Family resemblance can be striking!
Chapter 14.

Mendel & Genetics

2004-2005
He studied at the University of Vienna from 1851 to 1853 where he was influenced by a physicist who encouraged experimentation and the application of mathematics to science and a botanist who aroused Mendel’s interest in the causes of variation in plants. After the university, Mendel taught at the Brunn Modern School and lived in the local monastery.

The monks at this monastery had a long tradition of interest in the breeding of plants, including peas. Around 1857, Mendel began breeding garden peas to study inheritance.
Mendel’s work

- Bred pea plants
  - cross-pollinated true breeding parents (P)
  - raised seed & then observed traits (F₁)
  - allowed offspring to cross-pollinate & observed next generation (F₂)

P = parents
F = filial generation
Mendel collected data for 7 pea traits

<table>
<thead>
<tr>
<th>Character</th>
<th>Dominant Trait</th>
<th>Reccessive Trait</th>
<th>F2 Generation</th>
<th>Dominant/Recessive Ratio</th>
</tr>
</thead>
<tbody>
<tr>
<td>Flower color</td>
<td>Purple</td>
<td>White</td>
<td>185/229</td>
<td>3:1</td>
</tr>
<tr>
<td>Flower position</td>
<td>Axial</td>
<td>Terminal</td>
<td>461/207</td>
<td>3:1</td>
</tr>
<tr>
<td>Seed color</td>
<td>Yellow</td>
<td>Green</td>
<td>602/186</td>
<td>3:1</td>
</tr>
<tr>
<td>Seed shape</td>
<td>Round</td>
<td>Wisked</td>
<td>547/185</td>
<td>3:1</td>
</tr>
<tr>
<td>Pod shape</td>
<td>Indented</td>
<td>Conventional</td>
<td>882/284</td>
<td>3:1</td>
</tr>
<tr>
<td>Pod color</td>
<td>Green</td>
<td>Yellow</td>
<td>438/153</td>
<td>3:1</td>
</tr>
<tr>
<td>Stem length</td>
<td>Tall</td>
<td>Dwarf</td>
<td>187/177</td>
<td>3:1</td>
</tr>
</tbody>
</table>
Looking closer at Mendel’s work

In a typical breeding experiment, Mendel would cross-pollinate (hybridize) two contrasting, true-breeding pea varieties.

The true-breeding parents are the P generation and their hybrid offspring are the F₁ generation.

Mendel would then allow the F₁ hybrids to self-pollinate to produce an F₂ generation.
What did Mendel’s findings mean?

- Traits come in alternative versions
  - purple vs. white flower color
  - alleles
    - different alleles vary in the sequence of nucleotides at the specific locus of a gene

purple-flower allele & white-flower allele are 2 DNA variations at flower-color locus
Traits are inherited as discrete units

- For each character, an organism inherits 2 alleles, 1 from each parent
  - **diploid** organism inherits 1 set of chromosomes from each parent
    - homologous chromosomes
    - diploid = 2 sets of chromosomes
    - like having 2 editions of encyclopedia
      - Encyclopedia Britannica
      - Encyclopedia Americana

What's the advantage of being diploid?
What did Mendel’s findings mean?

- Some traits mask others
  - purple & white-flower colors are separate traits that do not blend
    - purple x white ≠ light purple
    - purple masked white
  - dominant allele
    - fully expressed
  - recessive allele
    - no noticeable effect
    - non-functional protein

AP Biology
Genotype vs. phenotype

- difference between how an organism “looks” & its genetics
  - description of an organism’s trait = phenotype
  - description of an organism’s genetic makeup = genotype

Explain Mendel’s results using...
dominant & recessive...
phenotype & genotype

AP Biology
Making crosses

- using representative letters
  - flower color alleles = P or p
  - true-breeding purple-flower peas = PP
  - true-breeding white-flower peas = pp

\[ PP \times pp \rightarrow Pp \]

All plants had purple flowers
Looking closer at Mendel’s work

**P**
true-breeding purple-flower peas

**X**
true-breeding white-flower peas

**F₁**
generation (hybrids)
100% purple-flower peas

self-pollinate

**F₂**
generation
75% purple-flower peas
25% white-flower peas

3:1
Punnett squares

Pp   x   Pp

male / sperm
P   p

PP  Pp  Pp  pp

Pp  Pp

female / eggs
p

% genotype  % phenotype
PP  25%  75%
Pp  50%  25%
PP  25%  25%

1:2:1  3:1

AP Biology

2004-2005
### Genotypes

- **Homozygous** = same alleles = PP, pp
- **Heterozygous** = different alleles = Pp

#### homozygous dominant

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>pp (homozygous)</td>
<td>Purple</td>
</tr>
<tr>
<td>Pp (heterozygous)</td>
<td>Purple</td>
</tr>
<tr>
<td>Pp (heterozygous)</td>
<td>Purple</td>
</tr>
</tbody>
</table>

#### homozygous recessive

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>pp (homozygous)</td>
<td>White</td>
</tr>
</tbody>
</table>

Ratio: 1:2:1

Ratio: 3:1

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2004-2005
**Phenotype vs. genotype**

- 2 organisms can have the same phenotype but have different genotypes

<p>| | |</p>
<table>
<thead>
<tr>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>PP</td>
<td>homozygous dominant</td>
</tr>
<tr>
<td>Pp</td>
<td>heterozygous</td>
</tr>
</tbody>
</table>
Mendel chose peas wisely

- Pea plants are good for genetic research
  - available in many varieties with distinct heritable features with different variations
    - flower color, seed color, seed shape, etc.
  - Mendel had strict control over which plants mated with which
    - each pea plant has male & female structures
    - pea plants can self-fertilize
    - Mendel could also cross-pollinate plants: moving pollen from one plant to another
Mendel chose peas luckily

- Pea plants are good for genetic research
  - relatively simple genetically
    - most characters are controlled by a single gene
    - each gene has only 2 alleles, one of which is completely dominant to the other
Mendel’s laws of heredity (#1)

- Law of segregation
  - when gametes (eggs & sperm) are produced during meiosis, homologous chromosomes separate from each other
  - each allele for a trait segregates (is packaged) into a separate gamete

```
PP  P  p  P  p
PP  P  p
PP  P
PP  P
PP  P
PP  P
PP  P
PP  P
PP  P
PP  P
PP  P
PP  P
```

AP Biology
2004-2005
Law of Segregation

- What meiotic event creates the law of segregation

and Mendel didn’t even know DNA or genes existed!

AP Biology
Dominant phenotypes

- It is not possible to predict the genotype of an organism with a dominant phenotype

So how do you figure out the phenotype?

PP?
Pp?
Test cross

- Cross-breed the dominant phenotype — unknown genotype — with a homozygous recessive (pp) to determine the identity of the unknown allele.

\[ \text{X} \]

AP Biology
2004-2005
Test cross

100% 1:1
Monohybrid cross

- Some of Mendel’s experiments followed the inheritance of single characters
  - flower color
  - seed color
  - monohybrid crosses
Dihybrid cross

- Some of Mendel’s experiments followed the inheritance of 2 different characters
  - seed color & seed shape
  - dihybrid crosses

- Helped him understand other genetic mechanisms ("rules")
Inheritance of seed shape

- Seed shape
  - wrinkled peas
  - smooth peas
Wrinkled seeds in pea plants with two copies of the recessive allele are due to the accumulation of monosaccharides and excess water in seeds because of the lack of a key enzyme. The seeds wrinkle when they dry.

Both homozygous dominants and heterozygotes produce enough enzyme to convert all the monosaccharides into starch and form smooth seeds when they dry.
Making gametes

- How are the alleles handed out?
  - together or separately?

YyRr → YR, yr
YyRr → YR, Yr, yR, yr

Which system explains the data?
**Dihybrid cross**

<table>
<thead>
<tr>
<th></th>
<th>YR</th>
<th>Yr</th>
<th>yR</th>
<th>yr</th>
</tr>
</thead>
<tbody>
<tr>
<td>YR</td>
<td>YYRR</td>
<td>YYRr</td>
<td>YyRR</td>
<td>YyRr</td>
</tr>
<tr>
<td>Yr</td>
<td>YYRr</td>
<td>YYrr</td>
<td>YyRr</td>
<td>Yyrr</td>
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<tr>
<td>yR</td>
<td>YyRr</td>
<td>Yyrr</td>
<td>yyRr</td>
<td>yyrr</td>
</tr>
<tr>
<td>yr</td>
<td>YyRr</td>
<td>Yyrr</td>
<td>yyRr</td>
<td>yyrr</td>
</tr>
</tbody>
</table>

- **9/16** yellow round
- **3/16** green round
- **3/16** yellow wrinkled
- **1/16** green wrinkled
Mendel’s laws of heredity (#2)

- Law of independent assortment
  - each pair of alleles — for each trait — segregates into gametes independently
    = independent assortment
  - 4 classes of gametes — YR, Yr, yR, yr — are produced in equal amounts
Law of Independent Assortment

- What meiotic event creates the law of independent assortment?

and Mendel didn’t even know DNA or genes existed!
Review: Mendel’s laws of heredity

- Law of segregation
  - monohybrid cross = single trait
  - each allele for a trait segregates (is packaged) into separate gametes
    - established by Meiosis 1

- Law of independent assortment
  - dihybrid (or more) cross
    - 2 or more traits
  - each pair of alleles for each trait segregates into gametes independently
    - established by Meiosis 1
Some interesting historical facts

- While Mendel was acknowledged by his contemporaries as an outstanding plant breeder, his revolutionary genetics work was overlooked for 34 years.

- Mendel published “Experiments on Plant Hybrids” in 1865. In 1900, 16 years after Mendel’s death, a number of scientists independently rediscovered his work.
Some interesting historical facts

- Charles Darwin, a contemporary of Mendel, proposed that evolution by natural selection was dependent on variation in the population.
- But Darwin was unable to propose a mechanism for how this variation was transmitted.
- The key was Mendel’s work, and nearly a century after Mendel published his findings, historians found a copy of Mendel’s paper in Darwin’s study. He presumably never read it.