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 phenotype
• **Tay-Sachs disease** is fatal; a dysfunctional enzyme causes an accumulation of lipids in the brain
  
  – At the *organismal* level, the allele is recessive
  
  – At the *biochemical* level, the phenotype (i.e., the enzyme activity level) is incompletely dominant
  
  – At the *molecular* level, the alleles are codominant
Frequency of Dominant Alleles

- Dominant alleles are not necessarily more common in populations than recessive alleles
- For example, one baby out of 400 in the United States is born with extra fingers or toes
• The allele for this unusual trait is dominant to the allele for the more common trait of five digits per appendage

• In this example, the recessive allele is far more prevalent than the population’s dominant allele
Multiple Alleles

• Most genes exist in populations in more than two allelic forms

• For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme (I) that attaches A or B carbohydrates to red blood cells: $I^A$, $I^B$, and $i$.

• The enzyme encoded by the $I^A$ allele adds the A carbohydrate, whereas the enzyme encoded by the $I^B$ allele adds the B carbohydrate; the enzyme encoded by the $i$ allele adds neither.
(a) The three alleles for the ABO blood groups and their associated carbohydrates

<table>
<thead>
<tr>
<th>Allele</th>
<th>Carbohydrate</th>
</tr>
</thead>
<tbody>
<tr>
<td>$\mathbf{A}$</td>
<td>A $\bigtriangleup$</td>
</tr>
<tr>
<td>$\mathbf{B}$</td>
<td>B $\bigcirc$</td>
</tr>
<tr>
<td>$i$</td>
<td>none</td>
</tr>
</tbody>
</table>

(b) Blood group genotypes and phenotypes

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Red blood cell appearance</th>
<th>Phenotype (blood group)</th>
</tr>
</thead>
<tbody>
<tr>
<td>$\mathbf{A}\mathbf{A}$ or $\mathbf{A}i$</td>
<td>![Red blood cell A]</td>
<td>A</td>
</tr>
<tr>
<td>$\mathbf{B}\mathbf{B}$ or $\mathbf{B}i$</td>
<td>![Red blood cell B]</td>
<td>B</td>
</tr>
<tr>
<td>$\mathbf{A}\mathbf{B}$</td>
<td>![Red blood cell AB]</td>
<td>AB</td>
</tr>
<tr>
<td>ii</td>
<td>![Red blood cell O]</td>
<td>O</td>
</tr>
</tbody>
</table>
Pleiotropy

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

• For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as cystic fibrosis and sickle-cell disease
Extending Mendelian Genetics for Two or More Genes

• Some traits may be determined by two or more genes
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.
Fig. 14-12

Sperm

1/4 BC

1/4 bC

1/4 Bc

1/4 bc

Eggs

1/4 BC

BBCC

BbCC

BBCc

BbCc

1/4 bc

BbCC

bbCC

BbCc

bbCc

1/4 BC

BBCc

BbCc

BBcc

Bbcc

1/4 bc

BbCc

bbCc

Bbcc

bbcc

9 : 3 : 4
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.
Eggs × Sperm

Phenotypes: Number of dark-skin alleles:

<table>
<thead>
<tr>
<th>0</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
<th>6</th>
</tr>
</thead>
<tbody>
<tr>
<td>( \frac{1}{64} )</td>
<td>( \frac{6}{64} )</td>
<td>( \frac{15}{64} )</td>
<td>( \frac{20}{64} )</td>
<td>( \frac{15}{64} )</td>
<td>( \frac{6}{64} )</td>
<td>( \frac{1}{64} )</td>
</tr>
</tbody>
</table>
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.
• 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
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
Fig. 14-15

Key
- Male
- Female
- Affected male
- Affected female
- Mating
- Offspring, in birth order (first-born on left)

1st generation (grandparents)
- Ww
- ww
- Ww

2nd generation (parents, aunts, and uncles)
- Ww
- ww
- Ww
- Ww

3rd generation (two sisters)
- WW or Ww
- ww

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

1st generation (grandparents)
- Ff
- Ff
- Ff
- Ff

2nd generation (parents, aunts, and uncles)
- FF or Ff
- ff
- Ff
- Ff

3rd generation (two sisters)
- ff
- FF or Ff

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

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Fig. 14-15a

Key

- Male
- Female
- Affected male
- Affected female

Mating
Offspring, in birth order (first-born on left)
(a) Is a widow’s peak a dominant or recessive trait?

Widow’s peak

No widow’s peak

1st generation (grandparents)

2nd generation (parents, aunts, and uncles)

3rd generation (two sisters)
1st generation (grandparents)

2nd generation (parents, aunts, and uncles)

3rd generation (two sisters)

Attached earlobe

Free earlobe

(b) Is an attached earlobe a dominant or recessive trait?
• Pedigrees can also be used to make predictions about future offspring

• We can use the multiplication and addition rules to predict the probability of specific phenotypes
Recessively Inherited Disorders

• Many genetic disorders are inherited in a recessive manner
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 (i.e., pigmented)

- Albinism is a recessive condition characterized by a lack of pigmentation in skin and hair
### Sperm

<table>
<thead>
<tr>
<th>Egg</th>
<th>Sperm</th>
<th>Parents</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>Aa</td>
<td>Normal</td>
<td>AA</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Normal</td>
</tr>
<tr>
<td>a</td>
<td>Aa</td>
<td>Normal</td>
<td>Aa</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Normal</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Aa</td>
</tr>
<tr>
<td></td>
<td>a</td>
<td></td>
<td>Normal</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>aa</td>
</tr>
</tbody>
</table>

**Outcome:***

- **AA**: Normal
- **Aa**: Normal (carrier)
- **Aa**: Normal (carrier)
- **aa**: Albino

*Fig. 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.

• 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 400 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

• *Achondroplasia* is a form of dwarfism caused by a rare dominant allele
Fig. 14-17

Parents

Dwarf

Normal

Dd

×

dd

Sperm

Eggs

D

d

Dd Dwarf

Dd Dwarf

dd Normal

dd Normal

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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
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
Genetic Testing and Counseling

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease
Counseling Based on Mendelian Genetics and Probability Rules

• Using family histories, genetic counselors help couples determine the odds that their children will have genetic disorders.
Tests for Identifying Carriers

• For a growing number of diseases, tests are available that identify carriers 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 **ultrasound** and **fetoscopy**, allow fetal health to be assessed visually in utero.
Amniotic fluid withdrawn

Centrifugation

Fluid

Fetal cells

Several hours

Several weeks

Biochemical tests

Several weeks

Karyotyping

(a) Amniocentesis

Suction tube inserted through cervix

Fetal cells

Several hours

Several weeks

(b) Chorionic villus sampling (CVS)
Fig. 14-18a

(a) Amniocentesis

- Amniotic fluid withdrawn
- Centrifugation
- Fetus
- Placenta
- Uterus
- Cervix
- Fluid
- Fetal cells
- Several hours
- Several weeks
- Biochemical tests
- Several weeks
- Karyotyping

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(b) Chorionic villus sampling (CVS)

Biochemical tests

Suction tube inserted through cervix

Several hours

Fetal cells

Karyotyping

Several hours

Fetus

Chorionic villi

Placenta

Suction tube

Fig. 14-18b

Chorionic villus sampling (CVS)
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
<table>
<thead>
<tr>
<th>Degree of dominance</th>
<th>Description</th>
<th>Example</th>
</tr>
</thead>
<tbody>
<tr>
<td>Complete dominance of one allele</td>
<td>Heterozygous phenotype same as that of homozygous dominant</td>
<td><img src="image1" alt="PP" /> <img src="image2" alt="Pp" /></td>
</tr>
<tr>
<td>Incomplete dominance of either allele</td>
<td>Heterozygous phenotype intermediate between the two homozygous phenotypes</td>
<td><img src="image3" alt="C^R C^R" /> <img src="image4" alt="C^R C^W" /> <img src="image5" alt="C^W C^W" /></td>
</tr>
<tr>
<td>Codominance</td>
<td>Heterozygotes: Both phenotypes expressed</td>
<td><img src="image6" alt="μ^A μ^B" /></td>
</tr>
<tr>
<td>Multiple alleles</td>
<td>In the whole population, some genes have more than two alleles</td>
<td>ABO blood group alleles <img src="image7" alt="μ^A, μ^B, i" /></td>
</tr>
<tr>
<td>Pleiotropy</td>
<td>One gene is able to affect multiple phenotypic characters</td>
<td>Sickle-cell disease</td>
</tr>
</tbody>
</table>

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### Relationship among genes

<table>
<thead>
<tr>
<th>Relationship</th>
<th>Description</th>
<th>Example</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Epistasis</strong></td>
<td>One gene affects the expression of another</td>
<td><img src="image" alt="Epistasis Example" /></td>
</tr>
<tr>
<td><strong>Polygenic inheritance</strong></td>
<td>A single phenotypic character is affected by two or more genes</td>
<td><img src="image" alt="Polygenic Inheritance Example" /></td>
</tr>
</tbody>
</table>

**Epistasis Example**

- **Parental Genotypes**: $BbCc$ and $BbCc$
- **Genotypes of Offspring**:
  - $BC$, $bC$, $Bc$, $bc$
- **Ratio**: $9 : 3 : 4$

**Polygenic Inheritance Example**

- **Parental Genotypes**: $AaBbCc$ and $AaBbCc$
- **Genotypes of Offspring**:
  - $AABBCC$, $AABbCc$, $AaBbCc$, $aabbCC$, $AaBBcc$, $aaBbCc$, $AAbbCc$, $aabbcc$
- **Ratio**: $1 : 4 : 6 : 4 : 1$
Fig. 14-UN4

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If dependent assortment:

Parents
YyRr × yyrr

Sperm from YyRr plant
1/2 YR 1/2 yr

Eggs from yyrr plant
1/2 yellow-round : 1/2 green-wrinkled
1 : 1 Phenotypic ratio

If independent assortment:

Parents
YyRr × yyrr

Sperm from YyRr plant

Eggs from yyrr plant
1/4 YR 1/4 Yr 1/4 yr 1/4 yr

1/4 yellow-round : 1/4 yellow-wrinkled:
1/4 green-round : 1/4 green-wrinkled
1 : 1 : 1 : 1 Phenotypic ratio
Parents

\[ Ii \times ii \]

Sperm from
\[ ii \text{ plant} \]

Eggs from
\[ II \text{ plant} \]

\[
\begin{array}{cc}
Ii & Ii \\
ii & ii \\
\end{array}
\]

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

Phenotypic ratio: 1 inflated : 1 constricted (2:2 is equivalent)
Parents
AaTt × AaTt

Eggs from AaTt plant

Sperm from AaTt plant

<table>
<thead>
<tr>
<th>AT</th>
<th>At</th>
<th>aT</th>
<th>at</th>
</tr>
</thead>
<tbody>
<tr>
<td>AATT</td>
<td>AAT+</td>
<td>AaTT</td>
<td>AaTt</td>
</tr>
<tr>
<td>AAT+</td>
<td>AAT+</td>
<td>AaTT</td>
<td>Aatt</td>
</tr>
<tr>
<td>AaTT</td>
<td>AaTt</td>
<td>aaTT</td>
<td>aaTt</td>
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<tr>
<td>AaTt</td>
<td>Aatt</td>
<td>aaTt</td>
<td>aatt</td>
</tr>
</tbody>
</table>
\[
\begin{align*}
ppyy ii & \quad \frac{1}{2} (\text{probability of } pp) \times \frac{1}{4} (yy) \times \frac{1}{2} (ii) = \frac{1}{16} \\
ppYy ii & \quad \frac{1}{2} (pp) \times \frac{1}{2} (Yy) \times \frac{1}{2} (ii) = \frac{2}{16} = \frac{1}{8} \\
PpYY ii & \quad \frac{1}{2} (Pp) \times \frac{1}{4} (YY) \times \frac{1}{2} (ii) = \frac{1}{16} \\
ppYY ii & \quad \frac{1}{2} (pp) \times \frac{1}{4} (YY) \times \frac{1}{2} (ii) = \frac{1}{16} \\
ppyy ii & \quad \frac{1}{2} (pp) \times \frac{1}{4} (yy) \times \frac{1}{2} (ii) = \frac{1}{16}
\end{align*}
\]

Fraction predicted to have at least two recessive traits = \frac{6}{16} or \frac{3}{8}
Fig. 14-UN11

<table>
<thead>
<tr>
<th>Eggs</th>
<th>GI</th>
<th>Gi</th>
<th>gi</th>
<th>Gi</th>
</tr>
</thead>
<tbody>
<tr>
<td>GI</td>
<td>GGII</td>
<td>GGI</td>
<td>GgII</td>
<td>GgIi</td>
</tr>
<tr>
<td>Gi</td>
<td>GGIi</td>
<td>GGIi</td>
<td>GgII</td>
<td>Ggii</td>
</tr>
<tr>
<td>gi</td>
<td>GgII</td>
<td>GgII</td>
<td>ggII</td>
<td>ggII</td>
</tr>
<tr>
<td>gi</td>
<td>GgII</td>
<td>GgII</td>
<td>ggII</td>
<td>ggii</td>
</tr>
</tbody>
</table>

9 green-inflated: 3 green-constricted:
3 yellow-inflated: 1 yellow-constricted
You should now be able to:

1. Define the following terms: true breeding, hybridization, monohybrid cross, P generation, F\textsubscript{1} generation, F\textsubscript{2} generation

2. Distinguish between the following pairs of terms: dominant and recessive; heterozygous and homozygous; genotype and phenotype

3. Use a Punnett square to predict the results of a cross and to state the phenotypic and genotypic ratios of the F\textsubscript{2} generation
4. Explain how phenotypic expression in the heterozygote differs with complete dominance, incomplete dominance, and codominance

5. Define and give examples of pleiotropy and epistasis

6. Explain why lethal dominant genes are much rarer than lethal recessive genes

7. Explain how carrier recognition, fetal testing, and newborn screening can be used in genetic screening and counseling