19.1B: Population Genetics

Population genetics is the study of the distributions and changes of allele frequency in a population.

Learning Objectives

• Define a population gene pool and explain how the size of the gene pool can affect the evolutionary success of a population

Key Points

• A gene pool is the sum of all the alleles (variants of a gene) in a population.
• Allele frequencies range from 0 (present in no individuals) to 1 (present in all individuals); all allele frequencies for a given gene add up to 100 percent in a population.
• The smaller a population, the more susceptible it is to mechanisms like natural selection and genetic drift, as the effects of such mechanisms are magnified when the gene pool is small.
• The founder effect occurs when part of an original population establishes a new population with a separate gene pool, leading to less genetic variation in the new population.

Key Terms

• allele: one of a number of alternative forms of the same gene occupying a given position on a chromosome
• gene pool: the complete set of unique alleles that would be found by inspecting the genetic material of every living member of a species or population
• founder effect: a decrease in genetic variation that occurs when an entire population descends from a small
Population Genetics

A gene for a particular characteristic may have several variations called alleles. These variations code for different traits associated with that characteristic. For example, in the ABO blood type system in humans, three alleles ($I^A$, $I^B$, or $i$) determine the particular blood-type protein on the surface of red blood cells. A human with a type $I^A$ allele will display A-type proteins (antigens) on the surface of their red blood cells. Individuals with the phenotype of type A blood have the genotype $I^A I^A$ or $I^A i$, type B have $I^B I^B$ or $I^B i$, type AB have $I^A I^B$, and type O have $i i$.

![ABO blood type in humans](image)

Figure 1: ABO blood type in humans: In humans, each blood type corresponds to a combination of two alleles, which represent the type of antigens displayed on the outside of a red blood cell. Human blood types are A, B, AB, and O.

A diploid organism can only carry two alleles for a particular gene. In human blood type, the combinations are composed of two alleles such as $I^A I^A$ or $I^A I^B$. Although each organism can only carry two alleles, more than those two alleles may be present in the larger population. For example, in a population of fifty people where all the blood types are represented, there may be more $I^A$ alleles than $i$ alleles. Population genetics is the study of how selective forces change a population through changes in allele and genotypic frequencies.

Allele Frequency

The allele frequency (or gene frequency) is the rate at which a specific allele appears within a population. In population genetics, the term evolution is defined as a change in the frequency of an allele in a population. Frequencies range from 0, present in no individuals, to 1, present in all individuals. The gene pool is the sum of all the alleles at all genes in a population.

Using the ABO blood type system as an example, the frequency of one of the alleles, for example $I^A$, is the number of copies of that allele divided by all the copies of the ABO gene in the population, i.e. all the alleles. Allele frequencies can be expressed as a decimal or as a percent and always add up to 1, or 100 percent, of the total population. For example, in a sample population of humans, the frequency of the $I^A$ allele might be 0.26, which would mean that 26% of the chromosomes in that population carry the $I^A$ allele. If we also know that the frequency of the $I^B$ allele in this population is 0.14, then the frequency of the $i$ allele is 0.6, which we obtain by subtracting all the known allele frequencies from 1 (thus: $1 – 0.26 – 0.14 = 0.6$). A change in any of these allele frequencies over time would constitute evolution in the population.
Population Size and Evolution

When allele frequencies within a population change randomly with no advantage to the population over existing allele frequencies, the phenomenon is called genetic drift. The smaller a population, the more susceptible it is to mechanisms such as genetic drift as alleles are more likely to become fixed at 0 (absent) or 1 (universally present). Random events that alter allele frequencies will have a much larger effect when the gene pool is small. Genetic drift and natural selection usually occur simultaneously in populations, but the cause of the frequency change is often impossible to determine.

Natural selection also affects allele frequency. If an allele confers a phenotype that enables an individual to better survive or have more offspring, the frequency of that allele will increase. Because many of those offspring will also carry the beneficial allele and, therefore, the phenotype, they will have more offspring of their own that also carry the allele. Over time, the allele will spread throughout the population and may become fixed: every individual in the population carries the allele. If an allele is dominant but detrimental, it may be swiftly eliminated from the gene pool when the individual with the allele does not reproduce. However, a detrimental recessive allele can linger for generations in a population, hidden by the dominant allele in heterozygotes. In such cases, the only individuals to be eliminated from the population are those unlucky enough to inherit two copies of such an allele.

The Founder Effect

The founder effect occurs when part of a population becomes isolated and establishes a separate gene pool with its own allele frequencies. When a small number of individuals become the basis of a new population, this new population can be very different genetically from the original population if the founders are not representative of the original. Therefore, many different populations, with very different and uniform gene pools, can all originate from the same, larger population. Together, the forces of natural selection, genetic drift, and founder effect can lead to significant changes in the gene pool of a population.

Figure 1: The Founder Effect: Here are three possible outcomes of the founder effect, each with gene pools separate from the original populations.