**Chapter 6  
The ways of change: drift and selection**

Genetic Variation

* The biological sciences now generally define **evolution** as being the sum total of the genetically inherited changes in the individuals who are the members of a population's gene pool.  It is clear that the effects of evolution are felt by individuals, but it is the population as a whole that actually evolves.

Population genetics

* Study of the distribution of alleles in populations and causes of allele frequency changes

Allele frequency

* **Allele frequency** is the proportion of all copies of a gene that is made up of a particular gene variant (allele).
* In other words, it is the number of copies of a particular allele divided by the number of copies of all alleles at the locus in a population.
* It can be expressed for example as a percentage. In population genetics, allele frequencies are used to depict the amount of genetic diversity at the individual, population, and species level. It is also the relative proportion of all alleles of a gene that are of a designed type.
* In the early 20th century by **Godfrey Hardy**, an English mathematician, and **Wilhelm Weinberg**, a German physician developed an equation to at population genetics:

**Hardy-Weinberg equilibrium**

* Population allele frequencies do not change if:
  + Population is infinitely large
  + Genotypes do not differ in fitness
  + There is no mutation
  + Mating is random
  + There is no migration
* Through mathematical modeling based on probability, they concluded in 1908 that gene pool frequencies are inherently stable but that evolution should be expected in all populations virtually all of the time.
* Hardy-Weinberg theorem proves that allele frequencies do not change in the absence of drift, selection, mutation, and migration
* Mechanisms of evolution are forces that change allele frequencies

Predictions from Hardy-Weinberg

* Allele frequencies predict genotype frequencies
* In this equation (p² + 2pq + q² = 1), p is defined as the frequency of the dominant allele and q as the frequency of the recessive allele for a trait controlled by a pair of alleles (A and a).
* In other words, p equals all of the alleles in individuals who are homozygous dominant (AA) and half of the alleles in people who are heterozygous (Aa) for this trait in a population
* The Hardy-Weinberg equation allows us to predict which ones they are.  Since p = 1 - q and q is known, it is possible to calculate p as well.  Knowing p and q, it is a simple matter to plug these values into the Hardy-Weinberg equation (p² + 2pq + q² = 1).
* This then provides the predicted frequencies of all three genotypes for the selected trait within the population.

Examples of how H-W can be used:

MN locus on erythrocyte: two alleles (M & N), and 3 genotypes (MM,MN,NN).

* Counting 320 people yielded the following:
  + MM 187, MN 114, NN19
* Genotype frequency:
  + Frequency of MM = 187/320 = 0.584
  + Frequency of MN = 114/320 = 0.356
  + Frequency NN = 19/320 = 0.059
* Allele frequency:
  + 320 people = 640 gene copies (diploid)
  + # of M alleles = (187 x 2) + (144) = 518
  + Frequency of M = 488/640 = 0.81

1 in 1700 US Caucasian newborns have cystic fibrosis. C for normal is dominant over c for cystic fibrosis. What percent of the above population have cystic fibrosis (cc or q2)?

* Calculate "q" first?
* Since you know q2 (cc), you can easily find q by calculating the square root of q2

q2= 1/1700

= 0.00059 (0.059% are cc)

q = 0.024

(2.4 % frequency of cystic fibrosis allele)

* Now solve for p:

p = 1 – q

p = 1 – 0.024

p = 0.976

* Now that you know that p =.976 and q = .024. The following genotypes can be found:

(*f*)CC- Normal homozygous dominant =

p2 = 0.953 95.3% frequency for normal

(*f*)Cc -carriers of cystic fibrosis =

2pq = 2(0.976)(0.024)

= 0.0468 46.8% frequency for carriers

* How many of the 1700 of the population are homozygous Normal?

0.953 (1700) = 1620.1 (round to 1620)

* How many of the 1700 in the population are heterozygous (carrier)?

0.0468 (1700) = 79.56 (round to 80)

Populations evolve through a variety of mechanisms

|  |  |  |
| --- | --- | --- |
| Mechanism | Action | Result |
| H-W equilibrium (no evolution) | random mating, no migration, mutation, genetic drift, and no natural selection | Allelic frequencies do not change |
| Genetic Drift | Imperfect sampling causes some alleles to be underrepresented in relative to others | Allele is lost or decreased (others increase) |
| Natural selection | Environmental factors are either favorable or unfavorable for allele | Allele is lost or decreased (others increase) |
| Migration (gene flow) | Individuals with new allele enter population (immigration) | New allele becomes more common |

Hardy-Weinberg serves as the fundamental null model in population genetics

**Genetic Drift**

* Genetic drift—along with natural selection, mutation, and migration—is one of the basic mechanisms of evolution.
* Genetic drift—is simply the evolutionary equivalent of a sampling error.
* The genes of the next generation will be the genes of the “lucky” individuals, not necessarily the healthier or “better” individuals. That, in a nutshell, is genetic drift. It happens to ALL populations—there’s no avoiding the vagaries of chance.
* In each generation, some individuals may, just by chance, leave behind a few more descendents (and genes, of course!) than other individuals.
* This drifting happens in populations of organisms. Due to many random factors, the genes in one generation do not wind up in identical ratios in the next generation, and this is evolution.
* It is possible for the frequency of genes for to increase in a population of without the help of natural selection. While this is evolution, it is evolution due to chance, not selection.
* **Genetic drift causes evolution in finite populations**
* Drift reduces genetic variation in a population
* Alleles are lost at a faster rate in small populations
* Alternative allele is fixed
* **So….**
  + **Genetic drift has several important effects on evolution:**
  + **Drift reduces genetic variation in populations, potentially reducing a population’s ability to evolve in response to new selective pressures.**
  + **Genetic drift acts faster and has more drastic results in smaller populations. This effect is particularly important in rare and endangered species.**
  + **Genetic drift can contribute to speciation. For example, a small isolated population may diverge from the larger population through genetic drift.**

**Bottleneck and Founder Effects**

* Genetic drift can be affected by things such as the bottleneck effect or the founder effect.
* Bottlenecks reduce genetic variation
* **Population bottlenecks** occur when a population’s size is reduced for at least one generation. Because genetic drift acts more quickly to reduce genetic variation in small populations, undergoing a bottleneck can reduce a population’s genetic variation by a lot, even if the bottleneck doesn’t last for very many generations.
* A **founder effect** occurs when a new colony is started by a few members of the original population. This small population size means that the colony may have:
  + reduced genetic variation from the original population.
  + a non-random sample of the genes in the original population
* When a small part of a population moves to a new locale, or when the population is reduced to a small size because of some environmental change, the genes of the "founders" of the new society are disproportionately frequent in the resulting population.
* If individuals in the group tend to marry within it, there's a greater likelihood that the recessive genes of the founders will come together in the cells that produce offspring.
* For example, the Afrikaner population of Dutch settlers in South Africa is descended mainly from a few colonists. Today, the Afrikaner population has an unusually high frequency of the gene that causes Huntington’s disease, because those original Dutch colonists just happened to carry that gene with unusually high frequency.
* **Founder effect is NOT the same as genetic drift, since drift is cumulative change over many generations.**
* **Both drift and founder effect result from random sampling**

**The concept of fitness**

* **Fitness**: the reproductive success of an individual with a particular phenotype
* Components of fitness:
  + Survival to reproductive age
  + Mating success
  + Fecundity
* Relative fitness: fitness of a genotype standardized by comparison to other genotypes

**Natural selection more powerful in large populations**

* Drift weaker in large populations
* Small advantages in fitness can lead to large changes over the long term
* Alleles that lower fitness experience **negative selection**
* Alleles that increase fitness experience **positive selection**

Relationships among alleles at a locus

* **Additive:** allele yields twice the phenotypic effect when two copies present
* **Dominance:** dominant allele masks presence of recessive in heterozygote

Mutation generates variation

* Mutation rates for any given gene are low
* But, considering genome size and population size *many* new mutations arise each generation
  + Estimate in humans: 9.8 billion new mutations
* Source of variation for selection and drift to act

Mutation-selection balance

* Equilibrium frequency reached through tug-of-war between negative selection and new mutation
* Explains persistence of rare deleterious mutations in populations

**Frequency-dependent selection**

* **Frequency-dependent selection** is the term given to an evolutionary process where the fitness of a phenotype is dependent on its frequency relative to other phenotypes in a given population.
* In **positive** (or **diversifying**) frequency-dependent selection, the fitness of a phenotype increases as it becomes more common.
* In **negative** (or **purifying**) frequency-dependent selection, the fitness of a phenotype increases as it becomes rarer. This is an example of balancing selection.

Balancing selection

* Some forms of selection maintain diversity in populations:
  + Negative frequency-dependent selection
    - In human pathogens, such as the flu virus, once a particular strain has become common, most individuals have developed an immune response to that strain. But a rare, novel strain of the flu virus is able to spread quickly to almost any individual, causing continual evolution of viral strains
  + Heterozygote advantage
    - Heterozygote advantage and sickle-cell anemia

Inbreeding coefficient

* Probability that two alleles are identical by descent
* Inbreeding depression results in reduced fitness
* Rare deleterious alleles more likely to combine in homozygotes