Chapter 3

Transmission of Genes From Generation to Generation
3.1 Heredity: How are Traits Inherited?

- Why do we begin examining inheritance by discussing Gregor Mendel and pea plants?

- Before Mendel experimented with the inheritance of traits in garden peas there was no clear understanding of how traits were inherited and passed from one generation to the next.

- There was, however, a good supply of data on garden peas and how to grow them.
3.3 Crossing Pea Plants: Single Traits

- Mendel’s initial crosses studied the inheritance of a single trait such as shape or seed color and each trait had only two varieties. In all, he studied seven simple traits, for example…

<table>
<thead>
<tr>
<th>Trait</th>
<th>variety 1</th>
<th>variety 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>pea shape</td>
<td>smooth</td>
<td>wrinkled</td>
</tr>
<tr>
<td>pea color</td>
<td>yellow</td>
<td>green</td>
</tr>
<tr>
<td>pod shape</td>
<td>full</td>
<td>constricted</td>
</tr>
<tr>
<td>flower position</td>
<td>axial</td>
<td>terminal</td>
</tr>
</tbody>
</table>

**true breeding—when self pollinated offspring always exhibited the same variety of that trait**
Mendel’s Terminology

- P1 = parental generation
- F1 = first generation (F stands for *filial* or son in Latin.)
- F2 = second generation

Example experiment
- P1: smooth x wrinkled
- F1: offspring all smooth
- F2: offspring 5,474 smooth (75%)
  1,850 wrinkled (25%)  

- For each cross the F1 and F2 generation always showed the same results
Mendel’s’ Conclusions

- In these crosses there were two inherited factors that were responsible for the trait (these factors are now referred to as genes).

- In the F1 generation, one factor recessed into the background, but returned in the F2 generation. (recessive)

- For a specific trait, F1 plants must carry two factors, one from each parent
Combinations of Gene Forms (Alleles)

- **Allele**
  - Alternative forms of a gene (P or p)
  - There may be many alleles within a population, but each individual has only two alleles for each gene

- **Homozygous**
  - Having identical alleles for one gene (PP or pp)

- **Heterozygous**
  - Having two different alleles for one gene (Pp)
Recessive and Dominant Alleles

- **Dominant Allele**
  - Will mask the phenotype of the recessive
  - Allele expressed in the F1 (heterozygous) condition

- **Recessive Allele**
  - Need two recessive alleles to express the trait
Phenotype and Genotype

- **Genotype**
  - The specific genetic make up of an organism
    - PP, Pp, or pp

- **Phenotype**
  - Observable properties of an organism or how it looks
    - Smooth, round, wrinkled, short, tall
Mendel’s Principle of Segregation

- For each trait, the pair of factors (alleles) separate from each other during gamete formation.

\[ \text{meiosis} \]

\[ \text{A} \quad \text{a} \]
Using the Principle of Segregation in a Punnett Square

**F₁ cross**

Gamete formation by F₁ parents

Set up Punnett square

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 SS</td>
<td>3/4 Smooth</td>
</tr>
<tr>
<td>2 Ss</td>
<td>Smooth</td>
</tr>
<tr>
<td>1 ss</td>
<td>1/4 wrinkled</td>
</tr>
</tbody>
</table>

Gamete combinations represent random fertilization
Using a punnett square to make predictions.

A purple-flowered plant (PP) is crossed with a white-flowered plant (pp). What is the probability of getting a white flowered offspring?
Using the Principle of Segregation in a Punnett Square

Probability of white flower = 0
Next, pause the presentation and try this one yourself before going on to the next slide.

A purple-flowered plant (Pp) is crossed with a white-flowered plant (pp). What is the probability of getting a white flowered offspring?
Using the Principle of Segregation in a Punnett Square

Probability of white flower = \( \frac{1}{2} \) or 50%
Mendelian Traits in Humans

Some traits in humans are caused by a single gene with alleles that are either dominant or recessive:

- Cleft chin (dominant) vs. smooth chin
- Hitchhiker’s thumb (dominant) vs. straight thumb
- Free (dominant) vs. attached earlobes
- Freckles (dominant) vs. none
- Albinism (recessive)
3.4 More Pea Plants, Multiple Traits: The Principle of Independent Assortment

**But what happens when two traits are under study…**

- Mendel’s later experiments showed that alleles of different genes segregate independently from those of other gene pairs
Mendel’s Principle of Independent Assortment
Dihybrid Cross Problems

Cross two parents:

RrYy x RRYy

- What is the probability of having offspring with round, yellow seeds?
- What is the probability of having offspring with wrinkled, yellow seeds?
Dihybrid Cross Problems

Cross two parents:

RrYy x RRYy

- What is the probability of having offspring with round, yellow seeds?
- Complete a Punnett square for each gene, then multiply the probabilities together:

RrYy x RRYy

<table>
<thead>
<tr>
<th></th>
<th>R</th>
<th>r</th>
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<tbody>
<tr>
<td>R</td>
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<td>Rr</td>
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<td>R</td>
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<table>
<thead>
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<th></th>
<th>Y</th>
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<tr>
<td>Y</td>
<td>YY</td>
<td>Yy</td>
</tr>
<tr>
<td>y</td>
<td>Yy</td>
<td>yy</td>
</tr>
</tbody>
</table>
Dihybrid Cross Problems

Cross two parents:

RrYy x RRYy

- What is the probability of having offspring with wrinkled, yellow seeds?
Mendel’s Principle of Independent Assortment

- **Independent assortment**
  - The random distribution of alleles of *different genes* into gametes during meiosis
  - Yields all possible combinations of gametes with equal probability in a cross between two individuals
Meiosis I

Metaphase II

Gametes

Fig. 3-12, p. 55
Mendel’s Contribution

- Mendel’s principle of segregation and principle of independent assortment are fundamental to our understanding of the science of heredity (genetics).

- We can identify genetic traits because they have a predictable pattern of inheritance worked out by Gregor Mendel.
3.5 Meiosis Explains Mendel’s Results: Genes are on Chromosomes

…in all living organisms

- Genes pairs (alleles) are located on chromosome pairs
- The position occupied by a gene on a chromosome is referred to as a locus
- The behavior of chromosomes in meiosis causes segregation and independent assortment of alleles
Pedigrees

- Traits in humans are traced by constructing pedigrees that follow traits through generations.

- A full description of pedigree analysis will be covered in the presentation for chapter 4. Please read the introductory information in chapter 3.
3.7 Variations from Mendel

- Alleles can interact in ways other than dominant/recessive
  - Incomplete dominance
  - Codominance
  - Multiple alleles

- Different genes can interact with one another in creating one phenotype
  - Epistasis
Incomplete Dominance

- The expression of a phenotype that is **intermediate** to those of the parents.
- An example is the inheritance of flower color in snapdragons:
  - $R^1R^1$ (red) x $R^2R^2$ (white) = $R^1R^2$ (pink)

- In humans: curly, wavy and straight hair
  - CC: Curly
  - Cc: Wavy
  - cc: straight
Codominance

- Full phenotypic expression of both alleles of a gene
- An example is the inheritance of the MN blood group in humans: (L is the gene for a glycoprotein found on the surface of red blood cells.)

<table>
<thead>
<tr>
<th>GENOTYPe</th>
<th>BLOOD TYPE (PHENOTYPe)</th>
</tr>
</thead>
<tbody>
<tr>
<td>$L^M L^M$</td>
<td>M</td>
</tr>
<tr>
<td>$L^M L^N$</td>
<td>MN</td>
</tr>
<tr>
<td>$L^N L^N$</td>
<td>N</td>
</tr>
</tbody>
</table>
Multiple Alleles

- Genes that have more than two alleles in the human population (Recall that each person can have just two alleles for any one gene.)

- An example if the inheritance of the ABO blood types in humans

- 3 alleles: $I^A$, $I^B$, i
  - (the $I^A$ and $I^B$ alleles are co-dominant)
<table>
<thead>
<tr>
<th>Genotypes</th>
<th>Phenotypes</th>
</tr>
</thead>
<tbody>
<tr>
<td>$I^A I^A$, $I^A i$</td>
<td>Type A</td>
</tr>
<tr>
<td>$I^B I^B$, $I^B i$</td>
<td>Type B</td>
</tr>
<tr>
<td>$I^A I^B$</td>
<td>Type AB</td>
</tr>
<tr>
<td>$ii$</td>
<td>Type O</td>
</tr>
</tbody>
</table>
Fig. 3-19, p. 63
Genes Can Interact in Complex Ways to Produce Phenotypes

- **Epistasis**
  - A form of gene interaction in which one gene masks or prevents expression of another gene.
  - An example is the Bombay blood type in humans.
    - Bombay gene, unrelated to the ABO blood type gene, when mutated, can block expression of blood types A and B.