

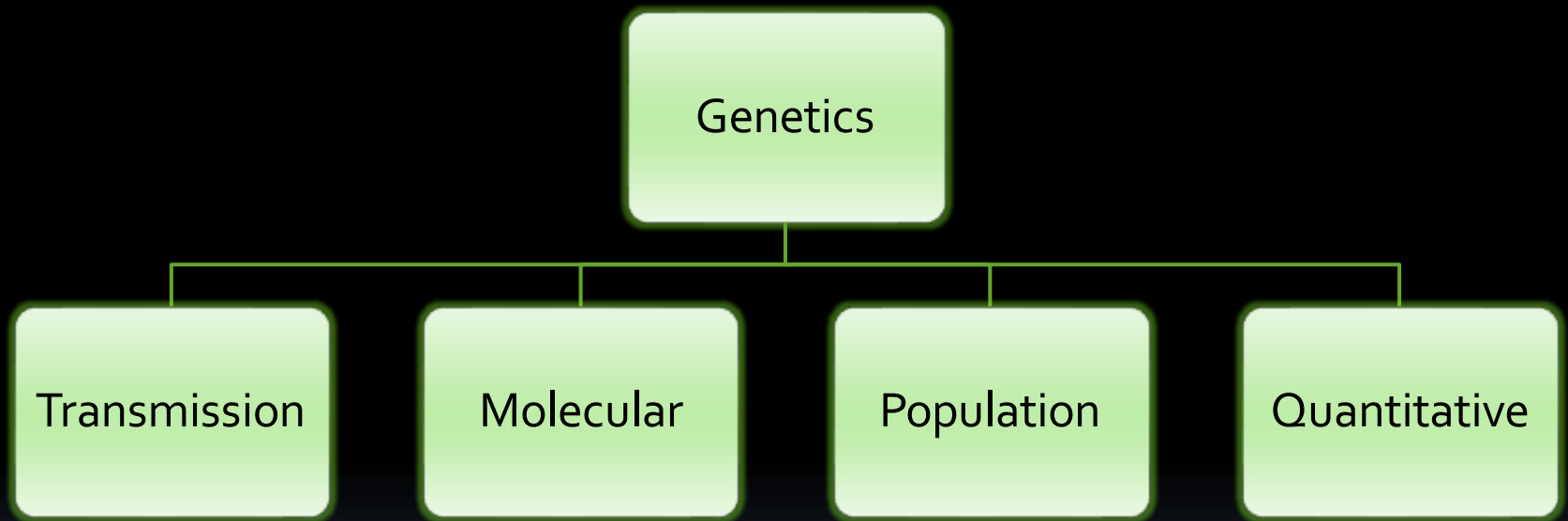
CC-14: Evolutionary Biology

UNIT-5

POPULATION GENETICS

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Science of Genetics



The principal aim of population genetics

- It investigate the patterns of genetic variation found among individuals within groups (the genetic structure of populations) and how these patterns vary geographically and change over time.
- To understand the genetics of evolution, a change in a population or species over time.
- The methods used focus on the **gene pool** of a **Mendelian population** rather than the genotypes of its individual members.
- perspective shifts away from the individual and the cell and focuses instead on a large group of individuals, a Mendelian population.

Mendelian population

- It is a group of local population or community of similar and sexually interbreeding or potentially interbreeding individuals (*who* share a common set of genes – gene pool) living within a circumscribed area at a given time.
- Its members have equal opportunity of mating with any other member of opposite sex.

Different Terminology

- Local breeding population or Mendelian population -----by Dobzhansky
- Panmictic populationby Sewall Wright
- Gamodeme.....by Gilmour & Gregor



GENE POOL

- Sumtotal of genes present in a Mendalian Population
- Gametic Pool

Gene Frequency

- It refers to the proportion of an allele in the gene pool as compared with other alleles at the same locus, with no regards to their distribution in organisms.
- Gene frequency can be calculated by dividing the number of a particular gene in question with the total number of genes present on that locus in the population.

Genotype Frequency

- It is the total number of a kind of individuals from a population all of which exhibit similar character with respect to the locus under consideration.
- It can be determined by dividing the number of individuals with that genotype by the total number of individuals in the population.

Box 21.1 Sample Calculation of Genotype and Allele Frequencies for Hemoglobin Variants Among Nigerians Where Multiple Alleles Are Present

Hemoglobin Genotypes

AA	AS	SS	AC	SC	CC	Total
2,017	783	4	173	14	11	3,002

Calculation of Genotype Frequencies

$$\text{Genotype frequency} = \frac{\text{Number of individuals with the genotype}}{\text{Total number of individuals}}$$

$$f(SS) = \frac{4}{3,002} = 0.0013 \quad f(AA) = \frac{2,017}{3,002} = 0.672 \quad f(AC) = \frac{173}{3,002} = 0.058$$
$$f(AS) = \frac{783}{3,002} = 0.261 \quad f(SC) = \frac{14}{3,002} = 0.0047 \quad f(CC) = \frac{11}{3,002} = 0.0037$$

Calculation of Allele Frequencies from the Number of Individuals with a Particular Genotype

$$\text{Allele frequency} = \frac{\text{Number of copies of a given allele in the population}}{\text{Sum of all alleles in the population}}$$

$$f(S) = \frac{(2 \times \text{number of SS individuals}) + (\text{number of AS individuals}) + (\text{number of SC individuals})}{2 \times \text{total number of individuals}}$$

$$f(S) = \frac{(2 \times 4) + 783 + 14}{(2 \times 3,002)} = \frac{805}{6,004} = 0.134$$

$$f(A) = \frac{(2 \times \text{number of AA individuals}) + (\text{number of AS individuals}) + (\text{number of AC individuals})}{2 \times \text{total number of individuals}}$$

$$f(A) = \frac{(2 \times 2,017) + 783 + 173}{(2 \times 3,002)} = \frac{4,990}{6,004} = 0.831$$

$$f(C) = \frac{(2 \times \text{number of CC individuals}) + (\text{number of AC individuals}) + (\text{number of SC individuals})}{2 \times \text{total number of individuals}}$$

$$f(C) = \frac{(2 \times 11) + 173 + 14}{(2 \times 3,002)} = \frac{209}{6,004} = 0.035.$$

Calculation of Allele Frequencies from the Frequencies of Particular Genotypes

$$f(S) = f(SS) + \frac{1}{2}f(AS) + \frac{1}{2}f(SC)$$


$$f(S) = 0.0013 + (\frac{1}{2} \times 0.261) + (\frac{1}{2} \times 0.0047) = 0.134$$

$$f(A) = f(AA) + \frac{1}{2}f(AS) + \frac{1}{2}f(AC)$$

$$f(A) = 0.672 + (\frac{1}{2} \times 0.261) + (\frac{1}{2} \times 0.058) = 0.831$$

$$f(C) = f(CC) + \frac{1}{2}f(SC) + \frac{1}{2}f(AC)$$

$$f(C) = 0.0037 + (\frac{1}{2} \times 0.0047) + (\frac{1}{2} \times 0.058) = 0.035$$



In 1908 Hardy and Weinberg each published papers describing a mathematical relationship between allele frequencies and genotype frequencies. This relationship, now called the **Hardy-Weinberg principle**, allows us to predict a population's genotype frequencies from its allele frequencies.



STATEMENT:

'The relative frequency of various kinds of genes in a large and randomly mating sexual panmictic population tend to remain constant from generation to generation in the absence of mutation , natural selection and gene flow.'

Table 21.1 Possible Combinations of *A* and *a* Gametes from Gametic Pools for a Population

		Male gametes	
		<i>A</i> (<i>p</i>)	<i>a</i> (<i>q</i>)
Female gametes	<i>A</i> (<i>p</i>)	<i>AA</i> (<i>p</i> ²)	<i>Aa</i> (<i>pq</i>)
	<i>a</i> (<i>q</i>)	<i>Aa</i> (<i>pq</i>)	<i>aa</i> (<i>q</i> ²)

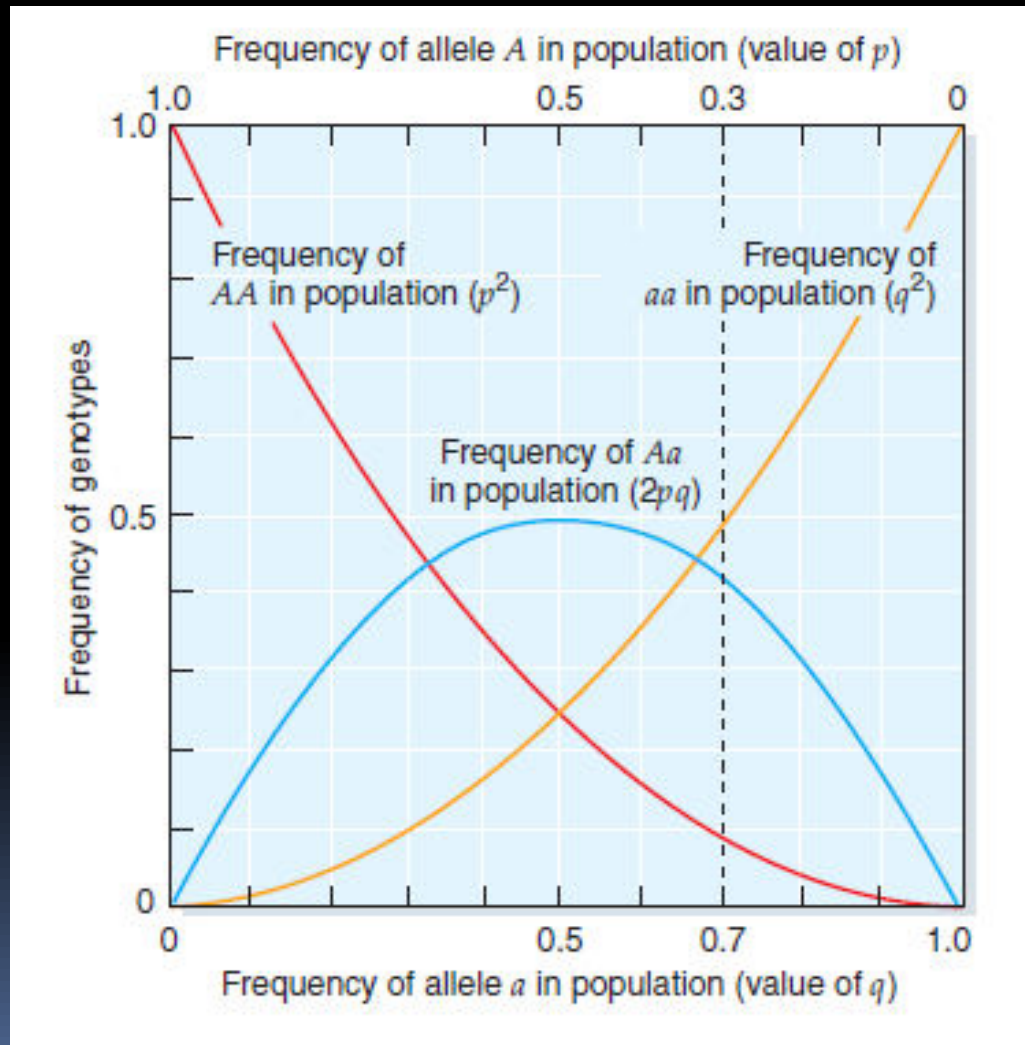
In sum, $p^2 AA + 2pq Aa + q^2 aa = 1.00$

Table 21.2 Algebraic Proof of Genetic Equilibrium in a Randomly Mating Population for One Gene Locus with Two Alleles

Type of Mating ♀ ♂		Mating Frequency	Offspring Frequencies Contributed to the Next Generation by a Particular Mating		
			AA	Aa	aa
$p^2 AA \times p^2 AA$		p^4	p^4	—	—
$p^2 AA \times 2pq Aa$ } $2pq Aa \times p^2 AA$ } ^a		$4p^3q$	$2p^3q$	$2p^3q$	—
$p^2 AA \times q^2 aa$ } $q^2 aa \times p^2 AA$ }		$2p^2q^2$	—	$2p^2q^2$	—
$2pq Aa \times 2pq Aa$		$4p^2q^2$	p^2q^2	$2p^2q^2$	p^2q^2
$2pq Aa \times q^2 aa$ } $q^2 aa \times 2pq Aa$ }		$4pq^3$	—	$2pq^3$	$2pq^3$
$q^2aa \times q^2aa$		q^4	—	—	q^4
Totals		$(p^2 + 2pq + q^2)^2 = 1$	$p^2(p^2 + 2pq + q^2) = p^2$	$2pq(p^2 + 2pq + q^2) = 2pq$	$q^2(p^2 + 2pq + q^2) = q^2$
Genotype frequencies = $(p + q)^2 = p^2 + 2pq + q^2 = 1$ in each generation afterward.					
Gene (allele) frequencies = $p(A) + q(a) = 1$ in each generation afterward.					

^aFor example, matings between AA and Aa will occur at $p^2 \times 2pq = 2p^3q$ for AA \times Aa and at $2pq \times p^2 = 2p^3q$ for Aa \times AA for a total of $4p^3q$. Two progeny types, AA and Aa, result in equal proportions from these matings. Therefore, offspring frequencies are $2p^3q$ (i.e., $1/2 \times 4p^3q$) for AA and for Aa.

Relationship of the frequencies of the genotypes AA , Aa , and aa to the frequencies of alleles A and a (in values of p [top abscissa] and q [bottom abscissa], respectively) in populations that meet the assumptions of the Hardy–Weinberg law. Any single population is defined by a single vertical line such as $p=0.3$ and $q=0.7$.



Conditions for Hardy-Weinberg Equilibrium

- Random Mating
- Large Population Size
- Biparental Mode Of Reproduction
- Homogeneous Age Structure
- Absence of Evolutionary Forces
 - No gene mutation
 - No Natural Selection
 - No gene Flow (Immigration / emigration)

Application of the Hardy-Weinberg Principle

- used as an initial test of whether evolution is occurring in a population
- medical applications - estimating the percentage of a population carrying the allele for an inherited disease

Example:

- consider phenylketonuria (PKU), a metabolic disorder that results from homozygosity for a recessive allele and occurs in about one out of every 10,000 babies born in the United States.
- Left untreated, PKU results in mental disability and other problems

To apply the Hardy-Weinberg equation

- condition 1 - no new PKU mutations are being introduced into the population
- condition 2 - that people neither choose their mates on the basis of whether or not they carry this gene nor generally mate with close relatives
- condition 3 - also ignore any effects of differential survival and reproductive success among PKU genotypes
- condition 4 – assume that there are no effects of genetic drift
- Condition 5 - no effects of gene flow from other populations into the United States

Evolution:

- A deviation from any of these conditions is a potential cause of evolution

- **violation of condition 1 - mutation**

- **violation of condition 2 – non random mating**

can affect the frequencies of homozygous and heterozygous genotypes but by itself usually has no effect on allele frequencies in the gene pool.

- **violations of conditions 3–5 - NS, Genetic drift, Gene flow**

three mechanisms alter allele frequencies directly and cause most evolutionary change

Basic Postulates of Darwinism

- Overproduction
- Struggle for Existence
 - Intra specific
 - Inter specific
 - With Environment
- Variation & Heredity
- Survival of the Fittest
- Origin of species

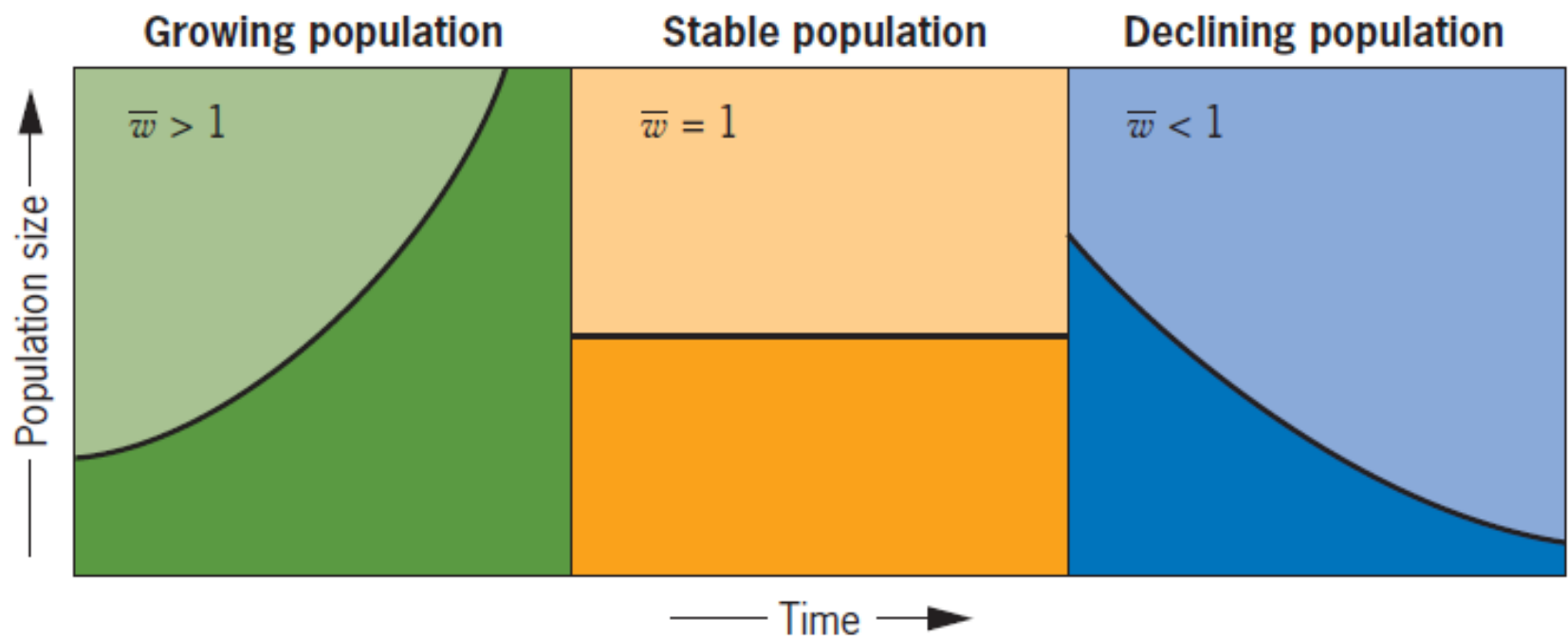
Natural Selection:

- Selection for survival and reproduction in the face of competition is the mechanism that changes the physical and behavioral characteristics of a species.

Darwin called this process **natural selection.**

THE CONCEPT OF FITNESS

- Geneticists refer the ability to survive and reproduce as fitness, a quantitative variable they usually symbolize by the letter w .
- Each member of a population has its own fitness value: 0 if it dies or fails to reproduce, 1 if it survives and produces 1 offspring, 2 if it survives and produces 2 offspring, and so forth.
- The average of all these values is the average fitness of the population, usually symbolized as \bar{w} .



■ **FIGURE 23.4** Significance of average fitness (\bar{w}) for population size as a function of time. Population size grows, is stable, or declines depending on the value of the average fitness.



Relative fitness

- The contribution an individual makes to the gene pool of the next generation **relative** to the contributions of other individuals

Illustration by example:

- A barnacle that is more efficient at collecting food than its neighbors may have greater stores of energy and hence be able to produce a larger number of eggs.
- A moth may have more offspring than other moths in the same population because its body colors more effectively conceal it from predators, improving its chance of surviving long enough to produce more offspring.
- These examples illustrate how in a given environment, certain traits can lead to greater **relative fitness**

Selection coefficient:

- Conventionally fitness of the competitively superior genotype(s) is expressed as 1 and the fitness of the inferior genotype(s) is expressed as a deviation from 1.
- This fitness deviation, usually symbolized by the letter s , is called the ***selection coefficient***
- it measures the intensity of natural selection acting on the genotypes in the population.

Relationship between W & S


$$W = 1 - S$$

Illustration:

- let's assume that fitness is determined by a single gene segregating two alleles, A and a , in a particular species of insect.
- Again assume that allele A causes the insects to be dark in color, that allele a causes them to be light in color, and that A is completely dominant to a .
- In a forest habitat, where plant growth is luxuriant, the dark form of the insect survives better than the light form. Consequently, the fitnesses of genotypes AA and Aa are greater than the fitness of genotype aa .
- By contrast, in open fields, where plant growth is scarce, the light form of the insect survives better than the dark form, and the fitness relationships are reversed

Illustration:

Genotype:	<i>AA</i>	<i>Aa</i>	<i>aa</i>
Phenotype:	dark	dark	light
Relative fitness in forest habitat:	1	1	$1 - s_1$
Relative fitness in field habitat:	$1 - s_2$	$1 - s_2$	1



Effect of natural selection on allele frequencies

- an insect population in the forest habitat with the initial frequency of A , $p = 0.5$, so the frequency of a, $q = 0.5$, and that $s_1 = 0.1$.
- Also assume that the population **mates randomly** and that the genotypes are present in **Hardy–Weinberg frequencies** at fertilization each generation.

Initial genetic composition of the population is:

Genotype:	<i>AA</i>	<i>Aa</i>	<i>aa</i>
Relative fitness:	1	1	$1 - 0.1 = 0.9$
Frequency (at fertilization):	$p^2 = 0.25$	$2pq = 0.50$	$q^2 = 0.25$

In forming the next generation, each genotype will contribute gametes in proportion to its frequency and relative fitness. Thus, the relative contributions of the three genotypes will be:

Genotype:	<i>AA</i>	<i>Aa</i>	<i>aa</i>
Relative contribution to next generation:	$(0.25) \times 1 = 0.25$	$(0.50) \times 1 = 0.50$	$(0.25) \times (0.9) = 0.225$

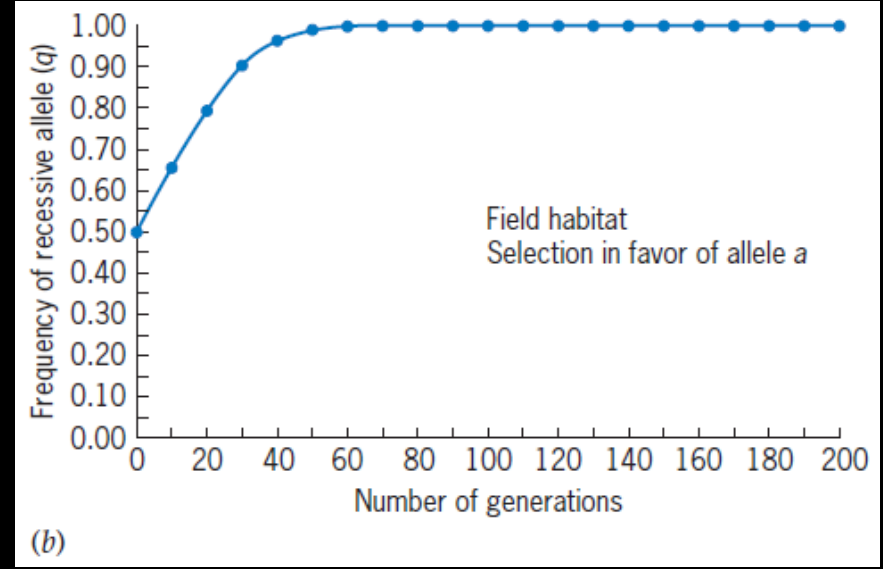
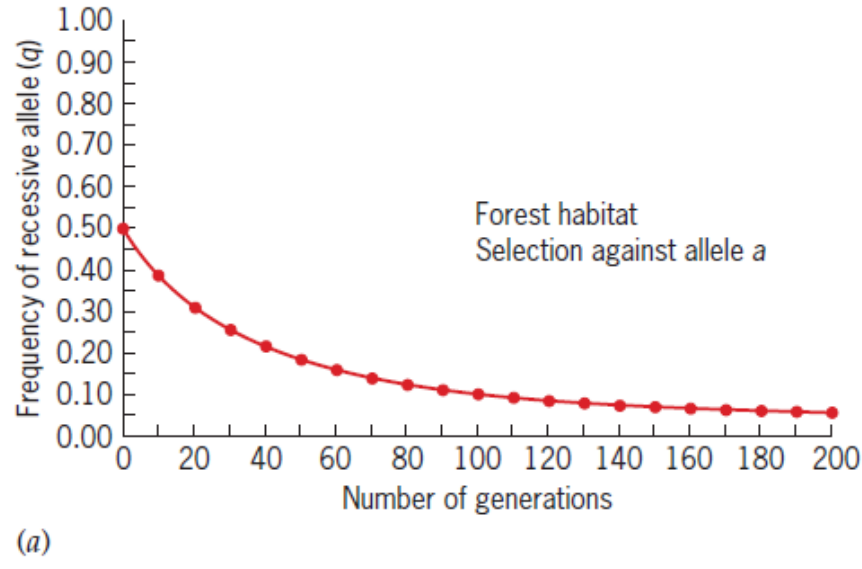
Dividing each of these relative contributions by their sum ($0.25 + 0.50 + 0.225 = 0.975$), we will obtain the proportional contributions of each of the genotypes to the next generation:

Genotype:	<i>AA</i>	<i>Aa</i>	<i>aa</i>
Proportional contribution to next generation:	0.256	0.513	0.231

From these numbers we can calculate the frequency of the *a* allele after one generation of selection simply by noting that all the genes transmitted by the *aa* homozygotes are *a* and that half the genes transmitted by the *Aa* heterozygotes are *a*.

In the next generation, the frequency of *a*, symbolized q' , will be

$$q' = 0.231 + (1/2)(0.513) = 0.487$$



■ **FIGURE 23.5** (a) Selection against the recessive allele *a* in the forest habitat. (b) Selection in favor of the recessive allele *a* in the field habitat.

In the field habitat, *aa* homozygotes are selectively superior to the other two genotypes. Thus, starting with $q = 0.5$, Hardy–Weinberg genotype frequencies, and the selection coefficient $s = 0.1$, we have:

Genotype:	<i>AA</i>	<i>Aa</i>	<i>aa</i>
Relative fitness:	$1 - 0.1 = 0.9$	$1 - 0.1 = 0.9$	1
Frequency:	0.25	0.50	0.25

After one generation of selection in the field habitat, the frequency of *a* will be $q' = 0.513$, which is slightly greater than the starting frequency.

Every generation afterward, the frequency of *a* will rise, and eventually it will equal 1, at which point we can say that the allele has been fixed in the population.




(a)



(b)

■ **FIGURE 23.6** (a) The dark form of the peppered moth on tree bark covered with lichens. (b) The light form of the peppered moth on tree bark covered with soot from industrial pollution.



Evolution by natural selection is a blend of chance and “sorting”:

- chance in the creation of new genetic variations (as in mutation) and sorting as natural selection favors some alleles over others. Because of this favoring process, the outcome of natural selection is not random. Instead, natural selection consistently increases the frequencies of alleles that provide reproductive advantage and thus leads to adaptive evolution.

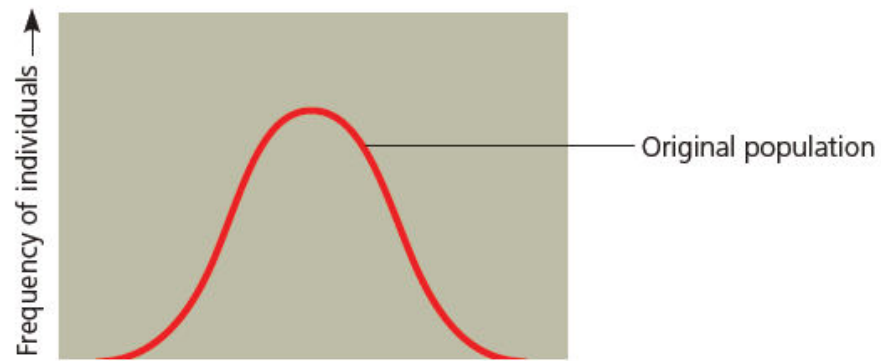


Modes of selection

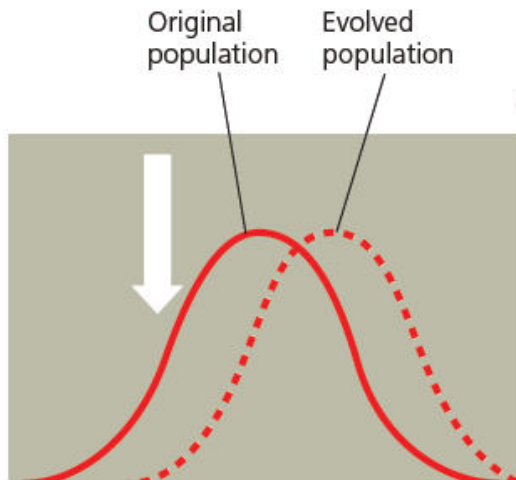
Directional selection

Disruptive selection

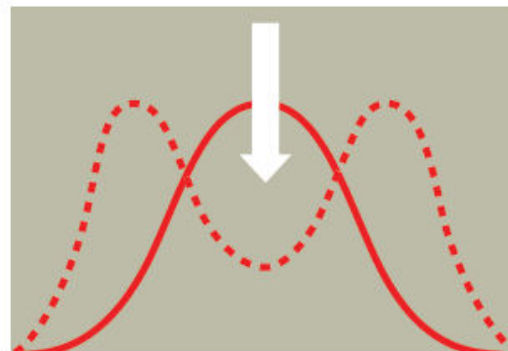
Stabilizing selection



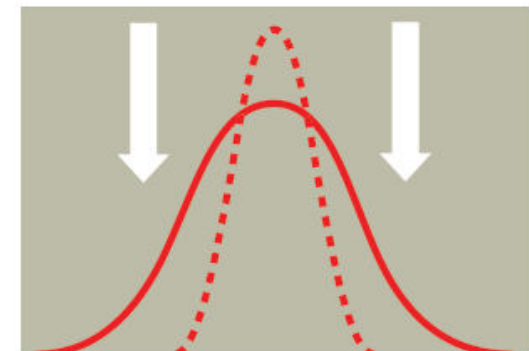
Phenotypes (fur color)



(a) Directional selection shifts the overall makeup of the population by favoring variants that are at one extreme of the distribution. In this case, lighter mice are selected against because they live among dark rocks, making it harder for them to hide from predators.



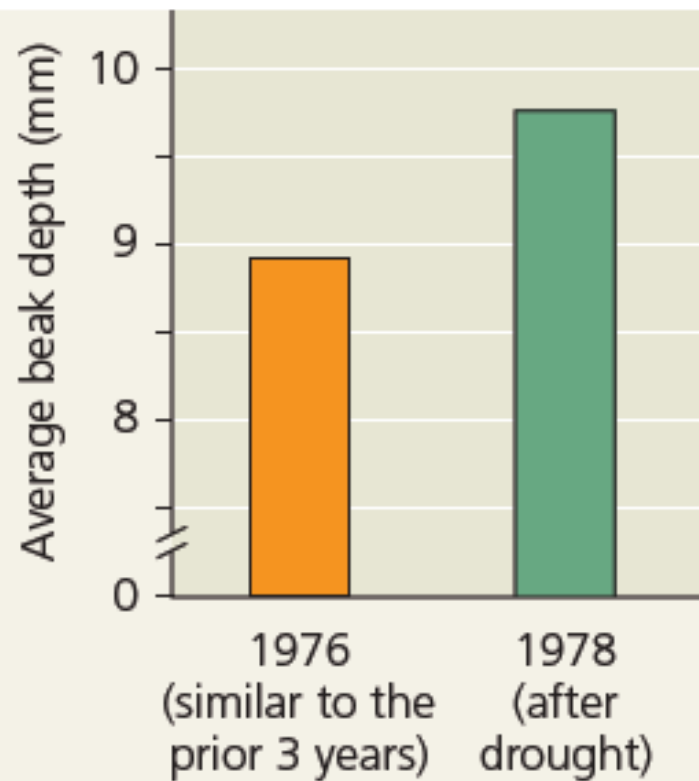
(b) Disruptive selection favors variants at both ends of the distribution. These mice have colonized a patchy habitat made up of light and dark rocks, with the result that mice of an intermediate color are selected against.



(c) Stabilizing selection removes extreme variants from the population and preserves intermediate types. If the environment consists of rocks of an intermediate color, both light and dark mice will be selected against.

Directional selection

- occurs when conditions favor individuals exhibiting one extreme of a phenotypic range, thereby shifting a population's frequency curve for the phenotypic character in one direction or the other
- Ex: an increase in the relative abundance of large seeds over small seeds led to an increase in beak depth in a population of Galápagos finches



◀ **Figure 23.2 Evidence of selection by food source.**

The data represent adult beak depth measurements of medium ground finches hatched in the generations before and after the 1977 drought. Beak sizes remained large until 1983, when changing conditions no longer favored large-beaked birds.

Disruptive selection


- occurs when conditions favor individuals at both extremes of a phenotypic range over individuals with intermediate phenotypes.
- Ex: members of population of black-bellied seedcracker finches in Cameroon display two distinctly different beak sizes. Small-billed birds feed mainly on soft seeds, whereas large-billed birds specialize in cracking hard seeds. It appears that birds with intermediate-sized bills are relatively inefficient at cracking both types of seeds and thus have lower relative fitness

Stabilizing selection

- Acts against both extreme phenotypes and favors intermediate variants.
- This mode of selection reduces variation and tends to maintain the status quo for a particular phenotypic character
- Ex: the birth weights of most human babies lie in the range of 3–4 kg (6.6–8.8 pounds); babies who are either much smaller or much larger suffer higher rates of mortality.

The Preservation of Genetic Variation

- The tendency of directional and stabilizing selection is to reduce variation.
- But variation is also found at loci affected by selection.



What prevents
natural selection from reducing
genetic variation at those
loci by culling all unfavorable
alleles?

CONCEPT OF **Balancing Selection**

- Selection itself may preserve variation at some loci.
- **Balancing selection** occurs when natural selection maintains two or more forms in a population.
- This type of selection includes
 - ❖ Heterozygote advantage
 - ❖ Frequency-dependent selection

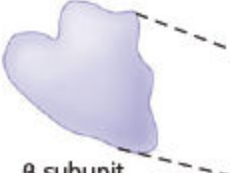
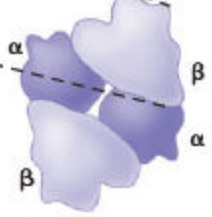

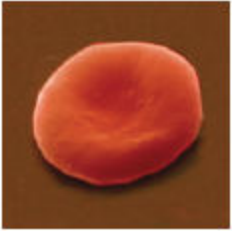

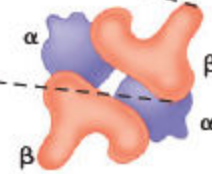
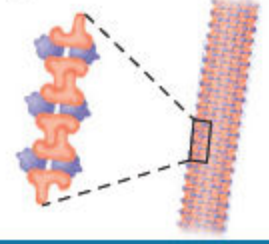

HETEROZYGOUS SUPERIORITY:

- If individuals who are heterozygous at a particular locus have greater fitness than do both kinds of homozygotes, they exhibit **heterozygote advantage**.
- In such a case, natural selection tends to maintain two or more alleles at that locus.
- heterozygote advantage is defined in terms of genotype, not phenotype

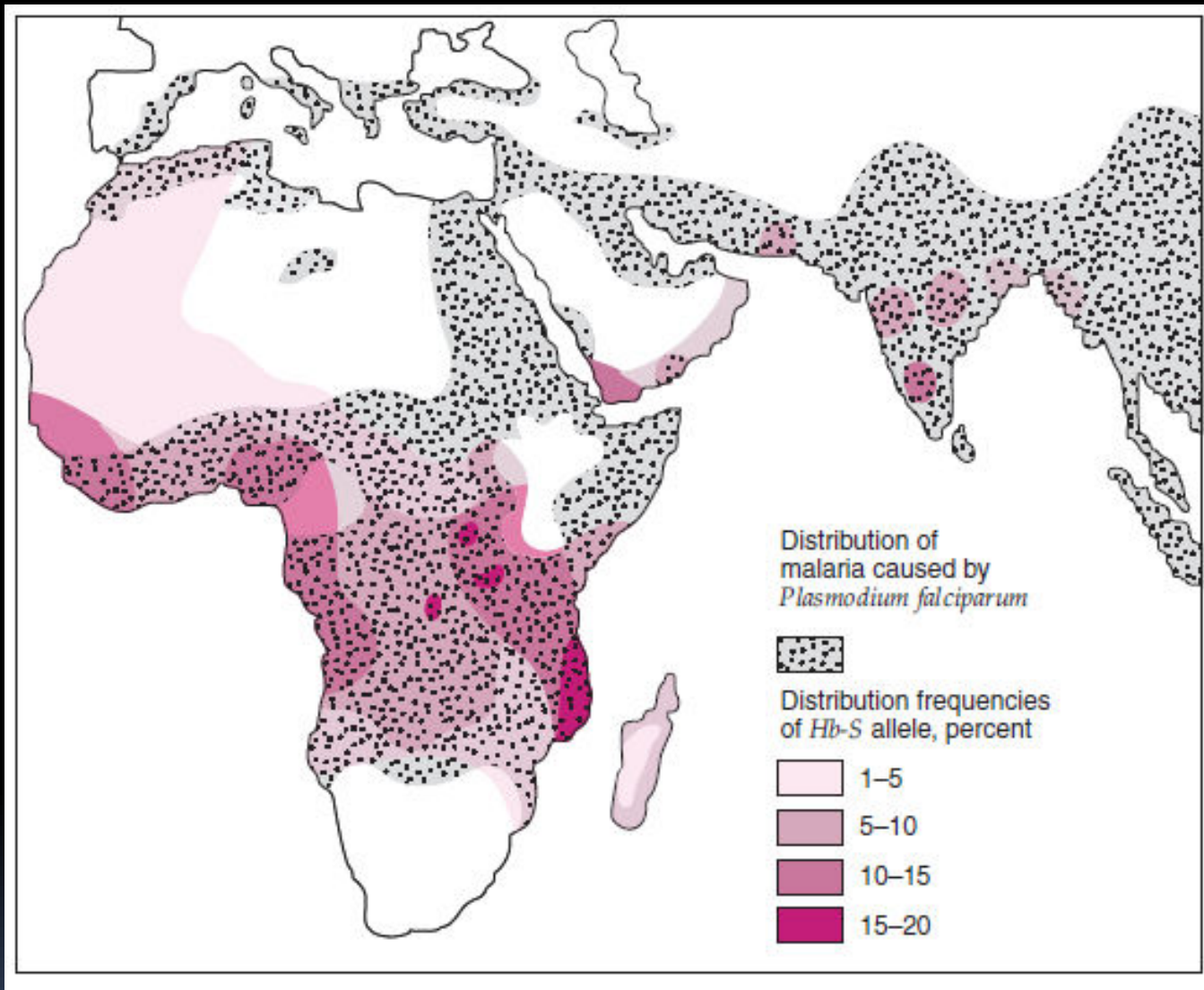
Example:

- In an environment in which malaria is common, the heterozygotes are at a selective advantage over the two homozygotes ($Hb-A/Hb-A$ & $Hb-S/Hb-S$).
- Although some red blood cells become sickled in heterozygotes ($Hb-A/Hb-S$ individuals have sickle-cell trait, a mild form of sickle-cell anemia), not enough become sickled to cause sickle-cell disease
- Heterozygotes for the sickle-cell allele are protected against the most severe effects of malaria

Despite the problems faced by Hb-S homozygotes, natural selection cannot eliminate this allele from the population, because the allele has beneficial effects in the heterozygote state.

	Primary Structure	Secondary and Tertiary Structures	Quaternary Structure	Function	Red Blood Cell Shape
Normal hemoglobin	1 Val 2 His 3 Leu 4 Thr 5 Pro 6 Glu 7 Glu	 <p>β subunit</p>	Normal hemoglobin 	Molecules do not associate with one another; each carries oxygen. 	Normal red blood cells are full of individual hemoglobin molecules, each carrying oxygen.  10 μm
Sickle-cell hemoglobin	1 Val 2 His 3 Leu 4 Thr 5 Pro 6 Val 7 Glu	Exposed hydrophobic region  <p>β subunit</p>	Sickle-cell hemoglobin 	Molecules interact with one another and crystallize into a fiber; capacity to carry oxygen is greatly reduced. 	Fibers of abnormal hemoglobin deform red blood cell into sickle shape.  10 μm

▲ **Figure 5.21** A single amino acid substitution in a protein causes sickle-cell disease.



The distribution of malaria caused by the parasite *Plasmodium falciparum* coincides with distribution of the *Hb-S* allele for sickle-cell anemia



THANK YOU