

#### Objectives

- Explain why the majority of point mutations are harmless.
- Explain how sexual recombination generates genetic variability.
- Define the terms population, species, gene pool, relative fitness, and neutral variation.

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- List the five conditions that must be met for a population to remain in Hardy-Weinberg equilibrium.
- Apply the Hardy-Weinberg equation to a population genetics problem.



## The Smallest Unit of Evolution

 One common misconception about evolution is that individual organisms evolve, in the Darwinian sense, during their lifetimes

- Natural selection acts on individuals, but populations evolve
- Genetic variations in populations contribute to evolution



## Population Genetics and The Modern Synthesis

- Microevolution is change in the genetic makeup of a population from generation to generation
- Population genetics provides a foundation for studying evolution
  - Population genetics is the study of how populations change genetically over time
    - Reconciled Darwin's and Mendel's ideas

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- The modern synthesis integrates Mendelian genetics with the Darwinian theory of evolution by natural selection
  - Focuses on populations as units of evolution

## Gene Pools and Allele Frequencies



- A population is a localized group of individuals that are capable of interbreeding and producing fertile offspring
- The gene pool is the total aggregate of genes in a population at any one time

   Consists of all gene loci in all individuals of the population





## The Hardy-Weinberg Theorem

 The Hardy-Weinberg theorem describes a population that is not evolving

 States that the frequencies of alleles and genotypes in a population's gene pool remain constant from generation to generation provided that only Mendelian segregation and recombination of alleles are at work

Mendelian inheritance preserves genetic variation in a population

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 In a given population where gametes contribute to the next generation randomly, allele frequencies will not change









## Hardy-Weinberg Equilibrium



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- Hardy-Weinberg equilibrium:
  - Describes a population in which random mating occurs
  - Describes a population where allele frequencies do not change

 If p and q represent the relative frequencies of the only two possible alleles in a population at a particular locus, then:
 p + q = 1, and

- $-p^{2} + 2pq + q^{2} = 1$ 
  - p<sup>2</sup> and q<sup>2</sup> represent the frequencies of the homozygous genotypes and 2pq represents the frequency of the heterozygous genotype

## Conditions for Hardy-Weinberg Equilibrium

- The Hardy-Weinberg theorem describes a hypothetical population
  - Five conditions for non-evolving populations
    - Extremely large population size
    - infinitely large
    - No gene flow - no migration
    - No mutations
    - do not alter gene pool
    - Random mating
    - mates randomly selected
    - No natural selection
       equal reproductive success





• We can use the Hardy-Weinberg equation to estimate the percentage of the human population carrying the allele for an inherited disease

- example:

 PKU occurs in 1 in 10000 babies (q<sup>2</sup>=0.0001), then q=0.01, p=0.99 and 2pq=0.0198

### The Origin of Variation

 Two processes, mutation and sexual recombination, produce the variation in gene pools that contributes to differences among individuals

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### Mutation

- Mutations are changes in the nucleotide sequence of DNA
  - Cause new genes and alleles to arise
- A point mutation is a change in one base in a gene

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- Can have a significant impact on phenotype
  - · Is usually harmless, but may have an adaptive impact







## Mutations That Alter Gene Number or Sequence

- Chromosomal mutations that affect many loci are almost certain to be harmful

   May be neutral and even beneficial
- Gene duplication duplicates chromosome segments

#### **Mutation Rates**

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- Mutation rates tend to be low in animals and plants
  - Average about one mutation in every 100,000 genes per generation
  - Are more rapid in microorganisms

#### Sexual Recombination

 In sexually reproducing populations, sexual recombination is far more important than mutation in producing the genetic differences that make adaptation possible

## **Changing Allele Frequencies**

 Three major factors alter allele frequencies and bring about most evolutionary change

 Natural selection

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- Genetic drift
- Gene flow

## Natural Selection

 Differential success in reproduction results in certain alleles being passed to the next generation in greater proportions

### Genetic Drift

- Statistically, the smaller a sample the greater the chance of deviation from a predicted result
- Genetic drift describes how allele frequencies can fluctuate unpredictably from one generation to the next
  - Tends to reduce genetic variation





### The Bottleneck Effect

- In the bottleneck effect a sudden change in the environment may drastically reduce the size of a population
  - The gene pool may no longer be reflective of the original population's gene pool

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• Understanding the bottleneck effect can increase understanding of how human activity affects other species





Greater prairie chicken	Pre-bo (Illinoi: Bange of greater prairie chicken	ittleneck Po s, 1820) (III	st-bottleneck inois, 1993)
Location	Population size	Number of alleles per locus	Percentage of eggs hatched
Illinois 1930–1960s 1993	1,000–25,000 <50	5.2 3.7	93 <50
Kansas, 1998 (no bottleneck)	750,000	5.8	99


## The Founder Effect

- The founder effect occurs when a few individuals become isolated from a larger population
  - Can affect allele frequencies in a population

## Gene Flow

- Gene flow causes a population to gain or lose alleles
  - Results from the movement of fertile individuals or gametes
  - Tends to reduce differences between populations over time

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## Natural Selection

- Natural selection is the primary mechanism of adaptive evolution
  - Natural selection accumulates and maintains favorable genotypes in a population

## **Genetic Variation**

- Genetic variation occurs in individuals in populations of all species
  - Is not always heritable

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## Variation Within a Population

• Both discrete and quantitative characters contribute to variation within a population

- Discrete characters can be classified on an eitheror basis
- Quantitative characters vary along a continuum within a population

### Polymorphism

- Phenotypic polymorphism describes a population in which two or more distinct morphs for a character are each represented in high enough frequencies to be readily noticeable
- Genetic polymorphisms are the heritable components of characters that occur along a continuum in a population

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## Measuring Genetic Variation



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• Average heterozygosity measures the average percent of loci that are heterozygous in a population

## Variation Between Populations

 Most species exhibit geographic variation differences between gene pools of separate populations or population subgroups









## **Evolutionary Fitness**

 The phrases "struggle for existence" and "survival of the fittest" are commonly used to describe natural selection

 Can be misleading

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• Reproductive success is generally more subtle and depends on many factors

 Relative fitness is the contribution an individual makes to the gene pool of the next generation, relative to the contributions of other individuals

 Relative fitness measures, indirectly, the contribution of a genotype to the next generation as compared to the contributions of alternative genotypes for the same locus

## Directional, Disruptive, and Stabilizing Selection

- Selection favors certain genotypes by acting on the phenotypes of certain organisms
- Three modes of selection are:
  - Directional
  - Disruptive
  - Stabilizing

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- Directional selection favors individuals at one end of the phenotypic range
- Disruptive selection favors individuals at both extremes of the phenotypic range
- Stabilizing selection favors intermediate variants and acts against extreme phenotypes





## Diploidy

• Diploidy maintains genetic variation in the form of hidden recessive alleles

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## **Balancing Selection**

 Balancing selection occurs when natural selection maintains stable frequencies of two or more phenotypic forms in a population

 Leads to a state called balanced polymorphism

## Heterozygote Advantage

- Some individuals who are heterozygous at a particular locus have greater fitness than homozygotes
  - Natural selection will tend to maintain two or more alleles at that locus
- The sickle-cell allele causes mutations in hemoglobin but also confers malaria resistance
  - Exemplifies the heterozygote advantage











### **Neutral Variation**

 Neutral variation is genetic variation that appears to confer no selective advantage

## **Sexual Selection**



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- Sexual selection is natural selection for mating success
  - Can result in sexual dimorphism, marked differences between the sexes in secondary sexual characteristics
- Intrasexual selection is a direct competition among individuals of one sex for mates of the opposite sex

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- Intersexual selection occurs when individuals of one sex (usually females) are choosy in selecting their mates from individuals of the other sex
  - May depend on the showiness of the male's appearance





## The Evolutionary Enigma of Sexual Reproduction

- Sexual reproduction produces fewer reproductive offspring than asexual reproduction, a so-called reproductive handicap
- If sexual reproduction is a handicap, why has it persisted?

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 It produces genetic variation that may aid in disease resistance





## Why Natural Selection Cannot Fashion Perfect Organisms

• Selection can only edit existing variations

- Evolution is limited by historical constraints
- Adaptations are often compromises
- Chance and natural selection interact

