# Introductory Genetics

## Genetic Variation

Results when homologous segments of DNA (genes) have two or more different nucleotide sequences.

Results in inheritable differences among individuals in a population.

Genetic variation can be discussed in an hierarchical fashion because variation at a lower level is required for variation at a higher level.

DNA, gene, multiple genes, individual, population, among populations, species

Commonly used terms:

**Phenotype:** Appearance, state, or composition of some particular **character** of an individual. What constitutes a character can differ widely: tall-short, hair color, amino acid sequence of a polypeptide.

**Genotype:** Genetic information carried by a pair of genes on homologous chromosomes. In genetics we are often interested in how strongly the genotype influences the phenotype.

**Allele:** Form of a gene. Different forms are usually named for having a specific phenotypic affect: albino-normal, finespot-largespot.

**Locus:** Physical location occupied by a gene on a DNA molecule (chromosome). For all intents and purposes often used interchangeably with gene. (Plural for locus os loci.)

**Homozygous:** Individual having two copies of the same allele at a locus (AA or aa). Is genetically invariant because can produce only one kind of gamete.

**Heterozygous:** Individual that has two different alleles at a locus (Aa). Is genetically variable because can produce two different gametes.

From a genetics perspective traits are usually broken into two categories:

**Single gene traits:** Those whose phenotype is largely controlled by a single gene. Usually fall into discrete or qualitative categories, normal-albino (Table 3).

**Multiple gene traits:** Those whose phenotype is affected by many genes. Also called **polygenic** traits. Often form continuous distributions, length, weight, condition factor.

## Introductory Population Genetics and Evolution

**Evolution** has a variety of definitions with a common underlying theme; the genetic characteristics of a population change through time.

1. Change in morphology through time.
2. Change in allele frequencies through time.
3. Change in population **gene pool** through time.

Results in spatial and temporal genetic diversification and speciation.

The basic unit of evolution is the **population**; a group of **conspecific** individuals that are spatially and temporally connected through interbreeding. In reality, boundaries between populations are often "fuzzy."

Evolution is a fact or something known with certainty. Fossil record clearly indicates spatial and temporal diversification and speciation. Studies using morphological characters, analysis of proteins and analysis of DNA have also provided clear evidence that the genetic characteristics of populations change through time.

Theory is a scientifically acceptable explanation of facts. Theory is involved in the study of evolution when one attempts to offer explanations and processes for observed change; that is, when one tries to explain evolution.

### The nonevolving or equilibrium population

It is convenient to first examine the characteristics of a hypothetical population that is not evolving in order to examine how the processes of evolution affect a population's genetic composition.

In order for a population to be non-evolving the following conditions must hold:

1. Infinite population size.
2. Random mating or union of gametes.
3. No selection.
4. No mutation.
5. No migration or gene flow.

The genetic composition of a nonevolving population does not change. Allele and genotype frequencies in the population do not change.

The genetic composition of a nonevolving population is completely described by the **Hardy-Weinberg equilibrium** model.

Consider a single locus with two alleles; A and a

p = frequency of A in the population
q = frequency of a in the population
p + q = 1 since p and q must account for all genes in the population

With infinite population size there is no sampling error so the frequency of alleles in gametes is the same as in the population;

frequency A = p  frequency a = q

An AA zygote can be formed only one way; the union of an A sperm and an A egg. In probability calculations, terms connected by "and" are combined by multiplication. The probability of an AA zygote, therefore, is: $p \times q = p^2$

Likewise, the probability of an aa zygote is $q^2$.

A heterozygous zygote (Aa) can be formed two ways:

A sperm and a egg **or** a sperm and A egg

The probability of the first is pq, and the second qp. In probability calculations, terms connected by "**or**" are combined by addition so we have:

$$pq + qp = 2pq$$

In the nonevolving population, therefore, the frequency of genotypes in the next generation is strictly a function of allele frequencies:

$$AA = p^2$$
$$Aa = 2pq$$
$$aa = q^2$$

Since these must account for all individuals then:

$$p^2 + 2pq + q^2 = 1 \qquad \text{or} \qquad (p + q)^2 = 1$$

Simplify by taking the square root of each side: $p + q = 1$

First let's consider a codominant locus.The population has the following numbers of each genotype.

| AA | Aa | aa |
|---|---|---|
| 5 | 10 | 5 |

The frequency of $A = q = \frac{2(5) + 1(10) + 0(5)}{40} = 0.50$

The frequency of $a = q = \frac{0(5) + 1(10) + 2(5)}{40} = 0.50$

Note the observed genotype distribution conforms exactly to the expected Hardy-Weinberg distribution.

| AA | Aa | aa |
|---|---|---|
| $p^2$ | 2pq | $q^2$ |
| 0.25 | 0.50 | 0.25 |

Note an alternative way of getting the frequency of a is to take the square root of the aa genotype frequency ($\sqrt{0.25} = 0.5$). Then the frequency of A is $1 - 0.5 = 0.5$