Hardy Weinberg Review

The H-W Law predicts genotype frequencies (t+1) from allele frequencies (t).

Combined with the relationship between allele and genotype frequencies within generations, we can solve ALL H-W problems.

\[
\begin{align*}
\text{Allele Frequency} & \\
\text{Genotype Frequency} & \\
\text{Time t} & \\
\text{Time t+1} & \\
F_{11} & = p^2 \\
F_{12} & = 2pq \\
F_{22} & = q^2
\end{align*}
\]

Factors That Drive Change in Allele Frequency

1. 
2. 
3. 
4. 
5.
Mutants!

More Mutants!

Even more mutants!
Fun Facts About Mutation in Us!

- Each of us is born with 300 new mutations that make us different from our parents
- At least 4500 human genetic defects have been found that cause inherited disease
- In 500,000 humans, there will be 800,000 mutations each generation

Mutation

- New alleles arise by mutation
- Mutation is change in the nucleotide sequence of DNA

- Mutation may involve alteration of
  - 1 base pair
  - Several bases
  - Part of the chromosome
  - Whole chromosomes
Causes of Mutations

- Errors in DNA replication
- Physical chromosome damage
- Insertion of transposable element
- UV radiation
- Radiation at other wavelengths (gamma, x-ray)
- Chemicals of various sorts

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### Source of Exposure

<table>
<thead>
<tr>
<th>Source of Exposure</th>
<th>Dose</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dental X-ray</td>
<td>0.005 mSv*</td>
</tr>
<tr>
<td>135g bag of Brazil nuts</td>
<td>0.005 mSv</td>
</tr>
<tr>
<td>Chest X-ray</td>
<td>0.02 mSv</td>
</tr>
<tr>
<td>Transatlantic flight</td>
<td>0.07 mSv</td>
</tr>
<tr>
<td>Nuclear power station worker average annual occupational exposure</td>
<td>0.18 mSv</td>
</tr>
<tr>
<td>UK annual average radiation dose</td>
<td>1.3 mSv</td>
</tr>
<tr>
<td>CT scan of the head</td>
<td>1.4 mSv</td>
</tr>
<tr>
<td>UK average annual radiation dose</td>
<td>2.7 mSv</td>
</tr>
<tr>
<td>USA average annual radiation dose</td>
<td>6.2 mSv</td>
</tr>
<tr>
<td>CT scan of the chest</td>
<td>6.6 mSv</td>
</tr>
<tr>
<td>Average annual radiation dose to people in Cornwall</td>
<td>7.6 mSv</td>
</tr>
<tr>
<td>Whole body CT scan</td>
<td>10 mSv</td>
</tr>
<tr>
<td>Annual exposure limit for nuclear industry employees</td>
<td>20 mSv</td>
</tr>
<tr>
<td>Level at which changes in blood cells can be readily observed</td>
<td>100 mSv</td>
</tr>
<tr>
<td>Acute radiation effects including nausea and a reduction in white blood cell count</td>
<td>1000 mSv</td>
</tr>
<tr>
<td>Dose of radiation which would kill about half of those receiving it in a month</td>
<td>5000 mSv</td>
</tr>
</tbody>
</table>

*unit = millisieverts

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### Definitions

- Haplotype - a particular DNA sequence that differs from homologous sequences by one or more mutations
- Genetic markers - specific mutations geneticists use to recognize certain genes or genetic 'lines'
We define $\mu$ as the fraction of $A_1$ alleles that mutate to $A_2$ each generation.

Then $1-\mu$ is the fraction that remain $A_1$.

Then:

Mutation Model (cont’d)
How long will it take to change $p$ from 0.9 to 0.1 (a substantial evolutionary change)?

Evolution by Mutation
Molecular Clocks

- Slow accumulation of neutral mutations allows us to establish a biological clock.
- Knowing the rate of substitution of DNA base pairs within genes, we can estimate times since two lineages diverged, and reconstruct phylogenies.

Mutation and Migration History

- Accumulation of mutations in maternally inherited DNA (e.g., mitochondrial) or paternally inherited DNA (Y-chromosome) can help us understand past migration.


The Genographic Project
National Geographic Society

“Millions of Men May Be Descended From Irish King, Study Says”

23 & me!
Effect of Mutation

- Constant level of mutation produces very slow change in allele frequency, assuming these changes are neutral (do not affect fitness)
- Mutation is evolutionarily important as a source of variation.
Mutation Summary
- Mutation produces slow evolutionary change
- Mutations are largely neutral or detrimental
- Mutation is a source of variation
- Steady mutational forces allow us to gauge time since divergence of lineages with a "molecular clock"
- Mutational legacies allow us to trace migration pathways by using unique marker genes

Next Lecture
- Genetic drift - random genetic change in small populations