LAB #6: Mendelian Genetics
Lab Manual Exercise 13
Genotype & Phenotype

• Genotype refers to particular genes an individual carries

• Phenotype refers to an individual’s observable traits

• Cannot always determine genotype by observing phenotype
Alleles

- Different forms of a gene
  - distinct in DNA sequences

- Originally arose by mutation
  - which changes DNA sequence

- Alleles come in dominant and recessive genotypes
  - Dominant allele masks a recessive allele of a gene
  - We denote dominant alleles with CAPITAL letters

Homologous Chromosomes
When talking about genotypes:

- **Homozygous**
  - two identical alleles at a locus
  - *AA*: homozygous dominant
  - *aa*: homozygous recessive

- **Heterozygous**
  - two different alleles at a locus - *Aa*
Mendel’s Theory of Segregation

• An individual inherits a particulate unit of information (allele) about a trait from each parent.
• During gamete formation, the two alleles of a gene in an organism segregate from each other. (MEIOSIS!)
• Genes are inherited as discrete “particles.”
• Blending does not occur, so that an allele that is present in the parental generation and is not evident in the F$_1$ generation can reappear in the F$_2$ generation.
Law of Independent Assortment

• Mendel concluded that the two “units” (alleles) for the first trait were to be assorted into gametes independently of the two “units” (alleles) for the other trait

• The two chromosome in each homologous pair are sorted into gametes during meiosis
Nucleus of a diploid (2n) Reproductive cell with two pairs of homologous chromosomes

Possible alignments of the two homologous chromosomes during metaphase I of meiosis

The resulting alignments at metaphase II:

Allelic combinations possible in gametes:

1/4 AB
1/4 ab
1/4 Ab
1/4 aB
The Garden Pea Plant

- Used to study genetics
- Self-pollinating
- True breeding
- Can be experimentally cross-pollinated
Mendel’s Experiments

Monohybrid Cross: A cross between two individuals in the same species in which **ONE** genetic trait is documented.

- **purple flowers**: AA
- **white flowers**: aa
TRUE-BREEDING PARENTS:

**purple flowers**: AA × **white Flowers**: aa

GAMETES:

AA → A
AA → A
aa → a
aa → a

F1 HYBRID OFFSPRING:

Aa
Monohybrid Punnett Square

True-breeding homozygous recessive parent plant

aa

True-breeding homozygous dominant parent plant

A

a

F1 PHENOTYPES

Aa

Aa

Aa

Aa

AA

Aa

Aa

Aa

Aa

Aa

Aa

Aa
An F1 plant self-fertilizes and produces gametes:

- Aa
- A
- a
- AA
- Aa
- aa

F2 PHENOTYPES:
- AA
- Aa
- Aa
- aa
Dihybrid Cross

Dihybrid Cross: A cross between two individuals in the same species in which **TWO** genetic traits are documented.

- **purple flowers**: AA
- **white flowers**: aa
- **tall**: BB
- **dwarf**: bb

What are parental genotypes?
TRUE-BREEDING PARENTS:

AABB

purple Flowers, Tall  x  white Flowers, dwarf

GAMETES:

AB

AB

ab

ab

AaBb

F1 HYBRID OFFSPRING:

What is the F1 generation phenotype?
**F₁ Generation:** Purple-flowered, tall

<table>
<thead>
<tr>
<th>Meiosis, gamete formation</th>
<th>Meiosis, gamete formation</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/4</td>
<td>1/4</td>
</tr>
<tr>
<td><strong>AB</strong></td>
<td><strong>Ab</strong></td>
</tr>
<tr>
<td><strong>aB</strong></td>
<td><strong>ab</strong></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>1/4</th>
<th>1/16</th>
<th>1/16</th>
<th>1/16</th>
<th>1/16</th>
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</thead>
<tbody>
<tr>
<td><strong>AB</strong></td>
<td><strong>AABB</strong></td>
<td><strong>AABb</strong></td>
<td><strong>AaBB</strong></td>
<td><strong>AaBb</strong></td>
</tr>
<tr>
<td>1/4</td>
<td>1/16</td>
<td>1/16</td>
<td>1/16</td>
<td>1/16</td>
</tr>
<tr>
<td><strong>Ab</strong></td>
<td><strong>AABB</strong></td>
<td><strong>AAbb</strong></td>
<td><strong>AaBb</strong></td>
<td><strong>Aabb</strong></td>
</tr>
<tr>
<td>1/4</td>
<td>1/16</td>
<td>1/16</td>
<td>1/16</td>
<td>1/16</td>
</tr>
<tr>
<td><strong>aB</strong></td>
<td><strong>AABB</strong></td>
<td><strong>AAbb</strong></td>
<td><strong>aaBB</strong></td>
<td><strong>aaBb</strong></td>
</tr>
<tr>
<td>1/4</td>
<td>1/16</td>
<td>1/16</td>
<td>1/16</td>
<td>1/16</td>
</tr>
<tr>
<td><strong>ab</strong></td>
<td><strong>AABB</strong></td>
<td><strong>AAbb</strong></td>
<td><strong>aaBb</strong></td>
<td><strong>aabb</strong></td>
</tr>
</tbody>
</table>

**Adding up the F₂ combinations possible:**

- 9/16 or 9 purple-flowered, tall
- 3/16 or 3 purple-flowered, dwarf
- 3/16 or 3 white-flowered, tall
- 1/16 or 1 white-flowered, dwarf
<table>
<thead>
<tr>
<th>Trait Studied</th>
<th>Dominant Form</th>
<th>Recessive Form</th>
<th>F&lt;sub&gt;2&lt;/sub&gt; Dominant-to-Recessive Ratio</th>
</tr>
</thead>
<tbody>
<tr>
<td>SEED SHAPE</td>
<td>5,474 round</td>
<td>1,850 wrinkled</td>
<td>2.96:1</td>
</tr>
<tr>
<td>SEED COLOR</td>
<td>6,022 yellow</td>
<td>2,001 green</td>
<td>3.01:1</td>
</tr>
<tr>
<td>POD SHAPE</td>
<td>882 inflated</td>
<td>299 wrinkled</td>
<td>2.95:1</td>
</tr>
<tr>
<td>POD COLOR</td>
<td>428 green</td>
<td>152 yellow</td>
<td>2.82:1</td>
</tr>
<tr>
<td>FLOWER COLOR</td>
<td>705 purple</td>
<td>224 white</td>
<td>3.15:1</td>
</tr>
<tr>
<td>FLOWER POSITION</td>
<td>651 long stem</td>
<td>207 at tip</td>
<td>3.14:1</td>
</tr>
<tr>
<td>STEM LENGTH</td>
<td>787 tall</td>
<td>277 dwarf</td>
<td>2.84:1</td>
</tr>
</tbody>
</table>
Monohybrid Cross
Exercise 13.1 B pg. 183
Monohybrid Cross
Lab handout exercise

- Tobacco Seedlings
  - Make individual counts of white and green phenotypes
  - DO NOT PULL PLANTS OUT OF PETRI DISH
  - Pool class data
  - Data analysis
- NOTE: Difference between corn and seedling counts = one piece of corn is THE population, while each Petri dish of seedlings is a population.
- TAKE HOME: each seed is an individual (whether its part of an ear of corn or laying in a Petri dish).
Dihybrid Cross

Exercise 13.2 B pg. 189
Observable human traits

Exercise 13.3, pg. 190

• There are many traits that follow the Mendelian pattern of inheritance.
• Ex: Earlobe Anatomy depends on a single gene with two alleles
  - Dominant allele specifies detached earlobes - E
  - Recessive allele specifies attached lobes - e

In today’s lab, we will examine the genotypes for several human traits.
We'll Use Statistics Today

- Scientists use statistical tests to examine the likelihood that their results are due to random events (vs. “real” results)

- For example, if you flip a coin 100 times, you probably won’t get a clean 50 heads, 50 tails. The same problem arises for genetic ratios (e.g., you might get 2.81:1.03 ratio, instead of 3:1 ratio)

- Chi-square ($X^2$) is one such statistical procedure that examines the difference between your actual data versus some prediction

- If you calculate a probability less than or equal to 5% ($p \leq 0.05$), then your results significantly differ from the predicted genetic ratio

- Don’t get bogged down in the math behind chi-squares (e.g., there are many electrical tools we use in our day-to-day life without knowing the actual electrical circuitry...but they still get the job done for us!)
Chi Squared Analysis
Lab and handout exercise

• Scientists use statistical tests to examine the likelihood that their results are due to random events (vs. “real” results)

• Otherwise stated: Chi-square ($\chi^2$) tests if an observed frequency distribution fits an expected distribution

$$X^2 = \sum \frac{(O - E)^2}{E}$$
Monohybrid Chi-square ($\chi^2$)

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Genotype</th>
<th>Observed (O)</th>
<th>Expected (E)</th>
<th>$(O-E)^2$</th>
<th>$(O-E)^2/E$</th>
</tr>
</thead>
<tbody>
<tr>
<td>Purple</td>
<td>P_</td>
<td>64</td>
<td>60.75</td>
<td>10.56</td>
<td>0.174</td>
</tr>
<tr>
<td>White</td>
<td>pp</td>
<td>17</td>
<td>20.25</td>
<td>10.56</td>
<td>0.522</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>81</td>
<td>81</td>
<td>0</td>
<td>6.96</td>
</tr>
</tbody>
</table>

Expected ratio in a monohybrid cross? 3:1

How do we calculate $E$?

\[
\begin{align*}
(3/4) \times 81 &= 60.75 \\
(1/4) \times 81 &= 20.25
\end{align*}
\]
Chi-square ($\chi^2$)

- Once you have calculated your chi-squared compare it to the table on page 186.

- If your chi-squared is GREATER than the .05 cutoff = chance alone cannot explain the deviation

- Your number is LESS than the .05 cutoff = variation is due to chance.
Monohybrid Chi-square ($\chi^2$)

- What is the expected ratio?
  - 9:3:3:1

- How do we calculate expected values?
  - \((9/16) \times \text{Total} = E\)
  - \((3/16) \times \text{Total} = E\)
  - \((3/16) \times \text{Total} = E\)
  - \((1/16) \times \text{Total} = E\)
TODAY’S PLAN

• Work in pairs, but complete the post-lab BY YOURSELF!

• Follow the lab book and do all of the exercises in order, except SKIP section 13.1.C1, 2, & 3 (all Fern exercises).

• Look at the tobacco seedlings & count number of green vs. albino. As soon as you have your tobacco data, give it to your TA. Do the $X^2$ test for these data... **Show all work!**

• Answer all the questions in your lab book.

• Post-lab
  - Have TA check off your lab book work
  - Turn in post-lab sheet with completed tables and problems

• When you’re all finished turn in the post-lab assignment.
Inheritance of X-Linked Genes

- X and Y chromosomes function as a homologous pair.
- X is chromosome on which disease-causing allele is found.
- No comparable allele is found on the Y chromosome.