Charles Darwin (1809-1882)

• Developed the theory of Evolution by Natural Selection
• Published *The Origin of Species by Means of Natural Selection, or The Preservation of Favored Races in the Struggle for Life* (1859).
  • Variation exists and is heritable
  • More organisms are born than can possibly survive on the available supply of food
  • Organisms with variations best suited to the environment survive more often
  • New “races” (species) are occasionally formed.
• Acceptance of Darwin.
  • The theory of evolution was readily accepted. Buffon, Lamark, Erasmus Darwin all produced prior theories of evolution
  • Evolution by natural selection elicited a sharp negative reaction, largely on non-scientific grounds.
• Darwin also struggled to developed a “provisional” theory of inheritance called **pangenesis** in 1868
  • gemmules (particles) were “thrown off” by each part of an organism, and collected in the reproductive apparatus.
  • gemmules were the hereditary agents.
  • Fleeming Jenkin in 1867: blending reduces variability.
• Doubts by Frances Galton and Thomas Huxley.
  • Inherited characters are discontinuous
• By the late 1800s, Darwin’s theory of evolution by natural selection had become something of a footnote--a mechanism that might account for some aspects of evolution.
Gregor Mendel (1822-1884)

- Performed a series of plant breeding experiments over a 10 year period
- Formulated our most fundamental laws of heredity
- Reported them to a scientific society in 1865
- Published his results in 1866
- The results were circulated by the society and by Mendel
- Mendel’s work went largely unrecognized for many years

- At the time, the cell theory had been generally established
- Aspects of fertilization were known. (Plants and animals.)
- Blending theory of inheritance was popular (Aristotle)
  - Maternal and paternal characters were “mixed”
  - Deficiencies were recognized, but nothing better.
- Rediscovery of Mendel:
  - 1900, de Vries, Correns and von Tschermak independently discovered “Mendel’s” laws.
  - Also, Mendel’s papers were “discovered”.

- Why did it take so long for Mendel’s results to be recognized?
  1) Biologists generally thought about continuous traits
  2) They were unprepared for Mendel's mathematical approach
  3) Mendel was using unfamiliar terminology to describe things like Traits, Dominant, Recessive, and so forth
  4) Mendel was not known by biologists or plant breeders.
Law of Segregation (or Mendel’s First Law).

- Hereditary characters (such as round or wrinkled seeds) are determined by particulate factors (now called alleles).

- These factors occur in pairs (one from each parent)

- During the formation of the gametes these factors are segregated so that only one of the pair is transmitted by a gamete.

- When the male and female gametes fuse to form the zygote, the double number is restored.

Mendel also discovered the concept of Dominance

Developed the notation of a capital letter for a dominant allele and lower case for a recessive allele.
ALTERNATE FORMS

Seeds
1. Round
   - Wrinkled
2. Yellow cotyledons
   - Green cotyledons
3. White coat (white flowers)
   - Gray coat (violet flowers)

Pods
4. Full
   - Constricted
5. Yellow
   - Green

Stem
6. Axial pods and flowers along stem
   - Terminal pods and flowers on top of stem
7. Tall (6–7 ft)
   - Dwarf (¾–1 ft)
$P_1$  

Tall  
Dwarf  

$F_1$  

X Self  
Tall  

$F_2$  

Tall  
Dwarf  

3 : 1
Law of Independence (or Mendel’s Second Law).

- Members of one pair of alleles segregate independently of other pairs. (Or genes segregate independently.)
- This independence means that the frequencies of one trait arise independently from the frequencies of another.
- Or, if we look at two characteristics simultaneously, the probabilities are multiplied. This 2nd law has not held up! Mendel was lucky (or very careful) with the traits he picked.
- We now know that many traits are not independent because they are located near each other on the same chromosome.
- That is, some genes are linked.

Why didn’t Mendel discover linkage?
Mendel’s great contributions:

1. He replaced a theory of blending inheritance with a particulate mechanism.

2. Organisms have two alleles for a trait—one from each parent.

3. He verified his ideas by careful experimentation.

---

Table 1: Relationship between modern genetic terminology and character pairs used by Mendel

<table>
<thead>
<tr>
<th>Character pair used by Mendel</th>
<th>Alleles in modern terminology</th>
<th>Located in chromosome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Seed colour, yellow–green</td>
<td>$I-i$</td>
<td>1</td>
</tr>
<tr>
<td>Seed coat and flowers, coloured–white</td>
<td>$A-a$</td>
<td>1</td>
</tr>
<tr>
<td>Mature pods, smooth expanded–wrinkled indented</td>
<td>$V-v$</td>
<td>4</td>
</tr>
<tr>
<td>Inflorescences, from leaf axils–umbellate in top of plant</td>
<td>$Fa-fa$</td>
<td>4</td>
</tr>
<tr>
<td>Plant height, &gt;1m–around 0.5 m</td>
<td>$Le-le$</td>
<td>4</td>
</tr>
<tr>
<td>Unripe pods, green–yellow</td>
<td>$Gp-gp$</td>
<td>5</td>
</tr>
<tr>
<td>Mature seeds, smooth–wrinkled</td>
<td>$R-r$</td>
<td>7</td>
</tr>
</tbody>
</table>

Here is what he observed:
round v. wrinkled
yellow v. green

Pure-bred plants were crossed (P generation):
round yellow  x  wrinkled green.

All offspring of the P generation were round and yellow.

Next the F1 plants were allowed to self-fertilize and produce a second generation of plants (F2).

Result:
- 9/16: round and yellow
- 3/16: round and green
- 3/16: wrinkled and yellow
- 1/16: wrinkled and green
  (a 9:3:3:1 ratio)
**P₁**
Round, Yellow
\((RRYY)\)
Wrinkled, Green
\((rryy)\)

**F₁**
X Self
Round, Yellow
\((RrYy)\)

**F₂**
Round, Yellow
\((315)\)
\((RRYY; RRYy; RrYY; RrYy)\)
Round, Green
\((108)\)
\((RRyy; Rryy)\)
Wrinkled, Yellow
\((101)\)
\((rrYY; rrYy)\)
Wrinkled, Green
\((32)\)
\((rryy)\)
Transmission probabilities.

Autosomal inheritance

Maternal factors

- $F_1$
- $F_2$

\[
\begin{align*}
\Pr(F_1) &= 0.5 \\
\Pr(F_2) &= 0.5
\end{align*}
\]

Paternal factors

- $M_1$
- $M_2$

\[
\begin{align*}
\Pr(M_1) &= 0.5 \\
\Pr(M_2) &= 0.5
\end{align*}
\]

(Assuming segregation)

\[
\begin{align*}
\Pr(F_1 M_1) &= \Pr(F_1) \times \Pr(M_1) = 0.5 \times 0.5 = 0.25 \\
\Pr(F_1 M_2) &= \Pr(F_1) \times \Pr(M_2) = 0.5 \times 0.5 = 0.25 \\
\Pr(F_2 M_1) &= \Pr(F_2) \times \Pr(M_1) = 0.5 \times 0.5 = 0.25 \\
\Pr(F_2 M_2) &= \Pr(F_2) \times \Pr(M_2) = 0.5 \times 0.5 = 0.25
\end{align*}
\]

(Assuming independent pairing)
• Some terms
  • *Genotype*: the particular genetic makeup. The two alleles at a particular gene.
  • *Phenotype*: The observed characteristics expressed by a genotype.
• Examples: some *Mendelian traits* in humans (over 4000 known)
  • PTC (phenylthiocarbamide).
    Alleles: $T$  $t$

    | Genotype | Phenotype |
    |----------|-----------|
    | *homozygous* $TT$ | Taster |
    | *heterozygous* $Tt$ | Taster |
    | *homozygous* $tt$ | Non-taster |

  • Tongue-rolling
    Alleles: $R$  $r$

    | Genotype | Phenotype |
    |----------|-----------|
    | *homozygous* $RR$ | Can roll |
    | *heterozygous* $Rr$ | Can roll |
    | *homozygous* $rr$ | Cannot roll |

• These traits show *Complete dominance*
• Rhesus blood type
  Alleles:  \( D \quad d \)

<table>
<thead>
<tr>
<th></th>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>homozygous</td>
<td>( DD )</td>
<td>Rh positive</td>
</tr>
<tr>
<td>heterozygous</td>
<td>( Dd )</td>
<td>Rh positive</td>
</tr>
<tr>
<td>homozygous</td>
<td>( dd )</td>
<td>Rh negative</td>
</tr>
</tbody>
</table>

• Alleles can also exhibit partial dominance (also, incomplete dominance or intermediate dominance)
  \( HH \)     phenotype 1
  \( Hh \)     intermediate phenotype
  \( hh \)     phenotype 2

• A trait can have more than 2 types of alleles segregating in a population. But any individual only has only one pair of alleles.
A trait can have more than 2 types of alleles

E.g. ABO blood group in humans

<table>
<thead>
<tr>
<th>Alleles:</th>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>homozygous:</td>
<td>AA</td>
<td>type A</td>
</tr>
<tr>
<td></td>
<td>BB</td>
<td>type B</td>
</tr>
<tr>
<td></td>
<td>OO</td>
<td>type O</td>
</tr>
<tr>
<td>heterozygous</td>
<td>AB</td>
<td>type AB</td>
</tr>
<tr>
<td></td>
<td>AO</td>
<td>type A</td>
</tr>
<tr>
<td></td>
<td>BO</td>
<td>type B</td>
</tr>
</tbody>
</table>

The $A$ and $B$ alleles are said to be codominant because both alleles can be detected in an $AB$ individual.

Most traits are codominant at very low levels (protein, molecular).