Biol 304: General Genetics

Extensions & Modification of Basic Principles of Heredity

Under basic principles of heredity we expect:

\[ \text{Aa x Aa} = \frac{3}{4} \text{ phenotypic ratio} \]

\[ \text{AaBb x AaBb} = \frac{9}{16} : \frac{3}{16} : \frac{3}{16} : \frac{1}{16} \text{ phenotypic ratio} \]

However, under certain circumstances we get deviations from expected ratios:

1) Incomplete & partial dominance
2) Sex limited & sex-influenced traits
3) Multiple alleles
4) Lethals
5) Gene interaction/epistasis
6) Pleiotropy

Table 5.1 Differences between dominance, incomplete dominance, and codominance

<table>
<thead>
<tr>
<th>Type of Dominance</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dominance</td>
<td>Phenotype of the heterozygote is the same as the phenotype of one of the homozygotes.</td>
</tr>
<tr>
<td>Incomplete dominance</td>
<td>Phenotype of the heterozygote is intermediate (falls within the range) between the phenotypes of the two homozygotes.</td>
</tr>
<tr>
<td>Codominance</td>
<td>Phenotype of the heterozygote includes the phenotypes of both homozygotes.</td>
</tr>
</tbody>
</table>
Under basic principles of heredity we expect:

\[ \text{Aa} \times \text{Aa} = 3:1 \text{ phenotypic ratio} \]

\[ \text{AaBb} \times \text{AaBb} = 9:3:3:1 \text{ phenotypic ratio} \]

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Sex-limited characteristics

Genes that encode sex-influenced traits are inherited according to Mendel’s principles but are expressed differently in males and females.
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Mendel’s principle of segregation applies to crosses with multiple alleles

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### Lethals

Alleles, which when present in the homozygous state, are lethal.

Can be autosomal or sex-linked.

**Ex.** Spider syndrome in sheep:
- Abnormal transformation of cartilage to bone (i.e., ends of bones are calcified)
- SS=normal, Ss=normal, ss= spider syndrome

Yellow coat color in mice is caused by a recessive lethal gene, producing distorted phenotypic ratios in the progeny of two yellow mice:

![Image of mice showing phenotypic ratios](image-url)

*Figure 7: Mendelian Inheritance, Fourth Edition, Sinauer Associates, Inc.*
Under basic principles of heredity we expect:

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\[ AaBb \times AaBb = 9:3:3:1 \] phenotypic ratio

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**Gene Interaction/Epistasis**

**Epistasis** - interaction of two genes at different loci controlling discrete traits. Usually the interaction is nonreciprocal such that one gene interferes with or prevents the expression of another.
Gene interaction in which two loci determine a single characteristic

A multistep biochemical pathway synthesizes the carotenoid pigments responsible for color variation in peppers.

Yellow pigment in summer squash is produced in a two-step pathway.
Pigment is produced in a two-step pathway in snails

1. A dominant allele at the A locus is required to produce enzyme A, which converts compound A into compound B.
2. A dominant allele at the B locus is required to produce enzyme B, which converts compound B into compound C (pigment).
3. Pigmented snails must produce enzymes A and B, which requires genotype A_A_B_.
4. Albino snails arise from the absence of enzyme A (a) or compound B is never produced...
5. ...or from the absence of enzyme B (b) or compound B is never produced, or from the absence of both enzymes (aabb).

Recessive Epistasis (labrador retrievers)

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>B_E_</td>
<td>black</td>
</tr>
<tr>
<td>bbE_</td>
<td>brown</td>
</tr>
<tr>
<td>B_ee</td>
<td>yellow</td>
</tr>
<tr>
<td>bbee</td>
<td>yellow</td>
</tr>
</tbody>
</table>

P  BBEE x bbee
F1 BbEe (BbEe x BbEe)
F2 9/16 B_E_ (black)
     3/16 bbE_ (brown)
     3/16 B_ee (yellow)
     1/16 bbee (yellow)  9:3:4 ratio

Table 5.2: Modified dihybrid phenotypic ratios due to gene interaction

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>A_A_</td>
<td>black</td>
</tr>
<tr>
<td>A_a_</td>
<td>yellow</td>
</tr>
<tr>
<td>a_a_</td>
<td>albino</td>
</tr>
</tbody>
</table>

*Each ratio is produced by a dihybrid cross (AaBb x AaBb). Shaded bars represent combinations of genotypes that give the same phenotype.
Modified Mendelian Ratios in other Dihybrid crosses

<table>
<thead>
<tr>
<th>Case</th>
<th>Organism</th>
<th>Character</th>
<th>T_2 Phenotypes</th>
<th>Modified ratio</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Mouse</td>
<td>Coat color</td>
<td>agouti, albino, black, albino</td>
<td>9:3:3:1</td>
</tr>
<tr>
<td>2</td>
<td>Squash</td>
<td>Color</td>
<td>white, yellow, green</td>
<td>12:3:1</td>
</tr>
<tr>
<td>3</td>
<td>Pea</td>
<td>Flower color</td>
<td>purple, white</td>
<td>9:7</td>
</tr>
<tr>
<td>4</td>
<td>Squash</td>
<td>Fruit shape</td>
<td>disc, sphere, long</td>
<td>9:6:1</td>
</tr>
<tr>
<td>5</td>
<td>Chicken</td>
<td>Color</td>
<td>white, colored, white</td>
<td>15:5</td>
</tr>
<tr>
<td>6</td>
<td>Mouse</td>
<td>Color</td>
<td>white-spotted, colored, white</td>
<td>10:9:9</td>
</tr>
<tr>
<td>7</td>
<td>Shearwater</td>
<td>Pore capsule</td>
<td>triangular, oval</td>
<td>15:1</td>
</tr>
<tr>
<td>8</td>
<td>Flour beetles</td>
<td>Color</td>
<td>red, savoy, red savoy, black, jet</td>
<td>9:3:3:1</td>
</tr>
</tbody>
</table>

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Pleiotropy - situation which exists when a single gene influences more than one trait

Ex. Phenylketonuria - an autosomal recessive genetic disorder characterized by a deficiency in the enzyme phenylalanine hydroxylase (PAH). This enzyme is necessary to metabolize the amino acid phenylalanine to the amino acid tyrosine. PAH is found on chromosome number 12.

If left untreated, mental retardation, blue eyes, and light colored skin. A diet low in phenylalanine and high in tyrosine can be a very effective treatment.

Penetrance & Expressivity

Concept of Penetrance

Degree of expression of a trait can be studied quantitatively

**Penetrance** - % of individuals that show some degree of expression of the mutant genotype

If 15% of (genotypically) mutant flies exhibit the wildtype phenotype, then the mutant gene is said to have a penetrance of 85%.

Polydactyly - occasionally people possess the dominant allele for polydactyly, but have a normal phenotype. We know they have it because their children inherit the polydactyly.
**Concept of Expressivity**

Expressivity - variation in phenotype/range of expression of individuals with a given genotype (Ex. Polydactyly)

Human polydactyly exhibits incomplete penetrance and variable expressivity

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**Genetic Maternal Effect**

The phenotype of the offspring is determined by the genotype of the mother and arise when substances present in the cytoplasm of an egg (encoded by the mother’s nuclear genes) are pivotal in early development.
Cytoplasmic Inheritance

The phenotype of the offspring is determined by genes that are not found in the nucleus, but in the cytoplasm.

EX. Mitochondria and chloroplasts

A zygote inherits nuclear genes from both parents, but usually all of its cytoplasmic organelles/genes from only one parent...usually the egg
Genomic Imprinting

Although a zygote inherits autosomal genes from both parents, the effect of gene expression can be affected by their parental origin. This phenomenon is called genomic imprinting.

A gene that exhibits genomic imprinting in both mice and humans is Igf2, which encodes a protein called insulin-like growth factor II. Offspring inherit one allele from both parents; however, the paternal allele is expressed in the fetus and placenta, but the maternal copy is “silent”.

Ex. Prader-willi and Angelman syndromes - see page 120 of your text for explanations

Genes and the environment

Temp-sensitive alleles

Phenocopy - situation where environmental factors alone can produce a phenotype that is the same phenotype produced by a specific genotype