CHAPTER 21

THE EVOLUTION OF POPULATIONS

OUTLINE

I. The modern evolutionary synthesis integrated Darwinism and Mendelism: *science as a process*

II. A population has a genetic structure defined by its gene pool's allele and genotype frequencies

III. The Hardy-Weinberg Theorem describes a nonevolving population

IV. Microevolution is a generation–to–generation change in a population's allele or genotype frequencies: *an overview*

V. Genetic drift can cause evolution via chance fluctuation in a small population's gene pool: *a closer look*
   A. Bottleneck Effect
   B. Founder Effect

VI. Gene flow can cause evolution by transferring alleles between populations: *a closer look*

VII. Mutations can cause evolution by substituting one allele for another in a gene pool: a closer look

VIII. Nonrandom mating can cause evolution by shifting the frequencies of genotypes in a gene pool: *a closer look*
   A. Inbreeding
   B. Assortative Mating

IX. Natural selection can cause evolution via differential reproductive success among varying members of a population: *a closer look*

X. Genetic variation is the substrate for natural selection
   A. How Extensive *Is* Genetic Variation Within and Between Populations?
   B. How *Is* Genetic Variation Generated?
   C. How *Is* Genetic Variation Preserved?
   D. Does All Genetic Variation Affect Survival and Reproductive Success?

XI. Natural selection is the mechanism of adaptive evolution
   A. Fitness
   B. What Does Selection Act On?
   C. Modes of Natural Selection
   D. Sexual Selection

XII. Does evolution fashion perfect organisms?

OBJECTIVES

After reading this chapter and attending lecture, the student should be able to:

1. Explain what is meant by the "modern synthesis".
2. Explain how microevolutionary change can affect a gene pool.
3. In their own words, state the Hardy-Weinberg theorem.
4. Write the general Hardy-Weinberg equation and use it to calculate allele and genotype frequencies.
5. Explain the consequences of Hardy-Weinberg equilibrium.
6. Demonstrate, with a simple example, that a disequilibrium population requires only one generation of random mating to establish Hardy-Weinberg equilibrium.
7. Describe the usefulness of the Hardy-Weinberg model to population geneticists.
8. List the conditions a population must meet in order to maintain Hardy-Weinberg equilibrium.
9. Explain how genetic drift, gene flow, mutation, nonrandom mating and natural selection can cause microevolution.
10. Explain the role of population size in genetic drift.
11. Distinguish between the bottleneck effect and the founder effect.
12. Explain why mutation has little quantitative effect on a large population.
13. Describe how inbreeding and assortative mating affect a population's allele frequencies and genotype frequencies.
14. Explain, in their own words, what is meant by the statement that natural selection is the only agent of microevolution which is adaptive.
15. Describe the technique of electrophoresis and explain how it has been used to measure genetic variation within and between populations.
16. List some factors that can produce geographical variation among closely related populations.
17. Explain why even though mutation can be a source of genetic variability, it contributes a negligible amount to genetic variation in a population.
18. Give the cause of nearly all genetic variation in a population.
19. Explain how genetic variation may be preserved in a natural population.
20. In their own words, briefly describe the neutral theory of molecular evolution and explain how changes in gene frequency may be nonadaptive.
21. Explain what is meant by "selfish" DNA.
22. Explain the concept of relative fitness and its role in adaptive evolution.

23. Explain why the rate of decline for a deleterious allele depends upon whether the allele is dominant or recessive to the more successful allele.
24. Describe what selection acts on and what factors contribute to the overall fitness of a genotype.
25. Give examples of how an organism's phenotype may be influenced by the environment.
26. Distinguish among stabilizing selection, directional selection and diversifying selection.
27. Define sexual dimorphism and explain how it can influence evolutionary change.
28. Give at least four reasons why natural selection cannot breed perfect organisms.

KEY TERMS

species
gene flow
gene pool
fixed allele
microevolution
Hardy-Weinberg equilibrium
nonadaptive
genetic drift
sampling error
bottleneck effect
founder effect
inbreeding
assortative mating
polygenic traits
morphs
polymorphic
electrophoresis
geographical variation
cline
step cline
point-mutation
supergenes
sexual recombination
balanced polymorphism
heterozygote advantage
sickle-cell anemia
frequency-dependent selection
Batesian mimicry
neutral variation
selfish DNA
adaptive evolution
relative fitness
selection coefficient
pleiotropy
coadapted gene complex
norm of reaction
stabilizing selection
directional selection
diversifying selection
sexual dimorphism
sexual selection

LECTURE NOTES

Natural selection works on individuals, but it is the *population* that evolves. Darwin understood this, but was unable to determine its genetic basis.

I. The modern evolutionary synthesis integrated Darwinism and Mendelism: *science as a process*

Shortly after the publication of *The Origin of Species*, most biologists were convinced that species evolved. Darwin was less successful in convincing them that natural selection was the mechanism for evolution, because little was known about inheritance.

- An understanding about inheritance was necessary to explain:
  - ⇒ How chance variations arise in populations.
  - ⇒ How these variations are precisely transmitted from parents to offspring.
- Though Gregor Mendel was a contemporary of Darwin's, Mendel's principles of inheritance went unnoticed until the early 1900's.

For Darwin, the raw material for natural selection was variation in quantitative characters that vary along a continuum in a population.

- We now know that continuous variation is usually determined by many segregating loci (polygenic inheritance).
- As did Mendel, geneticists in the early 1900's recognized only discrete characters inherited on an either-or basis. Thus, for them, there appeared to be no genetic basis for the subtle variations that were central to Darwin's theory.

During the 1920's, genetic research focused on mutations, and a widely accepted alternative to Darwin's theory was that evolution occurs in rapid leaps as a result of radical phenotypic changes caused by mutations.

- This idea contrasted with Darwin's view of gradual evolution due to environmental selection acting on continuous variation among individuals of a population.
- Another popular theory was *orthogenesis*, the idea that evolution has been a predictable progression to more and more elite forms of life.

In the 1930's, the science of *population genetics* emerged, which:

- Emphasized genetic variation within populations and recognized the importance of quantitative characters.
- Was an important turning point for evolutionary theory, because it reconciled Mendelian genetics with Darwinian evolution.

In the 1940's, the genetic basis of variation and natural selection was worked out, and *the modern synthesis* was formulated. This comprehensive theory:

- Integrated discoveries from different fields (i.e. paleontology, taxonomy, biogeography, and population genetics).
- Was collectively developed by many scientists including:
  - ⇒ Theodosius Dobzhansky – geneticist
  - ⇒ Ernst Mayr – biogeographer and systematist
  - ⇒ George Gaylord Simpson – paleontologist
  - ⇒ G. Ledyard Stebbins – botanist
- Emphasized the following:
  - ⇒ Importance of populations as units of evolution.
  - ⇒ The central role of natural selection as the primary mechanism of evolutionary change.
  - ⇒ Gradualism as the explanation of how large changes can result from an accumulation of small changes occurring over long periods of time.

Most of Darwin's ideas persist in the modern synthesis although many evolutionists are challenging some generalizations of the modern synthesis.

- This debate focuses on the rate of evolution and on the relative importance of evolutionary mechanisms other than natural selection.
- These debates do not question the fact of evolution, only what mechanisms are most important in the process.
- Such disagreements indicate that the study of evolution is very lively and that it continues to develop as a science.

II. A population has a genetic structure defined by its gene pool's allele and genotype frequencies

Population = Localized group of organisms which belong to the same *species*.

Species = Groups of actually or potentially interbreeding natural populations, which are reproductively isolated from other such groups.

Most species are not evenly distributed over a geographical range, but are concentrated in several localized population centers.

- Each population center is isolated to some extent from other population centers with only occasional gene flow among these groups.
- Obvious examples are isolated populations found on widely separated islands or in unconnected lakes.
- Some populations are not separated by such sharp boundaries.
  - ⇒ For example, a species with two population centers may be connected by an intermediate sparsely populated range.
  - ⇒ Even though these two populations are not absolutely isolated, individuals are more likely to interbreed with others from their population center. Gene flow between the two population centers is thus reduced by the intermediate range.

Gene pool = The total aggregate of genes in a population at any one time.

- Consists of all the alleles at all gene loci in all individuals of a population. Alleles from this pool will be combined to produce the next generation.
- In a diploid species, an individual may be homozygous or heterozygous for a locus since each locus is represented twice.
- An allele is said to be *fixed* in the gene pool if all members of the population are homozygous for that allele.
- Normally there will be two or more alleles for a gene, each having a relative frequency in the gene pool.

III. The Hardy-Weinberg Theorem describes a nonevolving population

NOTE: The Hardy-Weinberg model is so much easier to teach if the students calculate gene frequencies along with the instructor. This means that you must pause frequently to allow plenty of time for students to actively process the information and practice the calculations.

In the absence of other factors, the segregation and recombination of alleles during meiosis and fertilization will not alter the overall genetic makeup of a population.

- The frequencies of alleles in the gene pool will remain constant unless acted upon by other agents; this is known as the *Hardy-Weinberg Theorem*.
- The Hardy-Weinberg model describes the genetics of nonevolving populations. This theorem can be tested with theoretical population models.

To test the Hardy-Weinberg theorem, imagine an isolated population of wildflowers with the following characteristics: (See Campbell, Figure 21.3)

- It is a diploid species with both pink and white flowers.
- The population size is 500 plants: 480 plants have pink flowers, 20 plants have white flowers.
- Pink flower color is coded for by the dominant allele "A," white flower color is coded for by the recessive allele "a."
- Of the 480 pink-flowered plants, 320 are homozygous (AA) and 160 are heterozygous (Aa). Since white color is recessive, all white flowered plants are homozygous aa.
- There are 1000 genes for flower color in this population, since each of the 500 individuals has two genes (this is a diploid species).
- A total of 320 genes are present in the 160 heterozygotes (Aa): half are dominant (160 A) and half are recessive (160 a).
- 800 of the 1000 total genes are dominant.
- The frequency of the A allele is 80% or 0.8 (800/1000).

Genotypes	# of plants		# of A alleles per individual		Total # A alleles
AA plants	320	×	2	=	640
Aa plants	160	×	1	=	160
					800

- 200 of the 1000 total genes are recessive.
- The frequency of the a allele is 20% or 0.2 (200/1000).

Genotypes	# of plants		# of A alleles per individual		Total # A alleles
aa plants	20	×	2	=	40
Aa plants	160	×	1	=	160
					200

Assuming that mating in the population is completely random (all male-female mating combinations have equal chances), the frequencies of A and a will remain the same in the next generation.

- Each gamete will carry one gene for flower color, either A or a.
- Since mating is random, there is an 80% chance that any particular gamete will carry the A allele and a 20% chance that any particular gamete will carry the a allele.

The frequencies of the three possible genotypes of the next generation can be calculated using the rule of multiplication: (See Campbell, Chapter 13)

- The probability of two A alleles joining is $0.8 \times 0.8 = 0.64$; thus, 64% of the next generation will be AA.
- The probability of two a alleles joining is $0.2 \times 0.2 = 0.04$; thus, 4% of the next generation will be aa.
- Heterozygotes can be produced in two ways, depending upon whether the sperm or ovum contains the dominant allele (Aa or aA). The probability of a heterozygote being produced is thus $(0.8 \times 0.2) + (0.2 \times 0.8) = 0.16 + 0.16 = 0.32$.

The frequencies of possible genotypes in the next generation are 64% AA, 32% Aa and 4% aa.

- The frequency of the A allele in the new generation is $0.64 + (0.32/2) = 0.8$, and the frequency of the a allele is $0.04 + (0.32/2) = 0.2$. Note that the alleles are present in the gene pool of the new population at the *same* frequencies they were in the original gene pool.
- Continued sexual reproduction with segregation, recombination and random mating would *not alter* the frequencies of these two alleles: the gene pool of this population would be in a state of equilibrium referred to as *Hardy-Weinberg equilibrium*.
- If our original population had not been in equilibrium, only one generation would have been necessary for equilibrium to become established.

From this theoretical wildflower population, a general formula, called the *Hardy-Weinberg equation*, can be derived to calculate allele and genotype frequencies.

- The Hardy-Weinberg equation can be used to consider loci with three or more alleles.
- By way of example, consider the simplest case with only two alleles with one dominant to the other.
- In our wildflower population, let p represent allele A and q represent allele a, thus $p = 0.8$ and $q = 0.2$.
- The sum of frequencies from all alleles must equal 100% of the genes for that locus in the population: $p + q = 1$.
- Where only two alleles exist, only the frequency of one must be known since the other can be derived:

$$1 - p = q \quad \text{or} \quad 1 - q = p$$

When gametes fuse to form a zygote, the probability of producing the AA genotype is p^2; the probability of producing aa is q^2; and the probability of producing an Aa heterozygote is 2pq (remember heterozygotes may be formed in two ways).

- The sum of these frequencies must equal 100%, thus:

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

The Hardy-Weinberg equation permits the calculation of allelic frequencies in a gene pool, if the genotype frequencies are known. Conversely, the genotype can be calculated from known allelic frequencies.

For example, the Hardy-Weinberg equation can be used to calculate the frequency of inherited diseases in humans (e.g. phenylketonuria):

- 1 of every 10,000 babies in the United States is born with phenylketonuria (PKU) which can result in mental retardation if untreated.
- The allele for PKU is recessive, so babies with this disorder are homozygous recessive = q^2.
- Thus $q^2 = 0.0001$, with $q = 0.01$ (the square root of 0.0001).
- The frequency of p can be determined since $p = 1 - q$:

$$p = 1 - 0.01 = 0.99$$

- The frequency of carriers (heterozygotes) in the population is 2pq.

$$2pq = 2(0.99)(0.01) = 0.0198$$

- Thus, about 2% of the U.S. population are carriers for PKU.

IV. Microevolution is a generation–to–generation change in a population's allele or genotype frequencies: *an overview*

The Hardy-Weinberg equilibrium is important to the study of evolution since it tells us what will happen in a *nonevolving* population.

- This equilibrium model provides a base line from which evolutionary departures take place.
- It provides a reference point with which to compare the frequencies of alleles and genotypes of natural populations whose gene pools may be changing.

For Hardy-Weinberg equilibrium to be maintained, five conditions *must* be met:

1. *Very large population size*.
2. *Isolation from other populations*. There is no migration of individuals into or out of the population.
3. *No mutations*.
4. *Random mating*.
5. *No natural selection*. All genotypes are equal in survival and reproductive success. Differential reproductive success can alter gene frequencies.

In real populations, several factors can upset Hardy-Weinberg equilibrium and cause *microevolutionary* change.

Microevolution = Small scale evolutionary change represented by a generational shift in a population's relative allelic frequencies.

- Microevolution can be caused by *genetic drift*, *gene flow*, *mutation*, *nonrandom mating*, and *natural selection*; each of these conditions is a deviation from the criteria for Hardy-Weinberg equilibrium.
- Of these five possible agents for microevolution, only natural selection generally leads to an accumulation of favorable adaptations in a population.
- The other four are nonadaptive and are usually called non-Darwinian changes.

V. Genetic drift can cause evolution via chance fluctuation in a small population's gene pool: *a closer look*

Genetic drift = Changes in the gene pool of a small population due to chance.

- If a population is small, stochastic events have a greater impact on gene frequencies.
- Chance events may cause the frequencies of alleles to drift randomly from generation to generation, since the existing gene pool may not be accurately represented in the next generation.

For example, assume our theoretical wildflower population contains only 25 plants, and the genotypes for flower color occur in the following numbers: 16 AA, 8 Aa and 1 aa. In this case, a chance event could easily change the frequencies of the two alleles for flower color.

- A rock slide or passing herbivore which destroys three AA plants would immediately change the frequencies of the alleles from A = 0.8 and a = 0.2, to A = 0.77 and a = 0.23.
- Although this change does not seem very drastic, the frequencies of the two alleles were changed by a chance event.

The larger the population, the less important is the effect of genetic drift.

- Even though natural populations are not infinitely large (in which case genetic drift could be completely eliminated as a cause of microevolution), most are so large that the effect of genetic drift is negligible.
- However, some populations are small enough that genetic drift can play a major role in microevolution, especially when the population has less than 100 individuals.

Two situations which result in populations small enough for genetic drift to be important are the *bottleneck effect* and the *founder effect*.

A. Bottleneck Effect

The size of a population may be reduced drastically by such natural disasters as volcanic eruptions, earthquakes, fires, floods, etc. which kill organisms nonselectively.

- The small surviving population is unlikely to represent the genetic makeup of the original population.
- Genetic drift which results from drastic reduction in population size is referred to as the *bottleneck effect*.

- By chance some individuals survive. In the small remaining population, some alleles may be overrepresented, some under-represented, and some alleles may be totally absent.
- Genetic drift which has occurred may continue to affect the population for many generations, until it is large enough for random drift to be insignificant.

The bottleneck effect reduces overall genetic variability in a population since some alleles may be entirely absent.

- For example, a population of northern elephant seals was reduced to just 20 individuals by hunters in the 1890's.
  - ⇒ Since this time, these animals have been protected and the population has increased to about 30,000 animals.
  - ⇒ Researchers have found that no variation exists in the 24 loci examined from the present population. A single allele has been fixed at each of the 24 loci due to genetic drift by the bottleneck effect.
  - ⇒ This contrasts sharply with the large amount of genetic variation found in southern elephant seal populations which did not undergo the bottleneck effect.
- A lack of genetic variation in South African cheetahs may also have resulted from genetic drift, since the large population was severely reduced during the last ice age and again by hunting to near extinction.

B. Founder Effect

When a few individuals colonize a new habitat, genetic drift is also likely to occur. Genetic drift in a new colony is called the *founder effect*.

- The smaller the founding population, the less likely its gene pool will be representative of the original population's genetic makeup.
- The most extreme example would be when a single seed or pregnant female moves into a new habitat.
- If the new colony survives, random drift will continue to affect allele frequencies until the population reaches a large enough size for its influence to be negligible.
- No doubt, the founder effect was instrumental in the evolutionary divergence of the Galapagos finches.

The founder effect probably resulted in the high frequency of *retinitis pigmentosa* (a progressive form of blindness that affects humans homozygous for this recessive allele) in the human population of Tristan da Cunha, a group of small Atlantic islands.

- This area was colonized by 15 people in 1814, and one must have been a carrier.
- The frequency of this allele is much higher on this island than in the populations from which the colonists came.

Although inherited diseases are obvious examples of the founder effect, this form of genetic drift can alter the frequencies of any alleles in the gene pool.

VI. Gene flow can cause evolution by transferring alleles between populations: *a closer look*

Gene flow = The migration of fertile individuals, or the transfer of gametes, between populations.

- Natural populations may gain or lose alleles by gene flow, since they do not have gene pools which are closed systems required for Hardy-Weinberg equilibrium.
- Gene flow tends to reduce between-population differences which have accumulated by natural selection or genetic drift.
- An example of gene flow would be if our theoretical wildflower population was to begin receiving wind blown pollen from an all white-flower population in a neighboring field. This new pollen could greatly increase the frequency of the white flower allele, thus also altering the frequency of the red flower allele.
- Extensive gene flow can eventually group neighboring populations into a single population.

VII. Mutations can cause evolution by substituting one allele for another in a gene pool: *a closer look*

A new mutation which is transmitted in gametes immediately changes the gene pool of a population by substituting one allele for another.

In our theoretical wildflower population, if a mutation in a white flowered plant caused that plant to begin producing gametes which carried a red flower allele, the frequency of the white flower allele is reduced and the frequency of the red flower allele is increased.

Mutation itself has little quantitative effect on large populations in a single generation, since mutation at any given locus is very rare.

- Mutation rates of one mutation per 10^5 to 10^6 gametes are typical, but vary depending on the species and locus.
- An allele with a 0.50 frequency in the gene pool that mutates to another allele at a rate of 10^{-5} mutations per generation would take 2000 generations to reduce the frequency of the original allele from 0.50 to 0.49.
- The gene pool is effected even less, since most mutations are reversible.
- If a new mutation increases in frequency, it is because individuals carrying this allele are producing a larger percentage of offspring in the population due to genetic drift or natural selection, not because mutation is producing the allele in abundance.

Mutation is important to evolution since it is the original source of genetic variation, which is the raw material for natural selection.

VIII. Nonrandom mating can cause evolution by shifting the frequencies of genotypes in a gene pool: *a closer look*

Nonrandom mating increases the number of homozygous loci in a population, but does not in itself alter frequencies of alleles in a population's gene pool. There are two kinds of nonrandom mating: *inbreeding* and *assortative mating*.

A. Inbreeding

Individuals of a population usually mate with close neighbors rather than with more distant members of a population, especially if the members of the population do not disperse widely.

- This violates the Hardy-Weinberg criteria that an individual must choose its mate at random from the population.
- Since neighboring individuals of a large population tend to be closely related, inbreeding is promoted.
- Self-fertilization, which is common in plants, is the most extreme example of inbreeding.

Inbreeding results in relative genotypic frequencies (p^2, 2pq, q^2) that deviate from the frequencies predicted for Hardy-Weinberg equilibrium, but does not in itself alter frequencies of alleles (p and q) in the gene pool.

Self-fertilization in our theoretical wildflower population would increase the frequencies of homozygous individuals and reduce the frequency of heterozygotes.

- Selfing of AA and aa individuals would produce homozygous plants.
- Selfing of Aa plants would produce half homozygotes and half heterozygotes.
- Each new generation would see the proportion of heterozygotes decrease, while the proportions of homozygous dominant and homozygous recessive plants would increase.
- Inbreeding without selfing would also result in a reduction of heterozygotes, although it would take much longer.

One effect of inbreeding is that the frequency of homozygous recessive phenotypes increases.

An interesting thing to note is that even if the phenotypic and genotypic ratios change, the values of p and q do not change in these situations, only the way they are combined. A smaller proportion of recessive alleles are masked by the heterozygous state.

B. Assortative Mating

Assortative mating is another type of nonrandom mating which results when individuals mate with partners that are like themselves in certain phenotypic characters. For example:

- Snow geese occur in a blue variety and a white variety, with the blue color allele being dominant. Birds prefer to mate with those of their own color, blue with blue and white with white; this results in a lower frequency of heterozygotes than predicted by Hardy-Weinberg.
- Blister beetles (*Lytla magister*) in the Sonoran Desert usually mate with a same-size individual.

IX. Natural selection can cause evolution via differential reproductive success among varying members of a population: *a closer look*

The Hardy-Weinberg equilibrium condition that all individuals in a population have equal ability to produce viable, fertile offspring is probably never met.

- In any sexually reproducing population, variation among individuals exists and some variants leave more offspring than others.
- *Natural selection* is this differential success in reproduction.

Due to selection, alleles are passed on to the next generation in disproportionate numbers relative to their frequencies in the present generation.

- If in our theoretical wildflower population, white flowers are more visible to herbivores than pink flowers, plants with pink flowers (both AA and Aa) would leave more offspring on the average.
- Genetic equilibrium would be disturbed and the frequency of allele A would increase and the frequency of the a allele would decrease.

Natural selection is the only agent of microevolution which is adaptive, since it accumulates and maintains favorable genotypes.

- Environmental change would result in selection favoring genotypes present in the population which can survive the new conditions.
- Variability in the population makes it possible for natural selection to occur.

X. Genetic variation is the substrate for natural selection

Members of a population may vary in subtle or obvious ways. It is the genetic basis of this variation that makes natural selection possible.

A. How Extensive *Is* Genetic Variation Within and Between Populations?

Darwin considered the slight differences between individuals of a population as raw material for natural selection.

While we are more conscious of the variation among humans, an equal if not greater amount of variation exists among the many plant and animals species.

- Phenotypic variation is a product of inherited genotype and numerous environmental influences.
- Only the genetic or inheritable component of variation can have adaptive impact as a result of natural selection.

Polygenic characters which vary quantitatively within a population are responsible for much of the inheritable variation.

- For example, the height of the individuals in our theoretical wildflower population may vary from very short to very tall with all sorts of intermediate heights.

Discrete characters which are determined by only one locus vary categorically, such as flower color in our wildflowers, without intermediates.

- In our wildflower population, the red and white flowers would be referred to as different *morphs* (contrasting forms of a Mendelian character).
- A population is referred to as *polymorphic* for a character if two or more morphs are present in noticeable frequencies. (See Campbell, Figure 21.8)
- Polymorphism is found in human populations not only in physical characters (e.g. presence or absence of freckles) but also in biochemical characters (e.g. ABO blood group).

Darwin did not realize the extent of genetic variation in populations, since much of the genetic variation can only be determined with biochemical methods.

- Electrophoresis has been used to determine genetic variation among individuals of a population. This technique allows researchers to identify variations in protein products of specific gene loci.
- Electrophoretic studies show that in *Drosophila* populations the gene pool has two or more alleles for about 30% of the loci examined, and each fly is heterozygous at about 12% of its loci.
- Thus, there are about 700 – 1200 heterozygous loci in each fly. Any two flies in a *Drosophila* population will differ in genotype at about 25% of their loci.
- Electrophoretic studies also show comparable variation in the human population.

Note that electrophoresis underestimates genetic variation:

- Proteins produced by different alleles may vary in amino acid composition and still have the same overall charge, which makes them indistinguishable by electrophoresis.
- Also, DNA variation not expressed as protein is not detected by electrophoresis.

Geographical variation in allele frequencies exists among populations of most species.

- Natural selection can contribute to geographical variation, since at least some environmental factors are different between two locales. For example, one population of our wildflowers may have a higher frequency of white flowers because of the prevalence in that area of pollinators that prefer white flowers.
- Genetic drift may cause chance variations among different populations.
- Also, subpopulations may appear within a population due to localized inbreeding resulting from a "patchy" environment.

Cline = One type of geographical variation that is a graded change in some trait along a geographic transect.

- Clines may result from a gradation in some environmental variable.
- It may be a graded region of overlap where individuals of neighboring populations interbreed.
- For example, the average body size of many North American mammal species gradually increases with increasing latitude. It is presumed that the reduced surface area to volume ratio associated with larger size helps animals in cold environments conserve body heat.
- Studies of geographical variation confirm that genetic variation affects spatial differences of phenotypes in some clines. For example, yarrow plants are shorter at higher elevations, and some of this phenotypic variation has a genetic basis. (See Campbell, Figure 21.9)

B. How *Is* Genetic Variation Generated?

Genetic variation results from mutation and sexual recombination.

1. Mutation

Mutations produce new alleles. They are rare and random events which usually occur in somatic cells and are thus not inheritable.

- Only mutations that occur in cell lines which will produce gametes can be passed to the next generation.
- Geneticists estimate that only an average of one or two mutations occur in each human gamete-producing cell line.

Point mutation = Mutation affecting a single base in DNA.

- Much of the DNA in eukaryotes does not code for proteins, and it is uncertain how a point mutation in these regions affect an organism.
- Point mutations in structural genes may cause little effect, partly due to the redundancy of the genetic code.

Mutations that alter a protein enough to affect the function are more often harmful than beneficial, since organisms are evolved products shaped by selection and a chance change is unlikely to improve the genome.

- Occasionally, a mutant allele is beneficial, which is more probable when environmental conditions are changing.
- The mutation which allowed house flies to be resistant to DDT was present in the population and under normal conditions resulted in reduced growth rate. It became beneficial to the house fly population only after a new environmental factor (DDT) was introduced and tipped the balance in favor of the mutant alleles.

Chromosomal mutations usually affect many gene loci and tend to disrupt an organism's development.

- On rare occasions, chromosomal rearrangement may be beneficial. These instances (usually by translocation) may produce a cluster of genes with cooperative functions when inherited together.

Duplication of chromosome segments is nearly always deleterious.

- If the repeated segment does not severely disrupt genetic balance, it may persist for several generations and provide an expanded genome with extra loci.
- These extra loci may take on new functions by mutation while the original genes continue to function.
- Shuffling of exons within the genome (single locus or between loci) may also produce new genes.

Mutation can produce adequate genetic variation in bacteria and other microorganisms which have short generation times.

- Some bacteria reproduce asexually by dividing every 20 minutes, and a single cell can produce a billion descendants in only 10 hours.
- With this type of reproduction, a beneficial mutation can increase in frequency in a bacterial population very rapidly.
- A bacterial cell with a mutant allele which makes it antibiotic resistant could produce an extremely large population of clones in a short period, while other cells without that allele are eliminated.
- Although bacteria reproduce primarily by asexual means, most increase genetic variation by occasionally exchanging and recombining genes through processes such as conjugation, transduction and transformation.

2. Recombination

The contribution of mutations to genetic variation is negligible.

- Mutations are so infrequent at a single locus that they have little effect on genetic variation in a large gene pool.
- Although mutations produce new alleles, nearly all genetic variation in a population results from new combinations of alleles produced by sexual recombination.

Gametes from each individual vary extensively due to crossing over and random segregation during meiosis.

- Thus, each zygote produced by a mating pair possesses a unique genetic makeup.
- Sexual reproduction produces new combinations of old alleles each generation.

Plants and animals depend almost entirely on sexual recombination for genetic variation which makes adaptation possible.

C. How *Is* Genetic Variation Preserved?

Natural selection tends to produce genetic uniformity in a population by eliminating unfavorable genotypes. This tendency is opposed by several mechanisms that preserve or restore variation.

1. Diploidy

Diploidy hides much genetic variation from selection by the presence of recessive alleles in heterozygotes.

- Since recessive alleles are not expressed in heterozygotes, less favorable or harmful alleles may persist in a population.
- This variation is only exposed to selection when two heterozygotes mate and produce offspring homozygous for the recessive allele.
- If a recessive allele has a frequency of 0.01 and its dominant counterpart 0.99, then 99% of the recessive allele copies will be protected in heterozygotes. Only 1% of the recessive alleles will be present in homozygotes and exposed to selection.
- The more rare the recessive allele, the greater its protection by heterozygosity. That is, a greater proportion are hidden in heterozygotes by the dominant allele.
- This type of protection maintains a large pool of alleles which may be beneficial if conditions change.

2. Balanced Polymorphism

Selection may also preserve variation at some gene loci.

Balanced polymorphism = The ability of natural selection to maintain diversity in a population.

One mechanism by which selection preserves variation is *heterozygote advantage*.

- Natural selection will maintain two or more alleles at a locus if heterozygous individuals have a greater reproductive success than any type of homozygote.
- An example is the recessive allele that causes sickle-cell anemia in homozygotes. The locus involved codes for one chain of hemoglobin.
- Homozygotes for this recessive allele develop sickle-cell anemia which is often fatal.
- Heterozygotes are resistant to malaria. Heterozygotes thus have an advantage in tropical areas where malaria is prevalent, since homozygotes for the dominant allele are susceptible to malaria and homozygous recessive individuals are incapacitated by the sickle-cell condition.
- In some African tribes from areas where malaria is common, 20% of the hemoglobin loci in the gene pool is occupied by the recessive allele.

Other examples of heterozygote advantage are found in crop plants (e.g. corn) where inbred lines become homozygous at more loci and show stunted growth and sensitivity to diseases.

- Crossbreeding different inbred varieties often produces hybrids which are more vigorous than the parent stocks.
- This *hybrid vigor* is probably due to:
  1. Segregation of harmful recessives that were homozygous in the inbred varieties.
  2. Heterozygote advantage at many loci in the hybrids.

Balanced polymorphism can also result from patchy environments where different phenotypes are favored in different subregions of a populations geographic range.

Frequency-dependent selection also causes balanced polymorphism.

- In this situation the reproductive success of any one morph declines if that phenotype becomes too common in the population.
- For example, in *Papilio dardanus*, an African swallowtail butterfly, males have similar coloration but females occur in several morphs.
- The female morphs resemble other butterfly species which are noxious to bird predators. *Papilio* females are not noxious, but birds avoid them because they look like distasteful species.
- This type of protective coloration (*Batesian mimicry*) would be less effective if all the females looked like the same noxious species, because birds would encounter good-tasting mimics as often as noxious butterflies and would not associate a particular color pattern with bad taste.

D. Does All Genetic Variation Affect Survival and Reproductive Success?

Some genetic variations found in populations confer no selective advantage or disadvantage. They have little or no impact on reproductive success. This type of variation is called *neutral variation*.

- Much of the protein variation found by electrophoresis is adaptively neutral.
- For example, 99 known mutations affect 71 of 146 amino acids in the beta hemoglobin chain in humans. Some, like the sickle-cell anemia allele, affect the reproductive potential of an individual, while others have no obvious effect.
- The *neutral theory* of molecular evolution states than many variant alleles at a locus may confer no selective advantage or disadvantage.
- Natural selection would not affect the relative frequencies of neutral variations. Frequency of some neutral alleles will increase in the gene pool and others will decrease *due the chance effects of genetic drift*.

Variation in DNA which does not code for proteins may also be nonadaptive.

- Most eukaryotes contain large amounts of DNA in their genomes which have no known function. Such noncoding DNA can be found in varying amounts in closely related species.

- Some scientists speculate that noncoding DNA has resulted from the inherent capacity of DNA to replicate itself and has expanded to the tolerance limits of the each species. The entire genome could exist as a consequence of being self-replicating rather than by providing an adaptive advantage to the organism.
- Transposons might fit this definition of "selfish DNA," although the degree of influence these sequences have on the evolution of genomes is not known.

Evolutionary biologists continue to debate how much variation, or even whether variation, is neutral.

- It is easy to show that an allele is deleterious to an organism.
- It is not easily shown that an allele provides no benefits, since such benefits may occur in immeasurable ways.
- Also, a variation may appear to be neutral under one set of environmental conditions and not neutral under other conditions.

We cannot know how much genetic variation is neutral, but if even a small portion of a population's genetic variation significantly affects the organisms, there is still a tremendous amount of raw material for natural selection and adaptive evolution.

XI. Natural selection is the mechanism of adaptive evolution

Adaptive evolution results from a combination of both:

- Chance events that produce new genetic variation (e.g. mutation and sexual recombination).
- Natural selection that favors propagation of some variations over others.

A. Fitness

Darwinian fitness is measured by the relative contribution an individual makes to the gene pool of the next generation.

- It is not a measure of physical and direct confrontation, but of the success of an organism in producing progeny.
- Organisms may produce more progeny because they are more efficient feeders, attract more pollinators (as in our wildflowers), avoid predators, etc.

Survival does not guarantee reproductive success, since a sterile organism may outlive fertile members of the population.

- A long life span may increase fitness if the organism reproduces over a longer period of time (thus leaving more offspring) than other members of the population.
- Even if all members of a population have the same life span, those that mature early and thus have a longer reproductive time span, have increased their fitness.
- Every aspect of survival and fecundity are components of fitness.

Relative fitness = The contribution of a genotype to the next generation compared to the contributions of alternative genotypes for the same locus.

- For example, if pink flower plants (AA and Aa) in our wildflower population produce more offspring than white flower plants (aa), then AA and Aa genotypes have a higher relative fitness.

Statistical estimates of fitness can be produced by the relative measure of selection *against* an inferior genotype. This measure is called the *selection coefficient*.

- For comparison, relative fitness of the most fecund variant (AA or Aa in our wildflower population) is set at 1.0.
- If white flower plants produce 80% as many progeny on average, then the white variant relative fitness is 0.8.
- The selection coefficient is the difference between these two values (1.0 – 0.8 = 0.2).
- The more disadvantageous the allele, the greater the selection coefficient.
- Selection coefficients can range up to 1.0 for a lethal allele.

The rate of decline in relative frequencies of deleterious alleles in a population depends on the magnitude of the selection coefficient working against it and whether the allele is dominant or recessive to the more successful allele.

- Deleterious recessives are normally protected from elimination by heterozygote protection.
- Selection against harmful dominant alleles is faster since they are expressed in heterozygotes.

The rate of increase in relative frequencies of beneficial alleles is also affected by whether it is a dominant or recessive.

- New recessive mutations spread slowly in a population (even if beneficial) because selection can not act in its favor until the mutation is common enough for homozygotes to be produced.
- New dominant mutations that are beneficial increase in frequency faster since even heterozygotes benefit from the allele's presence (for example, the mutant dark color producing allele in peppered moths).

Most new mutations, whether harmful or beneficial, probably disappear from the gene pool early due to genetic drift.

B. What Does Selection Act On?

Selection acts on phenotypes, indirectly adapting a population to its environment by increasing or maintaining favorable genotypes in the gene pool.

- Since it is the phenotype (physical traits, metabolism, physiology, and behavior) which is exposed to the environment, selection can only act indirectly on genotypes.

The connection between genotype and phenotype may not be as simple and definite as with our wildflower population where pink was dominant to white.

- *Pleiotropy* (the ability of a gene to have multiple effects) often clouds this connection. The overall fitness of a genotype depends on whether its beneficial effects exceed any harmful effects on the organism's reproductive success.
- Polygenic traits also make it difficult to distinguish the phenotype-genotype connection. Whenever several loci influence the same characteristic, the members of the population will not fit into definite categories, but represent a continuum along a range.

An organism is an integrated composite of many phenotypic features, and the fitness of a genotype at any one locus depends upon the entire genetic context. A number of genes may work cooperatively to produce related phenotypic traits.

C. Modes of Natural Selection

The frequency of a heritable characteristic in a population may be affected in one of three different ways by natural selection, depending on which phenotypes are favored. (See Campbell, Figure 21.12)

1. Stabilizing Selection

Stabilizing selection favors intermediate variants by selecting against extreme phenotypes.

- The trend is toward reduced phenotypic variation and greater prevalence of phenotypes best suited to relatively stable environments.
- For example, human birth weights are in the 3 – 4 kg range. Much smaller and much higher birth weight babies have a greater infant mortality.

2. Directional Selection

Directional selection favors variants of one extreme. It shifts the frequency curve for phenotypic variations in one direction toward rare variants which deviate from the average of that trait.

- This is most common when members of a species migrate to a new habitat with different environmental conditions or during periods of environmental change.
- For example, fossils show the average size of European black bears increased after periods of glaciation, only to decrease during warmer interglacial periods.

3. Diversifying Selection

In diversifying selection, opposite phenotypic extremes are favored over intermediate phenotypes.

- This occurs when environmental conditions are variable in such a way that extreme phenotypes are favored.
- For example, balanced polymorphism of *Papilio* where butterflies with characteristics between two noxious model species (thus not favoring either) gain no advantage from their mimicry.

D. Sexual Selection

Sexual dimorphism = Distinction between the secondary sexual characteristics of males and females.

- Often seen as differences in size, plumage, lion's manes, deer antlers, or other adornments in males.
- In vertebrates it is usually the male that is the "showier" sex.
- In some species, males use their secondary sexual characteristics in direct competition with other males to obtain female mates (especially where harem building is common). These males may defeat other males in actual combat, but more often they use ritualized displays to discourage male competitors.

Darwin viewed sexual selection as a separate selection process leading to sexual dimorphism.

- These enhanced secondary sexual characteristics usually have no adaptive advantage other than attracting mates.
- However, if these adornments increase a males ability to attract more mates, his reproductive success is increased and he contributes more to the gene pool of the next generation.

The evolutionary outcome is usually a compromise between natural selection and sexual selection.

- In some cases the line between these two types of selection is not distinct, as in male deer.
- A stag may use his antlers to defend himself from predators and also to attract females.

XII. Does Evolution Fashion Perfect Organisms?

Natural selection cannot breed perfect organisms because:

1. *Organisms are locked into historical constraints*. Each species has a history of descent with modification from ancestral forms.
   - Natural selection modifies existing structures and adapts them to new situations, it does not rebuild organisms.
   - For example, back problems suffered by some humans are in part due to the modification of a skeleton and musculature from the anatomy of four-legged ancestors which are not fully compatible to upright posture.
2. *Adaptations are often compromises*.
   - Each organism must be versatile enough to do many different things.
   - For example, seals spend time in the water and on rocks; they could walk better with legs, but swim much better with flippers.
   - Prehensile hands and flexible limbs allow humans to be very versatile and athletic, but they also make us prone to sprains, torn ligaments, and dislocations. Structural

reinforcement would prevent many of these disabling occurrences but would limit agility.

3. *Not all evolution is adaptive*.
    - Genetic drift probably affects the gene pool of populations to a large extent.
    - Alleles which become fixed in small populations formed by the founder effect may not be better suited for the environment than alleles that are eliminated.
    - Similarly, small surviving populations produced by bottleneck effect may be no better adapted to the environment or even less well adapted than the original population.
4. *Selection can only edit variations that exist*.
    - These variations may not represent ideal characteristics.
    - New genes are not formed by mutation on demand.

These limitations thus allow natural selection to operate on a "better than" basis and subtle imperfections are the best evidence for evolution.

REFERENCES

Campbell, N. *Biology*. 4th ed. Menlo Park, California: Benjamin/Cummings, 1996.

Doolittle, W.F. and C. Sapienza. "Selfish Genes, the Phenotype Paradigm and Genome Evolution." *Nature*, 284: 601-603, 1980.

Mettler, L.E., T.G. Gregg, and H.E. Schaffer. *Population Genetics and Evolution*. 2nd ed. Englewood Cliffs, New Jersey: Prentice Hall, 1988.

Milkman, R. *Perspectives on Evolution*. Sinauer, Massachusetts: Sunderland, 1982.