

1 **Quiz I Results**

2

3 **Mechanisms of Evolution**

Their Microevolutionary Effect

4 **Hardy-Weinberg Equilibrium**

- The Hardy-Weinberg Equilibrium Model maintains that certain conditions exist that contribute to population stability and if those conditions are maintained, gene frequencies will be invariable (unchanging) throughout any number of generations.

5 **Hardy-Weinberg Formula**

- Few populations meet these requirements but the Hardy-Weinberg Equilibrium Model sets up a reference for which population changes can be compared (using the Chi-squared statistic).

6 **Evolutionary Forces**

- There are a number of evolutionary forces that produce changes in gene frequencies at the population level (microevolution).

7 **Evolutionary Forces**

- Mutation
- Natural Selection
- Genetic Drift
- Gene Flow

- Non-Random Mating

8 **Mutation**

- A change in the genetic code

- Results in an alteration of its action (alters protein synthesis) and introduces new varieties of alleles

9 **Frequency of Mutations**

- Estimates of the frequency of mutations in human sex cells generally are about 1 per 10-100,000 for any specific gene. Since humans have approximately 32,000 genes, it is to be expected that most sex cells contain at least one gene mutation of some sort and that each

individual carries at least one mutation

10 **Characteristics of Mutations**

- Mutations are random
- Mutations must occur in the sex cells to affect allele frequencies
- Mutations increase variability in populations (the ultimate source of all variation within a population)
- Mutations caused by a variety of factors including errors in replication, different forms of radiation, certain ingested substances, and heat
- Mutation rates are low and by themselves do not lead to major changes in allele frequencies
- Most mutations are detrimental, some are lethal, and some are neutral (have no effect).

11 **Levels of Mutation**

- **DNA base – point mutation** (substitution, deletion, addition)
- **Larger sections of DNA**
- **Chromosomal level** (deletion or duplication of all or part of the chromosome)

12 **DNA Base Level Mutation**

- **Sickle Cell Anemia**
- Mutation in the sixth amino acid (out of 146 amino acids) of the beta chain of hemoglobin
- The DNA for normal hemoglobin contains the codon CTC at this location (codes for glutamic acid) and the DNA for sickle cell hemoglobin contains the codon CAC (codes for valine)
- This small change affects the entire structure of hemoglobin and in turn the well being of the individual

13 **Mutations in Large Sections of DNA**

- **Cri Du Chat Syndrome (cry of the cat)**
- Substantial loss of the short arm of chromosome 5 (1/25,000 to 1/50,000 births)
- Severe mental and physical pathologies including abnormal larynx development, microcephaly, respiratory problems

14 **Chromosomal Level Mutations**

- **Monosomy** (loss of an entire chromosome from a pair)
- **Trisomy** (addition of an entire chromosome)

15 **Down's Syndrome (Trisomy 21)**

- Duplication of one of the 21st pair of chromosomes
- Results in changes in cranial features, poor physical growth, and mental retardation (1/800 births)

16 **Mutations in the Sex Chromosomes**

- **Turner's Syndrome:** when an individual has only one X chromosome (have only 45 chromosomes and develop as females – generally short, undeveloped ovaries, and are generally sterile, 1/2000 female births)
- **Klinefelter's Syndrome:** when a male carries an extra X chromosome (XXY; have 47 chromosomes and are characterized by small testes and reduced fertility – 1/500 male births)

- **Jacob's Syndrome:** when a male carries an extra Y chromosome (XYY; have 47 chromosomes and are generally normal but some show delayed language development and cognitive problems – 1/1000 male births)

17 Mutation Summary

- Mutation rates are low and mutation by itself is not a major evolutionary force in changing allele frequencies at the population level

18 Natural Selection

- Differential survival and reproduction of organisms because of their biological characteristics
 - Some individuals have characteristics that are adaptive in certain environments and enable them to survive and reproduce at a higher rate than other individuals (**Darwinian Fitness**)
 - Over time these characteristics will become more predominate in a population

19 Characteristics of Natural Selection

- It is not random
- It limits and stabilizes genetic diversity (decreases variation within a population)

20 Types of Natural Selection

- **Simple Traits**
 - Selection Against the Recessive Homozygote
 - Selection For the Recessive Homozygote (selection against the dominant allele)
 - Selection For the Heterozygote (**Balancing Selection**)
- **Complex Traits**
 - **Stabilizing Selection**
 - **Directional Selection**
 - **Disruptive Selection**

21 Selection Against the Homozygous Recessive (First Generation)

	AA	Aa	aa
N (before selection) p=0.5; q=0.5	50	100	50
Fitness (% that survives)	100	100	0
N _{F1} (after selection) p=0.67; q=0.33	50	100	0

22 Selection Against the Homozygous Recessive (Second Generation)

	AA	Aa	aa
N _{F2} (before selection) p=0.67; q=0.33	89	89	22
Fitness (% that survives)	100	100	0

N_{F_2} (after selection)	89	89	0
-----------------------------	----	----	---

$p=0.75$; $q=0.25$

23 **Selection Against the Homozygous Recessive (Numerous Generations)**

24 **Selection Against the Homozygous Recessive**

Tay-Sachs Disease

- Metabolic disorder that causes blindness, mental retardation, and the destruction of the central nervous system
- Children with Tay-Sachs Disease generally die within the first few years of life
- The disease is caused by a recessive allele (single base mutation) and it occurs in individuals who are homozygous recessive
- Heterozygotes are normal but carry the recessive allele
- The occurrence of the allele is very low but is maintained by mutation but kept from increasing by natural selection

25 **Selection For the Homozygous Recessive (Selection Against the Dominant Allele)**

	AA	Aa	aa
N (before selection)	50	100	50
$p=0.5$; $q=0.5$			
Fitness (% that survives)	0	0	100
N (after selection)	0	0	50
$p=0$; $q=1.0$			

26 **Selection For the Homozygous Recessive (Selection Against the Dominant Allele)**

Achondroplastic Dwarfism

- Dwarfism (small body size and abnormal body proportions) caused by a dominant allele found in low frequencies ($p=0.00005$)
- Caused by mutation that ultimately affects skeletal growth
- Because of dominance individuals with one or two alleles have the disease
- Selection for the dominant allele occurs at two levels: homozygous dominant individuals die before or at birth; heterozygous individuals have reduced opportunities to mate

27 **Selection for the Heterozygote (Balancing Selection) – First Generation**

Codominant System

	AA	Aa	aa
N (before selection)	50	100	50
$p=0.5$; $q=0.5$			
Fitness (% that survive)	70	100	20
N_{F_1} (after selection)	35	100	10
$p=0.59$; $q=0.41$			

28 **Selection for the Heterozygote (Balancing Selection) – Second Generation**

Codominant System

	AA	Aa	aa
N_{F_2} (before selection) $p=0.59$; $q=0.41$	70	97	33
Fitness (% that survive)	70	100	20
N_{F_2} (after selection) $p=0.64$; $q=0.36$	49	97	7

29 **Selection for the Heterozygote (Balancing Selection)**

30 **Selection for the Heterozygote (Balancing Selection)**

Cystic Fibrosis

- Genetic disease caused by a recessive allele
- Homozygous recessives are affected by the excessive production of mucous in the lungs (prior to 1950 few survived passed infancy, today to the mid-twenties)
- Heterozygous individuals have higher resistances to other diseases such as cholera and typhoid fever

31 **Complex Traits and Selection: Stabilizing Selection**

- Selection against extreme values in a continuous trait

32 **Stabilizing Selection: Human Birth Weight**

33 **Complex Traits and Natural Selection Directional Selection**

- Selection against one extreme in a continuous trait and/or selection for the other extreme

34 **Complex Traits and Natural Selection Disruptive Selection**

- Selection against the mean and favoring both extremes

35 **Microevolution Simulations: Natural Selection**

- Microevolution Simulation Program

■ http://www.mhhe.com/mayfieldpub/relethford/student_resources/micro/index.htm

36 Genetic Drift

- Also referred to as sampling error, genetic drift is the random fluctuations in gene frequencies from one generation to the next

37 Genetic Drift at the Cross Level

38 Characteristics of Genetic Drift

- Most prevalent in small populations (where the number of matings is small enough that all genetic combinations may not be reproduced because of chance factors)
- Random
- Decreases variation within a population
- Can cause rapid evolution in small populations

39 Examples of Genetic Drift

- Initial population size of 10; $p=0.5$; $q=0.5$

40 Effect of Population Size on Genetic Drift

41 Special Examples of Genetic Drift

- **Founder's Effect:** A type of genetic drift caused by the founding of a new breeding population by a small number of founders; small probability that the founding population contains all of the variation present in the parent population

42 Special Examples of Genetic Drift

- **Bottleneck Effect:** A type of genetic drift in which a population loses large numbers of its population over a short time due to disease or disaster

43 Example of Genetic Drift:

Tristan da Cunha

- Island in the South Atlantic Ocean
- Garrison established by the English in 1816 included 15 individuals including soldiers, shipwrecked sailors, and a few women
- One man and women remained after abandonment of the garrison to be joined by a handful of settlers
- Only occasional immigration because of the isolated nature of the island
- In 1855 emigration reduced the population to 33 individuals from 103
- Boat disaster (15 males perished) and widows and children left the island reducing the population from 106 to 54
- By 1961 the population reached 270 (29% of the population can trace their ancestry to a couple in the original founding population)

44 Examples of Genetic Drift:

Pitcairn Islanders

- Mutineers of the HMS Bounty (9 sailors), six Tahitian men, and eight or nine Tahitian women founded a

population on Pitcairn Island in 1790

- Pitcairn Island is one of the most isolated islands in Polynesia
- Falling out between the British sailors and Tahitian men – leaving one sailor (Alexander Smith), the women (9), and some children (25)
- Island rediscovered in 1808
- Population size fluctuated never reaching above 200 with the current size at 56 in 1996 – many Pitcairn descendents have relocated to nearby islands

45 **Microevolution Simulations:
Genetic Drift**

- http://www.mhhe.com/mayfieldpub/relethford/student_resources/micro/index.htm

46 **Gene Flow**

- Movement of alleles from one population to another (don't confuse with migration)
- That is the exchange between different populations gene pools so that the next generation is a result of admixture of the parental populations

47 **Characteristics of Gene Flow**

- Distance has a major effect on gene flow (closer the populations the greater potential for significant gene flow)
- Ethnic and cultural differences also have a major effect on gene flow
- Gene flow increases genetic variation within a population and decreases genetic variation between populations

48 **Affect of Population Distance on Gene Flow**

49 **Effects of Language on Gene Flow**

50 **Example of Gene Flow**

- Exchange of 10% of genes each generation
- Population 1 ($p=1; q=0$) and Population 2 ($p=0; q=1$)

51 **Microevolution Simulations:
Gene Flow**

- http://www.mhhe.com/mayfieldpub/relethford/student_resources/micro/index.htm

52 **Interaction of Evolutionary Forces**

- Convenient to talk about them separately but they work in concert to alter gene frequencies
- They work sometimes together and sometimes opposed to one another

53 **Microevolution Simulations:
Natural Selection and Drift
Gene Flow and Drift**

- http://www.mhhe.com/mayfieldpub/relethford/student_resources/micro/index.htm

54 **Non-Random Mating**

- Patterns of mate choice that influence the distributions of genotype and phenotype frequencies

55 **Types of Non-Random Mating**

- **Positive Assortative Mating:** mating between individuals with similar phenotypes
- **Negative Assortative Mating:** mating between individuals with dissimilar phenotypes
- **Consanguineous Matings (Inbreeding):** mating between biologically related individuals

56 **Characteristics of Non-Random Mating**

- Results in changes to genotypic and phenotypic frequencies but not allelic frequencies
- Doesn't cause evolution but can affect the rate of evolutionary change
- Decreases variation within populations and results in more homozygotes and less heterozygotes

57 **Homozygosity and Inbreeding**

58 **Tristan da Cunha**

- Measure of Inbreeding (F: probability that both alleles at a locus are received from an identical ancestral source); **retinitis pigmentosa** (rare recessive allele) – 4/294 residents