Cycle 6: Families to Populations

Introduction to Phenotype and Genotype

Microevolution is the *change in the genetic makeup of a population from one generation to the next*. An example of this is the evolution of whiteflies to acquire resistance to pesticides. Another example is the decline in the population of Arabian Sea humpback whales due to the **bottleneck effect**: *a large decrease in population size due to environmental events*. The individuals that make up the current population have much more susceptibility to disease and are unlikely to survive in unfavourable conditions.

In any population, there are phenotypic and genotypic variations, as well as quantitative and qualitative variations. **Phenotypic variations** are *differences in appearance or function*, while **genotypic variations** are *differences in the actual DNA sequence of a gene*. **Quantitative variations** are *small numerical differences between individuals* (e.g. number of hair, lengths of toes, etc.) and **qualitative variations** are *categorical characteristics that exist in two or more discrete states and intermediate forms are absent* (e.g. snow geese that have either blue or white feathers and not both). The *existence of discrete variants of a character* is called **polymorphism**. For example, humans have polymorphic blood types (A, B, AB, O).

Phenotypic Variations and its Causes

Phenotypic variations and determining their causes (i.e., genetics or environmental) can be quite useful. Oftentimes, organisms with different genotypes will exhibit similar phenotypes, but can be differentiated based on the causes. For example, a farmer notices a grain of wheat which produces more wheat than others. By knowing the cause of this phenotypic variation, he can create more grains of wheat with this phenotype and can then increase his wheat yield.

The cause of the phenotypic variation can be determined through various methods. Breeding experiments can be used to demonstrate the genetic basis of phenotypic variations. For example, the breeding and crossing over of flowers can achieve the desired colour. Another method consists of conducting an experiment wherein an experimental variable is manipulated and the effects on genetically similar subjects are measured to isolate environmental factors. For example, an experiment where plants of the same genotype that are grown in full sunlight are compared with plants grown in the shade and the size of leaves and stems of both groups of plants are measured.

Population Genetics

Before delving into population genetics, we must first define allele, locus, and gene pool. An **allele** is a *version of a gene amongst other versions* (in haploids, there's one possible allele of gene that exists and in diploids, there are two alleles that are possessed per gene). A **locus** refers to the *location of a gene on a chromosome*. A **gene pool** refers to the *total genetic variability of the population represented by all the alleles at all gene loci in all individuals within a population*. With these terms, we can now define genotype frequency and allele frequency. A **genotype frequency** refers to the *prevalence/frequency of a genotype in a population*, while an **allele frequency** refers to the *prevalence/frequency of an allele in a population.* Using allele frequencies, we can calculate genotype frequencies and vice versa.

Example 1: Determine the allele frequency.
There are a total of 2,000 flowers, and among them, there are three different flower color phenotypes. The blue flowers (CBCB) have a genotype frequency of 0.50, the purple flowers (CBCR) have a genotype frequency of 0.40, and the red flowers (CRCR) have a genotype frequency of 0.10. What is the allele frequency of CB and CR?

Flower Phenotype	Genotype	Number of individuals	Genotype Frequency	Total Number of C^B alleles	Total Number of C^R alleles
Blue	C^BC^B	2000 x 0.50 = 1000	1000/2000 = 0.50	2 x 1000 = 2000	0 x 1000 = 0
Purple	C^BC^R	2000 x 0.40 = 800	800/2000 = 0.40	1 x 800 = 800	1 x 800 = 800
Red	C^RC^R	2000 x 0.10 = 200	200/2000 = 0.10	0 x 200 = 0	2 x 200 = 400
Total		2000	0.50 + 0.40 + 0.10 = 1.0	2800	1200
p = frequency of C^B allele = Total C^B allele/(Total alleles) = 2800/(2800 + 1200) = 0.7					
q = frequency of C^R allele = Total C^R allele/(Total alleles) = 1200/(2800 + 1200) = 0.3					

Note: The total genotype frequency should always equal to 1.

Hardy-Weinberg Principle

Hardy-Weinberg equilibrium (HWE) is a mathematical model that specifies the conditions that are necessary so that genetic equilibrium is achieved and *allele frequencies, genotype frequencies, and phenotype frequencies do not change from one generation to the nex*t. In order to be in HWE, five conditions must be met:

1. The population is closed to migration from other populations
2. The population is infinite in size
3. No mutation is occurring in the population
4. All genotypes in the population survive and reproduce equally well
5. Individuals in the population mate randomly with respect to genotype

Note that if succeeding generations are in HWE, there will be no changes as in frequencies. Thus, if a population is in HWE, it means that HWE conditions are met and there is no evolution or changes in allele frequencies in the population. On the other hand, if a population is not in HWE, its allele frequencies will change over time. Therefore, genotype frequencies in offspring generations can be predicted only if populations are in HWE.

Hardy Weinberg Equation
p2 + 2pq + q2

Example 2: Predict genotype frequency in the flowers from the previous example.

Recall from the previous example: CB = p = 0.7 and CR = q = 0.3
If the plants reproduce randomly in HWE, the equation p2 + 2pq + q2 predicts the genotype frequencies in the offspring generation:

- Frequency of CBCB = p2 = 0.72 = 0.49
- Frequency of CBCR = 2pq =2 x 0.7 x 0.3 = 0.42
- Frequency of CRCR = q2 = 0.32 = 0.09

Additionally, based on the genotype frequencies, it can be determined if a population is in HWE or not. This can be determined by calculating whether the allele frequencies are equal to the allele frequency of the parent generation. Again from the previous example: p = 0.7 and q = 0.3

- We must determine the allele frequencies from the predicted genotype frequencies under HWE
  - p = (2 x 980 + 840) / 4000 = 0.7
  - q = (2 x 600 + 840) / 4000 = 0.3
    - Frequencies were multiplied by the total population to give rise to the number of individuals (0.49 x 2000 = 980) and were divided by the total number of alleles

Since the allele frequencies are equal to the allele frequencies of the parent generation, the population is in HWE

Gene Flow and Genetic Variability

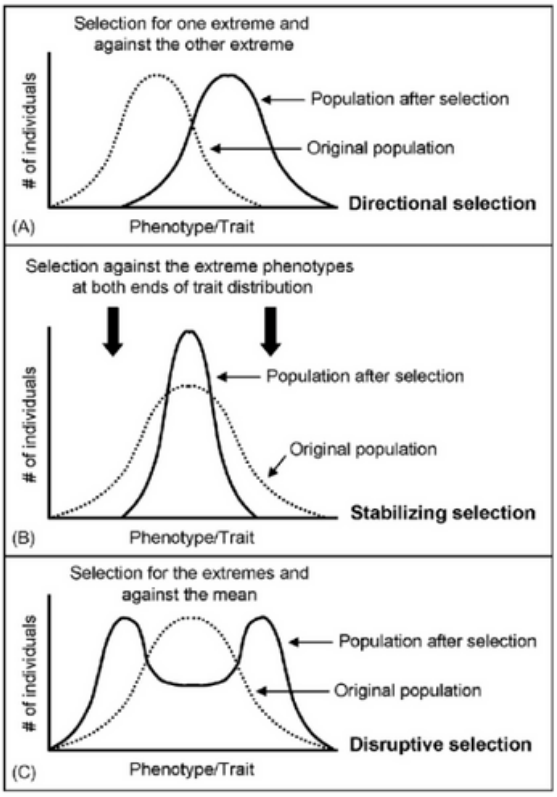
There are different methods that cause genetic variability such as: **mutations, selection, gene flow, genetic drift, and non-random mating**. These take populations out of HWE as they would violate the Hardy-Weinberg principle.

Term	Definition	Effect on Genetic Variation
Mutation	Heritable changes in DNA	Introduces new genetic variation into population; does not change the allele frequency quickly
Selection	Differential survivorship or reproduction of individuals with different genotypes	One allele can replace another or allelic variation can be preserved
Gene Flow	Change in allele frequencies as individuals join a population and reproduce	May introduce genetic variation from another population
Genetic Drift	Random changes in allele frequencies caused by chance events	Reduces genetic variation, especially in small population; can lead to the elimination of rare alleles
Non-Random Mating	When mating is non-random and is selected due to a particular phenotype/genotype	Can lead to an increase or decrease of genetic variation based on inbreeding or outbreeding Inbreeding or assortative mating will increase the frequency of homozygotes and reduce genetic variability (can expose the population to phenotyupes of harmful recessive alleles and result in a greater prevalence of these harmful phenotypes) Outbreeding/disassortative mating promotes heterozygosity (increases genetic variability)

Note: This table does not include the founder effect (reduction in genetic variability due to a small group of individuals being separated from the larger population) and population bottleneck. Due to the reduction in genetic variability, these will likely result in the increased frequency of deleterious alleles.

The Types and Directions of Natural Selection

Selection is a *naturally occurring mechanism of evolution* that leads to the selection of certain traits. There are three different types of selection: directional selection, stabilising selection, and disruptive selection, all of which affect genetic variation differently. **Directional selection** refers to when *individuals near one end of the phenotypic spectrum have the highest relative fitness*. This shifts a trait away from the existing mean and toward ONE favoured extreme. **Stabilizing selection** refers to when *individuals expressing intermediate phenotypes have the highest relative fitness*. This eliminates phenotypic extremes which reduces genetic and phenotypic variation and increases the frequency of intermediate phenotypes. Lastly, **disruptive selection** refers to when *extreme phenotypes have higher relative fitness than intermediate phenotypes*. This shifts a trait away from the mean and towards BOTH extremes.



https://www.researchgate.net/figure/Three-types-of-natural-selection-A-Directional-selection-B-stabilizing-selection_fig5_340583975

Absolute and Relative Fitness

Fitness refers to the *probability of reproductive success*. This can be further divided into absolute and relative fitness. **Absolute fitness** is the *number of offspring per parent*, while **relative fitness** refers to the *number of offspring of a parent of a particular genotype/phenotype compared to the number of offspring per parent of the most successful genotype/phenotype in that population*. In other words, the relative fitness of an individual is their absolute fitness divided by the absolute fitness of the most successful genotype (i.e. the maximal absolute fitness of that population).

Example 3: The absolute fitness of a black cat with genotype BB, an orange cat with genotype BR, and a red cat with genotype RR is 2, 2, and 6, respectively. Which of these cats has the greatest relative fitness?

- Relative fitness of BB = 2/6
- Relative fitness of BR = 2/6
- Relative fitness of RR = 6/6

BB = BR < RR

The red cat with genotype RR has the greatest relative fitness.

Selecting for Phenotypes

The phenotype and dominance status of an allele can lead to changes in allele frequencies and affect fitness. When there's selection against dominant phenotypes (i.e. selection for recessive phenotypes), the fitness of the dominant allele will be less than the fitness of the recessive allele and over time, the frequency of the recessive allele will reach 1, while the frequency of the dominant allele will reach 0. From the previous example, if B were the dominant allele and R were the recessive allele, then the fitness of BB will be equal to BR and both will be less than RR. Additionally, the frequency of R will be 1, while the frequency of B will be 0. When there's selection against recessive phenotypes (i.e., selection for dominant phenotypes), the opposite will occur. From the previous example, the relative fitness of BB will equal to BR and both will be greater than RR. Over time, the frequency of individuals with BB or BR phenotype will be 1, while the frequency of individuals with RR phenotype will be 0.

The fitness of heterozygotes and homozygotes can also affect the allele frequencies and genetic variation. **Heterozygote advantage** refers to when *heterozygotes have higher relative fitness than either homozygotes*. **Heterozygote disadvantage** refers to *selection for homozygotes*. If there's heterozygote advantage, the frequency of heterozygotes will reach 1, while the frequency of homozygotes will reach 0. However, the allele frequencies will both stabilise to 0.5 over time as the population will consists of only heterozygous individuals. Thus, rare alleles will increase in frequency, while common alleles will decrease in frequency. On the other hand, if there's heterozygote disadvantage, the frequency of heterozygotes will be equal to 0 over time. The frequency of homozygotes will vary based on the starting allele frequency. For example, if the starting frequency of allele A is higher than allele B, the frequency of homozygotes AA will be equal to 1 and the frequency of homozygotes BB will be 0, and vice versa.