

# Terms

You should have a working knowledge of the following terms:

- assortative mating
- bottleneck effect
- fitness
- founder effect
- inbreeding
- macroevolution
- microevolution
- mutation

## Introduction and Goals

Previously we examined how the genetic composition of a population is studied. In this tutorial we will examine the conditions that can alter genetic compositions. This theme is central to evolution. Genetically stable populations (those in Hardy-Weinberg equilibrium) do not evolve, however, genetically unstable populations do undergo evolutionary change. We will examine those conditions that affect genetic stability, and hence contribute to evolutionary change. By the end of this tutorial you should have a basic understanding of:

- How the founder effect and bottleneck effect relate to genetic drift
- How gene flow, mutations, and mating behavior can affect genetic stability
- How selection can influence allele frequency

## Microevolution versus Macroevolution

Evolution is a continuous process, but major evolutionary changes are rare events. Major evolutionary changes distinguish one population from another and result in the creation of a new species. Evolutionary changes that mark the appearance of a new species (or genera and higher) are **macroevolutionary** events.

However, evolution can occur within a species. Think about the human allele for sickle-cell disease; populations living in areas with high incidences of malaria are better adapted because of the high frequency of this allele. Hence, evolution of human populations has occurred in the past, is occurring today, and will continue to occur. Changes that lead to alterations in allele frequencies are **microevolutionary** events. At the most basic level, evolution is a change in allele frequencies and genotype frequencies in a population from generation to generation. A population is evolving even if allele frequencies are fluctuating at just one chromosomal location. At the population level, therefore, evolution is a nearly continuous process.

We have discussed how natural selection adapts organisms to their environment. As you learned from the example of the [sickle-cell allele](#), this adaptation results in a change in allele frequency. As you will soon learn, however, natural selection is only one mechanism that can change allele frequencies.

## Population Size

From a geneticist's point of view, a population composed of hundreds or thousands of individuals is less likely to evolve than a small population. The larger the population, the more buffering there is against random variations in allele frequencies; an infinite size population would be almost completely resistant to random fluctuations. As you will learn, small populations are especially vulnerable to the loss of genetic variability due to random events; in fact, they may evolve into extinction.

## Genetic Drift

Small populations are more likely to experience genetic drift (random fluctuations of allele and genotype frequencies). If a population has an allele frequency of  $p = 0.25$  or  $1/4$ , then the allele frequency of  $q$  is  $1 - 0.25 = 0.75$  or  $3/4$ . (Recall, the sum of all individual allele frequencies for a particular gene must add up to 1.) Therefore, for each  $p$  allele, there are 3  $q$  alleles in the population (or a 1:3 ratio). If a population consists of 1000 individuals and  $1000 \times 2 = 2000$  alleles, then 500 of the alleles in the gene pool would be  $p$  ( $2 \times 250$ ) and 1500 of the alleles would be  $q$  ( $2 \times 750$ ). If the alleles are counted in the next generation, they may have drifted by five alleles (e.g., to 505 and 1495), but the new allele frequencies would be 0.253 and 0.748, which is not dramatically different from the original values of 0.25 and 0.75. Although there is a measurable change, it would take many generations before one allele became fixed in the population and the other eliminated.

However, if the population in the example above contained only 10 individuals ( $2 \times 10 = 20$  alleles) and the initial allele frequencies were again  $p = 0.25$  and  $q = 0.75$ , then there would be 5  $p$  alleles ( $2 \times 2.5$ ) and 15  $q$  alleles ( $2 \times 7.5$ ) in the population. If the next generation also drifted by five alleles, then there would be 10  $p$  alleles ( $5 + 5$ ) and 10  $q$  alleles ( $15 - 5$ ). Therefore, in one generation the allele frequencies would go from 0.25 and 0.75 to 0.5 and 0.5. If both alleles are selectively neutral (have no effect on fitness), then each faces a probability of being lost in the course of several generations, and eventually one allele will become fixed in the population. Heterozygosity at that chromosomal location will then be equal to zero.

Next we'll examine the two major situations that restrict population sizes and lead to genetic drift.

## The Bottleneck Effect

One situation that can result in genetic drift is the **bottleneck effect**. A bottlenecking event (e.g., earthquakes, fires, over-hunting) decimates a population and results in only a small fraction of the population surviving. In a bottlenecking event, the remaining randomly selected survivors may not have the same allele and genotype frequencies as the original population. In the new population, some alleles may be found at higher frequencies, whereas others may be found at lower frequencies or even lost altogether.

In a small but rapidly reproducing population, the new allele frequencies may settle into a new equilibrium within one or a few generations and genetic drift will typically affect the population for a number of generations until the population size becomes so large that genetic drift is negligible. While natural disasters have historically been the cause of bottleneck effects, overzealous hunting can also cause this effect.



Many endangered species, such as this cheetah (shown here) and the American buffalo, were subject in recent history to overhunting. Although the cheetah is increasing in numbers, due to increased protective measures, all existing individuals are genetically similar, which weakens the surviving population. Even when a species, such as the cheetah, is protected following a bottlenecking event, the loss of genetic variability may make the species more likely to go extinct. Genetically homogeneous populations are more prone to catastrophes because a given insult (e.g., a disease or a predator) can sweep through the entire population quickly; without variation, there is no natural selection. Hence, conservation efforts must not only preserve numbers, they must also promote genetic variability. Only when some individuals are better suited to the environment than others, can evolution increase the adaptability of a population to its environment. Perhaps the potential to undergo natural selection is one of the best measures of a healthy population.

There is reason to think that bottleneck effects might play a major role in extinction. There is evidence that major catastrophic events (e.g., meteor strikes) might have led to major bottleneck effects in Earth's past. Such events might not have necessarily killed off all members of a species (e.g., dinosaurs), but might have decimated their population enough such that they became genetically weakened due to a loss in variability; eventually these populations died out. One book with a catchy title, "Extinction: Bad Genes or Bad Luck?" by David M. Raup, explores this idea.

A disease that kills only individuals with a genetic predisposition is not an example of a bottleneck event. This would be natural selection at work.

## The Founder Effect



Another situation that can result in genetic drift is the **founder effect**. A founder effect occurs when a few

individuals (founding individuals) become geographically separated from the original population and they form a new colony. The alleles present in the founding individuals make up the gene pool of the new population. Most likely, the founding individuals will not accurately represent the allele and genotype frequencies of the original population, nor will they possess all of the variation present in the original population. Because it would take several generations before their population would increase much in size, genetic drift would continue to influence the allele frequencies of the population. This effect is most commonly seen on remote islands.

An example of the founder effect in human populations occurs on the island of Tristan da Cunha. This island was settled in the 1800's by fifteen British immigrants. There have only been a couple of migrations since; today there are just seven surnames on this island that has about 250 inhabitants. Expectedly, this leads to problems associated with **inbreeding** (the mating of closely related individuals). One notable problem is the occurrence of retinitis pigmentosa (a rare form of blindness), which occurs at a much higher frequency in this population than in the original population.

## Gene Flow

Populations remain genetically stable if no alleles enter or leave the population. Recall, a change in allele frequencies due to interactions with outside populations is termed gene flow (introduced [here](#)). If a population were fixed for one allele at a certain chromosomal location and a gamete carrying an alternative allele were able to fertilize an individual from that population, then the allele frequencies would shift and the population would undergo microevolution. However, if the population were large and if the new allele was not selectively advantageous, then the population would quickly stabilize to a new equilibrium. Conversely, if the population were very small or if the new allele conferred a reproductive advantage, then the allele frequencies would probably continue to change.

## Mutations





A **mutation** is a change in an organism's DNA. Mutations may instantly introduce a new allele into a population, causing small but intermediate shifts in allele and genotype frequencies. Because mutations are the ultimate source of all genetic variation, they collectively are extremely powerful in influencing evolution. In general, however, most mutations have either no effect or no discernable effect on phenotypes. Because several different combinations of nucleotides may code for the same amino acid, a mutation may result in an unaltered protein. Conversely, there are many other types of mutations that can cause major disruptions. These mutations almost always result in a nonfunctional or less efficient product, but if the mutation strikes a region of nontranscribed DNA, then the mutation may have no detectable effect on the organism. Very rarely, a mutation may result in a protein that is slightly more efficient, or that performs a new function. In order for the frequency of the mutant allele to increase as a result of natural selection, the advantage would have to be great enough to affect the individual's reproductive success or survivorship. Therefore, although an assumption when applying the Hardy-Weinberg equation is that no net mutations occur, most mutations have little effect on the allele frequencies of a population overall.

## Random versus Nonrandom Mating

Random mate selection favors genetic stability. Nonrandom mate selection often occurs in natural populations because an individual is more likely to choose a mate from his or her vicinity than from more distant corners of the population. Frequent near-neighbor matings tend to subdivide the population's gene pool into subpopulations that have less genetic diversity than the gene pool at large.

Inbreeding (the mating of closely related individuals) was described earlier in this tutorial. Self-fertilization ("selfing" - introduced [here](#)) occurs when an individual fertilizes some of its own gametes (a common occurrence in plants). Both of these mating processes decrease the level of heterozygosity in their offspring. Normal healthy individuals can carry several or many deleterious alleles. Although each individual may carry some deleterious alleles, unrelated individuals are likely to be carriers of different alleles. Therefore, most of the resulting offspring will be healthy carriers as well. If the parents are closely related, however, then the level of homozygosity in the offspring will be higher. In this way, some of the deleterious alleles may be randomly lost, but it is more likely that some of the recessive deleterious alleles will occur in the homozygous state, exposing those individuals to selection.

Similarly, individuals may choose mates that have some of the same phenotypic traits (e.g., body size or height) as themselves. This is termed **assortative mating**. Although these kinds of characters are complex, involving numerous genes, they are still usually heritable. Mating of genetically similar (although not necessarily related) individuals tends to increase the amount of homozygosity in the population.

## Genetic Stability and Selection Pressure



This tutorial has focused on how random events can affect allele frequencies, hence affecting the genetic stability of populations. Even deleterious alleles may increase in frequency due to chance events (recall the Tristan da Cunha example).

One of the most important conditions for genetic stability is that all individuals have the same **fitness** (fitness is a measure of reproductive success - an individual who produces more offspring than another has greater fitness). Even minor deviations from this state can cause some individuals to produce more offspring than others, resulting in a steady change in allele frequencies. Any alleles that directly or indirectly affect an individual's ability to survive, mate, or reproduce may respond to selection pressure. Under very severe selection pressure (e.g., when bacteria are exposed to a strong antibiotic), a mutation conferring some form of resistance can increase in frequency in the population very quickly. In fact, [antibiotic resistance](#) (read "A Growing Threat") is becoming a major health risk, and [pesticide resistance](#) is a major concern to agriculturists. As you will learn, the example of antibiotic resistance is a result of natural selection because the surviving, resistant individuals are better adapted to their environment.

Similarly, an allele that makes an individual more vulnerable to disease or predation is likely to be under very strong selection pressure and may decrease in frequency very rapidly, until it is either very rare in the population or purged completely from the gene pool.

## Genetic Variation

Each of the phenomena discussed in this tutorial acts to change the frequency of alleles or genotypes in an evolving population. A population in which all of these forces are negligible or absent will remain in a state of equilibrium and allele frequencies will remain the same across generations. Few populations remain in equilibrium for very long, primarily due to the large amounts of genetic variation in natural populations. Frequently, individuals differ from one another in many ways (e.g., from neutral molecular differences in amino acid sequences to differences in appearance, ability, and behavior). Even under relatively strong selection pressure or very small population sizes, it can take many generations for an allele to become fixed. Even under controlled laboratory breeding programs, it can take many generations for a recessive deleterious allele to be removed from the population. Once an allele becomes sufficiently rare in the population, it is unlikely to be found in the homozygous state. Natural selection (one of the strongest and most directional of the evolutionary forces) can only act on the phenotype. Therefore, a recessive allele can linger for a very long time in a population if it is only rarely exposed to natural selection.

Similarly, although an individual may have a low fitness level, this may not be indicative of his or her genotype. Not all variation is heritable, and environmental influences (e.g., weather, nutrition, and availability of potential mates) may overwhelm subtle differences in genotypes. A well fed but genetically inferior animal may have greater reproductive potential than a genetically superior but undernourished animal. In this case, natural selection would not lead to a population better adapted to its environment.

# Summary

This tutorial examined population genetics, with reference to microevolution. Microevolution refers to the change in allele frequencies in a gene pool of a population over generations. A population is a group of interbreeding individuals. Over many generations, these changes can lead to new species. However, different populations of the same species can be undergoing different types of microevolutionary changes but still interbreed; hence, by definition, they are still members of the same species. (Think about the races of humans.) Keep in mind, microevolution describes small changes in a population over successive generations, and does not necessarily mean that two populations will eventually become new species. (Although this can happen over many generations.)

The Hardy-Weinberg equation can be used to determine the allele frequency in a population. Thus, it provides a useful tool to describe the degree of microevolution that is taking place over successive generations. Not all populations are undergoing microevolution at all times. In certain stable environments, many populations show no evidence for microevolution. (Although if the conditions change, then microevolution often commences.)

There are a number of conditions that predictably decrease microevolution; namely: large populations that are more genetically stable compared to smaller populations; no migration/immigration by which new alleles are introduced or removed from a population; no mutations to introduce new alleles; random matings that allow all individuals equal access to all alleles and; no natural selection that causes changes in allele frequencies due to fitness differences between individuals.

Conversely, opposing conditions increase microevolution; namely: small populations are genetically unstable because they are susceptible to genetic drift; migration/immigration that allows alleles to enter or leave a population; mutations that introduce new alleles; nonrandom matings that cause unequal access to all alleles; and natural selection that changes allele frequencies to favor individuals' reproductive capacity and fitness.