Bateson and Punnett (1906)

1. purple x red flowers → purple x purple → 3 purple, 1 red
2. long x round pollen → long x long → 3 long, 1 round

<table>
<thead>
<tr>
<th>F₂ Phenotypes</th>
<th>Genotype</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>purple, long</td>
<td>P-L-</td>
<td>284</td>
<td>215</td>
</tr>
<tr>
<td>purple, round</td>
<td>P-Ll</td>
<td>21</td>
<td>71</td>
</tr>
<tr>
<td>red, long</td>
<td>ppL-</td>
<td>21</td>
<td>71</td>
</tr>
<tr>
<td>red, round</td>
<td>PpLl</td>
<td>55</td>
<td>24</td>
</tr>
</tbody>
</table>

T.H. Morgan (1911)

red x purple eyes → red x red → 3 red, 1 purple
normal x vestigial wings → normal x normal → 3 normal, 1 vestigial

<table>
<thead>
<tr>
<th>F₂ Phenotype</th>
<th>Genotype</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>wild-type</td>
<td>pr-vg^-</td>
<td>1339</td>
<td>710</td>
</tr>
<tr>
<td>purple</td>
<td>prprvg</td>
<td>154</td>
<td>710</td>
</tr>
<tr>
<td>vestigial</td>
<td>pr-vgvg</td>
<td>151</td>
<td>710</td>
</tr>
<tr>
<td>purple,vestigial</td>
<td>prprvgvg</td>
<td>1195</td>
<td>710</td>
</tr>
</tbody>
</table>

T.H. Morgan (1911) “coupling”

red x purple eyes → red x red → 3 red, 1 purple
normal x vestigial wings → normal x normal → 3 normal, 1 vestigial

<table>
<thead>
<tr>
<th>F₂ Phenotype</th>
<th>Genotype</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>wild-type</td>
<td>pr-vg^-</td>
<td>1339</td>
<td>710</td>
</tr>
<tr>
<td>purple</td>
<td>prprvg</td>
<td>154</td>
<td>710</td>
</tr>
<tr>
<td>vestigial</td>
<td>pr-vgvg</td>
<td>151</td>
<td>710</td>
</tr>
<tr>
<td>purple,vestigial</td>
<td>prprvgvg</td>
<td>1195</td>
<td>710</td>
</tr>
</tbody>
</table>
T.H. Morgan (1911) “repulsion”
red x purple eyes → red x red → 3 red, 1 purple
normal x vestigial wings → normal x normal → 3
normal, 1 vestigial

pr+pr+vg x prprvg vg+ → pr+pr+vg x prprvg vg+ →

<table>
<thead>
<tr>
<th>F2 Phenotype</th>
<th>Genotype</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>wild-type</td>
<td>pr+vg-</td>
<td>157</td>
<td>584</td>
</tr>
<tr>
<td>purple</td>
<td>prprvg-</td>
<td>1067</td>
<td>584</td>
</tr>
<tr>
<td>vestigial</td>
<td>pr+-vgw</td>
<td>965</td>
<td>584</td>
</tr>
<tr>
<td>purple,vestigial</td>
<td>prprvgvg</td>
<td>146</td>
<td>584</td>
</tr>
</tbody>
</table>

Janssens’ Chiasmatype Theory (1909)

T.H. Morgan (1911) “coupling”
red x purple eyes → red x red → 3 red, 1 purple
normal x vestigial wings → normal x normal → 3
normal, 1 vestigial

pr+pr+vg x prprvg vg+ x prprvg vg+ →

<table>
<thead>
<tr>
<th>F2 Phenotype</th>
<th>Genotype</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>wild-type</td>
<td>pr+vg-</td>
<td>1339</td>
<td>710</td>
</tr>
<tr>
<td>purple</td>
<td>prprvg-</td>
<td>154</td>
<td>710</td>
</tr>
<tr>
<td>vestigial</td>
<td>pr+-vgw</td>
<td>151</td>
<td>710</td>
</tr>
<tr>
<td>purple,vestigial</td>
<td>prprvgvg</td>
<td>1195</td>
<td>710</td>
</tr>
</tbody>
</table>
T.H. Morgan (1911) “repulsion”

red x purple eyes → red x red → 3 red, 1 purple

normal x vestigial wings → normal x normal → 3 normal, 1 vestigial

pr+pr+vgvg x prprvgvg→ pr+pr+vgvg x prprvgvg→

<table>
<thead>
<tr>
<th>F2 Phenotype</th>
<th>Genotype</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>wild-type</td>
<td>pr−vg−</td>
<td>157</td>
<td>584</td>
</tr>
<tr>
<td>purple</td>
<td>prprvg−</td>
<td>1067</td>
<td>584</td>
</tr>
<tr>
<td>vestigial</td>
<td>pr−vgvg</td>
<td>965</td>
<td>584</td>
</tr>
<tr>
<td>purple, vestigial</td>
<td>prprvgvg</td>
<td>146</td>
<td>584</td>
</tr>
</tbody>
</table>

Interchromosomal meiotic recombination

Intrachromosomal meiotic recombination
Intra-chromosomal meiotic recombination

Linkage: Two genes on a single pair of homologs; exchange occurs between two non-sister chromatids

Stern (1936)

Mitotic recombination

Stern (1936)

mutation? No

Stern (1936)

mutation? No
Frequent revertants in ichthyosis with confetti (IWC).

(A and B) The backs of an 18-year-old female subject (103-1) and 42-year-old male (104-1) show background redness and scaling with hundreds of white, normal-appearing "confetti" spots.

(C) Histology of normal human skin showing basal layer (labeled B), stratum spinosum (S), granular layer (G), and stratum corneum (SC). (D) Affected skin shows loss of differentiation of all layers above the basal layer and hypercellularity with increased epidermal thickness. There is no granular layer, and marked perinuclear vacuolization in the suprabasal epidermis and retention of cell nuclei in the stratum corneum are seen. (E) Revertant skin shows normalization of epidermal thickness and architecture, with normal granular layer, normal spinous layer, and stratum corneum. Scale bars in C-E, 50 µm. (F) High-power (HP) view of spinous layer in normal epidermis with intercellular spines visible, overlying granular layer with purple keratohyalin granules and basket weave stratum corneum. (G) HP view of affected skin shows perinuclear vacuolization (black arrows), lack of keratohyalin granules, and retained nuclei (white arrows) in the stratum corneum. (H) HP view of revertant skin shows normal spinous layer with intercellular spines, granular layer with purple keratohyalin granules in keratinocytes, and basket weave stratum corneum. Scale bars in F-H, 25 µm.

S phase

\[ \text{S phase} \]

\[ \text{mitotic recombination} \]

Interchromosomal meiotic recombination

\[ \text{Interchromosomal meiotic recombination} \]

\[ P = R \]
Intrachromosomal meiotic recombination

P > R

T.H. Morgan (1911) “coupling”

red x purple eyes → red x red → 3 red, 1 purple

normal x vestigial wings → normal x normal → 3 normal, 1 vestigial

pr\(^+\)pr\(^+\)vg\(^+\)vg\(^+\) x prprvgvg → pr\(^+\)pr\(^+\)vg\(^+\)vg x prprvgvg →

<table>
<thead>
<tr>
<th>F(^2) Phenotype</th>
<th>Genotype</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>wild-type</td>
<td>pr(^+)vg(^-)</td>
<td>1339</td>
<td>710</td>
</tr>
<tr>
<td>purple</td>
<td>prprvg(^-)</td>
<td>154</td>
<td>710</td>
</tr>
<tr>
<td>vestigial</td>
<td>pr(^-)vg(^+)</td>
<td>151</td>
<td>710</td>
</tr>
<tr>
<td>purple,vestigial</td>
<td>prprvgvg</td>
<td>1195</td>
<td>710</td>
</tr>
</tbody>
</table>

T.H. Morgan (1911) “repulsion”

red x purple eyes → red x red → 3 red, 1 purple

normal x vestigial wings → normal x normal → 3 normal, 1 vestigial

pr\(^+\)pr\(^+\)vg\(^+\)vg\(^+\) x prprvgvg* → pr\(^+\)pr\(^+\)vg\(^+\)vg \* prprvgvg →

<table>
<thead>
<tr>
<th>F(^2) Phenotype</th>
<th>Genotype</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>wild-type</td>
<td>pr(^-)vg(^-)</td>
<td>157</td>
<td>584</td>
</tr>
<tr>
<td>purple</td>
<td>prprvg(^-)</td>
<td>1067</td>
<td>584</td>
</tr>
<tr>
<td>vestigial</td>
<td>pr(^+)vg(^+)</td>
<td>965</td>
<td>584</td>
</tr>
<tr>
<td>purple,vestigial</td>
<td>prprvgvg</td>
<td>146</td>
<td>584</td>
</tr>
</tbody>
</table>

Three-Point Testcross

• A technique for establishing gene linkage and for determining map positions of genes

• Sturtevant (1913)

AaBbCc x aabbcc

\[ \frac{1}{8} \text{ AaBbCc} \]

\[ \frac{1}{8} \text{ AaBbcc} \]

\[ \frac{1}{8} \text{ AabbCc} \]

\[ \frac{1}{8} \text{ aabBCc} \]

\[ \frac{1}{8} \text{ Aabbcc} \]

\[ \frac{1}{8} \text{ aaBbcc} \]

\[ \frac{1}{8} \text{ aabbCc} \]

\[ \frac{1}{8} \text{ aabbcc} \]
Interchromosomal meiotic recombination

\[ P = R \]

Intrachromosomal meiotic recombination

\[ P > R \]
genetic map unit
(aka centiMorgan)

- the distance between gene pairs for which 1 product of meiosis out of 100 is recombinant, i.e.

1mu/cM = 1% recombination

Analysis

1. Scute and echinus are 5.5mu apart on a chromosome (56/1008 = 5.5%)

2. Vestigial must be located on a different chromosome

<table>
<thead>
<tr>
<th>Phenotypes</th>
<th>Phenotypes</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>scute, echinus, crossveinless</td>
<td>scute, echinus, aveless</td>
<td>417</td>
<td>159</td>
</tr>
<tr>
<td>wild-type</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>scute</td>
<td>scute</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>echinus, crossveinless</td>
<td>echinus, aveless</td>
<td>54</td>
<td>122</td>
</tr>
<tr>
<td>scute, echinus</td>
<td>scute</td>
<td>echinus</td>
<td>+</td>
</tr>
<tr>
<td>crossveinless</td>
<td>+</td>
<td>aveless</td>
<td>+</td>
</tr>
<tr>
<td>scute, crossveinless</td>
<td>scute</td>
<td>+</td>
<td>aveless</td>
</tr>
<tr>
<td>echinus</td>
<td>+</td>
<td>echinus</td>
<td>+</td>
</tr>
</tbody>
</table>

Analysis

1. Scute and echinus are 5.5mu apart on a chromosome (56/1008 = 5.5%)

2. Echinus and crossveinless are 8.2mu apart on a chromosome (81/982 = 8.2%)

3. Scute and crossveinless are 13.7mu apart on a chromosome (135/982 = 13.7%)

4. Scute, echinus and crossveinless are located on the same chromosome

<table>
<thead>
<tr>
<th>Phenotypes</th>
<th>Phenotypes</th>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>scute, echinus, crossveinless</td>
<td>scute, echinus, aveless</td>
<td>417</td>
<td>159</td>
</tr>
<tr>
<td>wild-type</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>scute</td>
<td>scute</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>echinus, crossveinless</td>
<td>echinus, aveless</td>
<td>54</td>
<td>122</td>
</tr>
<tr>
<td>scute, echinus</td>
<td>scute</td>
<td>echinus</td>
<td>+</td>
</tr>
<tr>
<td>crossveinless</td>
<td>+</td>
<td>aveless</td>
<td>+</td>
</tr>
<tr>
<td>scute, crossveinless</td>
<td>scute</td>
<td>+</td>
<td>aveless</td>
</tr>
<tr>
<td>echinus</td>
<td>+</td>
<td>echinus</td>
<td>+</td>
</tr>
</tbody>
</table>

Analysis

1. Scute and echinus are 5.5mu apart on a chromosome (56/1008 = 5.5%)

2. Echinus and crossveinless are 8.2mu apart on a chromosome (81/982 = 8.2%)

3. Scute and crossveinless are 13.7mu apart on a chromosome (135/982 = 13.7%)

4. Scute, echinus and crossveinless are located on the same chromosome
Analysis

1. Crossveinless and cut are 6.4µm apart on a chromosome (93/1448 = 6.4%)
2. Cut and vermilion are 13.2µm apart on a chromosome (191/1448 = 13.2%)
3. Crossveinless and vermilion are 18.5µm apart on a chromosome (268/1448 = 18.5%)
4. Crossveinless, cut and vermilion are located on the same chromosome
Analysis

1. Crossveinless and cut are 6.4μm apart on a chromosome (93/1448 = 6.4%)
2. Cut and vermilion are 13.2μm apart on a chromosome (191/1448 = 13.2%)
3. Crossveinless and vermilion are 19.6μm apart on a chromosome (268 + 8 + 8 = 284; 284/1448 = 19.6%)
4. Crossveinless, cut and vermilion are located on the same chromosome

Interference

Interference \( I = 1 - C \) (coefficient of coincidence)

where,

\[ C = \frac{\text{observed \# double crossovers}}{\text{expected \# double crossovers}} \]

e.g.,

\[
\begin{align*}
\text{cv} & \quad \text{ct} \\
.064 & \quad .132 \\
\end{align*}
\]

therefore,

\[
I = 1 - \frac{8}{(.064 \times .132 \times 1448)} \\
I = 1 - (8/12) \\
I = +1/3
\]
Analysis

1. Scute and echinus are 5.5mu apart on a chromosome (56/1008 = 5.5%)  
2. Vestigial must be located on a different chromosome
Analysis

1. Scute and echinus are 5.5μm apart on a chromosome (56/1008 = 5.5%)

2. Vestigial must be located on a different chromosome (or at least 50μm apart on the same chromosome)

Mendel worked with three genes in chromosome 4, two genes in chromosome 1, and one gene in each of chromosome 5 and 7. It seems at first glance that, out of the 21 dihybrid combinations Mendel theoretically could have studied, no less than four (that is, a–i, v–fa, v–le, fa–le) ought to have resulted in linkages. As found, however, in hundreds of crosses and shown by the genetic map of the pea, a and i in chromosome 1 are so distantly located on the chromosome that no linkage is normally detected. The same is true for v or le and fa on chromosome 4. This leaves v–le, which ought to have shown linkage. Mendel, however, never did this dihybrid cross.

Why didn't Mendel detect sex linkage in peas?

Janssens' Chiasmatype Theory (1909)

Creighton and McClintock (1931)
Chromosomal knob positions in *Z. mays* ssp. *parviglumis* and ssp. *mexicana* (KATO 1976). The area of the circle is proportional to the size and frequency of the knob (knob index). The black internal knobs are unique to populations of *Z. mays* ssp. *parviglumis*, *mexicana*, and/or *mays*, while the gray terminal (telomeric) knobs are found in all Zea species. Ab10's chromosomal knob is larger than representation in this figure would allow.

---

**Creighton and McClintock (1931)**

**Figure 5.5**

**Table E2**

<table>
<thead>
<tr>
<th>Phenotype of <em>F1</em> Normal</th>
<th>Number of Kernels Analyzed</th>
<th>Cytological Appearance of Chromosome 9 in <em>F1</em> Offspring*</th>
<th>Did a Crossover Occur During Gamete Formation in Parent A?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Colored/waxy</td>
<td>3</td>
<td>Knobbed/translocation</td>
<td>Normal</td>
</tr>
<tr>
<td>Colorless/starchy</td>
<td>11</td>
<td>Knobbed/normal</td>
<td>Normal</td>
</tr>
<tr>
<td>Colorless/waxy</td>
<td>4</td>
<td>Knobless/normal</td>
<td>Normal</td>
</tr>
<tr>
<td>Colorless/starchy</td>
<td>2</td>
<td>Knobless/normal</td>
<td>Normal</td>
</tr>
<tr>
<td>Colorless/waxy</td>
<td>5</td>
<td>Knobbed/normal</td>
<td>Normal</td>
</tr>
<tr>
<td>Total</td>
<td>25</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

*In this table, the chromosome on the left was inherited from parent A, and the blue chromosome on the right was inherited from parent B.*

---

**Copy Choice Model**

**Breakage and Reunion Model**
Copy Choice Model

Breakage and Reunion Model

Meselson and Weigle (1961)

\[ ^{13}\text{C}^{15}\text{N} \rightarrow ^{12}\text{C}^{14}\text{N} \]

\[ h^+ c \times h^+ c^+ \rightarrow \rightarrow \rightarrow \rightarrow \rightarrow \rightarrow \rightarrow \rightarrow \rightarrow h^+ c \]

Results: recombinants were 97-99% “heavy”

“linkage groups”
FIGURE 5.13

- Formation of an ordered tube from a M. jannascha

- Membrane aggregate (I2)

- Membrane aggregate (II)

- Membrane aggregate (III)

- Membrane aggregate (IV)

- Membrane aggregate (V)

- Membrane aggregate (VI)

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>AAAaaaa</td>
<td>126</td>
</tr>
<tr>
<td>aaaaAAAA</td>
<td>132</td>
</tr>
<tr>
<td>AAaaaAAA</td>
<td>9</td>
</tr>
<tr>
<td>aaAAAAAA</td>
<td>11</td>
</tr>
<tr>
<td>AAaaaaAA</td>
<td>10</td>
</tr>
<tr>
<td>aaAAAAAaa</td>
<td>12</td>
</tr>
<tr>
<td></td>
<td>300</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>AAAaaaa</td>
<td>126</td>
</tr>
<tr>
<td>aaaaAAAA</td>
<td>132</td>
</tr>
<tr>
<td>AAaaaAAA</td>
<td>9</td>
</tr>
<tr>
<td>aaAAAAAA</td>
<td>11</td>
</tr>
<tr>
<td>AAaaaaAA</td>
<td>10</td>
</tr>
<tr>
<td>aaAAAAAaa</td>
<td>12</td>
</tr>
<tr>
<td></td>
<td>300</td>
</tr>
</tbody>
</table>

FDS
AAAaaaa  126  FDS
aaaaAAA  132  FDS
AAaaAaa  9  SDS
aaAAaaA  11  SDS
AAaaaaA  10  SDS
aaAAAAa  12  SDS
300

\[
\frac{42}{300} = 14\% = 14\text{mu}
\]

**genetic map unit** (aka centiMorgan)

- the distance between gene pairs for which 1 product of meiosis out of 100 is recombinant, i.e.

\[
1\text{mu} = 1\% \text{ recombination}
\]
42/300 = 14%
   = 14mu x ½
   = 7mu

Map distance =
100 (1/2) (#SDS asci/#total asci)

With unlinked loci,
PD = NPD and T may be large or small
With linked loci,

PD >> NPD unless the two genes are very far apart (T then would be very large, about 4X PD or NPD)

\[ PD = 24 \]
\[ NPD = 3 \]
\[ T = 27 \]
Genes must be linked which would account for the small NPD (recall DC produces them with linked genes). With unlinked genes PD=NPD (IA). 

Map distance = \(50 \times \frac{T + 6NPD}{\text{Total asci}}\)

= \(50 \times \frac{27 + 6(3)}{54}\)

= 41.67 \text{mu}

PD = 24
NPD = 3
T = 27

PD = 9
NPD = 11
T = 2

PD = 9
NPD = 11
T = 2
PD = 9
NPD = 11
T = 2

Genes must be unlinked as PD = NPD and T is small. For linked genes to have PD = NPD, T must be very large (i.e. the genes are far apart and numerous SC and DC produce numerous NPD and T). In this instance though T is small thus the genes must be unlinked.