**Patterns of Inheritance**

**Biological Principles Underlying Genetics**

- People have selected and mated dogs with preferred traits for more than 15,000 years.
- Over thousands of years, such genetic tinkering has led to the incredible variety of body types and behaviors in dogs today.
- The biological principles underlying genetics have only recently been understood.

**Heritable Variation and Patterns of Inheritance**

- **Heredity** is the transmission of traits from one generation to the next.
- **Genetics** is the scientific study of heredity.
- **Gregor Mendel**
  - worked in the 1860s,
  - was the first person to analyze patterns of inheritance, and
  - deduced the fundamental principles of genetics.
In an Abbey Garden

• Mendel studied garden peas because they
  – were easy to grow,
  – came in many readily distinguishable varieties,
  – are easily manipulated, and
  – can self-fertilize.
In an Abbey Garden

- **Mendel**
  - created purebred varieties of plants and
  - crossed two different purebred varieties.

- **Hybrids** are the offspring of two different purebred varieties.
  - The parental plants are the **P generation**.
  - Their hybrid offspring are the **F₁ generation**.
  - A cross of the F₁ plants forms the **F₂ generation**.
Mendel’s Law of Segregation

- Mendel performed many experiments.
- He tracked the inheritance of characters that occur as two alternative traits.

Monohybrid Crosses

- A monohybrid cross is a cross between purebred parent plants that differ in only one character.
Mendel developed four hypotheses from the monohybrid cross, listed here using modern terminology (including “gene” instead of “heritable factor”).

1. The alternative versions of genes are called **alleles**.
2. For each inherited character, an organism inherits two alleles, one from each parent.
   - An organism is **homozygous** for that gene if both alleles are identical.
   - An organism is **heterozygous** for that gene if the alleles are different.

3. If two alleles of an inherited pair differ,
   - then one determines the organism’s appearance and is called the **dominant allele** and
   - the other has no noticeable effect on the organism’s appearance and is called the **recessive allele**.

4. Gametes carry only one allele for each inherited character.
   - The two alleles for a character segregate (separate) from each other during the production of gametes.
   - This statement is called the **law of segregation**.

• Do Mendel’s hypotheses account for the 3:1 ratio he observed in the F₂ generation?

• **A Punnett square** highlights
  - the four possible combinations of gametes and
  - the four possible offspring in the F₂ generation.
Monohybrid Crosses

- Geneticists distinguish between an organism’s physical appearance and its genetic makeup.
  - An organism’s physical appearance is its **phenotype**.
  - An organism’s genetic makeup is its **genotype**.
Mendel’s Law of Independent Assortment

- A **dihybrid cross** is the mating of parental varieties differing in two characters.
- What would result from a dihybrid cross? Two hypotheses are possible:
  1. dependent assortment or
  2. independent assortment.
Mendel’s dihybrid cross supported the hypothesis that each pair of alleles segregates independently of the other pairs during gamete formation.

Thus, the inheritance of one character has no effect on the inheritance of another.

This is called Mendel’s law of independent assortment.

Independent assortment is also seen in two hereditary characters in Labrador retrievers.

Using a Testcross to Determine an Unknown Genotype

- A testcross is a mating between
  - an individual of dominant phenotype (but unknown genotype) and
  - a homozygous recessive individual.
The Rules of Probability

- Mendel’s strong background in mathematics helped him understand patterns of inheritance.
- The **rule of multiplication** states that the probability of a compound event is the product of the separate probabilities of the independent events.

Family Pedigrees

- Mendel’s principles apply to the inheritance of many human traits.
Figure 9.12

DOMINANT TRAITS

Freckles
Widow’s peak
Free earlobe

RECESSIVE TRAITS

No freckles
Straight hairline
Attached earlobe

Figure 9.12a

Freckles
No freckles

Figure 9.12b

Widow’s peak
Straight hairline

Figure 9.12c

Free earlobe
Attached earlobe
Family Pedigrees

- Dominant traits are not necessarily
  - normal or
  - more common.

- **Wild-type traits** are
  - those seen most often in nature and
  - not necessarily specified by dominant alleles.

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Family Pedigrees

- A family **pedigree**
  - shows the history of a trait in a family and
  - allows geneticists to analyze human traits.

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Human Disorders Controlled by a Single Gene

- Many human traits
  - show simple inheritance patterns and
  - are controlled by single genes on autosomes.
Most human genetic disorders are recessive.

Individuals who have the recessive allele but appear normal are carriers of the disorder.

<table>
<thead>
<tr>
<th>Table 9.1</th>
<th>Some Autosomal Disorders in People</th>
</tr>
</thead>
<tbody>
<tr>
<td>Disorder</td>
<td>Major Symptoms</td>
</tr>
<tr>
<td>Recessive Disorders</td>
<td></td>
</tr>
<tr>
<td>Albinism</td>
<td>Lack of pigment in skin, hair, and eyes</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>
</tr>
<tr>
<td>Phenylketonuria (PKU)</td>
<td>Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation unless treated</td>
</tr>
<tr>
<td>Sickle-cell disease</td>
<td>Sickle red blood cells; damage to many tissues</td>
</tr>
<tr>
<td>Tay/Sachs disease</td>
<td>Lipo accumulation in brain cells; mental deficiency; blindness; death in childhood</td>
</tr>
<tr>
<td>Dominant Disorders</td>
<td></td>
</tr>
<tr>
<td>Achondroplasia</td>
<td>Dwarfism</td>
</tr>
<tr>
<td>Alzheimer's disease (one type)</td>
<td>Mental deterioration; usually strikes late in life</td>
</tr>
<tr>
<td>Huntington's disease</td>
<td>Mental deterioration and uncontrollable movements; strikes in middle age</td>
</tr>
<tr>
<td>Hypercholesterolemia</td>
<td>Excess cholesterol in blood; heart disease</td>
</tr>
</tbody>
</table>

Recessive Disorders

- Cystic fibrosis is
  - the most common lethal genetic disease in the United States and
  - caused by a recessive allele carried by about one in 31 Americans.
**Recessive Disorders**

- Prolonged geographic isolation of certain populations can lead to **inbreeding**, the mating of close relatives.

- Inbreeding increases the chance of offspring that are homozygous for a harmful recessive trait.

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**Dominant Disorders**

- Some human genetic disorders are dominant.
  - **Achondroplasia** is a form of dwarfism.
    - The homozygous dominant genotype causes death of the embryo.
    - Thus, only heterozygotes have this disorder.
  - **Huntington’s disease**, which leads to degeneration of the nervous system, does not usually begin until middle age.
Genetic Testing

• Today many tests can detect the presence of disease-causing alleles.

• Most genetic tests are performed during pregnancy.
  – Amniocentesis collects cells from amniotic fluid.
  – Chorionic villus sampling removes cells from placental tissue.

• Genetic counseling helps patients understand the results and implications of genetic testing.