The Human Heritage:
Genes and Environment

Outline
• Sexual Reproduction and Genetic Transmission
• Genotype and Phenotype
• Mutations & Genetic Abnormalities
• Biology and Culture

We are all Unique - Why?
• Unique Genetics
  – Most of us
    • Exception: Identical Twins (MZ)
• Unique Environment
  – Varying degrees of similarity
• Combination of Genetics and Environment
  – All of us!
Sexual Reproduction and Genetic Transmission

- Mitosis: A Process of Cell Replication
- Meiosis: A Source of Variability
  - Crossing-over
- Sexual Determinism: A Case of Variability
Crossing-over

Crossing-over process

X and Y Chromosomes
23rd pair - determines sex
X X - Female
XY - Male

X Chromosome

Y Chromosome
Genotype and Phenotype

- **Genotype**: Set of genetic traits a person inherits; a person’s inborn capacity or potential
- **Phenotype**: Set of genetic traits a person inherits; a person’s inborn capacity or potential

The Laws of Genetic Inheritance

- **Simple Form**
  - Single pair of genes contributes to inherited characteristic (allele)
  - Alleles can be the same (homozygous) or different (heterozygous)
  - When the pair is the same - trait will be displayed

The Laws of Genetic Inheritance

- When alleles are different (heterozygous):
  - 1. Child will express characteristics of one of the alleles (Dominant allele)
  - 2. Child will express both (averaging, intermediate)
  - 3. Child is affected by both but the characteristic displayed is different from both - co-dominance
Inheritance of alleles for blood type

Type AB = Codominance
AO or BO = dominance

Inheritance of a Dominant Gene Disorder

<table>
<thead>
<tr>
<th>Affected Parent (Has the Disorder)</th>
<th>D</th>
<th>r</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dr</td>
<td></td>
<td>rr</td>
</tr>
<tr>
<td>Normal</td>
<td></td>
<td>rr</td>
</tr>
<tr>
<td>Father</td>
<td>r</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>D</th>
<th>Normal (50%)</th>
<th>Dr</th>
<th>Affected (25%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>r</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>D</th>
<th>Normal (50%)</th>
<th>rr</th>
<th>Normal (25%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>r</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

From Seifert/Hoffnung, Child and Adolescent Development, 5/e, Figure 3.10, p. 73. Used by permission of Houghton Mifflin Company.

Dominant Gene

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Polydactyly</td>
<td>1/300 - 1/100</td>
</tr>
<tr>
<td>Achondroplasia</td>
<td>1/2,300</td>
</tr>
<tr>
<td>Huntington disease</td>
<td>1/15,000 - 1/5,000</td>
</tr>
</tbody>
</table>
### Inheritance of a Recessive Gene Disorder

<table>
<thead>
<tr>
<th>Father</th>
<th>Mother</th>
<th>DD Normal (25%)</th>
<th>Dr Normal (25%)</th>
<th>rr Affected (25%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>rD</td>
<td>r</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>D</td>
<td>R</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

From Seifert/Hoffnung, Child and Adolescent Development, 5/e, Figure 3.11, p. 73. Used by permission of Houghton Mifflin Company.

### Recessive Gene

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cystic fibrosis</td>
<td>1/2,500 white persons (carrier risk: 1/25)</td>
</tr>
<tr>
<td>Sickle-cell disease</td>
<td>1/625 African Americans (carrier risk: 1/10)</td>
</tr>
<tr>
<td>Tay-Sachs disease</td>
<td>1/3,600 Eastern European Jews (carrier risk: 1/30 - 1/300)</td>
</tr>
</tbody>
</table>

### Sex-linked Genetic Effects

- Genes found only on the X or Y chromosome
- Most carried on the X chromosome (Why?)
- Males more susceptible to sex-linked genetic defects
Inheritance of Hemophilia, a Sex-Linked Disorder

<table>
<thead>
<tr>
<th>Carrier Mother</th>
<th>Carrier Father</th>
</tr>
</thead>
<tbody>
<tr>
<td>XX Normal Daughter (25%)</td>
<td>XY Normal Son (25%)</td>
</tr>
<tr>
<td>XX Carrier Daughter (25%)</td>
<td>XY Hemophilic Son (25%)</td>
</tr>
</tbody>
</table>

X-Linked Disorder

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hemophilia</td>
<td>1/2,500 male babies</td>
</tr>
<tr>
<td>Duchenne’s MD</td>
<td>1/3,500 male babies</td>
</tr>
</tbody>
</table>

Film: Tackling a Killer Disease

Things to look for:
1. Genetic and Chromosomal abnormalities
   X-linked gene disorders
2. Nature of experimentation
   A. Cause and effect relationships
   B. Independent and Dependent variables
   C. Experimental and Control conditions
   D. Double-blind Procedure
   E. Placebo
Multifactorial Conditions

- Normal variant of normal gene
- Indirect Genetic effects
  - Operate on risk factors not disease itself
  - Risk features continuously distributed attributes
  - Several genes involved
  - Genetic effects are probabilistic
- Example: Smoking

Multifactorial

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital heart disease</td>
<td>1/125</td>
</tr>
<tr>
<td>Neural tube defect</td>
<td>1 - 2/1,000</td>
</tr>
<tr>
<td>Cleft lip/cleft palate</td>
<td>1/1,000 - 1/5,000</td>
</tr>
</tbody>
</table>

Mutations & Genetic Abnormalities

- Additional Source of Variability
  - Can be inherited and passed on
    - Mutation affects the sperm or ovum
  - Can be limited to the individual
    - Mutation limited to specific types of cells
      - E.g., Exposure to environmental elements.
Mutations & Genetic Abnormalities

- Gene-Environmental Interaction
  - Sickle-Cell Anemia
- Chromosomal Error
  - Down Syndrome
- Sex-Linked Chromosomal Abnormalities
  - Phenylketonuria (PKU): A Treatable Genetic Disease

Down Syndrome: Chromosomal Error

A. Caused by:
1) Trisomy 21 - accounts for 95%
2) Translocation - accounts for 3-4%
3) Mosaicism - Accounts for 1%

B. Incidence
- 1/800 live births
- maternal age factor

Relationship Between Maternal Age and the Incidence of Down Syndrome
PKU - Single gene disorder

- Recessive pattern of inheritance (Chr. 12)
  - 1/8000 - 10,000 births
- Affects body’s metabolism of proteins
  - Lack enzyme that converts amino acids
  - Leads to toxic levels of phenylalanine
- Damage to nervous system
  - Untreated - effects evident by 3 - 5 months, retarded by 1 yr.
- Treatment - restricted diet

Examples of Sex Chromosome Abnormalities

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Incidence</th>
<th>Characteristics</th>
</tr>
</thead>
<tbody>
<tr>
<td>Turner syndrome (XO)</td>
<td>1/5,000 women</td>
<td>Short stature, limited secondary sex characteristics, infertile, near-average IQ, deficiencies in spatial abilities</td>
</tr>
<tr>
<td>Triple-X syndrome ( XXXX )</td>
<td>1/1,200</td>
<td>Can exhibit delays in speech and language development, coordination problems, academic and behavioral difficulties</td>
</tr>
<tr>
<td>Klinefelter syndrome (XXY)</td>
<td>1/600 men</td>
<td>Tall, female body contour, usually sterile, some evidence for short term memory and reading problems</td>
</tr>
<tr>
<td>“Supermale” (XYY)</td>
<td>1/1000</td>
<td>Above-average height, near-average IQ, some have learning disabilities</td>
</tr>
</tbody>
</table>

Prenatal Screening Tests

<table>
<thead>
<tr>
<th>Test</th>
<th>When Usually Administered (gestational age)</th>
<th>Typical Waiting Period for Results</th>
<th>Other Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Amniocentesis</td>
<td>15 – 18 weeks</td>
<td>About 2 weeks</td>
<td>Can be administered in weeks 11-14 but typically is not because the available supply of amniotic fluid is more limited.</td>
</tr>
<tr>
<td>Chorionic villus sampling</td>
<td>10 – 12 weeks</td>
<td>34 – 40 hours</td>
<td>Possibly a slightly greater risk than associated with amniocentesis, including link to Down’s syndrome.</td>
</tr>
<tr>
<td>Ultrasoundography</td>
<td>About 6 weeks and later</td>
<td>None</td>
<td>Provides picture of growing fetus. Not definitive for identifying rare disorders. Little evidence of any risk. Often used to accompany other test procedures.</td>
</tr>
</tbody>
</table>
The Process of Amniocentesis

Who Should Seek Prenatal Counseling?

1. Couples who already have a child with some serious defect such as Down syndrome, spina bifida, congenital heart disease, limb malformation, or mental retardation
2. Couples with a family history of a genetic disease or mental retardation
3. Couples who are blood relatives (first or second cousins)
4. African Americans, Ashkenazi Jews, Italians, Greeks, and other high-risk ethnic groups
5. Women who have had a serious infection early in pregnancy (rubella or toxoplasmosis) or who have been infected with HIV
6. Women who have taken potentially harmful medications early in pregnancy or habitually use drugs or alcohol
7. Women who have had X rays taken early in pregnancy
8. Women who have experienced two or more of the following: stillbirth, death of a newborn baby, miscarriage
9. Any woman thirty-five years or older

Source: Adapted from Fienbloom & Forman (1987) p. 129

Genes, The Organism, and the Environment

- Studying Gene-environment Interactions
- Range of Reaction
- Canalization
- The Study of Genetic Influences on Human Behavior
- Estimating Genetic Influence Through Kinship Studies
Gene-environment Interactions

- Two-way interaction
  - Genes predispose you to display characteristic
  - Environment can increase/decrease likelihood
- Genetic predisposition can change your reaction to the environment

The Effect of the Environment on Fur Color

1. Normally only feet, tail, ears and nose are black.

2. Remove fur on back & place icepack

3. New fur is black.

Range of Reaction

- Combination of genes and environments can lead to many possible outcomes
  - Example: If you have the genes that code for obesity but are in a time of famine, you will not show signs of obesity but if you are in a bountiful time you will.
- Researchers try to find the range of reaction for a given genotype by manipulating the environment (done with plants, animals)
  - For ethical reasons can’t be done with humans
The Concept of Range of Reaction for Intellectual Performance

Canalization

- Certain characteristics resistant to environmental input
- Narrow bandwidth of change regardless of a wide range of environmental contexts
  - Example: Language Acquisition

Gene-Environment Relationships

- **Passive links**: parents transmit traits through genes, the environments they provide, or both.
- **Evocative links**: people react to the characteristics of the child’s genotype.
- **Active links (niche-selection)**: children seek out environments compatible with their genotypes.
Studying Genetic Influence

• Difficult
  – can’t control environments thus no cause-effect analysis
  – Don’t know specific gene-behavior relationship
  – Can’t detail the environment of gene expression
  – Many behaviors controlled by multiple genes in interaction with the environment

Methods to estimate Genetic Influence

• Kinship Studies
  – Determine degree of genetic closeness
    • Parents-children - 50%
    • Siblings - 50%
    • MZ twins - 100%
    • Half siblings - 25%
  – Relate genetic closeness with trait similarity
    • If trait similarity increases with genetic similarity then evidence for heritability

Kinship Study Designs

• Family Studies
  – Relatives within a household are compared
  – Problem: Shared environment also
• Twin Studies
  – Comparison of MZ and DZ twins
• Adoption Studies
  – Comparison of child to biological and adopted parents, siblings
  – Comparison of twins reared apart v.s. together
Measuring the Effects of Nature and Nurture: Twin and Adoption Studies

<table>
<thead>
<tr>
<th>TYPE OF STUDY</th>
<th>OBJECTIVE</th>
<th>KEY COMPARISONS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Twin</td>
<td>Differences in genetic relatedness, same environment</td>
<td>Identical twins together</td>
</tr>
<tr>
<td></td>
<td>Fraternal twins together</td>
<td></td>
</tr>
<tr>
<td>Adoption</td>
<td>Same genetic relatedness, different environments</td>
<td>Identical twins together</td>
</tr>
<tr>
<td></td>
<td>Identical twins apart</td>
<td></td>
</tr>
</tbody>
</table>

Concordance rates for some Behavioral & Personality Disorders

<table>
<thead>
<tr>
<th>Twin Concordances</th>
<th>Identical Twins</th>
<th>Fraternal Twins</th>
</tr>
</thead>
<tbody>
<tr>
<td>Conduct disorder</td>
<td>.85</td>
<td>.70</td>
</tr>
<tr>
<td>Manic depression</td>
<td>.65</td>
<td>.50</td>
</tr>
<tr>
<td>Autism</td>
<td>.65</td>
<td>.50</td>
</tr>
<tr>
<td>Unipolar depression</td>
<td>.65</td>
<td>.50</td>
</tr>
<tr>
<td>Alcoholism—males</td>
<td>.40</td>
<td>.35</td>
</tr>
<tr>
<td>Schizophrenia</td>
<td>.40</td>
<td>.35</td>
</tr>
<tr>
<td>Alcoholism—females</td>
<td>.30</td>
<td>.25</td>
</tr>
</tbody>
</table>

* MZ twins raised apart are close to but below MZ twins raised together

What does this mean?

- There does seem to be a genetic influence on many traits (Evidence?)
- The environment still plays a major role. (Evidence?)