CHAPTER 9

Patterns of Inheritance

BIOLOGY AND SOCIETY:

TESTING YOUR BABY

- Genetic testing
  - Allows expectant parents to test for possibilities in their unborn child
  - Includes amniocentesis and CVS
  - Has risks associated with it.

HERITABLE VARIATION AND PATTERNS OF INHERITANCE

- Wild type traits are traits most commonly found in nature.
• Gregor Mendel
  – Was the first person to analyze patterns of inheritance
  – Deduced the fundamental principles of genetics.

In an Abbey Garden
• Mendel studied garden peas
  – These plants are easily manipulated
  – These plants can self-fertilize.
• Mendel carried out some cross-fertilization
• P, F₁, F₂, p.145

He also created true-breeding varieties of plants (self-fertilization produced offspring all identical to the parent)

Mendel then crossed two different true-breeding varieties, creating hybrids.

Mendel’s Principles of Segregation

• Mendel performed many experiments
  – He tracked several characteristics in pea plants from which he formulated several hypotheses.

During the production of gametes each ovum or sperm receives only one from each pair of chromosomes.
Monohybrid Crosses

A monohybrid cross is a cross between parent plants that differ in only one characteristic.

- Mendel developed four hypotheses from the monohybrid cross:
  - There are alternative forms of genes, now called alleles.
  - For each characteristic, each organism has two genes.
  - Gametes carry only one allele for each inherited characteristic.
  - Alleles can be dominant or recessive.
• Mendel’s principle of segregation
  – Pairs of alleles segregate (separate) during gamete formation; the fusion of gametes at fertilization creates allele pairs again.

• Phenotype
  – An organism’s physical traits ex. tall, short, green, etc.

• Genotype
  – An organism’s genetic makeup ex. TT, Tt, tt
**Genetic Alleles and Homologous Chromosomes**

- **Homologous chromosomes**
  - Have genes at specific loci
  - Have alleles of a gene at the same locus.

- **Homozygous**
  - When an organism has identical alleles for a gene

- **Heterozygous**
  - When an organism has different alleles for a gene.
Mendel’s Principle of Independent Assortment

- Two hypotheses for gene assortment in a dihybrid cross
  - Dependent assortment
  - Independent assortment.

**Mendel’s principle of independent assortment**

- Each pair of alleles segregates independently of the other pairs during gamete formation

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**Figure 9.10**

(a) Hypothesis: Dependent assortment
(b) Hypothesis: Independent Assortment

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**Figure 9.11**

Blind Blind

Phenotypes
Genotypes

- Black coat, normal vision (B_N_)
- Chocolate coat, blind (PRA) (bbN_)

Mating of heterozygotes (black, normal vision)

Phenotypic ratio of offspring

- 9 black coat, normal vision
- 3 black coat, blind
- 3 chocolate coat, normal vision
- 1 chocolate coat, blind
A dihybrid cross with two parents heterozygous for both genes gives nine genotypes and a 9:3:3:1 ratio of phenotypes.

See Fig. 9.8 p.149
**Family Pedigrees**

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

**Dominant Traits**
- Freckles

**Recessive Traits**
- No freckles

**Widow’s peak**
- Straight hairline

**Free earlobe**
- Attached earlobe

**A family pedigree**
- Shows the history of a trait in a family
- Allows researchers to analyze human traits.

**Human Disorders Controlled by a Single Gene**

- Many human traits
  - Show simple inheritance patterns
  - Are controlled by genes on autosomes.
Table 9.1  Some Autosomal Disorders in Humans

<table>
<thead>
<tr>
<th>Disease</th>
<th>Major Symptoms</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Recessive disorders</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Albinism</td>
<td>Lack of pigment in skin, hair, and eyes</td>
<td></td>
</tr>
<tr>
<td>Santa Vanze</td>
<td>Emesis, mucogingival erosions, periodontal disease</td>
<td></td>
</tr>
<tr>
<td>Malaria</td>
<td>Accumulation of parasites in tissues, mental retardation, and fever</td>
<td></td>
</tr>
<tr>
<td>Pneumocystis</td>
<td>Acquired immuno deficiency, death or lung infection</td>
<td></td>
</tr>
<tr>
<td>Tuberous sclerosis</td>
<td>Brain malformations, seizures, mental retardation and death</td>
<td></td>
</tr>
<tr>
<td>Dominant disorders</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Achondroplasia</td>
<td>Dwarfism</td>
<td></td>
</tr>
<tr>
<td>Marfan’s disease</td>
<td>Marfan syndrome, usually death from aortic aneurysm</td>
<td></td>
</tr>
<tr>
<td>Spinal dysraphism</td>
<td>Structural malformations, mental retardation, and death</td>
<td></td>
</tr>
</tbody>
</table>

Recessive Disorders

- Most human genetic disorders are recessive
  - Individuals can be carriers of these diseases.

![Diagram of Recessive Disorders](image)

Dominant Disorders

- Some human genetic disorders are dominant
  - Achondroplasia is a form of dwarfism.

![Diagram of Dominant Disorders](image)
BEYOND MENDEL

- Some patterns of genetic inheritance are not explained by Mendel’s principles.

Incomplete Dominance in Plants and People

- In incomplete dominance, F₁ hybrids have an appearance in between the phenotypes of the two parents.

Hypercholesterolemia

- Is a human trait that is incompletely dominant.
Multiple Alleles and Blood Type

• The ABO blood groups in humans are examples of multiple alleles.

<table>
<thead>
<tr>
<th>Blood Group Phenotype</th>
<th>Genotypes</th>
<th>Antibody Present in Blood</th>
</tr>
</thead>
<tbody>
<tr>
<td>O</td>
<td>A, a</td>
<td>Anti-A Anti-B</td>
</tr>
<tr>
<td>A</td>
<td>A, A, a</td>
<td>Anti-B</td>
</tr>
<tr>
<td>B</td>
<td>B, b</td>
<td>Anti-A</td>
</tr>
<tr>
<td>AB</td>
<td>A, B, a, b</td>
<td>—</td>
</tr>
</tbody>
</table>

Figure 9.20

• Two of the human blood type alleles exhibit codominance
  – Both alleles are expressed in the phenotype.

Pleiotropy and Sickle-Cell Disease

• Pleiotropy is the impact of a single gene on more than one characteristic
  – Sickle-cell disease is an example.
Figure 9.21

Individual homozygous for sickle-cell allele

Sickle-cell (abnormal) hemoglobin

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

Sickled cells

Breakdown of red blood cells

Changin of cells and stagnation of small blood vessels

Irreversible damage of sickled cells in system

Any signs

Hypoxia

Anemia

Heart failure

Physical weakness

Brain damage

Impaired mental function

Paralysis

Pneumonia and other infections

Rheumatism

Kidney failure

Accumulation of sickled cells in spleen

Clumping of cells

Spleen damage

Impaired

Organ failure

The Role of Environment

Many human characteristics result from a combination of heredity and environment.
THE CHROMOSOMAL BASIS OF INHERITANCE

- The chromosome theory of inheritance states that
  - Genes are located on chromosomes
  - The behavior of chromosomes during meiosis and fertilization accounts for inheritance patterns.

SEX CHROMOSOMES AND SEX-LINKED GENES

- Sex chromosomes
  - Influence the inheritance of certain traits.
Sex Determination in Human and Fruit Flies

- Sex chromosomes
  - Are designated X and Y
  - Determine an individual’s sex.

### Male

- 22 + X
- Sperm
- 22 + Y

### Female

- 22 + X
- Somatic cells
- 44 + XX
- Egg
- 44 + XY

Sex-Linked Genes

- Sex-linked genes
  - Are any genes located on a sex chromosome
  - Were discovered during studies on fruit flies.

**Sex-linked genes in fruit flies**

(a) (b)

**Inheritance patterns of a sex-linked gene**

- All females inherit two X chromosomes, one from each parent.
- All males inherit one X chromosome, always from the mother.

- R = red-eye allele
- r = white-eye allele

(a) Homozygous red-eyed female × white-eyed male

(b) Heterozygous female × red-eyed male

(c) Heterozygous female × white-eyed male
• Red-green color blindness
  – Is characterized by a malfunction of light-sensitive cells in the eyes.

• Hemophilia
  – Is a blood-clotting disease.

• The Y chromosome of human males is only about one-third the size of the X chromosome
  • Scientists believe that X and Y were once a fully homologous pair
  • Major episodes of change have rearranged pieces of the Y chromosome.
SUMMARY OF KEY CONCEPTS

• Mendel’s Principle of Segregation.

• Using a Testcross to Determine an Unknown Genotype.

• Incomplete Dominance in Plants and People.
• Pleiotropy and Sickle-Cell Disease.

• Polygenic Inheritance.