Genetics
Patterns of Inheritance
Biology 20
Genetics

- Study of heredity
- Aristotle
- Von Leewenhoek
- de Graff
- Pangenes
- Homunculus
- ovarian follicle is a miniature person
Blended Theory

• Genetic material mixes (intermediate between parents)
• Individuals in a pop. should reach a uniform appearance w/ time
1. Particulate Theory
   Gene idea
2. Character
3. Trait

1. Parents pass on discrete inheritable units of info to offspring
2. Detectable inheritable feature of an organism.
   Ex. seed color, hair color, seed shape, plant height
3. Each variant of a character
   Ex. seed color (green/yellow), hair color (brown/blonde)
   seed shape (round/wrinkled)
Mendel
1. Removed stamens from purple flower

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

3. Pollinated carpel matured into pod

4. Planted seeds from pod

### Flower color
- Purple
- White

### Flower position
- Axial
- Terminal

### Seed color
- Yellow
- Green

### Seed shape
- Round
- Wrinkled

### Pod shape
- Inflated
- Constricted

### Pod color
- Green
- Yellow

### Stem length
- Tall
- Dwarf

Copyright © 2003 Pearson Education, Inc., publishing as Benjamin Cummings.
Genetics Terms

- **Alleles**
  - Dominant
  - Recessive

- **Genotype**
  - Homozygous dominant:
  - Homozygous recessive:
  - Heterozygous:

- **Phenotype**

- **Alternative versions of gene**
- **Inherited feature**
- **Genetic make-up**
- **Arrangement of alleles**
  - 2 identical alleles (TT)
  - 2 identical alleles (tt)
  - 2 different alleles (Tt)

- **Physical appearance**
True breeding

• Produce offspring with the same trait as parents
True breeding

Homozygous tall
TT x TT

Homozygous short
tt x tt

All Tall

All Short
# P Generation Cross

Tall x short
TT x tt

<table>
<thead>
<tr>
<th></th>
<th>T</th>
<th>T</th>
</tr>
</thead>
<tbody>
<tr>
<td>t</td>
<td>Tt</td>
<td>Tt</td>
</tr>
<tr>
<td>t</td>
<td>Tt</td>
<td>Tt</td>
</tr>
</tbody>
</table>

**F₁ Genotype:** All Tt

**F₁ Phenotype:** All tall
Monohybrid Cross

• Principle of Segregation
  1. the pair of genes for each character:
     → segregate (separates) during gamete production
  2. fusion of gametes at fertilization:
     pairs genes once again
Principle of Segregation

Coat-color genes

- Brown: C
- White: c

Eye-color genes

- Black: E
- Pink: e

Tetrad in parent cell (homologous pair of duplicated chromosomes)

Chromosomes of the four gametes

Copyright © 2003 Pearson Education, Inc., publishing as Benjamin Cummings.
Two people that are heterozygous for brown eyes mate. What are the possible eye colors of their offspring?

B = brown
b = blue

P - Genotype: Bb x Bb

F₁ Genotype:
1 BB:2 Bb:1 bb

F₁ Phenotype:
3 Brown:1 blue
Dihybrid Crosses

• Mendel's Principle of Independent Assortment:

  each allele pair *segregates (separates) independently* of other gene pairs during gamete formation

• formula: $2^n$ where $n = \#$ of heterozygous pairs $=>$ to produce the unique gametes
How many unique gametes will be produced for the following genotypes?

What are the unique gametes?

a) RrYy
   a) 4
      RY, Ry, rY, ry

b) AaBbCcDD
   b) 4
      ABcD, AbcD, aBcD, abcD

c) AAbb
   c) 1
      Ab

d) AABBcCcDdeeFFggHHiiJJJKKLLmmnnooPPqqRRssTT
   d) 2
   What are they?
   • ABcD
   • ABcd
Hypothesis: Dependent assortment

P generation

\[ RRYY \quad rryy \]

Gametes

\[ RY \quad ry \]

F1 generation

\[ RrYy \]

Eggs

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

Sperm

F2 generation

Actual results contradict hypothesis

Hypothesis: Independent assortment

P generation

\[ RRYY \quad rryy \]

Gametes

\[ RY \quad ry \]

F1 generation

\[ RrYy \]

Eggs

\[ \frac{1}{4} RY \quad \frac{1}{4} RY \]
\[ \frac{1}{4} rY \quad \frac{1}{4} rY \]

Sperm

\[ \frac{1}{4} RY \quad \frac{1}{4} RY \]
\[ \frac{1}{4} rY \quad \frac{1}{4} rY \]

F2 generation

Actual results support hypothesis

Copyright © 2003 Pearson Education, Inc., publishing as Benjamin Cummings.
F<sub>1</sub> generation

Meiosis

Metaphase I (alternative arrangements)

Anaphase I

Metaphase II

Gametes

Fertilization among the F<sub>1</sub> plants

F<sub>2</sub> generation

9 : 3 : 3 : 1 (See Figure 9.5A)
Test Cross

• Identify genotype of dominant individual
• Cross with known genotype (recessive)

\[ P = \text{Purple flowers} \]
\[ p = \text{white flowers} \]

\[ PP \text{ or } Pp \times pp \]
Figure 9.5B_1

Test Cross

Phenotypes Genotypes

Black coat, normal vision $B_N$

Black coat, blind (PRA) $B_nn$

Chocolate coat, normal vision $bbN$

Chocolate coat, blind (PRA) $bbnn$
Rules of Probability

Figure 9.7

**F₁ genotypes**

- **Bb female**

**Formation of eggs**

- **Formation of sperm**

**F₂ genotypes**

- **Eggs**

<table>
<thead>
<tr>
<th>Sperm</th>
<th>Eggs</th>
</tr>
</thead>
<tbody>
<tr>
<td>(\frac{1}{2})</td>
<td>(\frac{1}{2})</td>
</tr>
<tr>
<td>(\frac{1}{2})</td>
<td>(\frac{1}{2})</td>
</tr>
<tr>
<td>(\frac{1}{4})</td>
<td>(\frac{1}{4})</td>
</tr>
<tr>
<td>(\frac{1}{4})</td>
<td>(\frac{1}{4})</td>
</tr>
</tbody>
</table>
Incomplete Dominance

- **P generation**
  - Red \( RR \)
  - White \( rr \)

- **F\(_1\) generation**
  - Pink \( Rr \)
  - Gametes \( \frac{1}{2} R, \frac{1}{2} r \)

- **F\(_2\) generation**
  - Red \( RR \)
  - Pink \( Rr \)
  - White \( rr \)
  - Gametes \( \frac{1}{2} R, \frac{1}{2} r \)
  - Eggs \( \frac{1}{2} R, \frac{1}{2} r \)
  - Sperm \( \frac{1}{2} R, \frac{1}{2} r \)

Copyright © 2003 Pearson Education, Inc., publishing as Benjamin Cummings.
Incomplete Dominance

- A mom has high blood cholesterol whereas her husband is normal.

<table>
<thead>
<tr>
<th>Genotypes</th>
<th>Phenotypes</th>
</tr>
</thead>
<tbody>
<tr>
<td>$HH$ Homozygous for ability to make LDL receptors</td>
<td>Normal</td>
</tr>
<tr>
<td>$Hh$ Heterozygous</td>
<td>Mild disease</td>
</tr>
<tr>
<td>$hh$ Homozygous for inability to make LDL receptors</td>
<td>Severe disease</td>
</tr>
</tbody>
</table>

What’s the probability that their child will have high blood cholesterol like their mother?
Blood Groups

• Co-dominance
  Where both alleles are expressed
  AB blood

• Multiple Alleles
  More than 2 alleles determine genotypes
  3 alleles → $I^A$, $I^B$ and $I^O$ or $i$
<table>
<thead>
<tr>
<th>Blood Group (Phenotype)</th>
<th>Genotypes</th>
<th>Carbohydrates Present on Red Blood Cells</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>$I^A I^A$ or $I^A i$</td>
<td><img src="image" alt="Carbohydrate A" /></td>
</tr>
<tr>
<td>B</td>
<td>$I^B I^B$ or $I^B i$</td>
<td><img src="image" alt="Carbohydrate B" /></td>
</tr>
<tr>
<td>AB</td>
<td>$I^A I^B$</td>
<td><img src="image" alt="Carbohydrate A and Carbohydrate B" /></td>
</tr>
<tr>
<td>O</td>
<td>$ii$</td>
<td><img src="image" alt="Neither" /></td>
</tr>
<tr>
<td>Blood Group (Phenotype)</td>
<td>Genotypes</td>
<td>Carbohydrates Present on Red Blood Cells</td>
</tr>
<tr>
<td>-------------------------</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="Carbohydrate A" /></td>
</tr>
<tr>
<td>B</td>
<td>$I^B I^B$ or $I^B i$</td>
<td><img src="image" alt="Carbohydrate B" /></td>
</tr>
<tr>
<td>AB</td>
<td>$I^A I^B$</td>
<td><img src="image" alt="Carbohydrate A and Carbohydrate B" /></td>
</tr>
<tr>
<td>O</td>
<td>$ii$</td>
<td><img src="image" alt="Neither" /></td>
</tr>
</tbody>
</table>
Blood Information

- Universal donor
- Universal recipient
- Antigens → cell identifier (CHO)
- Antibodies → defense (protein)
- Rh factor (Rhesus factor)
- Rh (+) or Rh (-)
- RhoGAM
Pleiotropy

- One gene $\rightarrow$ multiple affects
Epistasis

- One gene affects (controls) another gene’s expression

The eye color genes code for the production of a yellow-brown pigment

First Iris Layer Pigment
AA = Produce lots of pigment
Aa = Produce some pigment
aa = Do not produce pigment

Second Iris Layer Pigment
BB = Produce lots of pigment
Bb = Produce some pigment
bb = Do not produce pigment

Yellow overlay gene
- When combined with the basic pigment gene alters:
  - Light brown to hazel
  - Light blue to green
Epistasis

• Fur pigment
  \( E = \) dark pigment
  \( ee = \) no pigment

• Darkness & distribution of pigment
  \( B = \) dark
  \( b = \) light

\[
\begin{align*}
eebb & \quad eeB_\_ & \quad E\_bb & \quad E\_B\_
\end{align*}
\]
Epistasis

Phenotypes
Genotypes

Black coat, normal vision
$B_N$

Black coat, blind (PRA)
$B_{nn}$

Chocolate coat, normal vision
$bbN$

Chocolate coat, blind (PRA)
$bbnn$

Mating of heterozygotes
(black, normal vision)

Phenotypic ratio of offspring

$BbNn \times BbNn$

9 black coat, normal vision

3 black coat, blind (PRA)

3 chocolate coat, normal vision

1 chocolate coat, blind (PRA)
Polygenic Inheritance

- 2+ genes affects phenotype
- Examples:
  - Skin color
  - Height
  - Hair pigmentation
Phenocopy

- Environment affects phenotype
Recessively Inherited Disorders

- Heterozygous
  normal → no disorder
  “carrier”

- Albinism
- Chromosome 11
- 1/22,000
Recessively Inherited Disorders

• Tay Sachs Disease
• Chromosome 15
• Lipid accumulation in brain

• Occurs:
  – In utero → death by 5 yrs
  – Juvenile (~4 yrs) → death by 15
  – Adult (~30 yrs) → milder

• 1/3500 central European Jews
Cystic Fibrosis
• Chromosome 7
• Accumulation of Cl⁻
• Mucus build-up in lungs & liver
• 1/1,800 Caucasians
• 1/28 Caucasians → unknown carriers
Recessively Inherited Disorders

- Phenylketonuria (PKU)
- Phenylalanine
- Mental retardation
- 1/10,000 US & Europe
Dominantly Inherited Disorders

- Achondroplasia (dwarfism)
- Chromosome 7
  - Growth hormone deficient
- 1/25,000
Dominantly Inherited Disorders

- Phenylthiocarbamide (PTC)
- Polydactyly
  - Extra digits
Dominantly Inherited Disorders

- Huntington’s Disease
- Chromosome 4
- Begins in middle age
- Nervous degeneration
# 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>Very easily sunburned</td>
</tr>
<tr>
<td>Cystic fibrosis</td>
<td>Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in infancy unless treated</td>
<td>(\frac{1}{1,800}) Caucasians</td>
<td>See Modules 9.9 and 12.11</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}{500}) African Americans</td>
<td>Alleles are codominant; see Modules 9.13–9.15</td>
</tr>
<tr>
<td>(homozygous)</td>
<td></td>
<td></td>
<td></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 4.12</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 (one type)</td>
<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>See Modules 9.9 and 12.11</td>
</tr>
<tr>
<td>Hypercholesterolemia</td>
<td>Excess cholesterol in blood; heart disease</td>
<td>(\frac{1}{500}) are heterozygous</td>
<td>Incomplete dominance; see Module 9.12</td>
</tr>
</tbody>
</table>
Linked Genes

- Inherited together
- Do not assort independently

Experiment

<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

Most offspring: 3 purple long : 1 red round
Not accounted for: purple round and red long
Sex-linked Genes

- X-linked
- most X-linked genes have no homologous loci on the Y chromosome
- genes located on the sex chromosomes → X
- two sex chromosomes X & Y
- females XX, males XY
- fathers pass X-linked alleles to only all of their daughters

- Normal
  \[ X^A X^A \quad X^A Y \]
  \[ X^A X^a \]

- Sex-linked disorder
  \[ X^a X^a \quad X^a Y \]
Males have more sex-linked disorders than females. WHY?
Sex-linked Cross
Sex-linked Cross

$R = \text{red-eye allele}$

$r = \text{white-eye allele}$

Copyright © 2003 Pearson Education, Inc., publishing as Benjamin Cummings.
A man with normal vision marries a woman who has normal vision but whose father was colorblind.

1. What is the genotype for the man & woman?
2. What % of their children will have normal vision?
3. What % of their sons will have normal vision?
4. What % of their daughters will have normal vision?
5. Will they have any colorblind children? If so, whom?

1. $X^A Y$ and $X^A X^a$.

<table>
<thead>
<tr>
<th></th>
<th>$X^A$</th>
<th>Y</th>
</tr>
</thead>
<tbody>
<tr>
<td>$X^A$</td>
<td>$X^A X^A$</td>
<td>$X^A Y$</td>
</tr>
<tr>
<td>$X^a$</td>
<td>$X^A X^a$</td>
<td>$X^a Y$</td>
</tr>
</tbody>
</table>
A man with normal vision marries a woman who has normal vision but whose father was colorblind.

1. What is the genotype for the man & woman? 1. $X^A Y$ and $X^A X^a$.
2. What % of their children will have normal vision? 2. 75%
3. What % of their sons will have normal vision? 3. 50%
4. What % of their daughters will have normal vision? 4. 100%
5. Will they have any colorblind children? If so, whom? 5. Yes, males.
Red-green colorblindness
Hemophilia
Sex Influenced Trait

- Pattern Baldness
  - BB: Normal male, normal female
  - Bb: Bald male, normal female
  - bb: Bald male, thin female
  - BbXY: Balding male
  - BbXX: Normal female
Carrier recognition:

- Tests for heterozygous carriers for: Tay-Sachs allele, cystic fibrosis & sickle-cell
- Enables people to: Make informed decisions about having children
- Could also be abused: Ethical dilemmas about social implications of technology
Fetal Testing

- Alpha fetoprotein (AFP)
- Protein levels associated with Downs
- Amniocentesis
Fetal Testing

- Alpha fetoprotein (AFP)
- Amniocentesis
- Choronic villi sampling
Figure 9.10A

**Amniocentesis**

- Ultrasound transducer
- Fetus
- Placenta
- Uterus
- Amniotic fluid extracted
- Centrifugation
- Amniotic fluid
- Fetal cells
- Cultured cells
- Several hours
- Several weeks
- Biochemical and genetics tests
- Several weeks
- Karyotyping

**Chorionic Villus Sampling (CVS)**

- Ultrasound transducer
- Fetus
- Placenta
- Chorionic villi
- Tissue extracted from the chorionic villi
- Several hours
- Fetal cells
- Several weeks
Fetal Testing

• Alpha fetoprotein (AFP)
• Amniocentesis
• Choronic villi sampling
• Ultrasound