyme (phenylalanine hydroxylase)	Recessive	1/12,000
Hemophilia	Blood fails to clot	Defective blood-clotting factor VIII	Sex-linked recessive	1/10,000 (Caucasian males)
Huntington disease	Brain tissue gradually deteriorates in middle age	Production of an inhibitor of brain cell metabolism	Dominant	1/24,000
Muscular dystrophy (Duchenne)	Muscles waste away	Degradation of myelin coating of nerves stimulating muscles	Sex-linked recessive	1/3700 (males)
Hypercholesterolemia	Excessive cholesterol levels in blood lead to heart disease	Abnormal form of cholesterol cell surface receptor	Dominant	1/500

# Recessive Disorders

## Autosomal Recessive Inheritance: albinism



## **Autosomal Recessive Inheritance: albinism**



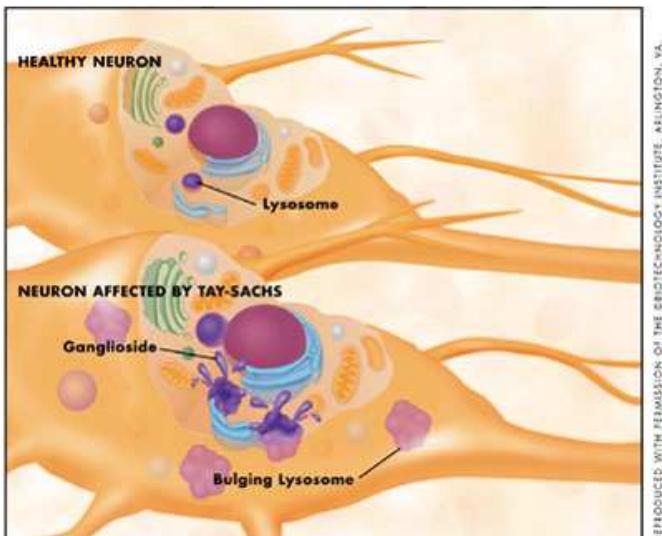


**An albino kangaroo.**



## Autosomal Recessive Alleles: Example #5

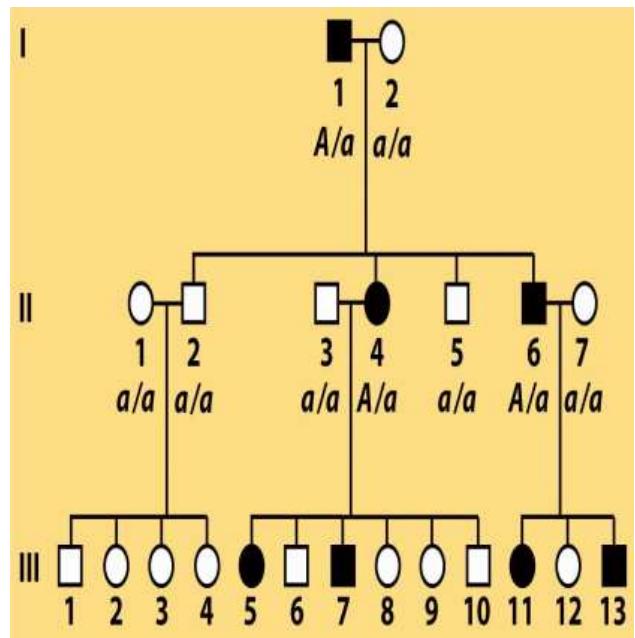
### Tay-Sachs disease

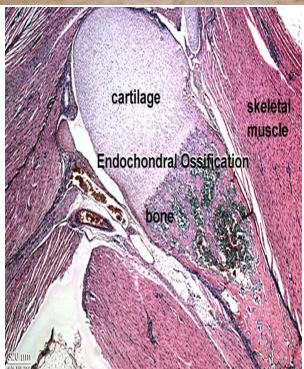
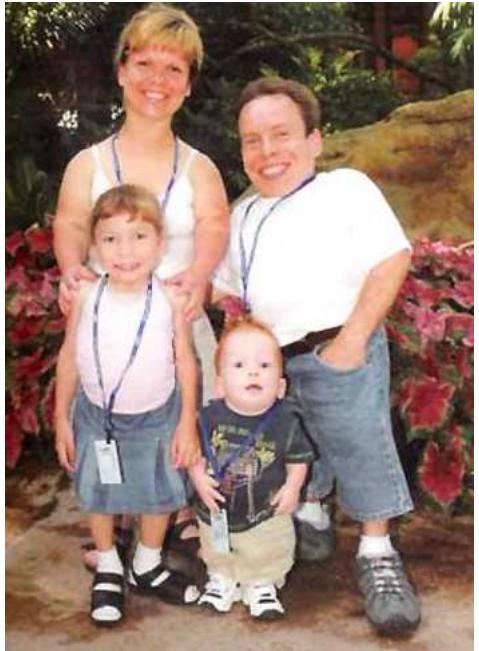


Lipid accumulation in brain cells which results in nervous system breakdown

# Dominant Disorders

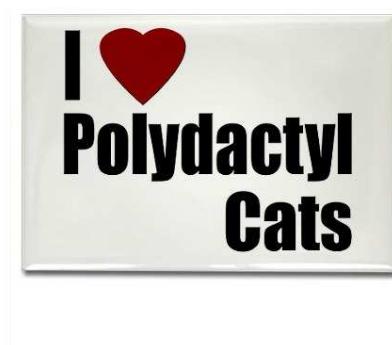
## Autosomal Dominant: Achondroplastic dwarfism





## Epiphyseal Growth Plates

Polydactyl = dominant disorder  
- common in cats





Polydactyl = dominant disorder  
- uncommon in humans



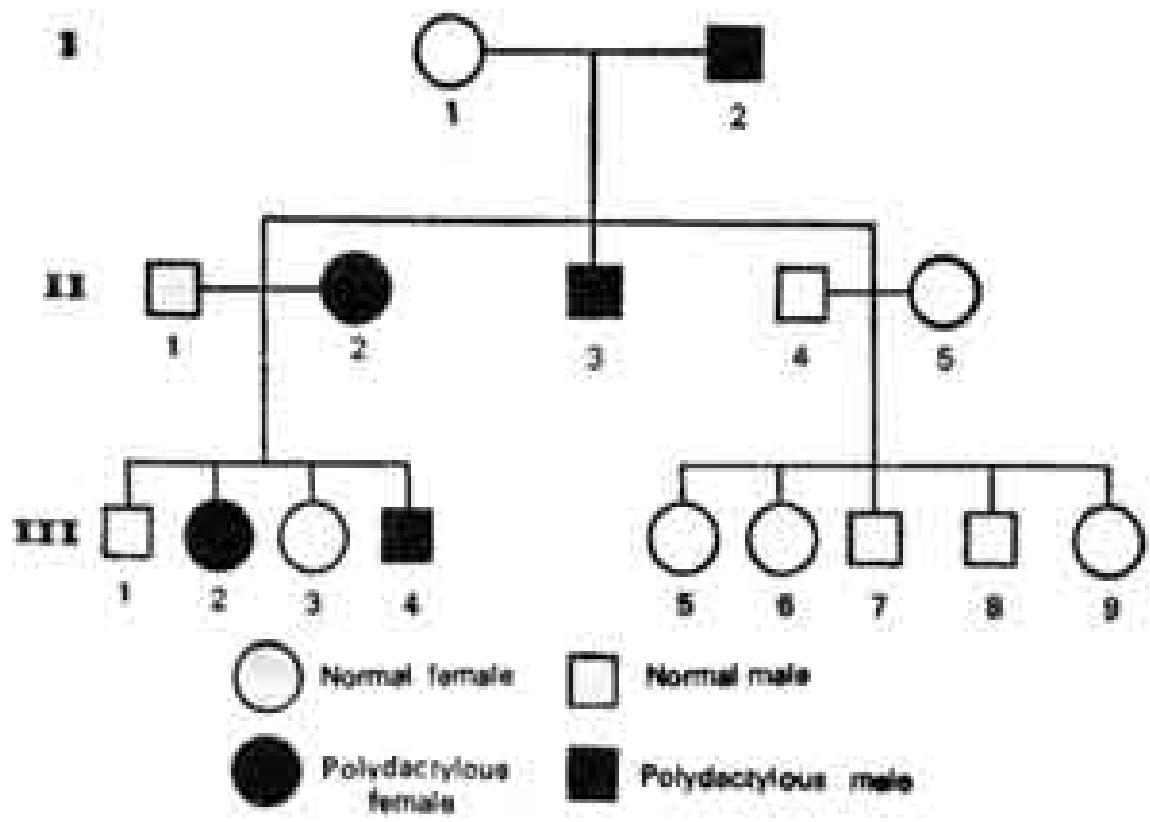


**Did you know that “Grey’s Anatomy” star Ellen Pompeo is polydactyl?**



31 fingers and toes!





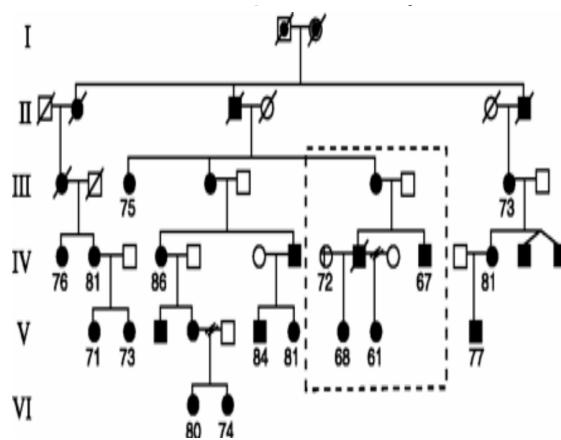
## **Autosomal Dominant - Huntington's disease**



A woman with Huntington disease.

# Autosomal Dominant FASPS

- Familial Advanced Sleep Phase Syndrome
- Autosomal dominant circadian rhythm variant
- Affected individuals are “morning larks”
  - They go to sleep at about 7:30 pm and awaken about 4:30 am
  - 4 hour sleep temperature and melatonin rhythm advance



# Beyond Dominant and Recessive

## **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 = Beyond Dominant and Recessive**

- 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

## The Relation Between

- 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

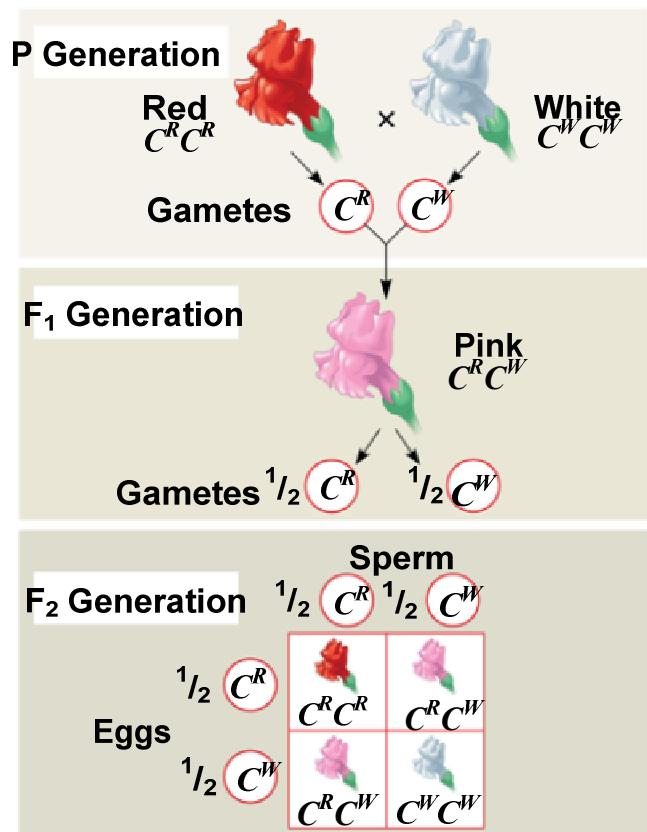
## **Frequency of Dominant Alleles**

- 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

## *Degrees of Dominance*

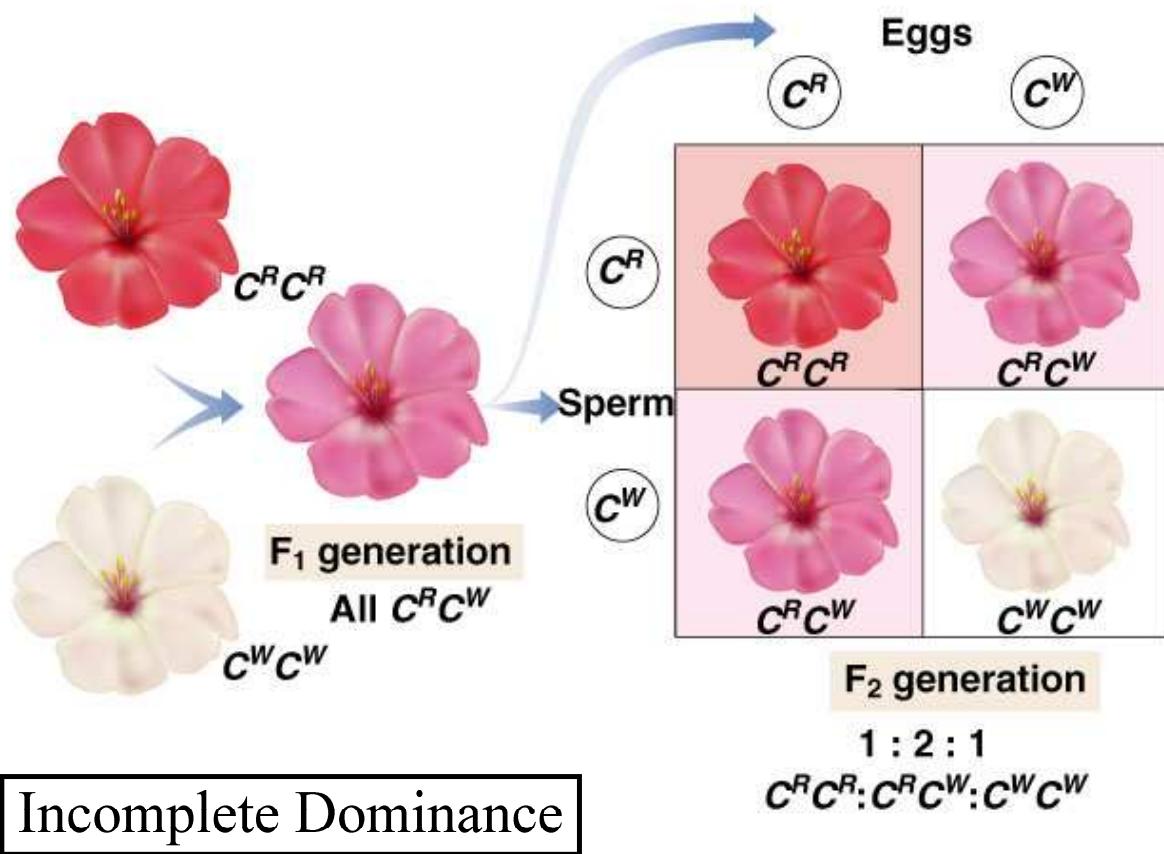
- **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are identical
  - 1.
- In **incomplete dominance**, the phenotype of  $F_1$  hybrids is somewhere between the phenotypes of the two parental varieties

Figure 14.10-3



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## Incomplete Dominance



# Hypercholesterolemia

= incomplete dominance

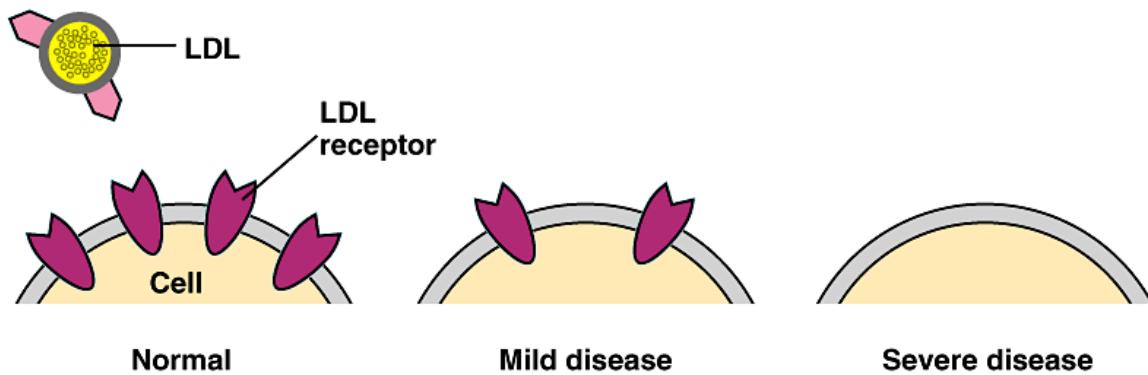
## Genotypes:

$HH$   
Homozygous  
for ability to make  
LDL receptors

$Hh$   
Heterozygous

$hh$   
Homozygous  
for inability to make  
LDL receptors

## Phenotypes:



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2.

- In **codominance**, two dominant alleles affect the phenotype in separate, distinguishable ways

## ABO Blood Groups

- this is an example of Multiple Alleles
  - what are they?

- It is also an example of Codominance - No single allele is dominant, and each allele has its own effect.

- ABO blood groups

- human gene that encodes enzyme that adds sugar molecules to lipids on the surface of red blood cells
    - $I^B$  adds galactose
    - $I^A$  adds galactosamine
    - i adds no sugar

## ABO Blood Groups

Possible alleles from female

Possible alleles from male

$I^A$	or	$I^B$	or	$i$
$I^A$		$I^A I^B$		$I^A i$
or		$I^B I^B$		$I^B i$
$I^B$				
or				
$i$		$I^A i$	$I^B i$	$ii$

Blood types

A

AB

B

O

Co-dominance is when both alleles contribute to the phenotype of the offspring

- examples:

1. - speckled chicken:

$F^B F^W$ ; both colors of feathers are expressed

2. - cattle - red and white allele for coat color

= Roan (pink) cow

3. - A and B portions of blood groups

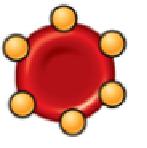
4. - Sickle-Cell Anemia

### ***3. 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	
Genotype	$I^A I^A$ or $I^A i$	$I^B I^B$ or $I^B i$	$I^A I^B$	$i i$
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

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Multiple alleles -when three or more alleles that code for the same

example: rabbit coat color (brown, gray, Himalayan, albino)

example: Human Blood!

- A, B, O, AB

- on the surface of blood cell membrane there are antigens which are used to determine blood type

- A and B are carbohydrates

- O blood type does not have an antigen

- A and B are both dominant (co-dominance) and O is recessive

- Genotype: A = I<sup>A</sup>I<sup>A</sup>, I<sup>A</sup>i

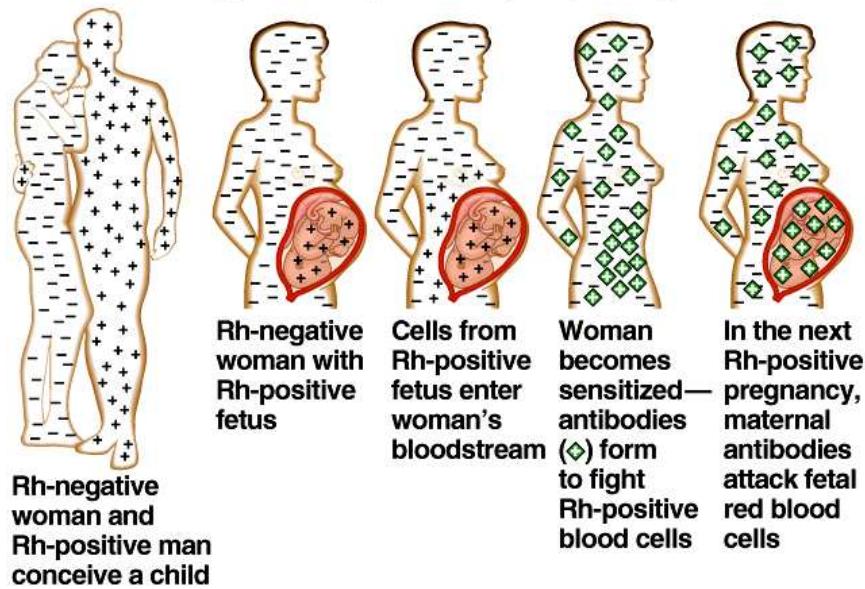
B = I<sup>B</sup>I<sup>B</sup>, I<sup>B</sup>i

AB = I<sup>A</sup>I<sup>B</sup> \*co-dominance!\*

O = ii

- other blood trait = Rh factor - it is simple dominant/recessive
  - first isolated in rhesus monkey
  - it is a protein
  - Rh+ = protein present, Rh - = protein absent
  - what is the true universal donor and recipient based on ABO blood groups and Rh factor

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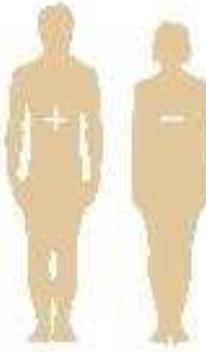


### Rh blood group

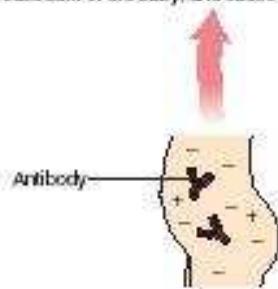
A reduction in red blood cells leads to anemia, a condition marked by weakness and fatigue. Severe anemia can lead to heart failure and death. The breakdown of red blood cells also causes the formation of bilirubin, the build up of which can lead to jaundice and possibly brain damage.



An Rh positive father and Rh negative mother may conceive an Rh positive baby.



In a subsequent pregnancy with an Rh positive baby there is the risk that it will develop Rh disease. Even though the blood circulation of the mother is separate from that of the child, the antibodies in her system can cross the placenta, enter the bloodstream of the baby, and cause its red blood cells to be killed.



The mother's immune system recognizes the cells as foreign and develops antibodies against them.

This usually isn't a problem if it's the mother's first pregnancy with an Rh positive child, because her blood circulation is separate from that of the baby.



At birth, or after an abortion or miscarriage, Rh positive blood cells from the baby enter the mother's bloodstream.

# ABO Blood Groups

Blood Group (Phenotype)	Genotypes	Antibodies Present in Blood	Reaction When Blood from Groups Below Is Mixed with Antibodies from Groups at Left			
			O	A	B	AB
O	$ii$	Anti-A Anti-B				
A	$I^A I^A$ or $I^A i$	Anti-B				
B	$I^B I^B$ or $I^B i$	Anti-A				
AB	$I^A I^B$	—				

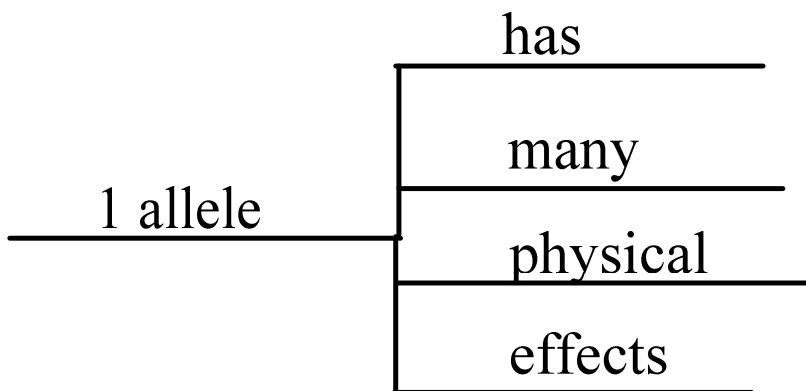
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## ***4. Pleiotropy***

- 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

## Pleiotropic effects

- Individual alleles often have more than one effect on the phenotype
  - e.g. sickle cell anemia
    - causes many problems



## **Extending Mendelian Genetics for Two or More Genes**

- Some traits may be determined by two or more genes

## Phenotypic Considerations

- Environmental effects
  - degree of allele expression may depend on the environment
- Epistasis
  - one gene interferes with the expression of another gene
  - coat color in Labrador retrievers



(a)



(b)

### Environmental effects on an allele's expression

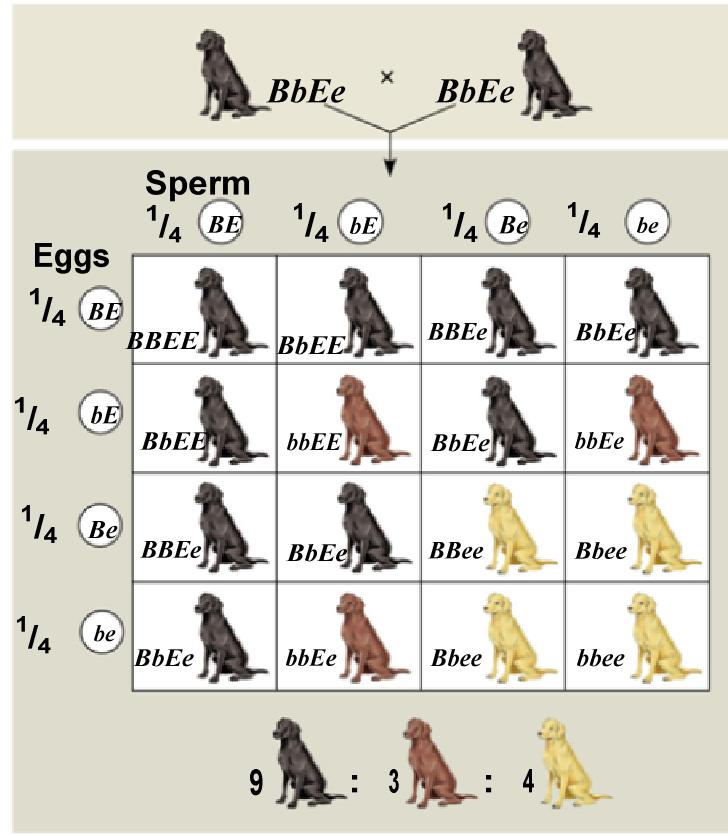
**Example 1:**  
**Arctic Fox - express fur pigment only when the weather is warm**  
**- these are the same animal!**

**Example 2:**  
**- the *ch* allele in Himalayan rabbits and Siamese cats is inactivated at temperatures above 33 degrees C**

## **6. Epistasis**

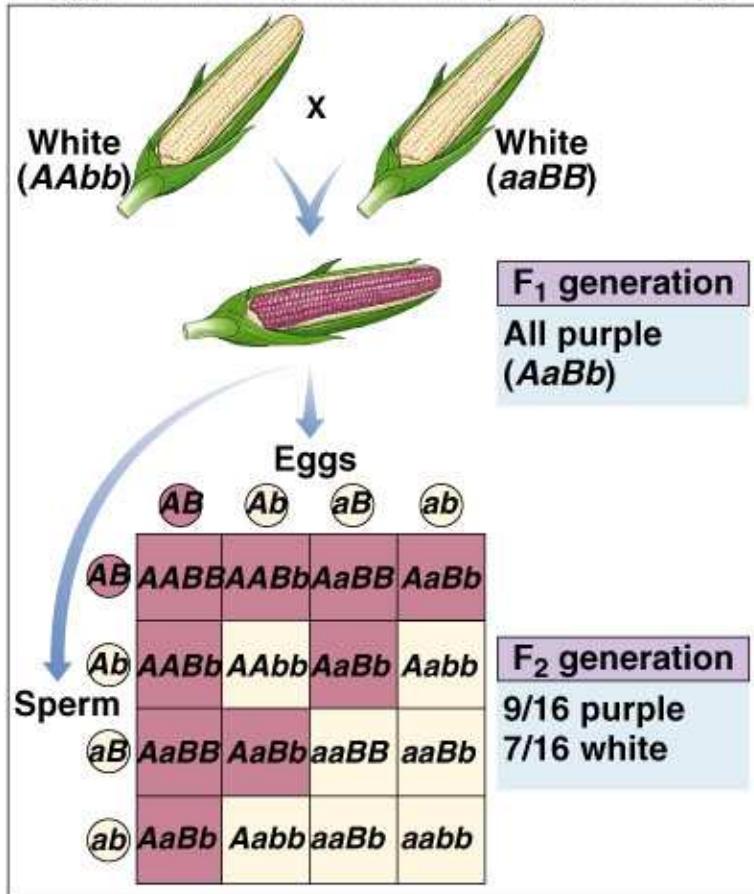
- In **epistasis**, a gene at one locus alters the phenotypic expression of a gene at a second locus
- For example, in Labrador retrievers and many other mammals, coat color depends on two genes
  - One gene determines the pigment color (with alleles *B* for black and *b* for brown)
  - The other gene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

Figure 14.12



Should be what? What does this mean?

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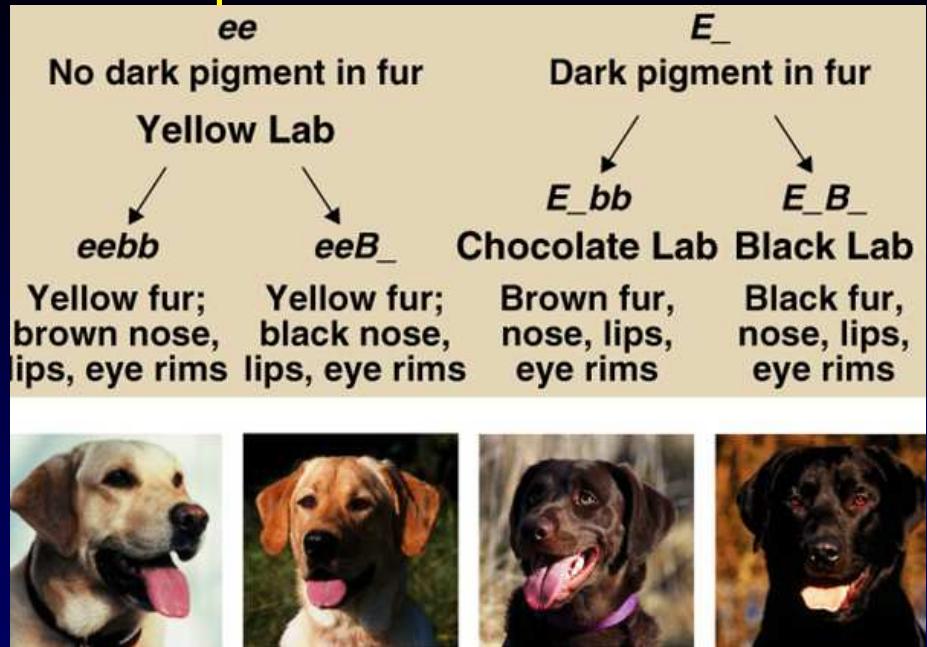


Epistasis affects grain color generates a Modified ratio in the F<sub>2</sub> generation of 9:6

- the purple pigment found in some varieties of corn is a product of a two-step biochemical pathway.

Unless both enzymes are active (the plant has a dominant allele for each of the two genes,  $A_B_$ ), no pigment is expressed.

## Epistatic Interactions



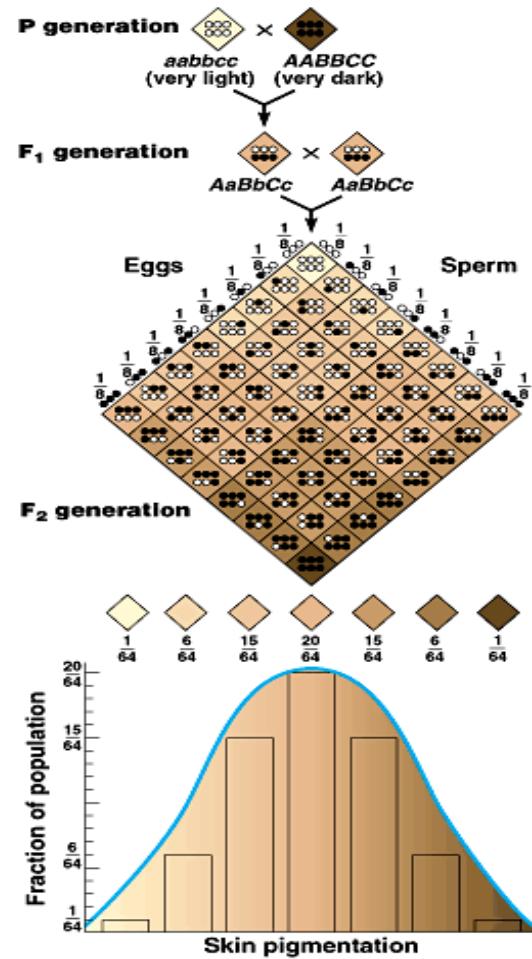
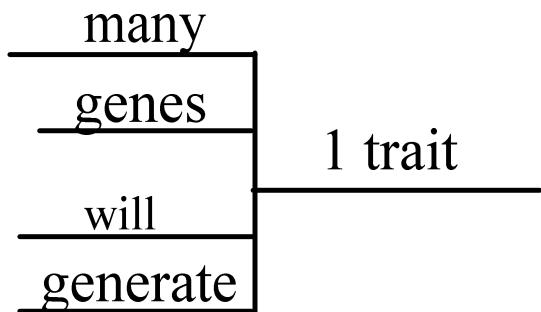
- coat color is the result of the interaction of 2 genes, each with 2 alleles
- the E gene determines if the pigment will be deposited in the fur, and the B gene determines how dark the pigment will be

## *7. 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

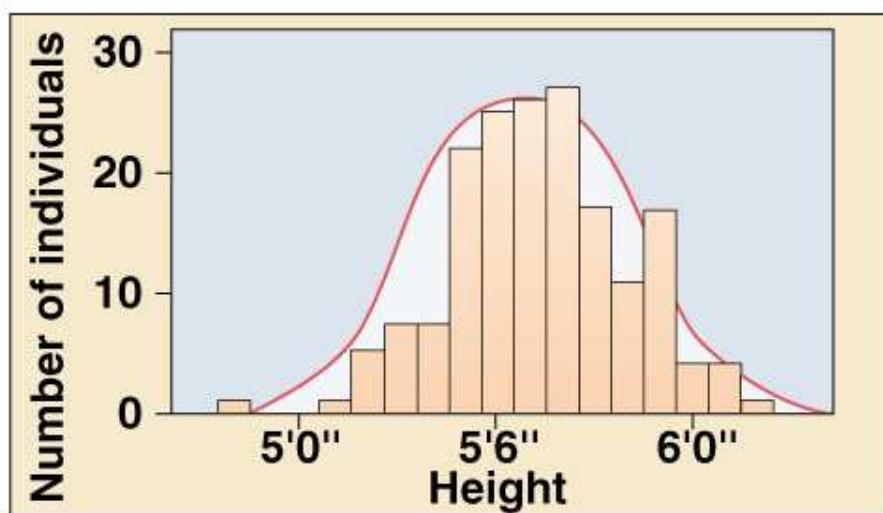
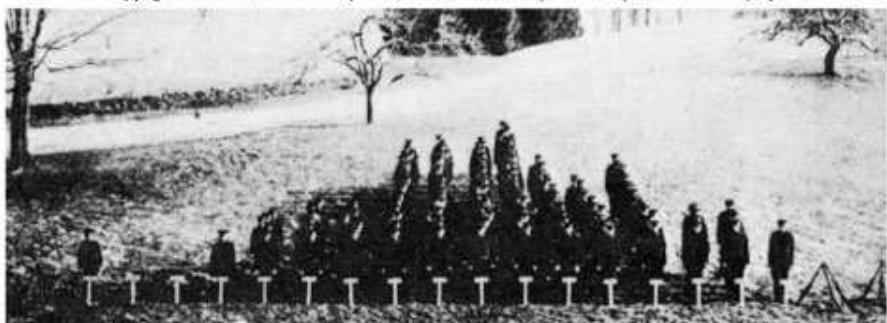
Polygenic Traits (Inheritance)

- trait produced by interaction of many genes
- e.g. = height, skin color



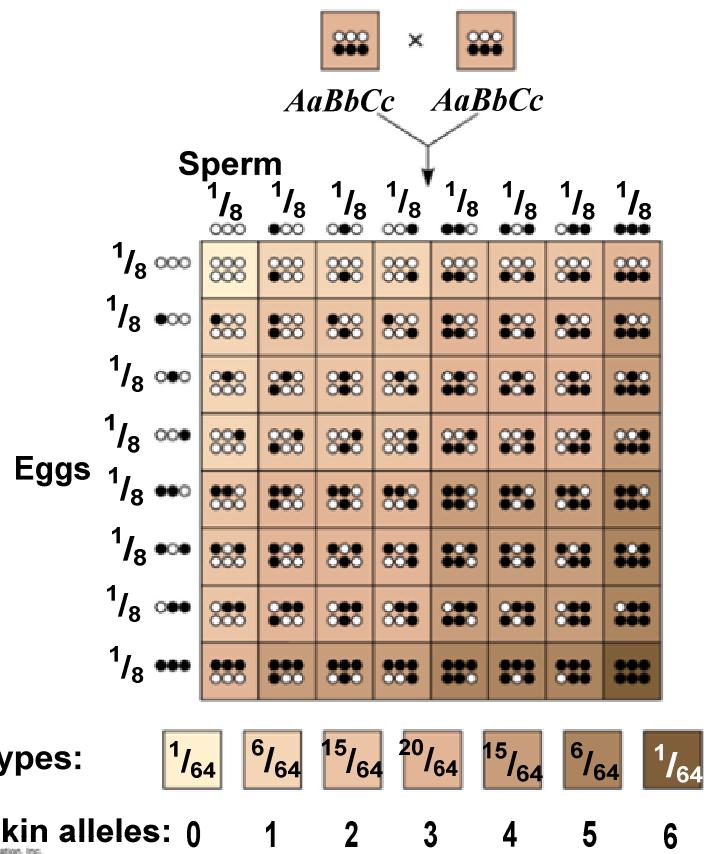
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Continuous Variation = polygenic

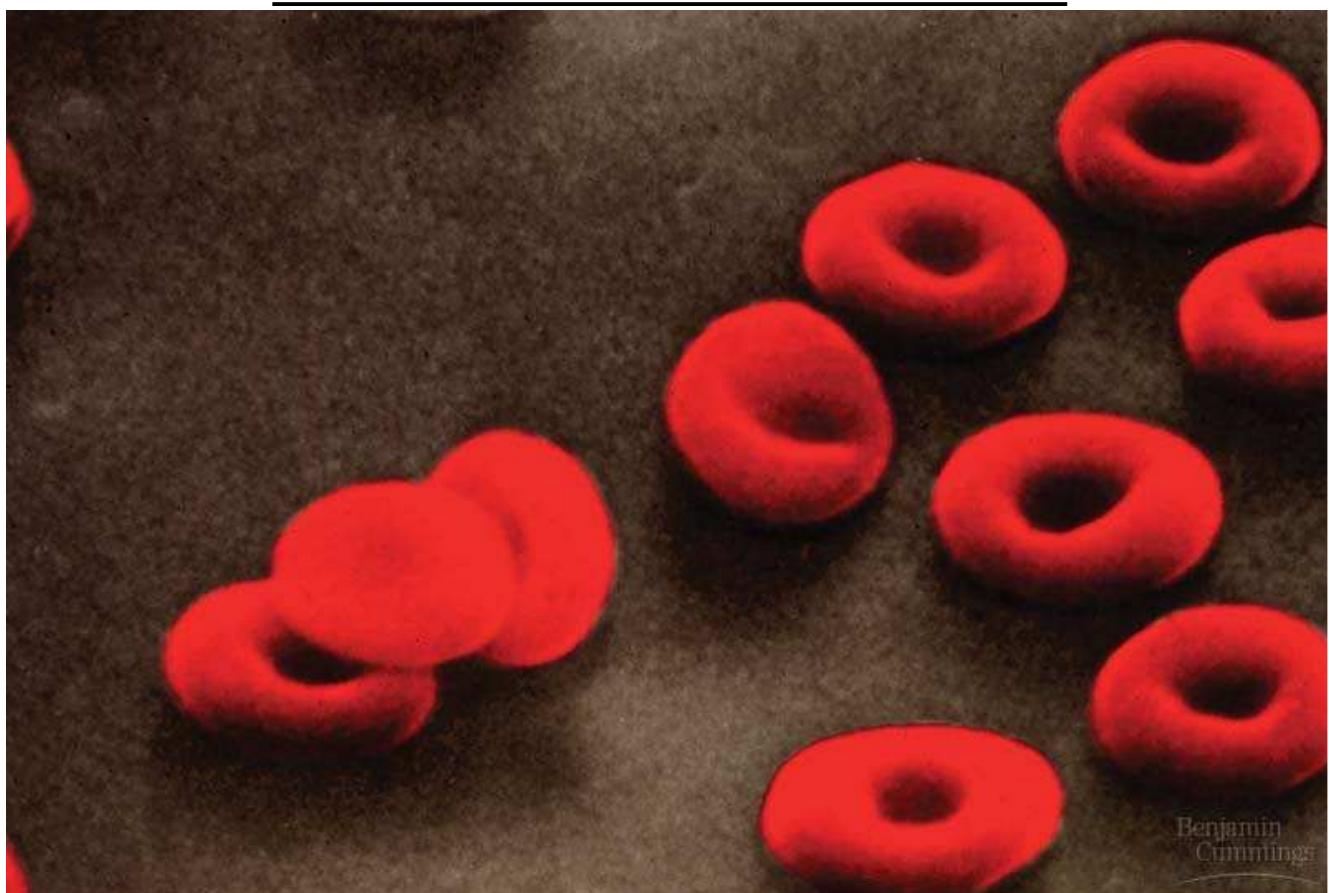
Figure 14.13



## Gene Disorders Due to Protein Alteration

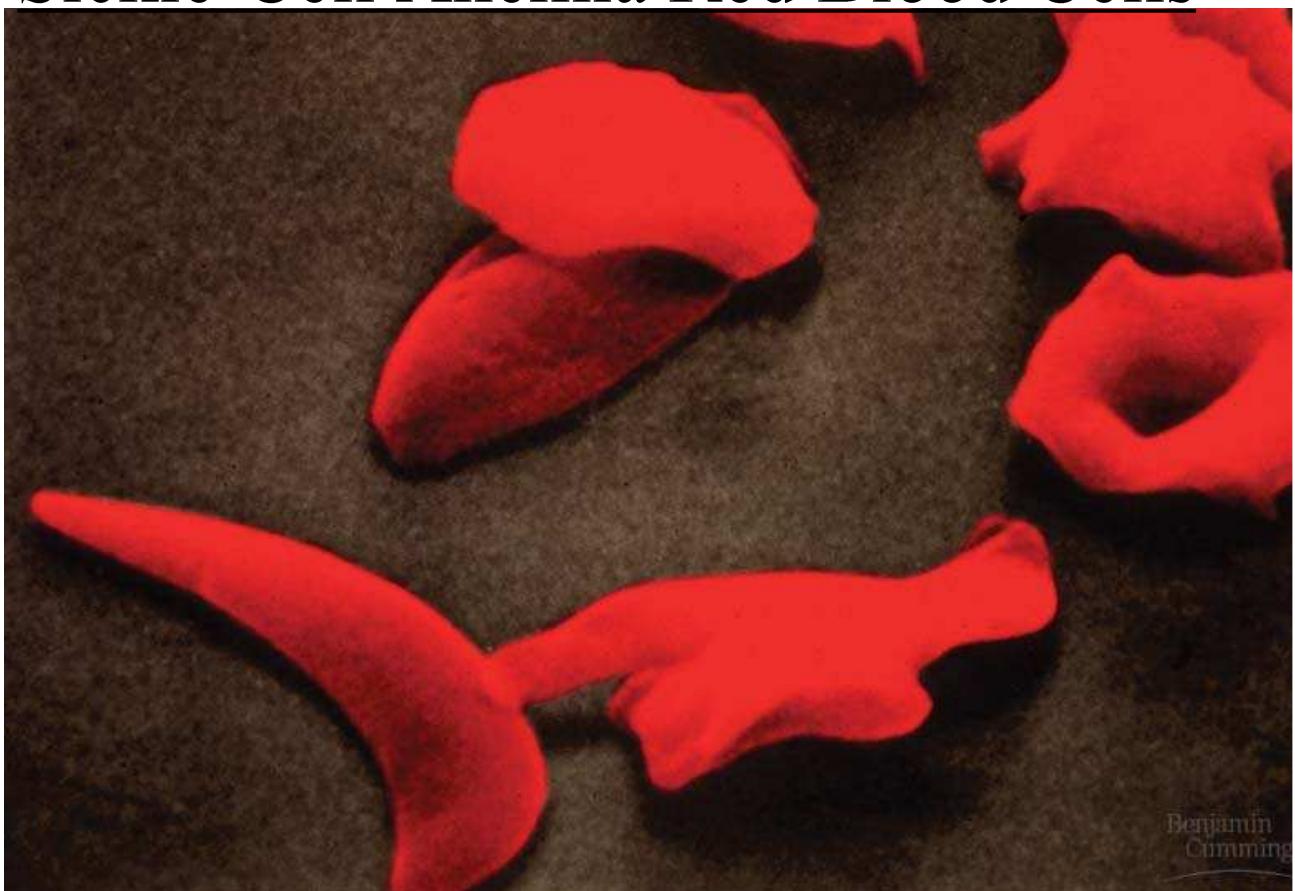
- Sickle-cell anemia is a recessive inherited disorder in which afflicted individuals have defective hemoglobin, and thus are unable to properly transport oxygen to tissues.
  - Homozygotes have sickle-cell
  - Heterozygotes usually appear normal, but are resistant to malaria
  - your book discusses SCA as being recessive
  - we will consider it Co-dominant

## Normal Red Blood Cells



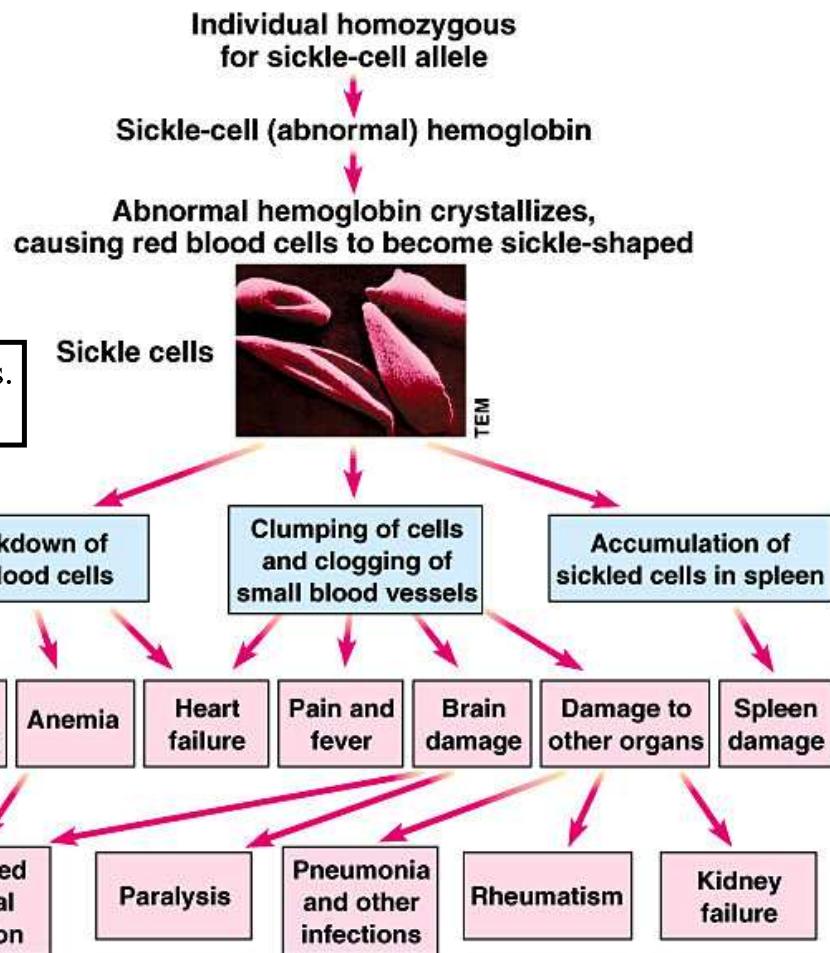
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## Sickle-Cell Anemia Red Blood Cells



# Sickle-Cell Anemia

One Gene causes many traits.  
What is this an example of?



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## **Sickle-Cell Anemia**

- causes red blood cells to bend = rigid = get stuck in capillaries = blockage = can be fatal
- Hemoglobin = protein that carries O<sub>2</sub> on rbc
  - change in 1 DNA base = SCA
  - valine substitutes glutamic acid  
= (substitution point mutation)
- SCA is recessive (per AP central = the way we will use it)  
or Co-dominant/semi-dominant  
and is also an example of Pleitropy

Why is the relationship with Malaria?

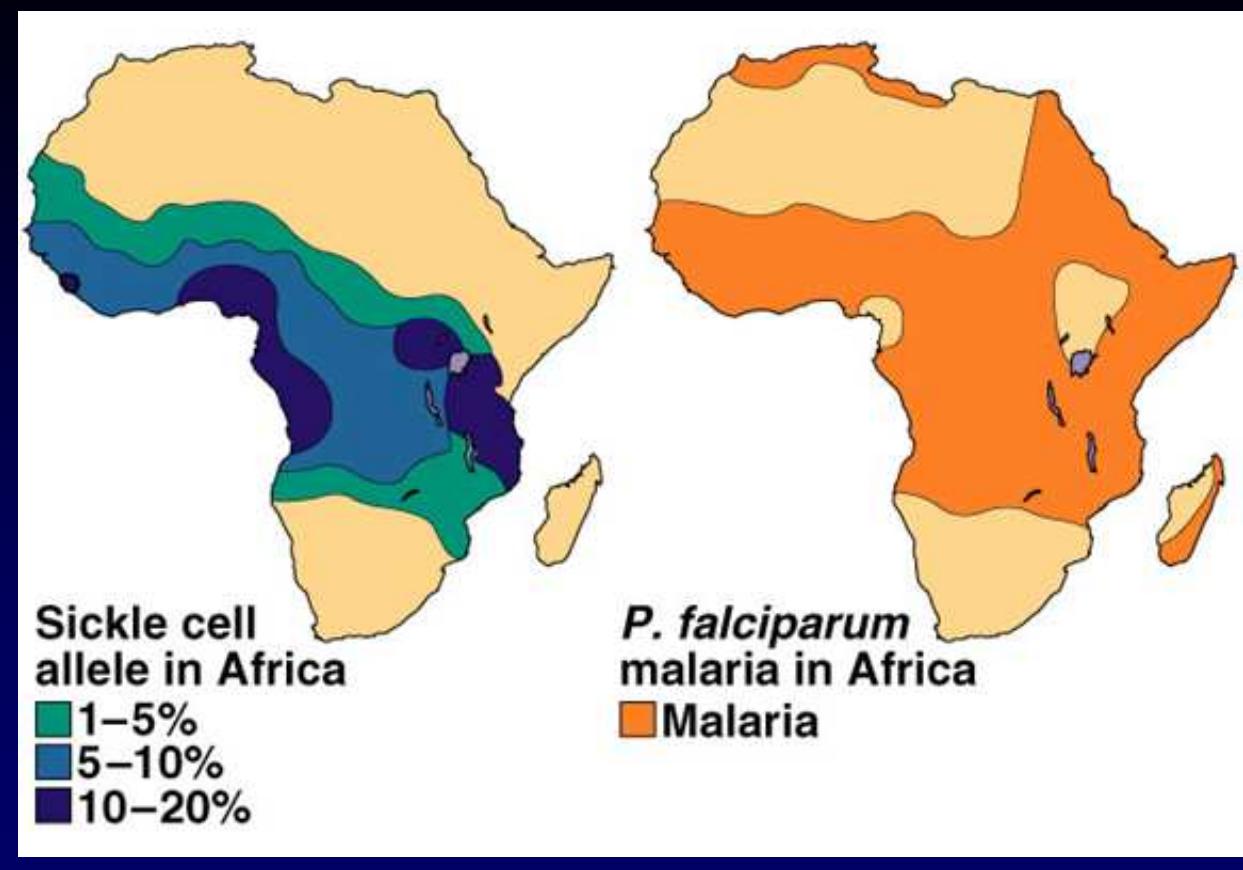
- SCA allows for resistance to the malaria protist.
- N = normal blood    n = SCA
- three genotypes = NN, Nn, nn
- What happens in each case?

nn =

Nn =

NN =

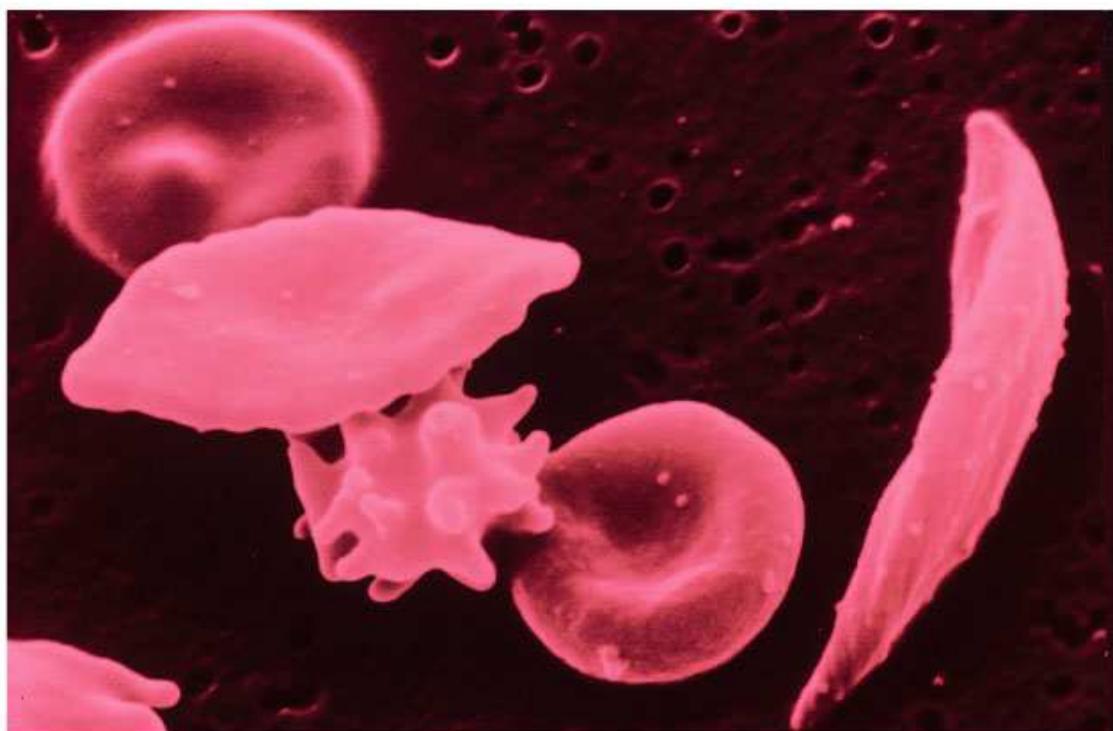
## Sickle Cell and Malaria



## Codominant alleles: sickle-cell

<http://animal.discovery.com/videos/monsters-inside-me-malaria-parasite.html>

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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

Figure 14.14



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Color variation due to range of soil pH

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

## **Integrating a Mendelian View of Heredity and Variation**

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

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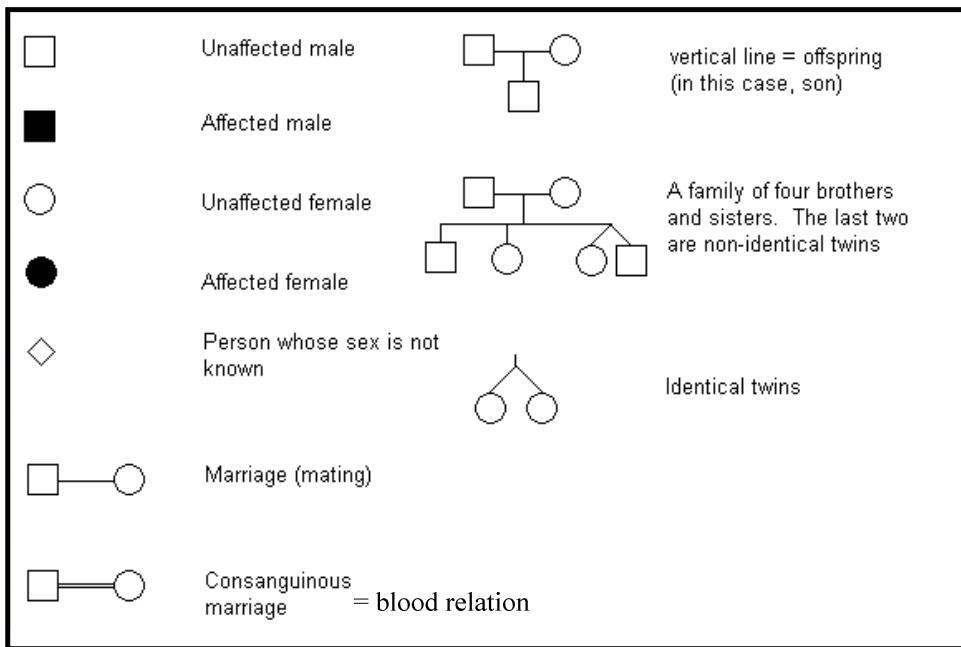
## **Concept 14.4: Many human traits follow Mendelian patterns of inheritance**

- 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

## Pedigree: a graphical representation of matings over multiple generations for a particular trait.

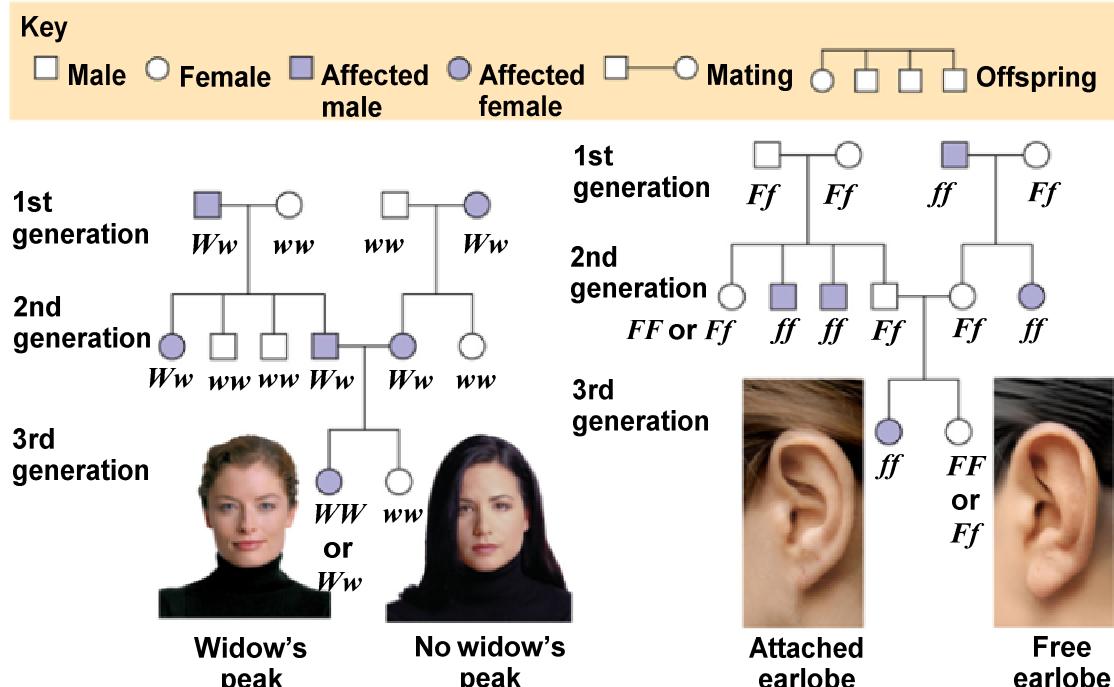


## Standardized Pedigree Symbols and Relationships

<input type="checkbox"/> Male		Couple (horizontal line connects mates)
<input type="checkbox"/> Female		Offspring (vertical line connects parents with offspring)
<input type="checkbox"/> Sex unspecified		Adopted in
<input type="checkbox"/> Proband		Adopted out
		Monozygotic twins
		Dizygotic twins
		Zygosity unknown

	Male	Female	Sex Unknown
Individual			
	b. 1925	30 y	4 mo
Affected individual (Define shading in key/legend)			
Affected individual (more than one condition)			
Multiple individuals, number known			
Multiple individuals, number unknown			
Deceased individual			
Stillbirth (SB)			
Pregnancy (P)			
Spontaneous abortion (SAB)			
Affected SAB			
Termination of pregnancy (TOP)			
Affected TOP			

Figure 14.15



(a) Is a widow's peak a dominant or recessive trait?

(b) Is an attached earlobe a dominant or recessive trait?

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Figure 14.15a



**Widow's  
peak**



**No  
widow's  
peak**

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Figure 14.15c



**Attached  
earlobe**

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**Free  
earlobe**

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- 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

## Sex Linkage

- A trait determined by a gene on the sex chromosome is said to be **sex-linked**.
  - In *Drosophila*, sex is determined by the number of copies of the x chromosome.
    - Mendelian traits assort independently because chromosomes assort independently.
  - 3 Sex-Linked Disorders where human males have a greater chance of the disorder - Why?  
Hemophilia  
Color Blindness  
Duchennes Muscular Dystrophy

## Pedigrees

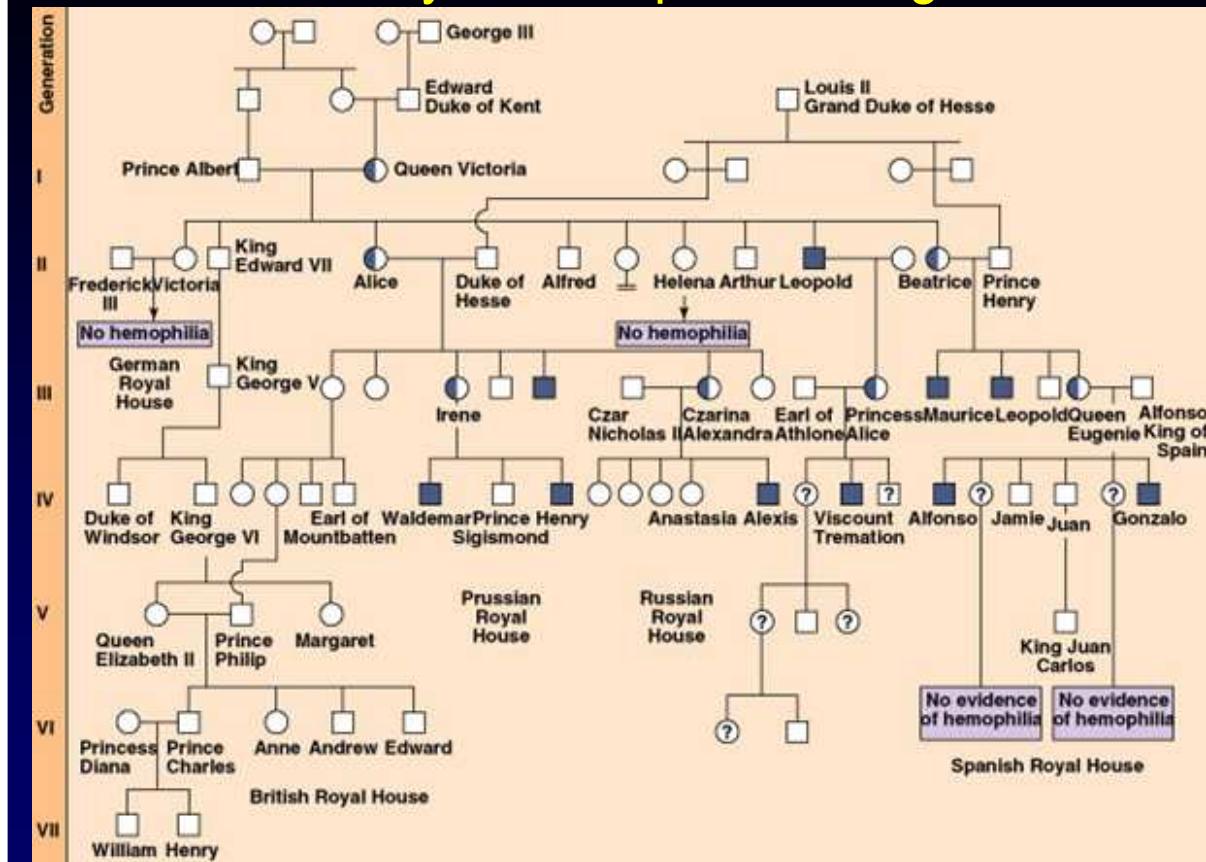
- Mutations are accidental changes in genes.
  - rare, random, and usually result in recessive alleles
    - pedigrees used to study heredity
  - **hemophilia** - inherited condition where blood is slow to clot or does not clot at all
    - only expressed when individual has no copies of the normal allele
    - Royal hemophilia - sex-linked

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Queen Victoria of England - 1894

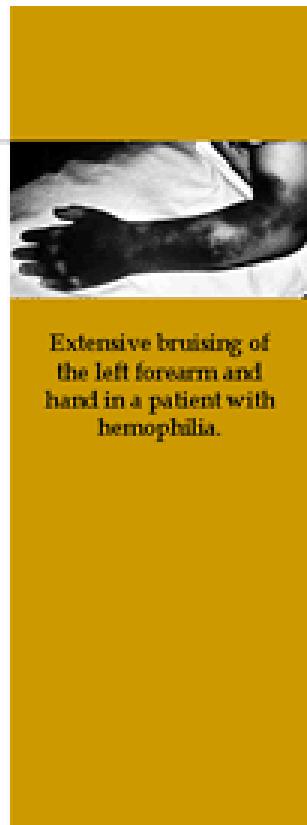
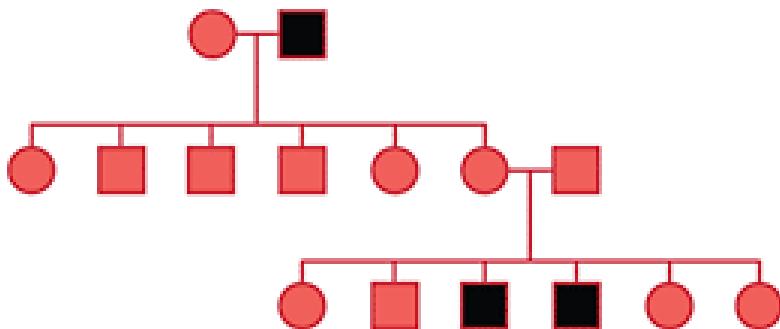
## Royal Hemophilia Pedigree



## Sex-Linkage:

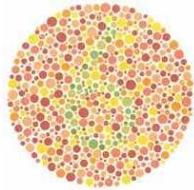
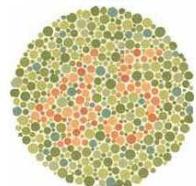
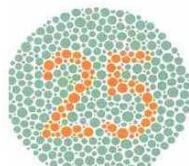
### Hemophilia: An Example

- In this pedigree, only males are affected, and sons do not share the phenotypes of their fathers.
  - Thus, hemophilia is linked to a sex chromosome—the X.
- Expression of hemophilia skips generations.
  - Thus, it is recessive.

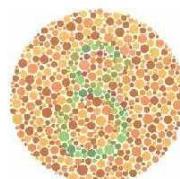
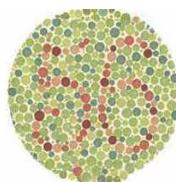
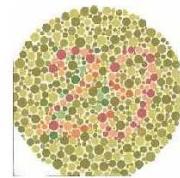


**Normal Color Vision**

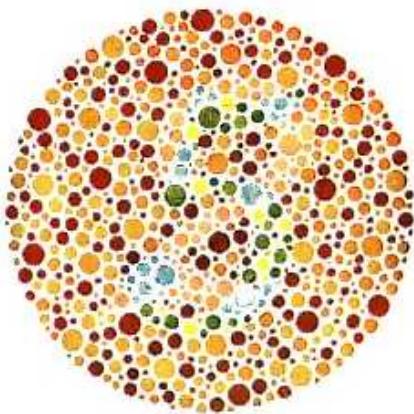
	Left	Right
Top	25	29
Middle	45	56
Bottom	6	8

**Red-Green Color Blind**

	Left	Right
Top	25	Spots
Middle	Spots	56
Bottom	Spots	Spots



**The test to the left is simpler.  
The individual with normal color vision will see a 5  
revealed in the dot pattern.  
An individual with Red/Green (the most common) color  
blindness will see a 2 revealed in the dots.**



## **Pedigree Problem:**

A normal vision man marries a woman who is a carrier for colorblindness. They have 6 children. The first, second, and fourth children are normal girls. The first and last sons have normal vision, but the second son born is colorblind. The second daughter marries a normal man and they have one child, a son, who is colorblind. Complete the pedigree of this family, including all proper symbols and genotypes.

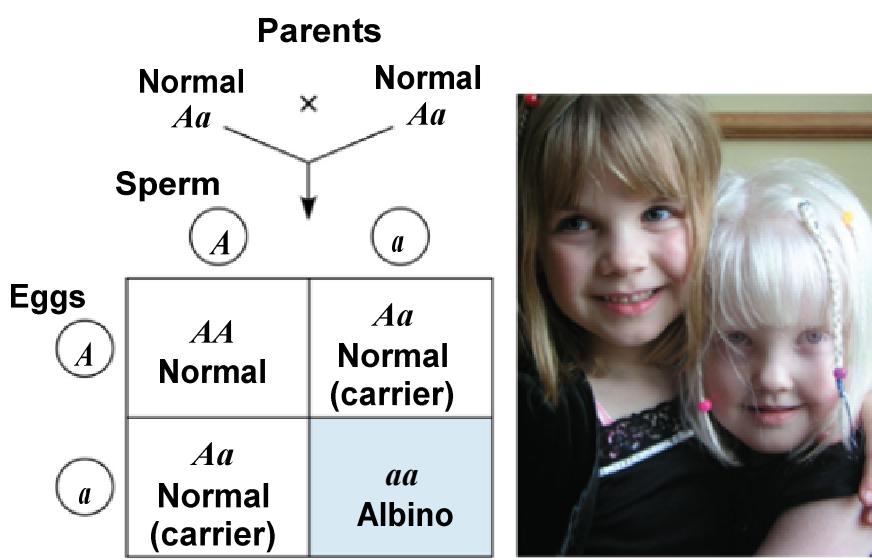
## **Recessively Inherited Disorders**

- 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



- 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

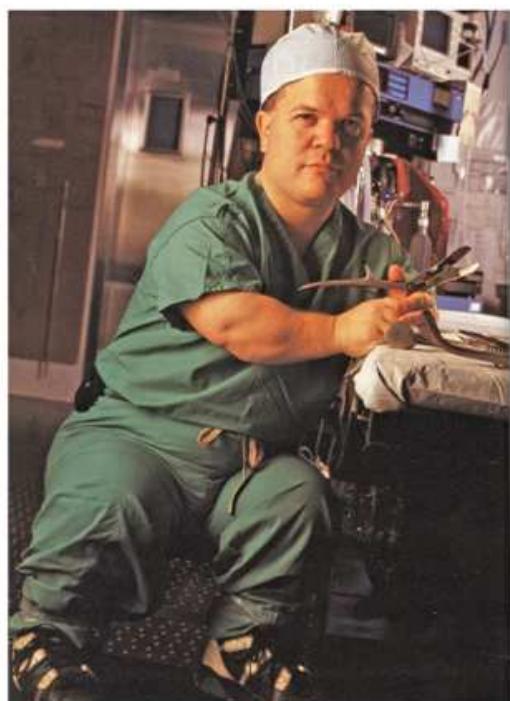
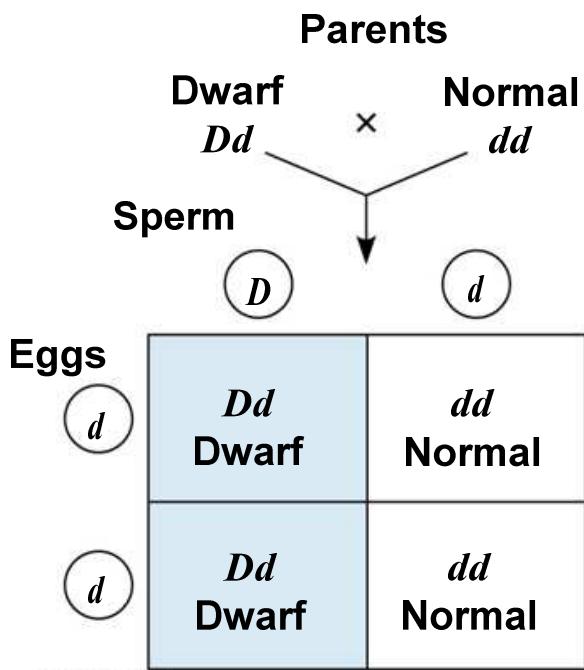
- Heterozygotes (said to have sickle-cell trait) are usually healthy but may suffer some symptoms
- About one out of ten African Americans has sickle cell trait, an unusually high frequency of an allele with detrimental effects in homozygotes
- Heterozygotes are less susceptible to the malaria parasite,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

## 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 14.17

### Achondroplasia = dominant



## ***Huntington's Disease: A Late-Onset Lethal Disease = dominant***

- 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
- Little is understood about the genetic contribution to most multifactorial diseases

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**Table 13.3** Some Important Genetic Disorders

Disorder	Symptom	Defect	Dominant/ Recessive	Frequency Among Human Births
Cystic fibrosis	Mucus clogs lungs, liver, and pancreas	Failure of chloride ion transport mechanism	Recessive	1/2500 (Caucasians)
Sickle cell anemia	Blood circulation is poor	Abnormal hemoglobin molecules	Recessive	1/625 (African Americans)
Tay-Sachs disease	Central nervous system deteriorates in infancy	Defective enzyme (hexosaminidase A)	Recessive	1/3500 (Ashkenazi Jews)
Phenylketonuria	Brain fails to develop in infancy	Defective enzyme (phenylalanine hydroxylase)	Recessive	1/12,000
Hemophilia	Blood fails to clot	Defective blood-clotting factor VIII	Sex-linked recessive	1/10,000 (Caucasian males)
Huntington disease	Brain tissue gradually deteriorates in middle age	Production of an inhibitor of brain cell metabolism	Dominant	1/24,000
Muscular dystrophy (Duchenne)	Muscles waste away	Degradation of myelin coating of nerves stimulating muscles	Sex-linked recessive	1/3700 (males)
Hypercholesterolemia	Excessive cholesterol levels in blood lead to heart disease	Abnormal form of cholesterol cell surface receptor	Dominant	1/500

## **Genetic Testing and Counseling**

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease
- Using family histories, genetic counselors help couples determine the odds that their children will have genetic disorders
- Probabilities are predicted on the most accurate information at the time; predicted probabilities may change as new information is available

## *Tests for Identifying Carriers*

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

Figure 14.18



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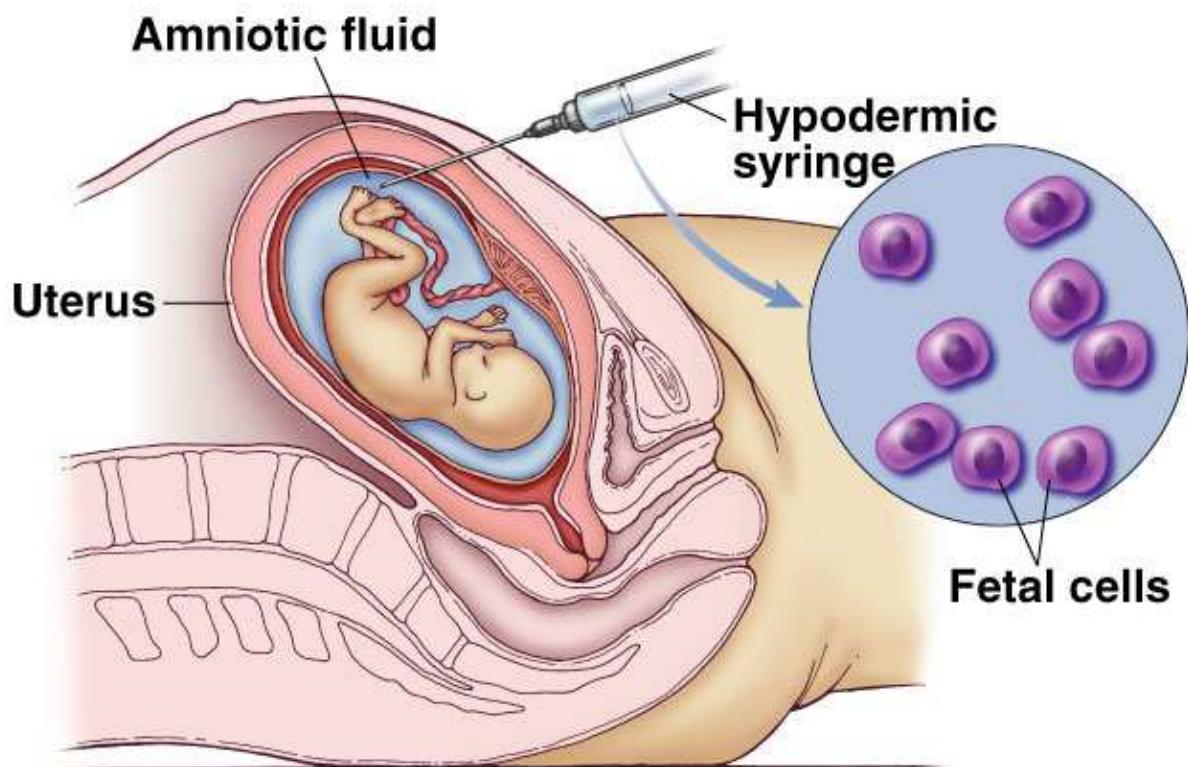
# Fetal Testing

## 3 Methods

## *Fetal Testing*

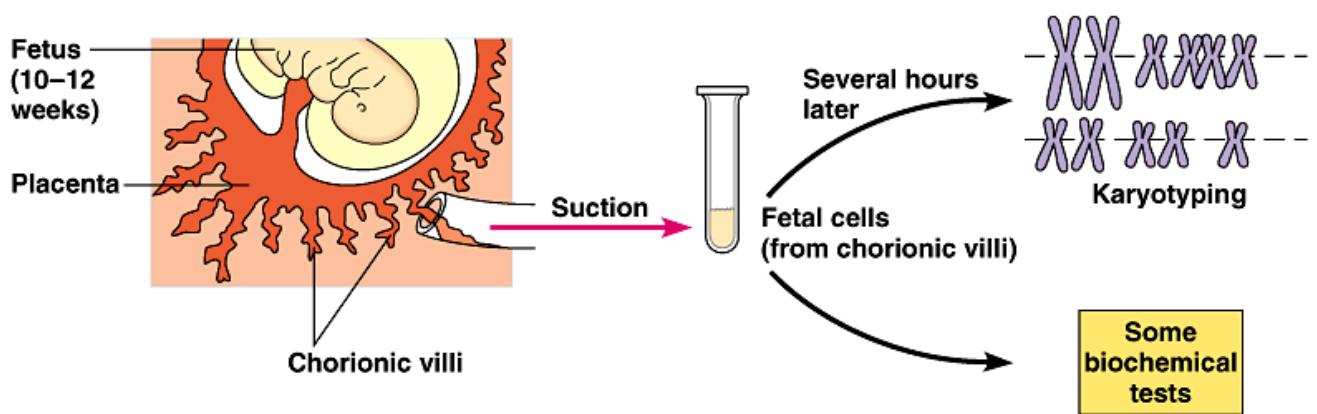
- In **amniocentesis**, the liquid that bathes the fetus is removed and tested
- In **chorionic villus sampling (CVS)**, a sample of the placenta is removed and tested
- Other techniques, such as *ultrasound* and *fetoscopy*, allow fetal health to be assessed visually in utero

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## Amniocentesis

# Chorionic Villus Biopsy (Sampling)



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# Ultrasound



## Ultrasound Image

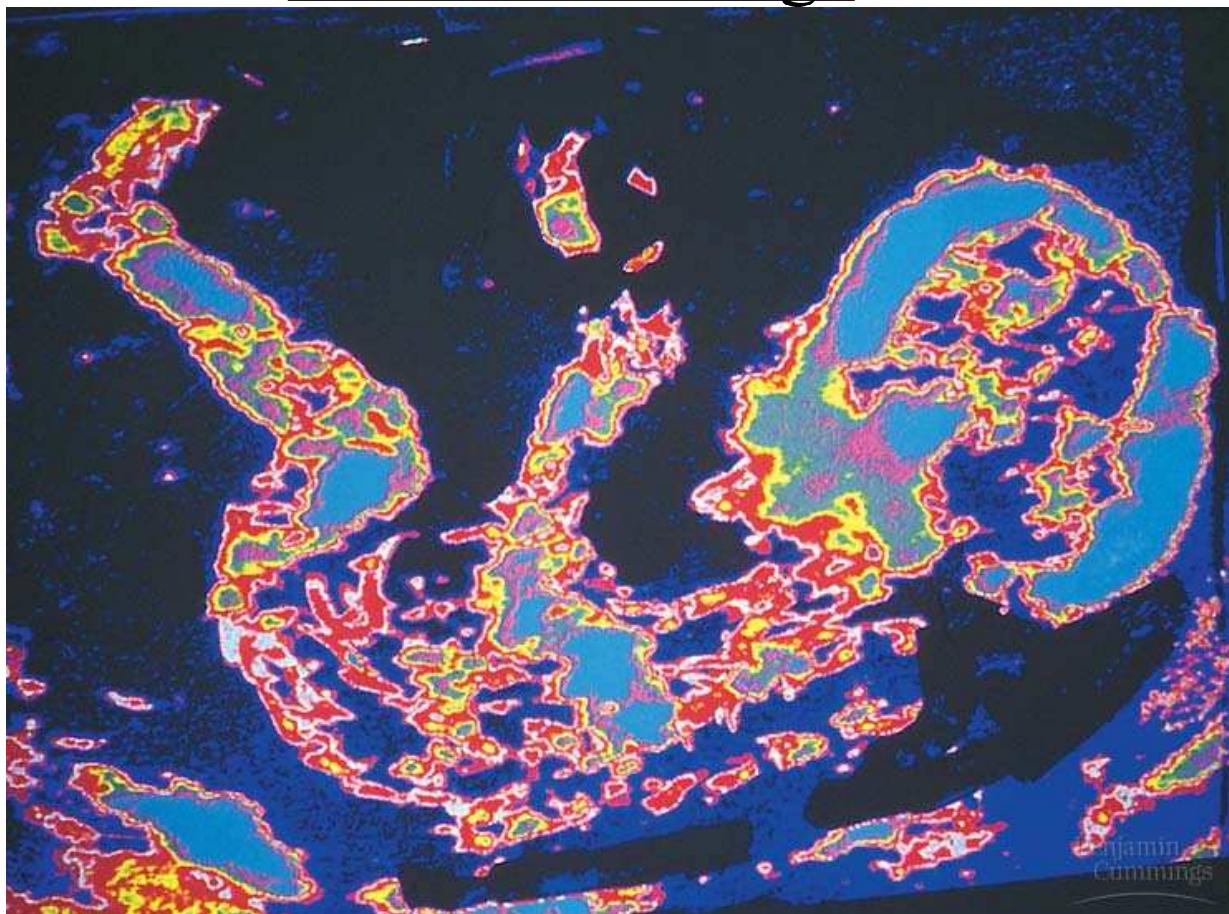
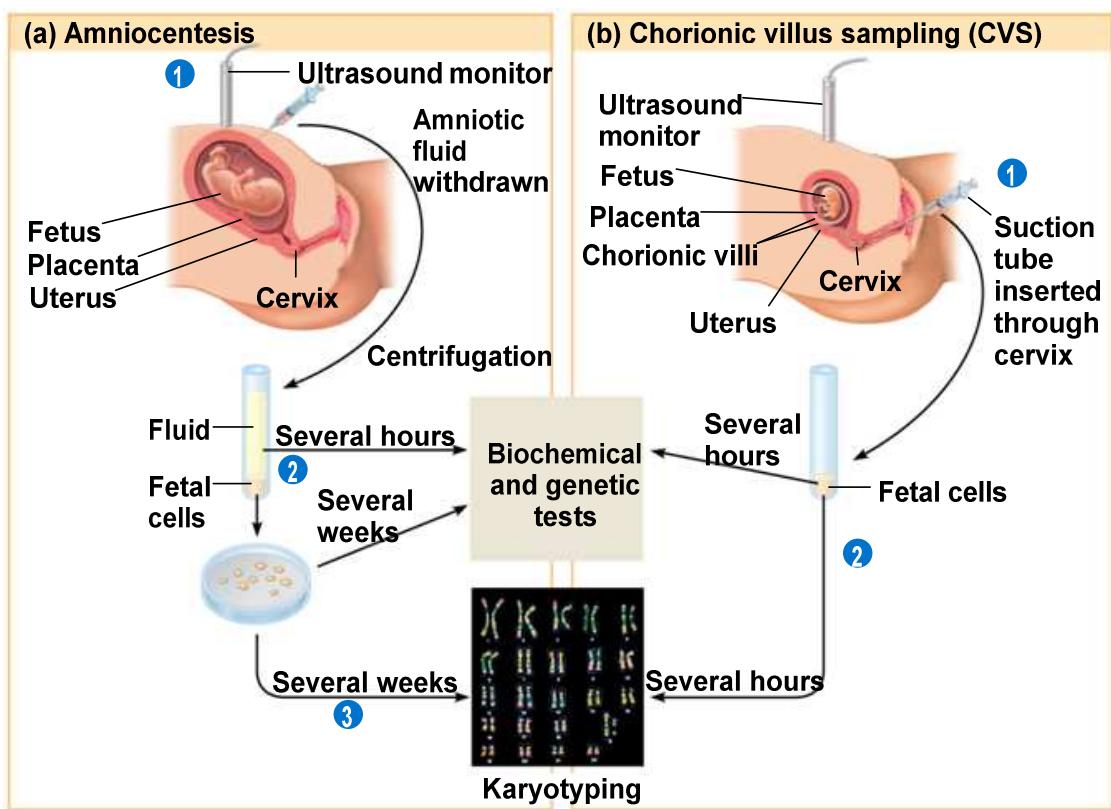


Figure 14.19



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## *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

Figure 14.UN03

Relationship among alleles of a single gene	Description	Example
Complete dominance of one allele	Heterozygous phenotype same as that of homozygous dominant	
Incomplete dominance of either allele	Heterozygous phenotype intermediate between the two homozygous phenotypes	 $C^R C^R$ $C^R C^W$ $C^W C^W$
Codominance	Both phenotypes expressed in heterozygotes	
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

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Figure 14.UN04

Relationship among two or more genes	Description	Example
<b>Epistasis</b>	<p>The phenotypic expression of one gene affects that of another</p>	<p><math>BbE \times BbE</math></p> <p>9 black : 3 brown : 4 tan</p>
<b>Polygenic inheritance</b>	<p>A single phenotypic character is affected by two or more genes</p>	<p><math>AaBbCc \times AaBbCc</math></p>

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additional reference pictures

Figure 14.UN05

<b><i>Character</i></b>	<b><i>Dominant</i></b>	<b><i>Recessive</i></b>
<b>Flower position</b>	<b>Axial (<i>A</i>)</b>	<b>Terminal (<i>a</i>)</b>
<b>Stem length</b>	<b>Tall (<i>T</i>)</b>	<b>Dwarf (<i>t</i>)</b>
<b>Seed shape</b>	<b>Round (<i>R</i>)</b>	<b>Wrinkled (<i>r</i>)</b>

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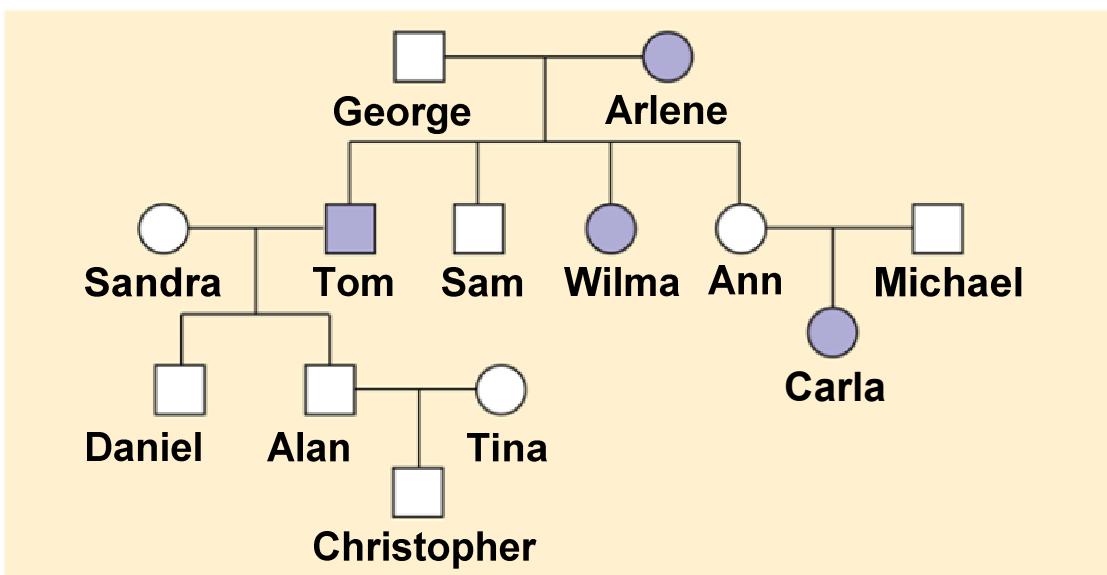
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Figure 14.UN06

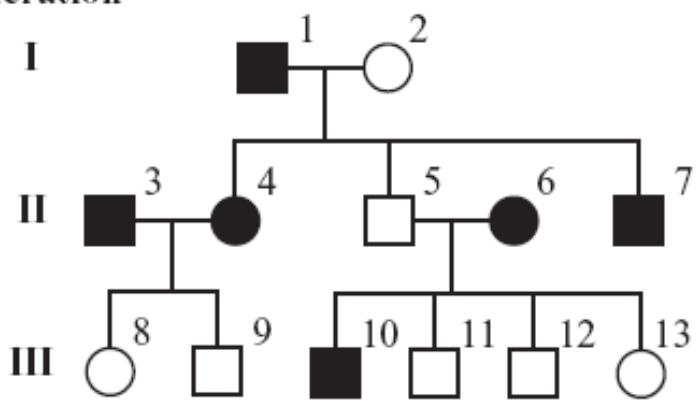


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Figure 14.UN07

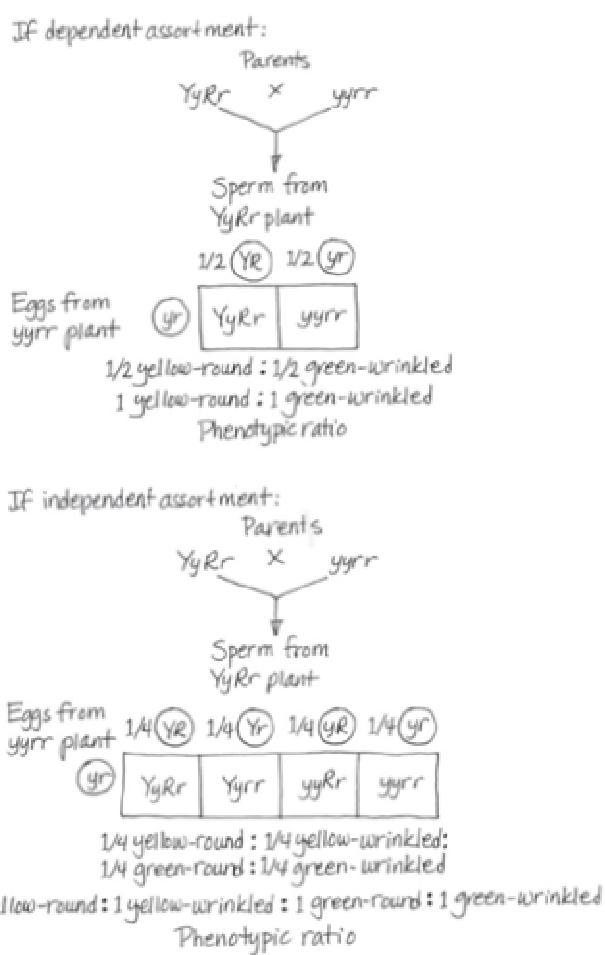


**Generation**



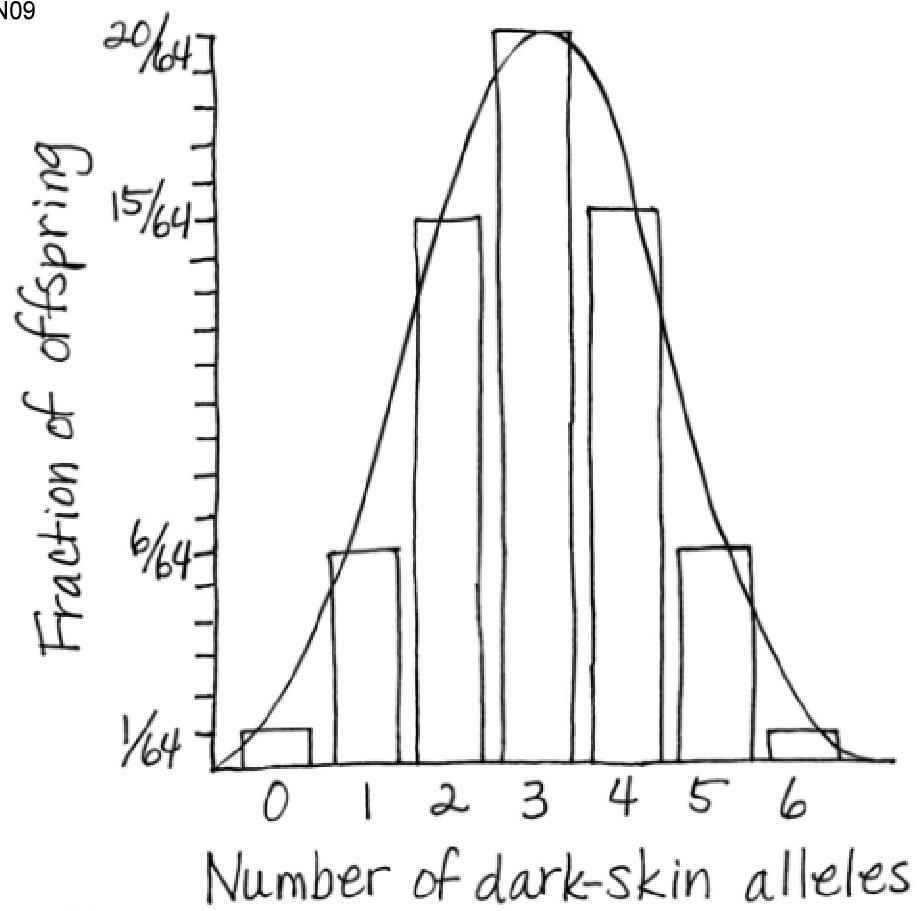
Key		
	Tune deaf	Normal tune perception
Male	■	□
Female	●	○

Figure 14.UN08



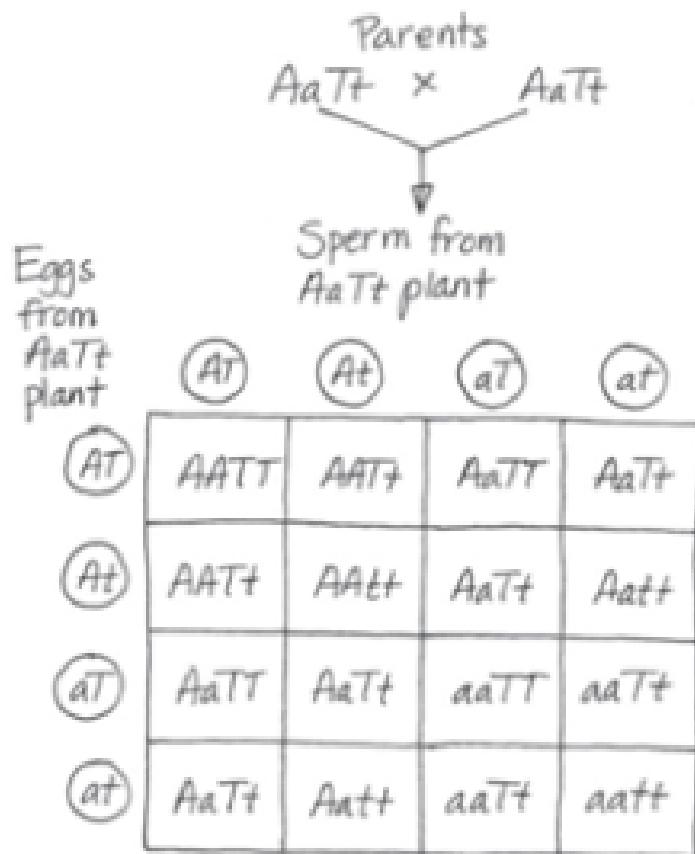
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Figure 14.UN09



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Figure 14.UN10



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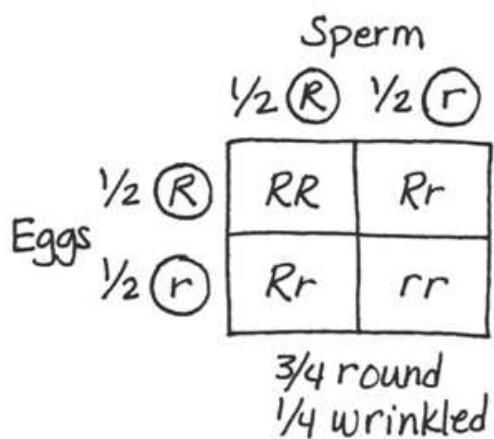
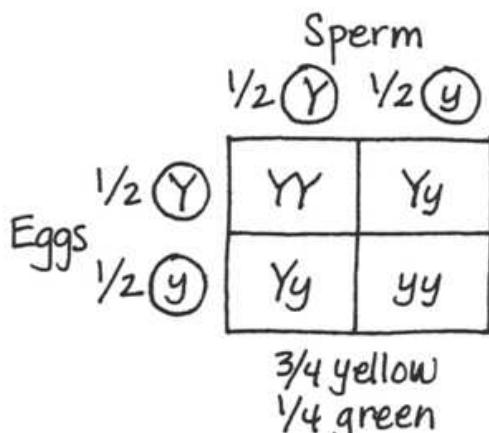
Figure 14.UN11

$ppyy\text{II}$	$\frac{1}{2}(\text{probability of } pp) \times \frac{1}{4}(yy) \times \frac{1}{2}(ii) = \frac{1}{16}$
$ppYy\text{ii}$	$\frac{1}{2}(pp) \times \frac{1}{2}(Yy) \times \frac{1}{2}(ii) = \frac{2}{16}$
$Pypy\text{ii}$	$\frac{1}{2}(Pp) \times \frac{1}{4}(yy) \times \frac{1}{2}(ii) = \frac{1}{16}$
$ppYY\text{ii}$	$\frac{1}{2}(pp) \times \frac{1}{4}(YY) \times \frac{1}{2}(ii) = \frac{1}{16}$
$ppyy\text{ii}$	$\frac{1}{2}(pp) \times \frac{1}{4}(yy) \times \frac{1}{2}(ii) = \frac{1}{16}$

---

Fraction predicted to have at least  
two recessive traits  $= \frac{6}{16}$  or  $\frac{3}{8}$

Figure 14.UN12



$$\frac{3}{4} \text{ yellow} \times \frac{3}{4} \text{ round} = \frac{9}{16} \text{ yellow-round}$$

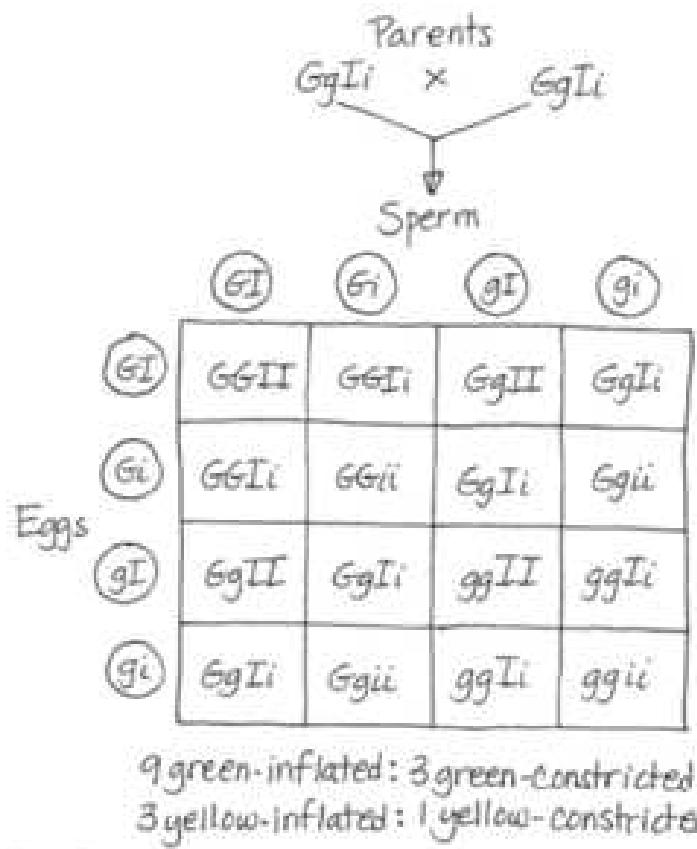
$$\frac{3}{4} \text{ yellow} \times \frac{1}{4} \text{ wrinkled} = \frac{3}{16} \text{ yellow-wrinkled}$$

$$\frac{1}{4} \text{ green} \times \frac{3}{4} \text{ round} = \frac{3}{16} \text{ green-round}$$

$$\frac{1}{4} \text{ green} \times \frac{1}{4} \text{ wrinkled} = \frac{1}{16} \text{ green-wrinkled}$$

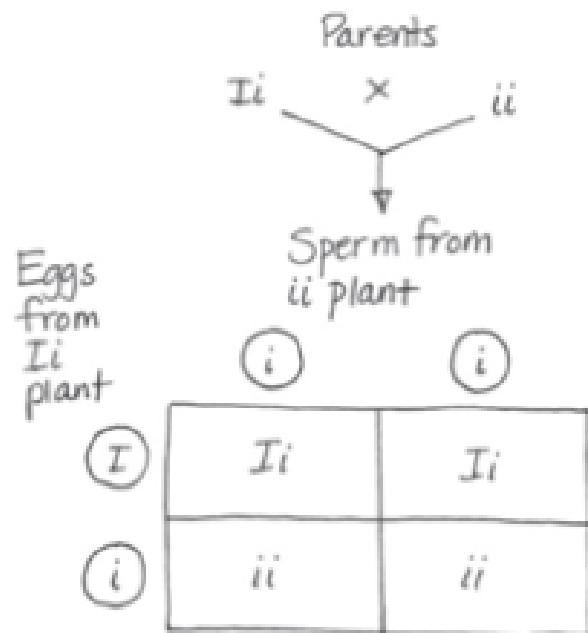
$= 9 \text{ yellow-round} : 3 \text{ yellow-wrinkled} : 3 \text{ green-round} : 1 \text{ green-wrinkled}$

Figure 14.UN13



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Figure 14.UN14



Genotypic ratio 1  $Ii$ : 1  $ii$   
(2:2 is equivalent)

Phenotypic ratio 1 inflated : 1 constricted  
(2:2 is equivalent)

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