Chapter 21
Lecture Outline

See separate PowerPoint slides for all figures and tables pre-inserted into PowerPoint without notes.
Patterns of Genetic Inheritance
Points to ponder

• What is the genotype and the phenotype of an individual?
• What are the genotypes for homozygous recessive and dominant individuals, and a heterozygous individual?
• Be able to draw a Punnett square for a one-trait cross, two-trait cross, and sex-linked cross.
• What are Tay-Sachs disease, Huntington disease, sickle-cell disease, and PKU? How are each of these inherited?
• What is polygenic inheritance?
• What is a multifactorial trait?
• What is sex-linked inheritance?
• Name three X-linked recessive disorders.
• What is codominance?
• What is incomplete dominance?
• What do you think about preimplantation genetic testing?
These traits are genetically inherited

Answer these questions about your inheritance.

• Do you have a widow’s peak or a straight hairline?
• Are your earlobes attached or unattached?
• Do you have short or long fingers?
• Do you have freckles?
Genotype

Genotype – specific genes for a particular trait written with symbols

– **Alleles** are alternate forms of a specific gene at the same position (locus) on a gene (e.g., allele for unattached earlobes and attached lobes); alleles occur in pairs.

– A **dominant gene** will be expressed and will mask a recessive gene (Tt or TT).

– A **recessive allele** is only expressed when a gene has two of this type of allele.
Genotype

- A **homozygous dominant genotype** consists of two dominant alleles (TT or AA).
- A **homozygous recessive genotype** consists of two recessive alleles (tt or aa).
- A **heterozygous genotype** consists of one dominant allele and one recessive allele (Tt or Aa).
Phenotype

Phenotype – the physical or outward expression of the genotype

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>EE</td>
<td>unattached earlobe</td>
</tr>
<tr>
<td>Ee</td>
<td>unattached earlobe</td>
</tr>
<tr>
<td>ee</td>
<td>attached earlobe</td>
</tr>
</tbody>
</table>

What are your genotype and phenotype?
21.1 Genotype and Phenotype

Understanding genotype and phenotype

Figure 21.1 Genetic inheritance affects our characteristics.

Allele Key
- \( A \) = Normal pigmentation
- \( a \) = Lack of pigmentation (albino)
21.2 One- and Two-Trait Inheritance

What about your inheritance?

Figure 21.2 Common inherited traits in humans.
Crosses

- **One-trait cross** – considers the inheritance of one characteristic
  
  e.g. WW $\times$ Ww

- **Two-trait cross** – considers the inheritance of two characteristics
  
  e.g. WWTT $\times$ WwTT

- Gametes only carry one allele, so if an individual has the genotype Ww, what are the possible gametes that this individual can pass on?

  Answer: either a $W$ or a $w$, but not both
Another example:

Parents

no freckles

ff

meiosis

gametes

Offspring

no freckles

ff

ff

no freckles

ff

ff

no freckles
Punnett squares

• **Punnett squares** are the use of a grid to diagram crosses between individuals by using the possible parental gametes.

• These allow one to determine the probability that an offspring will have a particular genotype and phenotype.
Figure 21.3 Expected results of a monohybrid cross.
Practicing Punnett squares

• What would a Punnett square involving a man (M) with a genotype Ff and a woman (F) with a genotype Ff look like?

F – freckles
f – no freckles

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<table>
<thead>
<tr>
<th></th>
<th>M/F</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>F</td>
<td>f</td>
<td></td>
</tr>
<tr>
<td>F</td>
<td>FF</td>
<td>Ff</td>
<td></td>
</tr>
<tr>
<td>f</td>
<td>Ff</td>
<td>ff</td>
<td></td>
</tr>
</tbody>
</table>
Practicing ratios

- Genotypic ratio is the number of offspring with the same genotype.
- Phenotypic ratio is the number of offspring with the same outward appearance.
Practicing ratios

- What is the genotypic ratio?
  1: 2: 1 (1 FF: 2 Ff: 1 ff)

- What is the phenotypic ratio?
  3: 1 (3 with freckles and 1 with no freckles)
Monohybrid crosses

Monohybrid cross – an experimental cross in which parents are identically heterozygous at 1 gene pair (e.g., Aa x Aa)

Figure 21.4 Determining if a dominant phenotype is homozygous or heterozygous.
Possible gametes for 2 traits

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Cell has two pairs of homologues.

Either

MEIOSIS I

ALLELE KEY

F = Freckles
f = No freckles
S = Short fingers
s = Long fingers

MEIOSIS II

Figure 21.5 Meiosis results in genetic diversity of gametes.
Dihybrid cross (a type of two-trait cross)

- **Dihybrid cross** – an experimental cross usually involving parents who are homozygous for different alleles of two genes
  - Results in a 9:3:3:1 genotypic ratio for the offspring

*Figure 21.6* Expected results of a dihybrid cross.
Punnett square for a dihybrid cross

- What would the Punnett square look like for a dihybrid cross between a male who is WWSS and a female who is wwss?

Figure 21.6 Expected results of a dihybrid cross.
21.2 One- and Two-Trait Inheritance

Figure 21.7 Two-trait cross.
21.2 One- and Two-Trait Inheritance

Phenotypic ratios of common crosses

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<table>
<thead>
<tr>
<th>Table 21.1</th>
<th>Phenotypic Ratios of Common Crosses</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Genotypes</strong></td>
<td><strong>Phenotypes</strong></td>
</tr>
<tr>
<td>Monohybrid $Aa \times$ monohybrid $Aa$</td>
<td>3:1 (dominant to recessive)</td>
</tr>
<tr>
<td>Monohybrid $Aa \times$ recessive $aa$</td>
<td>1:1 (dominant to recessive)</td>
</tr>
<tr>
<td>Dihybrid $AaBb \times$ dihybrid $AaBb$</td>
<td>9:3:3:1 (9 both dominant: 3 dominant for one of the traits: 3 dominant for other trait: 1 both recessive)</td>
</tr>
<tr>
<td>Dihybrid $AaBb \times$ recessive $aabb$</td>
<td>1:1:1:1 (all possible combinations in equal number)</td>
</tr>
</tbody>
</table>

Table 21.1 Phenotypic ratios of common crosses
Autosomal recessive disorder

- Individuals must be homozygous recessive to have the disorder.

**Key**
- $aa$ = affected
- $Aa$ = carrier (unaffected)
- $AA$ = unaffected
- $A?$ = unaffected (one allele unknown)

**Autosomal recessive disorders**
- Affected children can have unaffected parents.
- Heterozygotes ($Aa$) have an unaffected phenotype.
- Two affected parents will always have affected children.
- Affected individuals with homozygous unaffected mates will have unaffected children.
- Close relatives who reproduce are more likely to have affected children.
- Both males and females are affected with equal frequency.

**Figure 21.8** Autosomal recessive disorder pedigree.
Autosomal dominant disorder

- Individuals that are homozygous dominant and heterozygous will have the disorder.

**Figure 21.9**
Autosomal dominant disorder pedigree.

Autosomal dominant disorders
- Affected children will usually have an affected parent.
- Heterozygotes (Aa) are affected.
- Two affected parents can produce an unaffected child.
- Two unaffected parents will not have affected children.
- Both males and females are affected with equal frequency.
Autosomal recessive disorders of interest

- **Tay-Sachs disease** – lack of the enzyme that breaks down fatty acid proteins in lysosomes results in accumulation

- **Cystic fibrosis** – Cl⁻ ions do not pass normally through a cell membrane, resulting in thick mucus in lungs and other places, often causing infections
Autosomal recessive disorders of interest

- **Phenylketonuria (PKU)** – lack of an enzyme needed to make a certain amino acid; affects nervous system development

- **Sickle-cell disease** – red blood cells are sickle-shaped rather than biconcave, resulting in clogged blood vessels
Figure 21.10 Neuron affected by Tay–Sachs disease.
Autosomal dominant disorders of interest

• **Marfan syndrome** – defect in the production of the elastic connective tissue protein fibrillin; results in dislocated lens, long limbs and fingers, caved-in chest, and weak wall of aorta

• **Osteogenesis imperfecta** – defect in collagen synthesis; results in weakened, brittle bones

• **Huntington disease** – huntington protein has too many glutamine amino acids, leading to the progressive degeneration of brain cells
21.3 Inheritance of Genetic Disorders

Marfan syndrome

Chest wall deformities
- Long, thin fingers, arms, legs
- Scoliosis (curvature of the spine)
- Flat feet
- Long, narrow face
- Loose joints

Heart and blood vessels
- Mitral valve prolapse
- Enlargement of aorta
- Aneurysm
- Aortic wall tear
- Aneurysm
- Aortic wall tear

Eyes
- Lens dislocation
- Severe nearsightedness

Lungs
- Collapsed lungs

Skin
- Stretch marks in skin
- Recurrent hernias
- Dural ectasia: stretching of the membrane that holds spinal fluid

Figure 21.16 Marfan syndrome.
21.3 Inheritance of Genetic Disorders

Genetic disorders

Figure 21.11 Cystic fibrosis disease.

Figure 21.12 Huntington disease.

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Preimplantation genetic diagnosis

- If prospective parents carry an allele for a genetic disorder, they may seek assurance that their offspring will be free of the disorder.

- Following *in vitro* fertilization (IVF), the zygote divides.

- When the embryo has eight cells, one may be removed for genetic testing.

- Only embryos that will not have the genetic disorders of interest are placed in the uterus to continue developing.
Preimplantation genetic diagnosis

21.3 Inheritance of Genetic Disorders

**Figure 21A** The process of preimplantation genetic diagnosis.

- a. Testing the embryo
- b. Testing the egg

(both): © Brand X/SuperStock RF
Polygenic inheritance

- **Polygenic traits** – two or more sets of alleles govern one trait
  - Each dominant allele codes for a product, so these effects are additive.
  - This results in a continuous variation of phenotypes.
  - Environmental effects cause intervening phenotypes.
- e.g. skin color ranges from very dark to very light
- e.g. height varies among individual humans
Polygenic inheritance

- **Multifactorial trait** – a polygenic trait that is particularly influenced by the environment
  - e.g. skin color is influenced by sun exposure
  - e.g. height can be affected by nutrition
Polygenic inheritance

Figure 21.13 Height is a polygenic trait in humans.

Figure 21.14 Polygenic inheritance and skin color.
Demonstrating environmental influences on phenotype

- Himalayan rabbit’s coat color is influenced by temperature.
- There is an allele responsible for melanin production that appears to be active only at lower temperatures.
- The extremities have a lower temperature and thus the ears, nose, paws, and tail are dark in color.

Figure 21.15 Himalayan rabbit with temperature-susceptible coat color.
Incomplete dominance

- Occurs when the heterozygote phenotype is intermediate between phenotypes of the two homozygotes

- Example:

  \[(\text{curly hair}) \text{CC} \times \text{SS (straight hair)} \rightarrow \text{CS (wavy hair)}\]
Familial hypercholesterolemia

- Two mutated alleles lack LDL-cholesterol receptors.
- One mutated allele has half the normal number of receptors.
- Two normal alleles have the usual number of receptors.
- When receptors are completely absent, excessive cholesterol is deposited in various places in the body, including under the skin.
Familial hypercholesterolemia

Figure 21.17 The inheritance of familial hypercholesterolemia.
Codominance

• Occurs when the alleles are equally expressed in a heterozygote

• Example:

\[(\text{Type A blood}) \quad AA \quad \times \quad BB \quad (\text{Type B blood})\]

\[\quad AB \quad (\text{Type AB blood that has characteristics of both blood types})\]
Multiple allele inheritance

- The gene exists in several allelic forms.

- A person only has two of the possible alleles.

- A good example is the ABO blood system.

- A and B are codominant alleles.

- The O allele is recessive to both A and B; therefore, to have this blood type, you must have two recessive alleles.
Multiple allele inheritance

What type of blood would each of the following individuals have in a cross between Ao and Bo?

<table>
<thead>
<tr>
<th>Possible genotypes:</th>
<th>Phenotypes:</th>
</tr>
</thead>
<tbody>
<tr>
<td>AB</td>
<td>Type AB blood</td>
</tr>
<tr>
<td>Bo</td>
<td>Type B blood</td>
</tr>
<tr>
<td>Ao</td>
<td>Type A blood</td>
</tr>
<tr>
<td>oo</td>
<td>Type O blood</td>
</tr>
</tbody>
</table>
21.4 Beyond Simple Inheritance Patterns

Blood type inheritance

Parents

\[ \text{Blood type } A \]
\[ \text{Blood type } B \]
\[ \text{Blood type AB} \]
\[ \text{Blood type O} \]

Key

- Blood type A
- Blood type B
- Blood type AB
- Blood type O

Phenotypic Ratio

1:1:1:1

Offspring

\[ \text{Blood type A} \]
\[ \text{Blood type B} \]
\[ \text{Blood type AB} \]
\[ \text{Blood type O} \]

Figure 21.18 The inheritance of ABO blood types.
Sex-linked inheritance

- Traits are controlled by genes on the sex chromosomes.
  - **X-linked** inheritance – the allele is carried on the X chromosome
  - **Y-linked** inheritance – the allele is carried on the Y chromosome
  - Most **sex-linked** traits are X-linked.
21.5 Sex-Linked Inheritance

X-linked inheritance: Color blindness

Cross:
$X^B X^b \times X^B Y$

Possible offspring:
$X^B X^B$ normal vision female
$X^B X^b$ normal vision female
$X^B Y$ normal vision male
$X^b Y$ color-blind vision male

**Figure 21.19** Results of an X-linked cross.
21.5 Sex-Linked Inheritance

X-linked disorders

• More males than females are affected.
• An affected son can have parents who have the normal phenotype.
• For a female to have the characteristic, her father must also have it. Her mother must have it or be a carrier.
• The characteristic often skips a generation from the grandfather to the grandson.
• If a woman has the characteristic, all of her sons will have it.

Key
- $X^B X^B$ = Unaffected female
- $X^B X^b$ = Carrier female
- $X^b X^B$ = Color-blind female
- $X^B Y$ = Unaffected male
- $X^b Y$ = Color-blind male

Figure 21.20  X-linked recessive disorder pedigree.
21.5 Sex-Linked Inheritance

X-linked disorders

• These are more often found in males than females because recessive alleles are always expressed.

• Most X-linked disorders are recessive.
  – **Color blindness** is most often characterized by red-green color blindness.
  – **Duchenne muscular dystrophy** is characterized by wasting of muscles and death by age 20.
  – **Fragile X syndrome** is the most common cause of inherited mental impairment.
  – **Hemophilia** is characterized by the absence of particular clotting factors; blood clots very slowly or not at all.
21.5 Sex-Linked Inheritance

Duchenne muscular dystrophy

Figure 21.21 Muscular dystrophy.

(left, right): Courtesy Dr. Rabi Tawil, Director, Neuromuscular Pathology Laboratory, University of Rochester Medical Center; (center): Courtesy Muscular Dystrophy Association
21.5 Sex-Linked Inheritance

X-linked disorders: Hemophilia

Figure 21B  The royal families’ X-linked pedigree.