Chapter 19

Population Genetics
Populations as Genetic Reservoirs

- **Population**
  - A group of interbreeding organisms belonging to a single species

- **Gene pool**
  - Set of genetic information carried by the members of a sexually reproducing population

- **Allelic frequency**
  - Frequency of an allele is present in the population
Calculating Allelic Frequencies

<table>
<thead>
<tr>
<th>Population</th>
<th>Genotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>54 with Blood Type $M$</td>
<td>$MM$</td>
</tr>
<tr>
<td>26 with Blood Type $MN$</td>
<td>$MN$</td>
</tr>
<tr>
<td>20 with Blood Type $N$</td>
<td>$NN$</td>
</tr>
</tbody>
</table>

Total 100 individuals = 200 alleles

Freq. of $M = \frac{2(54)+26}{200} = .67$
Freq. of $N = \frac{2(20)+26}{200} = .33$
Allelic Frequencies

- Dominant and codominant alleles can be measured directly.
- Recessive allelic frequencies cannot be measured directly.
- Mathematical formulas such as Hardy-Weinberg can be used to determine allelic frequencies.
Using the Hardy-Weinberg Law

**Allele Frequencies**

\[ p = \text{frequency of all dominant alleles in population} \]

\[ q = \text{frequency of all recessive alleles in population} \]

\[ p + q = 1.0 \]
Calculating Allelic and Genotypic Frequencies

\[ p + q = 1 \]

<table>
<thead>
<tr>
<th></th>
<th>( A(p) )</th>
<th>( a(q) )</th>
</tr>
</thead>
<tbody>
<tr>
<td>( A(p) )</td>
<td>( AA ) ( (p^2) )</td>
<td>( Aa ) ( (pq) )</td>
</tr>
<tr>
<td>( a(q) )</td>
<td>( Aa ) ( (pq) )</td>
<td>( aa ) ( (q^2) )</td>
</tr>
</tbody>
</table>

Fig. 19.4
Using the Hardy-Weinberg Law

**Allele Frequencies**

- \( p \) = frequency of all dominant alleles in population
- \( q \) = frequency of all recessive alleles in population

\[
p + q = 1.0
\]

**Genotype Frequencies**

For gene with 2 alleles:

- \( p^2 \) = frequency of homozygous dominant individuals in population
- \( q^2 \) = frequency of homozygous recessive individuals in population
- \( 2pq \) = frequency of heterozygous individuals in population

\[
p^2 + 2pq + q^2 = 1.0
\]
Calculating Frequency of Alleles and Genotypes

<table>
<thead>
<tr>
<th>Eggs</th>
<th>Sperm</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>AA</td>
</tr>
<tr>
<td>(p = 0.6)</td>
<td>(p^2 = 0.36)</td>
</tr>
<tr>
<td>a</td>
<td>Aa</td>
</tr>
<tr>
<td>(q = 0.4)</td>
<td>(pq = 0.24)</td>
</tr>
</tbody>
</table>

Fig. 19.5
Assumptions of Hardy-Weinberg

• Large population
• No selection; all genotypes survive and reproduce equally
• Random mating
• No mutation or migration
# Frequency of Heterozygous Traits

<table>
<thead>
<tr>
<th>Trait</th>
<th>Heterozygote Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cystic fibrosis</td>
<td>1/22 whites; much lower in blacks, Asians</td>
</tr>
<tr>
<td>Sickle cell anemia</td>
<td>1/12 blacks; much lower in most whites and in Asians</td>
</tr>
<tr>
<td>Tay-Sachs disease</td>
<td>1/30 among descendants of Eastern European Jews; 1/350 among others of European descent</td>
</tr>
<tr>
<td>Phenylketonuria</td>
<td>1/55 among whites; much lower in blacks and those of Asian descent</td>
</tr>
<tr>
<td>Albinism</td>
<td>1/10,000 in Northern Ireland; 1/67,800 in British Columbia</td>
</tr>
</tbody>
</table>
Calculating the Probability of Having an Affected Child

For CF, 1/2000 are homozygous recessive. So, cc (genotype) frequency is 1/2,000 or 0.0005 = q^2
\[ \sqrt{q^2} = q = 0.022 \]
p = 1 - q = 0.978
2pq = heterozygote frequency = 2 × 0.978 × 0.022 = 0.043 = 1 in 23.

\[ \frac{1}{23} \times \frac{1}{23} \times \frac{1}{4} = \frac{1}{2,116} \]
Mutations Generate New Alleles

- Mutation alone has minimal impact on the genetic variability in the population.
- Drift, migration, and selection determine the frequency of alleles in the population.

Fig. 19.6
Changing Allelic Frequencies in a Population

- Genetic drift
  - Random fluctuations in allelic frequencies from generation to generation in a small population

- Founder effects
  - Allelic frequencies due to change in a population started by a small number of individuals

- Natural selection
  - Unequal reproductive success that is a result of differences in fitness
Natural Selection and Frequency of Genetic Disorders

Examples

- Lactose intolerance
- Duchenne muscular dystrophy
- Sickle cell anemia
- Tay-Sachs disease
Distribution of Sickle Cell Anemia and Malaria

Fig. 19.9
Measuring Genetic Diversity

Duffy blood group alleles

- \(FY^A\), \(FY^B\), and \(FY^O\)
- Frequency of \(FY^O\) in West Africans close to 100%
- Frequency of \(FY^O\) in Europeans close to 0%
- Measure the frequency of \(FY^A\) and \(FY^B\) in U.S. black population to estimate genetic mixing between populations
Frequency of FY*A

Fig. 19.10
Are There Races?

- Most genetic variation is present within populations
- Minimal variation among populations, including those classified as different racial groups
Genetic Variation

The variation within a population is greater than the variation between populations.

Fig. 19.11
**Homo sapiens**

- Combination of anthropology, paleontology, archaeology, and genetics used to study the dispersal of human populations
- Evidence suggests North and South America were populated by migrations during the last 15,000 or 30,000 years
Appearance and Spread of *Homo sapiens*

**European population**
Origin: 40,000 to 50,000 years ago

**Asian population**
Origin: 50,000 to 70,000 years ago

**Australo-Melanesian population**
Origin: 40,000 to 60,000 years ago

**New World population**
Origin: 20,000 to 30,000 years ago

**African populations**
Origin: 130,000 to 170,000 years ago
Population: 23,000 to 45,000

**Immigration from Africa**
About 137,000 years ago; 200 to 500 or more individuals

Fig. 19.12