Lecture 13

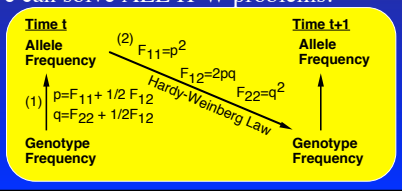
Mutation - Source of Variation

Intro to Drift - A Game of Chance

•See Article 13. Mutation

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.



Factors That Drive Change in Allele Frequency

1.
2.
3.
4.
5.


Mutants!


More Muta


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

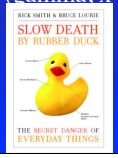
- New alleles arise by mutation
- Mutation is change in the nucleotide sequence of DNA

Mutation

- 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



Source of Exposure	*Dose*
Dental X-ray	*0.005 mSv**
135g bag of Brazil nuts	*0.005 mSv*
Chest X-ray	*0.02 mSv*
Transatlantic flight	*0.07 mSv*
Nuclear power station worker average annual occupational exposure	*0.18 mSv*
UK annual average radon dose	*1.3 mSv*
CT scan of the head	*1.4 mSv*
UK average annual radiation dose	*2.7 mSv*
USA average annual radiation dose	*6.2 mSv*
CT scan of the chest	*6.6 mSv*
Average annual radon dose to people in Cornwall	*7.8 mSv*
Whole body CT scan	*10 mSv*
Annual exposure limit for nuclear industry employees	*20 mSv*
Level at which changes in blood cells can be readily observed	*100 mSv*
Acute radiation effects including nausea and a reduction in white blood cell count	*1000 mSv*
Dose of radiation which would kill about half of those receiving it in a month	*5000 mSv*

**unit = millisieverts*

Definitions

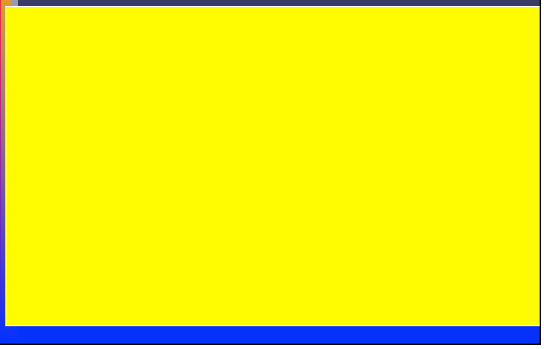
- Haplotype - a particular DNA sequence that differs from homologous sequences by one or more mutations
- Genetic markers - specific mutations geneticists use to recognize certain genes or genetic 'lines'

Mutation and Evolution at a Locus

Mutation Model

- We define μ as the fraction of A_1 alleles that mutate to A_2 each generation
- Then 1-μ is the fraction that remain A_1
- Then:

Mutation Model (cont'd)

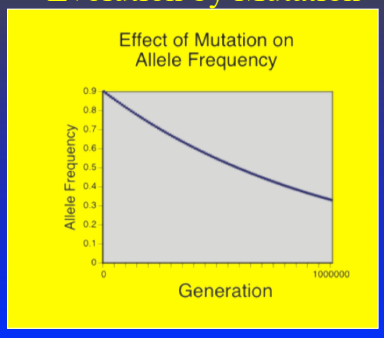


Prediction of Mutation Model

- How long will it take to change p from 0.9 to 0.1 (=a substantial evolutionary change)?

Mutation Model Prediction

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

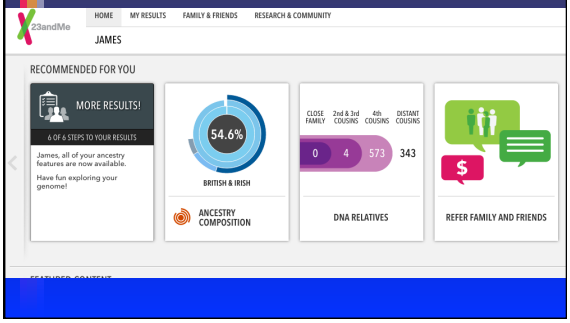
- Accumulation of mutations in maternally inherited DNA (e.g., mitochondrial) or paternally inherited DNA (Y-chromosome) can help us understand past migration.

https://www3.nationalgeographic.com/genographic/index.html

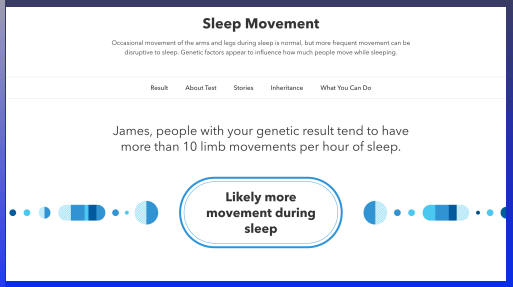
The Genographic Project
National Geographic Society

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

23 & me!



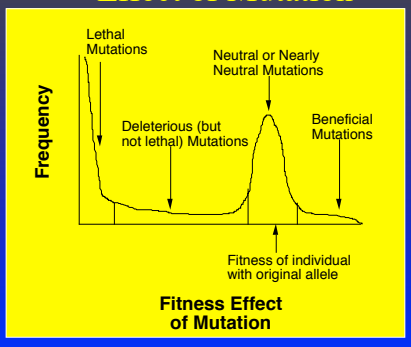
23&me (I just learned this!)



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.**

Effect of Mutation



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