Introduction: *Barking Up the Genetic Tree*

- Dogs are one of man’s longest genetics experiments
  - Dog breeds are the result of artificial selection
  - Populations of dogs became isolated from each other
  - Humans chose dogs with specific traits for breeding
  - Each breed has physical and behavioral traits due to a unique genetic makeup
- Sequencing of the dog’s genome shows evolutionary relationships between breeds
Ancestral canine

Wolf

Chinese Shar-Pei

Akita

Siberian Husky

Basenji

Alaskan Malamute

Afghan hound

Saluki

Rottweiler

Sheepdog

Retriever
MENDEL’S LAWS
9.1 The science of genetics has ancient roots

- Pangogenesis was an early explanation for inheritance
  - It was proposed by Hippocrates
  - Particles called pangenes came from all parts of the organism to be incorporated into eggs or sperm
  - Characteristics acquired during the parents’ lifetime could be transferred to the offspring
  - Aristotle rejected pangenesis and argued that instead of particles, the potential to produce the traits was inherited

- Blending was another idea, based on plant breeding
  - Hereditary material from parents mixes together to form an intermediate trait, like mixing paint
9.2 Experimental genetics began in an abbey garden

- Gregor Mendel discovered principles of genetics in experiments with the garden pea
  - Mendel showed that parents pass heritable factors to offspring (heritable factors are now called genes)
  - Advantages of using pea plants
    - Controlled matings
    - Self-fertilization or cross-fertilization
    - Observable characteristics with two distinct forms
    - True-breeding strains
Transferred pollen from stamens of white flower to carpel of purple flower.

Parents (P)

1. Removed stamens from purple flower.
2. Transferred pollen from stamens of white flower to carpel of purple flower.
Transferred pollen from stamens of white flower to carpel of purple flower

Parents (P)

Purple

1. Removed stamens from purple flower

Stamens

2. Transferred pollen from stamens of white flower to carpel of purple flower

Carpel

3. Pollinated carpel matured into pod

White


Pollinated carpel matured into pod
Transferred pollen from stamens of white flower to carpel of purple flower

Parents (P)

Purple

Removed stamens from purple flower

Pollinated carpel matured into pod

Planted seeds from pod

Offspring (F₁)
<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Purple</th>
<th>White</th>
</tr>
</thead>
<tbody>
<tr>
<td>Flower color</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Flower position</td>
<td>Axial</td>
<td>Terminal</td>
</tr>
<tr>
<td>Seed color</td>
<td>Yellow</td>
<td>Green</td>
</tr>
<tr>
<td>Seed shape</td>
<td>Round</td>
<td>Wrinkled</td>
</tr>
<tr>
<td>Pod shape</td>
<td>Inflated</td>
<td>Constricted</td>
</tr>
<tr>
<td>Pod color</td>
<td>Green</td>
<td>Yellow</td>
</tr>
<tr>
<td>Stem length</td>
<td>Tall</td>
<td>Dwarf</td>
</tr>
</tbody>
</table>
9.3 Mendel’s law of segregation describes the inheritance of a single character

- Example of a monohybrid cross
  - Parental generation: purple flowers × white flowers
  - F₁ generation: all plants with purple flowers
  - F₂ generation: 3/4 of plants with purple flowers, 1/4 of plants with white flowers

- Mendel needed to explain
  - Why one trait seemed to disappear in the F₁ generation
  - Why that trait reappeared in one quarter of the F₂ offspring
P generation (true-breeding parents)

Purple flowers × White flowers
P generation (true-breeding parents)

Purple flowers × White flowers

F₁ generation

All plants have purple flowers
P generation (true-breeding parents)

Purple flowers  White flowers

F₁ generation

All plants have purple flowers

F₂ generation

$\frac{3}{4}$ of plants have purple flowers  $\frac{1}{4}$ of plants have white flowers
9.3 Mendel’s law of segregation describes the inheritance of a single character

- Four Hypotheses

1. Genes are found in alternative versions called **alleles**; a genotype is the listing of alleles an individual carries for a specific gene.

2. For each characteristic, an organism inherits two alleles, one from each parent; the alleles can be the same or different.
   - A **homozygous** genotype has identical alleles.
   - A **heterozygous** genotype has two different alleles.
9.3 Mendel’s law of segregation describes the inheritance of a single character

- **Four Hypotheses**

  3. If the alleles differ, the dominant allele determines the organism’s appearance, and the recessive allele has no noticeable effect
    - The phenotype is the appearance or expression of a trait
    - The same phenotype may be determined by more than one genotype

  4. Law of segregation: Allele pairs separate (segregate) from each other during the production of gametes so that a sperm or egg carries only one allele for each gene
P plants

Genotypic ratio
1 \( PP \) : 2 \( Pp \) : 1 \( pp \)

Phenotypic ratio
3 purple : 1 white

\( F_1 \) plants (hybrids)

Gametes

Genetic makeup (alleles)
1 \( PP \) : 2 \( Pp \) : 1 \( pp \)

\( F_2 \) plants

Genotypic ratio
1 \( PP \) : 2 \( Pp \) : 1 \( pp \)

Phenotypic ratio
3 purple : 1 white

Gametes

Sperm

Eggs
9.4 Homologous chromosomes bear the alleles for each character

- For a pair of homologous chromosomes, alleles of a gene reside at the same locus
  - Homozygous individuals have the same allele on both homologues
  - Heterozygous individuals have a different allele on each homologue
Gene loci

Homozygous for the dominant allele

Homozygous for the recessive allele

Heterozygous

Genotype:

Genotype:  \( PP \)  Homozygous for the dominant allele

Genotype:  \( aa \)  Homozygous for the recessive allele

Genotype:  \( Bb \)  Heterozygous

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9.5 The law of independent assortment is revealed by tracking two characters at once

Example of a dihybrid cross

- Parental generation: round yellow seeds × wrinkled green seeds
- F₁ generation: all plants with round yellow seeds
- F₂ generation: 9/16 of plants with round yellow seeds
  3/16 of plants with round green seeds
  3/16 of plants with wrinkled yellow seeds
  1/16 of plants with wrinkled green seeds

Mendel needed to explain

- Why nonparental combinations were observed
- Why a 9:3:3:1 ratio was observed among the F₂ offspring
9.5 The law of independent assortment is revealed by tracking two characters at once

- **Law of independent assortment**
  - Each pair of alleles segregates independently of the other pairs of alleles during gamete formation.
  - For genotype $RrYy$, four gamete types are possible: $RY$, $Ry$, $rY$, and $ry$. 
Hypothesis: Dependent assortment

P generation

\[ RRYY \] \[ ryy \]

\[ \text{Gametes} \]

\[ RY \] \[ ry \]

\[ \text{Sperm} \]

\[ RrYy \]

\[ \text{Eggs} \]

\[ \frac{1}{2} RY \] \[ \frac{1}{2} ry \]

\[ \text{Hypothesized} \]

(not actually seen)

Hypothesis: Independent assortment

P generation

\[ RRYY \] \[ ryy \]

\[ \text{Gametes} \]

\[ RY \] \[ ry \]

\[ \text{Sperm} \]

\[ RrYy \]

\[ \text{Eggs} \]

\[ \frac{1}{4} RY \] \[ \frac{1}{4} rY \] \[ \frac{1}{4} Ry \] \[ \frac{1}{4} ry \]

\[ \text{Actual results} \]

(support hypothesis)

- **Yellow round**: \( \frac{9}{16} \)
- **Green round**: \( \frac{3}{16} \)
- **Yellow wrinkled**: \( \frac{3}{16} \)
- **Green wrinkled**: \( \frac{1}{16} \)
Phenotypes
Genotypes
Mating of heterozygotes (black, normal vision)

Phenotypic ratio of offspring

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9.6 Geneticists use the testcross to determine unknown genotypes

- **Testcross**
  - Mating between an individual of unknown genotype and a homozygous recessive individual
  - Will show whether the unknown genotype includes a recessive allele
  - Used by Mendel to confirm true-breeding genotypes
Testcross:

Genotypes

- $B_-$
- $bb$

Two possibilities for the black dog:

Gametes

- $BB ightarrow B$
- $b$

- $Bb ightarrow Bb$
- $bb$

Offspring

- All black
- 1 black : 1 chocolate
9.7 Mendel’s laws reflect the rules of probability

- The probability of a specific event is the number of ways that event can occur out of the total possible outcomes.

- **Rule of multiplication**
  - Multiply the probabilities of events that must occur together

- **Rule of addition**
  - Add probabilities of events that can happen in alternate ways
**F<sub>1</sub> genotypes**

**Formation of eggs**

*Bb* female

**Formation of sperm**

*Bb* male

**F<sub>2</sub> genotypes**

<table>
<thead>
<tr>
<th>B</th>
<th>b</th>
<th>B</th>
<th>b</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/4</td>
<td>1/4</td>
<td>1/4</td>
<td>1/4</td>
</tr>
</tbody>
</table>

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9.8 CONNECTION: Genetic traits in humans can be tracked through family pedigrees

- **A pedigree**
  - Shows the inheritance of a trait in a family through multiple generations
  - Demonstrates dominant or recessive inheritance
  - Can also be used to deduce genotypes of family members
Dominant Traits  Recessive Traits

Freckles  No freckles

Widow’s peak  Straight hairline

Free earlobe  Attached earlobe

Copyright © 2009 Pearson Education, Inc.
Widow’s peak

Straight hairline
First generation (grandparents)

Second generation (parents, aunts, and uncles)

Third generation (two sisters)

<table>
<thead>
<tr>
<th>Female</th>
<th>Male</th>
<th>Affected</th>
<th>Unaffected</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
9.9 CONNECTION: Many inherited disorders in humans are controlled by a single gene

- Inherited human disorders show
  - Recessive inheritance
    - Two recessive alleles are needed to show disease
    - Heterozygous parents are carriers of the disease-causing allele
    - Probability of inheritance increases with inbreeding, mating between close relatives
  - Dominant inheritance
    - One dominant allele is needed to show disease
    - Dominant lethal alleles are usually eliminated from the population
<table>
<thead>
<tr>
<th>Parents</th>
<th>Normal Dd</th>
<th>Normal Dd</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sperm</td>
<td>D</td>
<td>d</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Offspring</th>
<th>Eggs</th>
<th>Normal (carrier)</th>
<th>Deaf</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>DD Normal</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Dd Normal</td>
<td>dd</td>
</tr>
</tbody>
</table>

D: Normal

D: Normal (carrier)

d: Deaf
### Table 9.9 Some Autosomal Disorders in Humans

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Major Symptoms</th>
<th>Incidence</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Recessive disorders</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Albinism</td>
<td>Lack of pigment in skin, hair, and eyes</td>
<td>(\frac{1}{22,000})</td>
<td>Prone to skin cancer</td>
</tr>
<tr>
<td>Cystic fibrosis</td>
<td>Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated</td>
<td>(\frac{1}{2,500}) Caucasians</td>
<td>See Module 9.9</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>Accumulation of galactose in tissues; mental retardation; eye and liver damage</td>
<td>(\frac{1}{100,000})</td>
<td>Treated by eliminating galactose from diet</td>
</tr>
<tr>
<td>Phenylketonuria (PKU)</td>
<td>Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation</td>
<td>(\frac{1}{10,000}) in U.S. and Europe</td>
<td>See Module 9.10</td>
</tr>
<tr>
<td>Sickle-cell disease</td>
<td>Sickled red blood cells; damage to many tissues</td>
<td>(\frac{1}{400}) African-Americans</td>
<td>See Module 9.13</td>
</tr>
<tr>
<td>Tay-Sachs disease</td>
<td>Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood</td>
<td>(\frac{1}{3,500}) Jews from central Europe</td>
<td>See Module 4.11</td>
</tr>
<tr>
<td><strong>Dominant disorders</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Achondroplasia</td>
<td>Dwarfism</td>
<td>(\frac{1}{25,000})</td>
<td>See Module 9.9</td>
</tr>
<tr>
<td>Alzheimer’s disease</td>
<td>Mental deterioration; usually strikes late in life</td>
<td>Not known</td>
<td></td>
</tr>
<tr>
<td>(one type)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Huntington’s disease</td>
<td>Mental deterioration and uncontrollable movements; strikes in middle age</td>
<td>(\frac{1}{25,000})</td>
<td>See Module 9.9</td>
</tr>
<tr>
<td>Hypercholesterolemia</td>
<td>Excess cholesterol in blood; heart disease</td>
<td>(\frac{1}{500}) are heterozygous</td>
<td>See Module 9.11</td>
</tr>
<tr>
<td>Disorder</td>
<td>Major Symptoms</td>
<td>Incidence</td>
<td></td>
</tr>
<tr>
<td>-------------------------</td>
<td>--------------------------------------------------------------------------------</td>
<td>----------------------------</td>
<td></td>
</tr>
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</tr>
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<td></td>
</tr>
<tr>
<td>Tay-Sachs disease</td>
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<td>( \frac{1}{3,500} ) Jews from central Europe</td>
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</tr>
<tr>
<td><strong>Dominant disorders</strong></td>
<td></td>
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<td>Dwarfism</td>
<td>( \frac{1}{25,000} )</td>
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</tr>
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<td>Mental deterioration; usually strikes late in life</td>
<td>Not known</td>
<td></td>
</tr>
<tr>
<td>Huntington’s disease</td>
<td>Mental deterioration and uncontrollable movements; strikes in middle age</td>
<td>( \frac{1}{25,000} )</td>
<td></td>
</tr>
<tr>
<td>Hypercholesterolemia</td>
<td>Excess cholesterol in blood; heart disease</td>
<td>( \frac{1}{500} ) are heterozygous</td>
<td></td>
</tr>
</tbody>
</table>
9.10 CONNECTION: New technologies can provide insight into one’s genetic legacy

- Genetic testing of parents
- Fetal testing: biochemical and karyotype analyses
  - Amniocentesis
  - Chorionic villus sampling
- Maternal blood test
- Fetal imaging
  - Ultrasound
  - Fetoscopy
- Newborn screening
Amniocentesis

- Ultrasound monitor
- Needle inserted through abdomen to extract amniotic fluid
- Fetus
- Placenta
- Uterus

Chorionic villus sampling (CVS)

- Ultrasound monitor
- Suction tube inserted through cervix to extract tissue from chorionic villi
- Fetus
- Placenta
- Chorionic villi
- Cervix

Amniotic fluid
Fetal cells

Centrifugation

Several weeks

Biochemical tests

Karyotyping

Several hours
VARIATIONS ON MENDEL’S LAWS
9.11 Incomplete dominance results in intermediate phenotypes

- **Incomplete dominance**
  - Neither allele is dominant over the other
  - Expression of both alleles is observed as an intermediate phenotype in the heterozygous individual
P generation

Red

RR

White

rr

×

Gametes

R

r

F₁ generation

Pink

Rr

Gametes

\frac{1}{2} R \quad \frac{1}{2} r

Sperm

\frac{1}{2} R \quad \frac{1}{2} r

F₂ generation

Eggs

\frac{1}{2} R

\frac{1}{2} r

\frac{1}{2} R \quad \frac{1}{2} r

\frac{1}{2} Rr \quad \frac{1}{2} rr
**Genotypes:**

- **HH** Homozygous for inability to make LDL receptors
- **Hh** Heterozygous for inability to make LDL receptors
- **hh** Homozygous for inability to make LDL receptors

**Phenotypes:**

- Normal
- Mild disease
- Severe disease
Many genes have more than two alleles in the population

- **Multiple alleles**
  - More than two alleles are found in the population
  - A diploid individual can carry any two of these alleles
  - The ABO blood group has three alleles, leading to four phenotypes: type A, type B, type AB, and type O blood
9.12 Many genes have more than two alleles in the population

- **Codominance**
  - Neither allele is dominant over the other
  - Expression of both alleles is observed as a distinct phenotype in the heterozygous individual
  - Observed for type AB blood
<table>
<thead>
<tr>
<th>Blood Group (Phenotype)</th>
<th>Genotypes</th>
<th>Red Blood Cells</th>
<th>Antibodies Present in Blood</th>
<th>Reaction When Blood from Groups Below Is Mixed with Antibodies from Groups at Left</th>
</tr>
</thead>
<tbody>
<tr>
<td>O</td>
<td>ii</td>
<td><img src="image" alt="Red Blood Cells" /></td>
<td>Anti-A Anti-B</td>
<td><img src="chart" alt="Blood Types" /></td>
</tr>
<tr>
<td>A</td>
<td>I^A^A or I^A^i</td>
<td><img src="image" alt="Carbohydrate A" /></td>
<td>Anti-B</td>
<td><img src="chart" alt="Blood Types" /></td>
</tr>
<tr>
<td>B</td>
<td>I^B^B or I^B^i</td>
<td><img src="image" alt="Carbohydrate B" /></td>
<td>Anti-A</td>
<td><img src="chart" alt="Blood Types" /></td>
</tr>
<tr>
<td>AB</td>
<td>I^A^B</td>
<td><img src="image" alt="Red Blood Cells" /></td>
<td>—</td>
<td><img src="chart" alt="Blood Types" /></td>
</tr>
<tr>
<td>Blood Group (Phenotype)</td>
<td>Genotypes</td>
<td>Red Blood Cells</td>
<td></td>
<td></td>
</tr>
<tr>
<td>------------------------</td>
<td>-----------</td>
<td>-----------------</td>
<td></td>
<td></td>
</tr>
<tr>
<td>O</td>
<td>ii</td>
<td><img src="image" alt="Image of red blood cells with carbohydrate A" /> Carbohydrate A</td>
<td></td>
<td></td>
</tr>
<tr>
<td>A</td>
<td>$I^A I^A$ or $I^A i$</td>
<td><img src="image" alt="Image of red blood cells with carbohydrate A" /> Carbohydrate A</td>
<td></td>
<td></td>
</tr>
<tr>
<td>B</td>
<td>$I^B I^B$ or $I^B i$</td>
<td><img src="image" alt="Image of red blood cells with carbohydrate B" /> Carbohydrate B</td>
<td></td>
<td></td>
</tr>
<tr>
<td>AB</td>
<td>$I^A I^B$</td>
<td><img src="image" alt="Image of red blood cells with carbohydrate A" /> Carbohydrate A</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Blood Group (Phenotype)</td>
<td>Antibodies Present in Blood</td>
<td>Reaction When Blood from Groups Below Is Mixed with Antibodies from Groups at Left</td>
<td></td>
<td></td>
</tr>
<tr>
<td>------------------------</td>
<td>-----------------------------</td>
<td>---------------------------------------------------------------------------------</td>
<td></td>
<td></td>
</tr>
<tr>
<td>O</td>
<td>Anti-A, Anti-B</td>
<td><img src="image-url" alt="Image of reactions" /></td>
<td></td>
<td></td>
</tr>
<tr>
<td>A</td>
<td>Anti-B</td>
<td><img src="image-url" alt="Image of reactions" /></td>
<td></td>
<td></td>
</tr>
<tr>
<td>B</td>
<td>Anti-A</td>
<td><img src="image-url" alt="Image of reactions" /></td>
<td></td>
<td></td>
</tr>
<tr>
<td>AB</td>
<td>—</td>
<td><img src="image-url" alt="Image of reactions" /></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
9.13 A single gene may affect many phenotypic characters

- **Pleiotropy**
  - One gene influencing many characteristics
  - The gene for sickle cell disease
    - Affects the type of hemoglobin produced
    - Affects the shape of red blood cells
    - Causes anemia
    - Causes organ damage
    - Is related to susceptibility to malaria
Individual homozygous for sickle-cell allele

Sickle-cell (abnormal) hemoglobin

Abnormal hemoglobin crystallizes, causing red blood cells to become sickle-shaped

Sickle cells

Breakdown of red blood cells
- Physical weakness
- Anemia

Clumping of cells and clogging of small blood vessels
- Heart failure
- Pain and fever
- Brain damage

Accumulation of sickled cells in spleen
- Damage to other organs
- Spleen damage

Impaired mental function
- Paralysis
- Pneumonia and other infections
- Rheumatism
- Kidney failure
A single character may be influenced by many genes

- Polygenic inheritance
  - Many genes influence one trait
  - Skin color is affected by at least three genes
9.15 The environment affects many characters

- Phenotypic variations are influenced by the environment
  - Skin color is affected by exposure to sunlight
  - Susceptibility to diseases, such as cancer, has hereditary and environmental components
THE CHROMOSOMAL BASIS OF INHERITANCE
Mendel’s Laws correlate with chromosome separation in meiosis

- The law of segregation depends on separation of homologous chromosomes in anaphase I
- The law of independent assortment depends on alternative orientations of chromosomes in metaphase I
F<sub>1</sub> generation

All round yellow seeds

(RrYy)

Metaphase I of meiosis
(alternative arrangements)
F<sub>1</sub> generation

All round yellow seeds
(RrYy)

Metaphase I of meiosis
(alternative arrangements)

Anaphase I of meiosis

Metaphase II of meiosis

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Metaphase I of meiosis (alternative arrangements)

Anaphase I of meiosis

Metaphase II of meiosis

Gametes

Fertilization among the F₁ plants

F₂ generation

9 :3 :3 :1
9.17 Genes on the same chromosome tend to be inherited together

**Linked Genes**
- Are located close together on the same chromosome
- Tend to be inherited together

**Example studied by Bateson and Punnett**
- Parental generation: plants with purple flowers, long pollen crossed to plants with red flowers, round pollen
- The F$_2$ generation did not show a 9:3:3:1 ratio
- Most F$_2$ individuals had purple flowers, long pollen or red flowers, round pollen
Experiment

Purple flower

\[ PpLi \times PpLi \]

Long pollen

<table>
<thead>
<tr>
<th>Phenotypes</th>
<th>Observed offspring</th>
<th>Prediction (9:3:3:1)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Purple long</td>
<td>284</td>
<td>215</td>
</tr>
<tr>
<td>Purple round</td>
<td>21</td>
<td>71</td>
</tr>
<tr>
<td>Red long</td>
<td>21</td>
<td>71</td>
</tr>
<tr>
<td>Red round</td>
<td>55</td>
<td>24</td>
</tr>
</tbody>
</table>

Explanation: linked genes

Parental diploid cell

\[ PpLi \]

Meiosis

Most gametes

\[ PL \]

\[ pl \]

Fertilization

Sperm

\[ PL \]

\[ pl \]

Most offspring

Eggs

\[ PL \]

\[ PL \]

\[ pl \]

\[ pl \]

\[ PL \]

\[ pl \]

3 purple long : 1 red round

Not accounted for: purple round and red long
Experiment

$$PpLI \times PpLI$$

<table>
<thead>
<tr>
<th>Phenotypes</th>
<th>Observed offspring</th>
<th>Prediction (9:3:3:1)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Purple long</td>
<td>284</td>
<td>215</td>
</tr>
<tr>
<td>Purple round</td>
<td>21</td>
<td>71</td>
</tr>
<tr>
<td>Red long</td>
<td>21</td>
<td>71</td>
</tr>
<tr>
<td>Red round</td>
<td>55</td>
<td>24</td>
</tr>
</tbody>
</table>
Explanation: linked genes

Parental diploid cell
PpLl

Most gametes

Meiosis

Fertilization

Sperm

Most offspring

Eggs

3 purple long : 1 red round
Not accounted for: purple round and red long

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9.18 Crossing over produces new combinations of alleles

- Linked alleles can be separated by crossing over
  - Recombinant chromosomes are formed
  - Thomas Hunt Morgan demonstrated this in early experiments
  - Geneticists measure genetic distance by recombination frequency
Gametes

Tetrad

Crossing over

Gametes
Experiment

Parental phenotypes

Recombination frequency = 0.17 or 17%

Female

Male

Offspring

Gray long

Black vestigial

Gray vestigial

Black long

Recombinant phenotypes

Offspring

Female

Male

Explanation

Recombination frequency = \frac{391 \text{ recombinants}}{2,300 \text{ total offspring}} = 0.17 \text{ or } 17\%
Recombination frequency = \frac{391\text{ recombinants}}{2,300\text{ total offspring}} = 0.17\text{ or }17\%
Explanation

GgLl (female)

G L

G l

Eggs

G L  
g l

G l

Offspring

G L  
g l

g l

Sperm

g l

ggll (male)

g l

g l

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9.19 Geneticists use crossover data to map genes

- Genetic maps
  - Show the order of genes on chromosomes
  - Arrange genes into linkage groups representing individual chromosomes
Chromosome

Recombination frequencies

9.5%
9%
17%
9.5%
Mutant phenotypes

<table>
<thead>
<tr>
<th>Short aristae</th>
<th>Black body (g)</th>
<th>Cinnabar eyes (c)</th>
<th>Vestigial wings (l)</th>
<th>Brown eyes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Long aristae (appendages on head)</td>
<td>Gray body (G)</td>
<td>Red eyes (C)</td>
<td>Normal wings (L)</td>
<td>Red eyes</td>
</tr>
</tbody>
</table>

Wild-type phenotypes
SEX CHROMOSOMES AND SEX-LINKED GENES
9.20 Chromosomes determine sex in many species

- X-Y system in mammals, fruit flies
  - XX = female; XY = male

- X-O system in grasshoppers and roaches
  - XX = female; XO = male

- Z-W in system in birds, butterflies, and some fishes
  - ZW = female, ZZ = male

- Chromosome number in ants and bees
  - Diploid = female; haploid = male
**Parents’ diploid cells**

44 \(\text{XY}\) (male)  

44 \(\text{XX}\) (female)

**Sperm**

22 \(\text{XY}\)

22 \(\text{XX}\)

**Egg**

22 \(\text{Y}\)

22 \(\text{X}\)

**Offspring (diploid)**

44 \(\text{XY}\)

44 \(\text{XX}\)

**Diagram:**

- Male sperm: 22 \(\text{XY}\)  
- Female egg: 22 \(\text{XX}\)  
- Offspring: 44 \(\text{XY}\)
9.21 Sex-linked genes exhibit a unique pattern of inheritance

- **Sex-linked genes** are located on either of the sex chromosomes

  - Reciprocal crosses show different results
    - White-eyed female × red-eyed male → red-eyed females and white-eyed males
    - Red-eyed female × white-eyed male → red-eyed females and red-eyed males

  - X-linked genes are passed from mother to son and mother to daughter

  - X-linked genes are passed from father to daughter

  - Y-linked genes are passed from father to son
Female: $X^R X^R$
Male: $X^r Y$

Sperm: $X^r$ $Y$
Eggs: $X^R$ $X^R X^r$ $X^R Y$

$R =$ red-eye allele
$r =$ white-eye allele
Female: $X^R X^r$
Male: $X^r Y$

Sperm:
- $X^r$
- $Y$

Eggs:
- $X^R$
- $X^r$

Resulting Offspring:
- $X^R X^r$
- $X^R Y$
- $X^r X^r$
- $X^r Y$
9.22 CONNECTION: Sex-linked disorders affect mostly males

- Males express X-linked disorders such as the following when recessive alleles are present in one copy
  - Hemophilia
  - Colorblindness
  - Duchenne muscular dystrophy
9.23 EVOLUTION CONNECTION: The Y chromosome provides clues about human male evolution

- Similarities in Y chromosome sequences
  - Show a significant percentage of men related to the same male parent
  - Demonstrate a connection between people living in distant locations
Homologous chromosomes
Alleles, residing at the same locus
Meiosis
Paired alleles, alternate forms of a gene
Haploid gametes (allele pairs separate)
Fertilization
Gamete from other parent
Diploid zygote (containing paired alleles)
Incomplete dominance:

Red $RR$ x White $rr$ → Pink $Rr$

Pleiotropy:
- Multiple characters
- Single characters (such as skin color)

Polygenic inheritance:
- Single gene
- Multiple genes

Single gene:
- Red $RR$
- White $rr$
- Pink $Rr$
Genes located on chromosomes at specific locations called alternative versions called if both same, genotype called if different, genotype called.

In between called inheritance when phenotype expressed allele called unexpressed allele called heterozygous.
1. Explain and apply Mendel’s laws of segregation and independent assortment

2. Distinguish between terms in the following groups: allele—gene; dominant—recessive; genotype—phenotype; $F_1$—$F_2$; heterozygous—homozygous; incomplete dominance—codominance

3. Explain the meaning of the terms locus, multiple alleles, pedigree, pleiotropy, polygenic inheritance
4. Describe the difference in inheritance patterns for linked genes and explain how recombination can be used to estimate gene distances

5. Describe how sex is inherited in humans and identify the pattern of inheritance observed for sex-linked genes

6. Solve genetics problems involving monohybrid and dihybrid crosses for autosomal and sex-linked traits, with variations on Mendel’s laws