

**Chapter for today: Chap. XVIII****Major points for the day:**

1. Hardy–Weinberg equilibrium: stable allele frequencies
2. Causes of changes in allele frequency (“microevolution”)

**Review**

Since the 18th century scientists have increasingly found evidence challenging the static traditional theory of diversity

- Distribution of species on earth
- The existence of unexplainable similarities between species
- An apparent progressive series of fossils in the geological record

These observations suggested that species could change, challenging the previous assumptions of immutability. Various theories were advanced to explain the evidence:

- Catastrophism suggested that the geological record was misleading
- Lamarkianism suggested that change occurred by inheritance of acquired characteristics

Both theories are contradicted by evidence. Darwin proposed a theory in which theories change as a result of “natural selection”

- In populations competing for limiting resources some traits present in some individuals may allow them to compete more effectively
- Those individuals, and so their genetic traits, will tend to reproduce more than others
- As a consequence, populations can change over time as result of this selection

Today, we will look at the mechanisms which can cause populations to change

**Variation among individuals**

We have already seen how individuals in a population might differ

- Difference can arise among individuals as a result of mutation
- The differences can propagate through a population as a result of sexual reproduction
- Because of meiotic processes (chromosome segregation and recombination) alleles can be shuffled in a population, creating new combinations of alleles of multiple genes

Therefore, in a large population of individuals there can be great variation in alleles among individuals. This allelic variation provides the material for the process described by Darwin as natural selection

**Stable populations with variable traits**

It is possible for the variability of traits to be stable over generations. This occurs when the population is at “genetic equilibrium”

- The term “genetic equilibrium” means that the proportion of each allele in the population does not change from one generation to the next

In a population at genetic equilibrium the proportion of each allele, its “**allele frequency**” is constant, and the proportion of homozygous and heterozygous individuals in the population can be predicted. This derives from the **Hardy–Weinberg Rule**

In a population in which there are only two alleles at a given gene, where the frequency of one allele *A* is *p*, and the other *a* is *q*, the frequency of each of the three possible genotypes (*AA*, *Aa* and *aa*) is determined by this equation:

$$p^2 AA + 2 pq Aa + q^2 aa = 1$$

How is this equation derived?

We can think back to the Punnett square used to describe a monohybrid cross:

	A	a
A	AA	Aa
a	Aa	aa

- The Punnett square in this case describes the probability of each type of offspring from the cross of two heterozygotes
- The frequency of each of the alleles is the same in this cross; the probability that each parent will contribute *A* or *a* is the same, 50%
- In this situation, the frequency of *AA*, *Aa*, and *aa* is 25%, 50% and 25%, as derived from the Punnett square (1/4 *AA*, 2/4 *Aa*, and 1/4 *aa*)
- This same conclusion can be arrived at using the Hardy–Weinberg equation if you assign the frequencies 0.5 to both *p* and *q*
  - Frequency of *AA* =  $p \times p = (0.5)(0.5) = 0.25$
  - Frequency of *Aa* =  $2pq = 2(0.5)(0.5) = 0.5$
  - Frequency of *aa* =  $q \times q = (0.5)(0.5) = 0.25$

But what if the frequency of the two alleles is not equal, for example if the frequency of *A* is 0.7 and the frequency of *a* is 0.3. We can again use the Punnett square, but including the frequencies as well:

	A( <i>p</i> )	a( <i>q</i> )
A( <i>p</i> )	AA( $p^2$ )	Aa( $pq$ )
a( <i>q</i> )	Aa( $pq$ )	aa( $q^2$ )

- Given the values for *p* and *q* and the Hardy–Weinberg equation we can calculate the frequency of each genotype as 0.49 for *AA* ( $0.7 \times 0.7$ ), 0.42 for *Aa* ( $2 \times 0.7 \times 0.3$ ), and 0.09 for *aa* ( $0.3 \times 0.3$ )
  - Note that the three frequencies add up to 1 ( $0.49 + 0.42 + 0.09 = 1$ )

Suppose, as described in the book, a population of 1000 individuals with this frequency of alleles each produce two gametes. You would get:

- 980 *A* gametes from the 490 *AA* individuals
- 420 *A* gametes from the 420 *Aa* individuals
- 420 *a* gametes from the 420 *Aa* individuals
- 180 *a* gametes from the 90 *aa* individuals

Of the total of 2000 gametes there are 1400 *A* and 600 *a*, for frequencies of 0.7 and 0.3 respectively, the same as the original allele frequency in the population

The 2000 gametes could combine randomly to give 1000 individuals

490 AA + 420 Aa + 180 aa, the same frequency as the first population

In principal, this could continue for any number of generations, with the same population reproducing itself each time.

- This situation assumes five things:
  - 1 No genes are undergoing mutation (changes in allele frequency)
  - 2 The population is very large (sampling error)
  - 3 The population is isolated (changes in allele frequency)
  - 4 All members survive and reproduce (no natural selection)
  - 5 Mating is random

These assumptions essentially never occur in nature, so that the Hardy-Weinberg rule is in actuality an ideal, that is a hypothetical, situation to which a natural population can be compared to assess the factors which bias allele frequency

### Forces that skew a population away from Hardy-Weinberg equilibrium

We will now look at the kinds of effects that can force changes in allele frequencies over time.

- Changes in allele frequency are referred to as “**microevolution**”
- Using this word implies that these changes underlie the much more drastic changes that we associate with evolution.

**Natural selection:** the book states that “natural selection probably accounts for more changes in allele frequency than any other microevolutionary process”

- Natural selection occurs when environmental conditions affect the ability of individuals to survive or reproduce depending on their genotype
- Natural selection can affect allele frequency in three ways:

**Directional selection:** in directional selection the shift in allele frequencies occurs in a consistent direction

- The classic example of this is the peppered moth:
  - The moths come in a dark and light variety
  - Before the industrial revolution the light variety predominated
  - The pollution of the industrial revolution killed the light colored lichens covering tree trunks
  - This made the dark variety the better camouflaged, and as a result they became the predominant type
  - Later as pollution was reduced, and lichens returned, the light variety again predominated
- Mark-release-recapture experiments proved that the dark moths survived better in the polluted environment, while the light moths did in a less polluted location
- A second good example of directional selection is the predominance of antibiotic resistant bacteria as a direct result of widespread use of antibiotics in the years following their discovery

**Stabilizing selection:** in this case, selection against extremes stabilizes a population with an intermediate phenotype

- The book gives the example of birth weight; both low and high birth weight babies suffer higher mortality, which may stabilize the population with the a more viable intermediate size

**Destabilizing selection:** contrastingly, selection can disfavor an intermediate phenotype, stabilizing a population with two extreme phenotypes

- The example of a population of finches with either large or small bills that were selected for by a limiting supply of a particular type of seeds

**Balanced polymorphism:** this occurs when selection tends to stabilize a situation in which multiple alleles are present

- An example of this is the trait responsible for sickle-cell anemia
  - A mutation of the gene encoding hemoglobin causes red blood cells to collapse into a sickle shape
  - Sickle-cell anemia is endemic in Central Africa
  - The presence of this trait is stabilized because heterozygotes are resistant to severe effects of the disease malaria
  - Since malaria is endemic in this region, its effect has tended to select for retention of the sickle-cell trait, despite the severe effects it has on homozygotes

### Random forces can change allele frequency

Several mechanisms that do not necessarily distinguish between genotypes can also cause microevolutionary allele changes:

**Mutation:** this is the most obvious of the forces altering allele frequencies

- Mutation is defined in the book as “a heritable change in DNA that can alter gene expression”
- Mutations need not change gene expression, or have an effect on phenotype; because of the redundancy of the genetic code, some mutations change the DNA sequence without changing the sequence of the protein expressed from a gene
- Other mutations alter both the DNA and the protein it expresses
- Of these, some are **neutral**, meaning they have no effect on the viability or ability to reproduce of the organism
- Others are **beneficial** meaning that they are advantageous to the organism
- Still others are either **harmful** or **lethal** because the change they cause in the protein is detrimental to the organism

**Genetic drift:** “random change in allele frequencies through the generations owing to chance events alone”

- Especially in small populations, the ideal of the Hardy-Weinberg rule is violated
- By chance the individuals carrying one allele may either die more than expected or reproduce less than expected, leading to a loss of that allele in the next generation
- In this case, the increase of the other allele is not because it is better adapted (or “fittest”) but simply random chance; we know that this effect can have a major role in evolution

There are two forms of genetic drift that are particularly important:

**The founder effect:** when a small number of individuals break off and form a separate population (perhaps in a new geographic location) the ratio of alleles in the new population may be very different from the larger previous population. This is essentially a sampling difference; the larger the sub-population the more likely it will resemble the parent population

**Genetic bottlenecks:** sometimes a population (or in the worst cases, a species) will be reduced to a very small number; the genetic diversity of the small population may be much less than the previous population, leading to a great change in allele frequency.

- Often endangered species are reduced to very small numbers (the Hawaiian nene or the African cheetah)
- Both species were reduced to very small numbers (“bottlenecks”)

- The diversity of alleles present in those small populations were much less than the previous population
- The loss of variation in the populations may negatively affect the ability of the species to survive future natural selection

**Gene flow:** this is distinct from “genetic drift” in that it refers to movement of alleles between adjacent populations

- This flow may be either into a population (“immigration”) or out (“emigration”)
- This effect makes the populations more similar, and reduces the chance that the allele frequencies in the two populations will become very dissimilar