

Genetics Notes Chapter 18: Population Genetics

18.1-Detecting Genetic Variation

- The simplest type of variation is a difference in one nucleotide base or a **Single Nucleotide Polymorphism (SNP)**. Most widely studied variation.
- **Microsatellite loci polymorphisms**-short tandem repeat sequences are also studied. There is variation of the number of short repeats that there are.
- SNPs normally only have two alleles.
- For a SNP allele that occurs at a frequency of greater than 5% is considered a **common SNP**.
- For a SNP that occurs at a frequency of less than 5% is considered a **rare SNP**.
- SNPs are discovered through genome sequencing and comparison of multiple individuals.
- The number of alleles of a microsatellite variation is very high. Additionally, these regions are generally more highly mutated.
- Microsatellite loci can be identified either through screening through a previously sequenced genomes or by probing a cDNA library for repeats of interest.
- Other variations include indel mutations, inversions, relocations, deletions, and duplications.
- **Hapmap project** has set out to sequence thousands of people's genomes from different parts of the world to look at the variation amongst ourselves.

18.2-The Gene Concept and Hardy-Weinberg Equilibrium

- **Gene Pool**-The sum of the total alleles of the breeding members of a population at a given time.
- **Genotype frequencies**: the number of individuals with a certain genotype over the total number of individuals in a population. Genotype frequencies always sum to 1.
- **Allele frequencies** are the number of a certain allele over the total number of alleles.
Symbolized by p and q. $p+q=1$
- $p = f_{A/A} + \frac{1}{2}f_{A/a} = \text{frequency of } A$
- $q = f_{a/a} + \frac{1}{2}f_{A/a} = \text{frequency of } a$
- $p + q = f_{aa} + f_{aa} + f_{aa} = 1.0$
- $q = 1 - p$
- **Hardy-Weinberg Equation:** $p^2 + 2pq + q^2 = 1.0$
- Hardy-Weinberg Assumptions:
 - Random Mating
 - No genotype can reduce viability. All are equally viable.
 - There may be no subpopulations and no group within the population may be reproductively or genetically isolated in anyway.
 - Applies only to infinitely large populations.
- Populations that are in hardy-weinberg equilibrium have the same genotypic and allelic frequencies from generation to generations.

18.3-Mating Systems

- Populations with nonrandom mating will not follow Hardy-Weinberg frequencies.
- **Assortative Mating**- Mates are chosen by an individual based on resemblance to themselves.

- **Positive Assortative** mating occurs when similar types mate. Ex. Tall mate with tall and short with short. This increases the frequency of homozygotes in the gene pool.
- **Negative Assortative/Disassortative Mating**-“Opposites Attract” the unlike mate.
- Another form of mating bias is **geographic isolation** or distance. Mating is much more likely to occur between neighbors.
- This geographic isolation results in subpopulations that form a population structure. These have more homozygotes than predicted by Hardy-Weinberg.
- **Inbreeding**-mating between relatives.
- Inbred progeny are more likely to be homozygous at a given locus and therefore are more likely to have two deleterious recessive alleles.
- Inbreeding can lead to a reduction in vigor-**inbreeding depression**.
- The risk of having deleterious alleles increases with inbreeding, this risk depends on two factors.
 - The frequency of the deleterious allele in the population.
 - The degree of inbreeding.
- **Inbreeding coefficient**-the probability that two alleles in an individual trace back to the same copy of a common ancestor.
- If two copies of a gene in an individual are traced back to the same copy in an ancestor then the two copies are **identical by descent (IBD)**.
- Inbreeding coefficient calculated by: $F_I = \left(\frac{1}{2}\right)^n (1 + F_A)$
- In small populations, inbreeding is much more likely to occur.

18.4-Genetic Variation and its Measurement

- **Gene Diversity**-the probability that two alleles drawn at random from a gene pool will be different.
- **Nucleotide Diversity**-Gene diversity for a single nucleotide site.

18.5-The Modulation of Genetic Variation

- **Mutation Rate**-The probability that a copy of an allele changes to some other allelic form in one generation. Symbolized by the greek letter mu.
- Geneticists can use mutation rate and number of mutations to tell how long it has been since two species diverged from a common ancestor.
- Mutation rates are estimated by sequencing the genome of a founder individual and looking at the genomes of each generation of offspring in comparison.
- **Migration or gene flow**-the movement of individuals or gametes between populations.
- Recombination is another driving force behind variation, no alleles are gained or lost, but new haplotypes are formed.
- If the association of alleles at two loci are random then they are said to be in **linkage equilibrium**. Observed frequencies=Expected Frequencies.
- If the association of alleles at two loci are nonrandom then they are said to be at **linkage disequilibrium**. An allele at one locus is more often associated with a specific allele at a second locus.
- Linkage disequilibrium decays over time due to recombination.

- Changes in allele frequencies between generations due to sampling error is **random genetic drift**.
- It is weakest in larger populations.
- Genetic drift also depends on the initial frequency of the allele.
- Neutral alleles result in phenotypes that are equally viable.
- Genetic drift caused by random sampling of a population to create a newer much smaller population is called the **Founder's Effect**. New population's allelic frequencies may greatly differ from the original's.
- A sudden contraction of population size is a **bottleneck effect**.
- **Natural Selection**-the process by which certain heritable features allow an individual to be more likely to survive, reproduce, and pass down these features more than other individuals who lack these features.
- **Darwinian fitness**-the ability to survive and reproduce.
- **Absolute fitness**-the number of offspring an individual has.
- **Relative fitness**-An individual's fitness relative to another individual.
- **Directional Selection**-Allelic frequencies move in one direction until it either reaches fixation or is lost.
- **Positive Selection**-A favorable allele is brought to higher frequencies.
- **Purifying Selection**-Removes deleterious alleles from the population.
- If the heterozygous is the most fit, both alleles will stay maintained in the population, leading to **balancing selection** at an equilibrium point between the frequencies of the two alleles.
- Directional variation causes a decrease in genetic diversity and an increase in linkage disequilibrium.
- Balancing selection increases genetic diversity by increasing random genetic drift.