GENETIC DRIFT & NEUTRAL THEORY of EVOLUTION

There is much genetic variation within almost all species. The amount of genetic variation is too much to be maintained by selection.
Reading Assignment

► The sections relevant to this lecture are 25.3 Genetic Drift,
► Your lab on Population Genetics.
► A current major activity of biologists generating phylogenies which are based on the fact that allelic substitutions, presumably neutral, accumulate proportional to time since last common ancestor.
Mutations arise by chance (meaning the mutations are not directed to match environmental needs).

Favorable (= higher fitness) mutations increase in frequency via selection (changed fitness often associated with changed environment).

Deleterious (= lower fitness) mutations are reduced in frequency (many resistance mutations are deleterious unless toxic agent present).
Amounts of Genetic Variation

► When molecular techniques allowed scientists to measure genetic variation in proteins they found 1) many **polymorphic loci**, and 2) including some with many alleles.

► The amount of genetic variation was much greater than scientists expected from the population genetic models that existed.

► Selection models were ‘tweaked’, but ....
New Idea

► The **neutral theory** of evolution developed by Motoo Kimura (1\textsuperscript{st} publication 1968).

► The neutral theory departed from all existing models by using \( N \), the population size, as the most important population parameter.

► What is the neutral theory of evolution?
Sampling Variation

- Real populations have a finite number of members, N.
- Whenever you have a finite number of individuals you have sampling variation.
- Sampling leads to changes in frequency due to chance.
- Allele frequencies are not expected to stay the same in a real population.
Genetic drift

- Let the frequency of in next generation = $p'$
- $p$ will not = $p'$ if the population is finite, because of chance (sampling).
- The proportion of deviation from one generation to next should
  - Go up sometimes and down sometimes
  - Be inversely proportional to $N$, pop size
  - END when one allele reaches 100% of population, a state called fixation.
Allele frequency in finite populations

- One way to see that the infinite population size (=Hardy-Weinberg model) is not compatible with finite populations is to calculate expected frequencies and realize that the calculated value is impossible in real population. If 1 of 10 snails have the recessive phenotype, the estimated recessive allele frequency is 0.316, but 0.30 & 0.35 are the closest possible actual frequencies in population with only 20 (=2N) genes in ten diploid snails.
GENETIC DRIFT in populations differing by 2 orders of magnitude

Note fixation of both alleles
Experimental Study of Drift

(b) In most experimental populations, the forked bristle allele drifted to fixation or loss

Population size = 8 fruit flies (four males, four females)

- In 29 populations the forked allele became fixed:
  - $A_{forked} = 1.0$
  - $A_{normal} = 0.0$

- In 26 populations, both alleles were still present

- In 41 populations, the forked allele became lost:
  - $A_{forked} = 0.0$
  - $A_{normal} = 1.0$

In 70 of the 96 populations, genetic drift caused one allele to be lost
The Neutral Theory

1) There are no fitness differences between almost all of the molecular variation that is detected in populations.

Neutral is the word chosen to describe the lack of fitness differences (functionally equivalent alleles).

2) Amount of genetic variation in a population is determined by a balance between an increase due to mutation, rate = $\mu$, and a decrease due to finite population size (=genetic drift).
Fixation (of alleles)

- For allele frequencies the values of 0 and 1 are especially important, because a previously polymorphic population that had both $A$ and $a$ alleles now has only one kind, i.e. has no variation.
- Alleles that reach 100% are said to be fixed.
- Once lost, the only way genetic variation can be regained is through mutation.
Rate of loss of genetic variation in one generation

- If there are $N$ individuals in a population, there are $2N$ genes. Only if every gene were different could there be as many as $2N$ kinds.
- Sampling from an infinite gamete pool with $2N$ types, means there is a probability of $1/2N$ that the second allele drawn is identical to the first one drawn.
- $F$, a measure of genetic variation loss, $= 1/2N$
Genetic Variability Loss over time

- Proportion of variability retained from one generation to the next is: \((1 - \frac{1}{2N}) = 100\% \text{ minus that which is expected to be lost.}\)

- After \(t\) generations remaining variability = \((1 - \frac{1}{2N})^t\) which goes to zero as \(t\) increases.

- If \(N = 50\) then \(2N = 100\) so in one generation \((1 - \frac{1}{2N}) = 0.99 = 99\% \text{ retained}\)

- \((0.99)^{40} = 0.67\), so in 40 generations 1/3rd of initial variability is lost in population of 50 individuals (each generation).
Losses and gains of polymorphism

► Each polymorphic population has a possibility of becoming monomorphic (= completely homozygous) in the next generation.

► The rate that variability is lost is inversely proportional to population size.

► Once monomorphic (=only one allele) the only return to polymorphism is via mutation.
Bottlenecks

► When population is now large, but was very much smaller in the past we say it has gone thru a “genetic bottleneck”.

► The population will have less genetic variation than expected from its current population size, if, in the past, its population size was much smaller.
Probability of eventual fixation of neutral alleles

- Each neutral allele has an equal chance of being the one that will eventually become “fixed” via drift, i.e. reach 100% frequency.
- If the frequency of $a$ is 0.15, then the probability it will eventually become the only allele in the population is 0.15.
- If the initial frequency of $a$ is 0.7, then the probability it will eventually become fixed is 0.7.
GENETIC DRIFT recapitulation

► All populations have genetic drift.
► The average change in allele frequency (drift) from the value of the last generation is greater the smaller the population size.
► Thus drift is inversely proportional to population size.
Neutral Theory recapitulation

► Most evolution (change in allele frequency) is the result of:
  - Biological populations have a size, N.
  - Functionally equivalent alleles are expected to rise or fall by chance in finite populations.
  - Genetic drift results in a loss of genetic variation.
  - **Mutation** is the only ‘mechanism’ to increase genetic variation.
Dynamics of Genetic Variation

- Natural selection and drift are the main forces leading to allele frequency change.
  - Both are expected to lead to genetic uniformity.
- Mutation generates new alleles, variety.
- Separate populations is vary independently, except if migration between the populations is occurring. Migration makes the populations less differentiated.
Geographic structure & migration

- Species are composed of populations that are at least partially isolated from one another.
- Most of the geographical genetic structure that is seen in populations is the consequence of genetic drift.
- Migration between populations reduces the differences that tend to build up thru drift.
Vocabulary

- Genetic variation
- Mutation
- Neutral theory
- Selectively equivalent
- Drift
- Fixation
- Loss of variability

- $N$
- $1/2N$
- Monomorphic
- Genetic bottleneck
- Directional selection
- Stabilizing selection
- inversely proportional