Population Genetics
Introduction
Bio 36404
Branches of Genetics

- Classical=Transmission Genetics
- Cytological Genetics
- Molecular Genetics
- Population Genetics
Terms

• Population Genetics - study of genetic variation in natural populations.

• Population - group of individuals of the same species living in the same place and interbreeding.

• Population Size (N) - number of individuals in the population.

• Gene Pool - alleles carried by all individuals in a population.
Butterfly Population

N=10

Phenotype Frequency = # phenotype/N

White Frequency = #
8/10 = 0.8

Orange Frequency = #
2/10 = 0.2
Butterfly Population

White Dominant

AA - white
Aa - white
aa - orange
Genotype Frequency

Proportion of each genotype

\[
P \text{ freq. } AA = \frac{3}{10} = 0.3
\]

\[
H \text{ freq. } Aa = \frac{5}{10} = 0.5
\]

\[
Q \text{ freq. } aa = \frac{2}{10} = 0.2
\]

\[
P + H + Q = 1
\]
Allele Frequency

p = frequency of A allele
q = frequency of a allele

p + q = 1

Proportion of each allele

p = 11/20 = 0.55

q = 9/20 = 0.45

p + q = 1
Allele Frequency

alternate calculation

\[ p = P + \frac{1}{2} H \]
\[ q = Q + \frac{1}{2} H \]

\[ P = 0.3 \]
\[ H = 0.5 \]
\[ Q = 0.2 \]

\[ p = 0.3 + \frac{1}{2}(0.5) = 0.55 \]
\[ q = 0.2 + \frac{1}{2}(0.5) = 0.45 \]
Hardy-Weinberg Equilibrium

- Genotype and allele frequencies remain constant from one generation to the next if:
  - random mating
  - no migration
  - no mutation
  - no natural selection
  - large population size
If population in H-W equilibrium, then

\[(p+q)^2 = p^2 + 2pq + q^2\]

If population in H-W equilibrium, then

\[P = p^2\]
\[H = 2pq\]
\[Q = q^2\]
# Human Freckles

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Number</th>
<th>Ph. Frequency</th>
<th>Genotype</th>
<th>Genotype Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Freckles</td>
<td>120</td>
<td>120/200=0.60</td>
<td>AA</td>
<td>P=p^2=0.14</td>
</tr>
<tr>
<td>None</td>
<td>80</td>
<td>0.40</td>
<td>aa</td>
<td>Q = 0.4</td>
</tr>
<tr>
<td>Total</td>
<td>200</td>
<td>1.0</td>
<td>1.0</td>
<td></td>
</tr>
</tbody>
</table>

Assume HW  

\begin{align*}
Q &= q^2 = \text{proportion of aa} = 0.40 \\
q &= \sqrt{Q} = \sqrt{0.40} = 0.63 \\
p &= 1 - q = 0.37
\end{align*}
## Human Dimples

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Number</th>
<th>Phenotype Frequency</th>
<th>Genotype</th>
<th>Genotype Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dimples</td>
<td>20</td>
<td>0.20</td>
<td>AA</td>
<td>P=p²=0.0121</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Aa</td>
<td>H=2pq=0.19</td>
</tr>
<tr>
<td>None</td>
<td>80</td>
<td>0.80</td>
<td>aa</td>
<td>Q=0.80</td>
</tr>
<tr>
<td>Total</td>
<td>100</td>
<td>1.0</td>
<td></td>
<td>1.0</td>
</tr>
</tbody>
</table>

Assume HW 

\[ Q=q^2 = \text{proportion of } aa = 0.80 \]
\[ q = \sqrt{0.80} = 0.89 \]
\[ p=1-q = 0.11 \]
# Summary of Frequencies

<table>
<thead>
<tr>
<th>Alleles</th>
<th>Allele Freq.</th>
<th>Genotypes</th>
<th>Genotype Freq.</th>
<th>Genotype Freq. if H-W</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>p</td>
<td>AA</td>
<td>P</td>
<td>p^2</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Aa</td>
<td>H</td>
<td>2pq</td>
</tr>
<tr>
<td>a</td>
<td>q</td>
<td>aa</td>
<td>Q</td>
<td>q^2</td>
</tr>
</tbody>
</table>

Always:

\[
p = P + \frac{1}{2} H
\]

\[
q = Q + \frac{1}{2} H
\]

If H-W:

\[
q = \sqrt{q^2}
\]

\[
p = 1 - q
\]
X-Linked Trait

e.g. Color Blindness

Females XX

<table>
<thead>
<tr>
<th>CC or Cc - normal</th>
<th>99</th>
</tr>
</thead>
<tbody>
<tr>
<td>cc - color blind</td>
<td>1</td>
</tr>
</tbody>
</table>

100

Males Xy

<table>
<thead>
<tr>
<th>Cy - normal</th>
<th>92</th>
<th>0.92</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>cy - color blind</td>
<td>8</td>
<td>0.08</td>
<td>q</td>
</tr>
</tbody>
</table>

p = Cy = 0.92
q = cy = 0.08

P = CC = p^2 = (0.92)^2 = 0.846

H = Cc = 2pq = 2(0.92)(0.08) = 0.147

Q = cc = q^2 = (0.08)^2 = 0.0064
Activity 21