Lab Activity 36

Principles of Heredity

Portland Community College
BI 233
Terminology of Chromosomes

• **Homologous chromosomes**: A pair, of which you get one from mom, and one from dad.
  • Example: the pair of chromosomes 21 are homologous to each other
• **Sex Chromosomes**: The X and Y
  • These are homologous to each other
• **Autosomal Chromosomes**: The other 22 pairs of chromosomes that do not determine gender
• **Karyotype**: A chart of the chromosomes arranged in homologous pairs.
Karyotype of Human Chromosomes

• 22 pairs of autosomes
• 1 pair of sex chromosomes
<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gene</td>
<td>A genetic factor (region of DNA) that helps determine a characteristic</td>
</tr>
<tr>
<td>Allele</td>
<td>One of two or more alternate forms of a gene</td>
</tr>
<tr>
<td>Locus</td>
<td>Specific place on a chromosome occupied by an allele</td>
</tr>
<tr>
<td>Genotype</td>
<td>Set of alleles that an individual organism possesses</td>
</tr>
<tr>
<td>Heterozygote</td>
<td>An individual organism possessing two different alleles at a locus</td>
</tr>
<tr>
<td>Homozygote</td>
<td>An individual organism possessing two of the same alleles at a locus</td>
</tr>
<tr>
<td>Phenotype or trait</td>
<td>The appearance or manifestation of a character</td>
</tr>
</tbody>
</table>
Terminology of Genes: Alleles

- **Allele**: Alternative form of a gene at the same locus on homologous chromosomes
- **Homozygous**: Two alleles controlling a single trait are the same
- **Heterozygous**: The two alleles for a trait are different
- **Dominant**: An allele masks or suppresses the expression of its partner
- **Recessive**: The allele that is masked or suppressed
Allele $R$  

Allele $r$
Terminology

- **Genotype**: the genetic makeup
- **Phenotype**: the way one’s genotype is expressed
- **Punnett square**
  - Method of showing 4 possible genetic combinations in offspring of 2 individuals
<table>
<thead>
<tr>
<th></th>
<th>A</th>
<th>a</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>A</strong></td>
<td><strong>AA</strong></td>
<td><strong>Aa</strong></td>
</tr>
<tr>
<td></td>
<td>Homozygous Dominant</td>
<td>Heterozygous</td>
</tr>
<tr>
<td>a</td>
<td><strong>Aa</strong></td>
<td><strong>aa</strong></td>
</tr>
<tr>
<td></td>
<td>Heterozygous</td>
<td>Homozygous Recessive</td>
</tr>
</tbody>
</table>

Mother: Aa  
Father: Aa
Dominant and Recessive Genes

- **Homozygous**: The person has 2 copies of the dominant or 2 copies of the recessive gene (gets one from each parent)
- **Heterozygous**: The person has 1 copy of the dominant and 1 copy of the recessive gene
  - Genotype: Aa
  - Phenotype: Dominant Characteristic A
Example: Eye Color
Dominant and Recessive

- Dominant: Brown (B)
- Recessive: Blue (b)
- Parents:
  - Mom blue eyed (bb),
  - Dad: homozygous brown eyed (BB)
- 100% of children will be heterozygous (Bb) with brown eyes
Example: Eye Color
Dominant and Recessive

- Dominant: Brown (B)
- Recessive: Blue (b)
- Parents:
  - Mom blue eyed (bb),
  - Dad: heterozygous brown eyed (Bb)
- 50% of children will be heterozygous (Bb) with brown eyes
- 50% will be homozygous recessive (bb) with blue eyes
Dominant and Recessive

(a) P generation
Tall $Tt$ × Short $tt$

Gametes $T$ $t$ $t$ $t$

Fertilization

(b) F$_1$ generation

|$Tt$ | $Tt$
---|---|
|Tall | Tall
|tt | tt
|Short | Short

Conclusion: Genotypic ratio 1 $Tt$:1 $tt$
Phenotypic ratio 1 Tall:1 Short
Why Marrying Your Cousin is Bad!!!

• Inbreeding causes recessive alleles to become homozygous more often.
• If the recessive allele contains a genetic disease, it will show up in these children at a higher rate than in the normal population.
• Examples:
  • Tay-Sachs disease occurs primarily among Jews of Eastern European descent
(a) Alleles identical by descent
Incomplete Dominance

- Heterozygous individuals have a phenotype *intermediate* between homozygous dominant and homozygous recessive.
- Sickling is a human example when aberrant hemoglobin (Hb) is made from the recessive allele (s).

\[
\begin{align*}
SS &= \text{normal Hb is made} \\
Ss &= \text{sickle-cell trait (both aberrant and normal Hb is made)} \\
ss &= \text{sickle-cell anemia (only aberrant Hb is made)}
\end{align*}
\]
Incomplete Dominance

P generation

Purple fruit

Gametes

Fertilization

White fruit

Gametes

Fig_03-09-1 Genetics, Second Edition © 2005 W.H. Freeman and Company
Incomplete Dominance

**F₁ generation**

Violet fruit

\[ Pp \times Pp \]

Gametes

\[ P \quad p \quad p \quad P \]

Fertilization
Incomplete Dominance

**Conclusion:** Genotypic ratio 1 PP : 2 Pp : 1 pp
Phenotypic ratio 1 purple : 2 violet : 1 white
Multiple-Allele Inheritance

- Genes that exhibit more than two alternate alleles
- ABO blood grouping is an example
- Three alleles ($I^A$, $I^B$, $i$) determine the ABO blood type in humans
- $I^A$ and $I^B$ are codominant (both are expressed if present), and $i$ is recessive
  - Rh factor is complete dominance
Sex-Linked Inheritance

- Inherited traits determined by genes on the sex chromosomes
- X chromosomes bear over 2500 genes; Y carries about 15
- X-linked genes are:
  - Found only on the X chromosome
  - Typically passed from mothers to sons
  - Never masked or damped in males since there is no Y counterpart
Example: Color blindness
Sex-Linked Inheritance

- Dad is color blind ($X^OY$)
- Mom is homozygous normal ($XX$)
- No sons affected
- All daughters are carriers
Example: Color blindness
Sex-Linked Inheritance

- Dad is normal (XY)
- Mom is a carrier (X°X)
- 50% of sons affected
- 50% of daughters are carriers
Example: Color blindness
Sex-Linked Inheritance

- Dad is normal (XY)
- Mom is color blind (X°X°)
- 100% of sons affected
- 100% of daughters are carriers

<table>
<thead>
<tr>
<th></th>
<th>X</th>
<th>Y</th>
</tr>
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<tbody>
<tr>
<td>X°</td>
<td>X°X</td>
<td>X°Y</td>
</tr>
<tr>
<td>X°</td>
<td>X°X</td>
<td>X°Y</td>
</tr>
</tbody>
</table>
X-Chromosome Inactivation

- Females have double dose of X chromosome in all cells
- One X chromosome is randomly & permanently inactivated early in development
- Visible as dark-staining Barr body easily seen in nucleus of neutrophils as “drumstick”
  - Tightly coiled even in interphase cell
Example: X-Chromosome Inactivation
Polygenic Inheritance

- Depends on several different gene pairs at different loci acting in tandem
- Results in continuous phenotypic variation between two extremes
- Examples: skin color, eye color, and height
  - Although we think of eye color as simple dominant/recessive, there are many genes that code for eye color, which is why your eyes are not usually the exactly the same color as either of your parents.
Genomic Imprinting

• Genomic imprinting is the differential expression of a gene depending on whether it is inherited from the mother or the father

• Examples:

  • Igf2: Insulin-like growth factor 2
    • Only the paternal gene is expressed
    • Affects birth weight of human infants

• Prader-Willi and Angelman syndromes
Genomic Imprinting
Prader-Willi and Angelman Syndromes

- Caused by a small deletion on Chromosome 15
- Prader-Willi Syndrome: Paternal inheritance
  - Small hands and feet, Short stature
  - Voracious appetite
  - Mental retardation
- Angelman Syndrome: Maternal inheritance
  - Uncontrolled muscle movement
  - Feeding issues (poor eaters)
  - Very happy demeanor (laugh all the time)
  - Unusual seizures
  - Mental retardation
Cytoplasmic Inheritance

- A zygote inherits nuclear genes from both parents
- Cytoplasmic organelles are inherited only from the mother
  - Mitochondria have their own DNA
  - Contains 37 genes (nuclear DNA has 30,000 genes)
- Always passed from mother to all children
Abnormal Chromosomes: Trisomy

- Trisomy: Having 3 copies of a chromosome (or part of a chromosome) instead of 2
  - Can be caused by nondisjunction during Meiosis II when the sister chromatids are supposed to separate.
  - Can happen during prophase I crossing over (partial duplication of a chromosome)
  - Can happen during mitosis
  - Usually lethal
Trisomy 21

- Down’s syndrome
- Severe: 3 copies of chromosome 21
- Less severe: one chromosome 21 has an “extra” piece, so only some of the genes are in triplicate
- Less severe: if the defect happened during mitosis in the early embryo resulting in only some cells carrying 3 copies
Trisomy 18: Edward’s Syndrome

- Severely retarded
- Usually fatal shortly after birth.
Trisomy XXX

- 1 in 1,000 newborn girls
- Many girls and women with Triple X have no signs or symptoms. Signs and symptoms vary a lot between individuals, but can include:
  - Increased space between the eyes
  - Tall stature (height)
  - Small head
  - Learning disabilities
  - Delayed puberty
  - Infertility
  - Mental retardation
XXY
Klinefelter's Syndrome

• 1 in 1,000 male births
  • Sterility
  • Breast development
  • Incomplete masculine body build
• Social and/or school learning problems
Monosomy: XO Turner Syndrome

- Females with only one X
  - Short stature
  - Lack of ovarian development
  - Webbed neck
  - Arms turn out slightly at the elbow
  - Mentally normal
Pleiotropy

- The control by a single gene of several distinct and seemingly unrelated phenotypic effects.
- Example: PKU (phenylketonuria).
  - This disease causes mental retardation and reduced hair and skin pigmentation.
  - The cause is a mutation in a single gene that codes for the enzyme phenylalanine hydroxylase (PAH) that converts the phenylalanine to tyrosine.
  - The mutation results in no or reduced conversion of phenylalanine to tyrosine, and phenylalanine concentrations increase to toxic levels, causing damage at several locations in the body.
Pedigree

• A pedigree is a diagram of family relationships that uses symbols to represent people and lines to represent genetic relationships.

• These diagrams make it easier to visualize relationships within families, particularly large extended families.

• Pedigrees are often used to determine the mode of inheritance (dominant, recessive, etc.) of genetic diseases.
Factors to Consider in Pedigrees

- Is the trait located on a sex chromosome or an autosome?
  - Autosomal – not on a sex chromosome
  - Sex Linkage – located on one of the sex chromosomes
    - Y-linked - only males carry the trait.
    - X-linked (recessive) - sons inherit the disease from normal parents

- How is the trait expressed?
  - Dominant - the trait is expressed in every generation.
  - Recessive - expression of the trait may skip generations.
Pedigree Diagrams

- Male
- Female
- Affected individual

Mating

I
- Offspring in birth order; I and II are generations; offspring numbered II-1 and II-2

II

- Identical twins
- Non-identical twins
Autosomal Dominant Inheritance

- A disorder appears in several generations of a family.
- Affected parents have a 50% risk of an affected child with each pregnancy.

Points to consider:
1. Affected individuals have at least one affected parent
2. The phenotype generally appears every generation
3. Two unaffected parents only have unaffected offspring
Autosomal Recessive Inheritance

- Disorders often appear in only one generation of a family.
- Carrier couples have a 25% risk of an affected child with each pregnancy.

Points to consider:
1. Unaffected parents can have affected offspring
2. Affected progeny are both male and female
Example: Albinism

• Expressed in both sexes at approximately equal frequency, therefore it is autosomal.
• Not expressed in every generation, therefore it is recessive.
Albinism: Genotype of the Affected Individuals

- Assign codes for the alleles.
  - Code “A” for the dominant normal allele.
  - Code “a” for the recessive allele for albinism.
- Affected individuals must be homozygous for “a.”
- First generation parents must be “Aa” because they have normal phenotypes, but affected offspring.

```
Aa Aa
aa aa
```
Albinism: Genotype of the Normal Individuals

Normal individuals must have at least one “A.”
Albinism: Parent-Offspring Relationships

- “1” must transmit “a” to each offspring.
- The “A” in the offspring must come from the father.
- Normal father “2” could be either heterozygous or homozygous for an “A?”
X-linked Inheritance

- X-linked diseases are almost always recessive
- Male to male transmission of X-linked disorders is not seen.
- Carriers are designated with a dot.
Example: Hemophelia

- In this pedigree, only males are affected, and sons do not share the phenotypes of their fathers.
  - Thus, hemophilia is linked to a sex chromosome—the X.
- Expression of hemophilia skips generations.
  - Thus, it is recessive.
Hemophilia: Expression of the Female Sex Chromosomes

• All females are XX.
Hemophilia: Genotype the Affected Individuals

- Assign codes for the alleles.
  - Code “H” for the recessive hemophilia allele.
- Affected individuals must have an “H” on an X chromosome.
Y-Linked Inheritance

- Only males are affected.
- All sons of an affected father have the trait.
- Thus, the trait is Y-linked.
Cytoplasmic Inheritance

- Always passed from mother to all children
- Never passed through males to their offspring.
The End