

## Population Genetics

Social Patterns and Evolutionary Forces in Human Populations

## Population Genetics

- How do genes behave in populations
- What is a population?
  - A population is a subdivision of a species
  - A population is a community of individuals where mates are usually found
- A population shares a common gene pool

## Allele frequency

- An allele frequency is the proportion of one allele relative to all alleles at the locus in the population
  - Example: 20 Hb<sup>A</sup>/Hb<sup>A</sup>, 20 Hb<sup>A</sup>/Hb<sup>S</sup>, and 10 Hb<sup>S</sup>/Hb<sup>S</sup>
    - A allele frequency = 40 A alleles (in 20 Hb<sup>A</sup>/Hb<sup>A</sup> individuals) + 20 A alleles (in 20 Hb<sup>A</sup>/Hb<sup>S</sup> individuals) divided by 100 total alleles = 60/100 = **0.60 or 60%**
    - S allele frequency = 20 S alleles (in 20 Hb<sup>S</sup>/Hb<sup>S</sup> individuals) + 20 S alleles (in 20 Hb<sup>A</sup>/Hb<sup>S</sup> individuals) divided by 100 total alleles = 40/100 = **0.40 or 40%**

## Genotype frequency

- A genotype frequency is the proportion of a population that has one genotype relative to all genotypes at a specific locus
  - In the previous case, we had 10 homozygous sicklers, genotype Hb<sup>S</sup> / Hb<sup>S</sup>, out of 50 individuals for a genotype frequency of 10/50 or one-fifth or 0.20 or 20%

## Hardy-Weinberg Equilibrium

- The Hardy-Weinberg model describes a mathematical relationship that allows the prediction of the frequency of offspring genotypes based on parental allele frequencies
- It also predicts that allele frequencies will not change from one generation to the next, i.e., it is an equilibrium or non-evolutionary model

## Hardy-Weinberg Model

- If the frequency of an allele frequency (A) = p, and the frequency of the other allele at the locus frequency (B) = q, then the next generation will have:
  - The frequency of the AA genotype = p<sup>2</sup>
  - The frequency of the AB genotype = 2pq
  - The frequency of the BB genotype = q<sup>2</sup>

## Hardy-Weinberg Example

- At the MN blood group locus the frequency of the M allele (p) equals 0.4 and the frequency of the N allele (q) equals 0.6, the offspring in the next generation will have:
  - The frequency of the MM genotype =  $p^2 = (0.4)^2 = (0.4) \times (0.4) = 0.16$
  - The frequency of the MN genotype =  $2pq = 2 \times (0.4) \times (0.6) = 2 \times (0.24) = 0.48$
  - The frequency of the NN genotype =  $q^2 = (0.6)^2 = (0.6) \times (0.6) = 0.36$

## Hardy-Weinberg Requirements

- **Random mating**
- No **mutation**
- Closed population, no **gene flow** (or migration of individuals) in or out
- Infinite size, no stochastic effects or **genetic drift**
- Equal fertility for all genotype groups-- meaning no **selection** is occurring

## Random Mating

- The H-W model requires that mating be random with regard to the locus being considered
  - The frequency of mating between males of one genotype and females of another should be equal to the product of the two genotype frequencies

## Random Mating

- If the frequency of the AA genotype in males is 0.7 and in females is also 0.7, then about half of all matings ( $0.7 \times 0.7 = 0.49$ ) should be between AA males and females.
  - If the frequency of mating is much different from the prediction, (how much different is projected by a statistical Chi-square test) then there is some deviation from random, and the H-W model will not be accurate

## Assortative Mating

- If substantially more than half of the matings, say 2/3 of all matings are between AA males and females, this would be an example of **positive assortative mating**
  - Positive assortative mating is the occurrence of mating between similar individuals at higher than random frequencies, resulting in more homozygotes than the H-W model predicts

## Positive Assortative Mating

- As with most mammals, humans tend to mate with like individuals, particularly for visible or noticeable traits.

Trait	Spouse Correlation
I.Q.	0.47
Ear lobe length	0.40
Waist circumference	0.38
Height	0.28

### Negative Assortative Mating

- Mating between individuals with dissimilar genotypes will increase heterozygotes
  - Appears to be rare, but a study of Hutterites indicates that individuals with similar immune system genes are less likely than random to mate with one another
    - 24 mates sharing five genes were found when the allele frequencies predicted there should be 68
    - May be reduced fertility when the immune system genes are shared by both mates

### Inbreeding

- Mating with biologically related individuals will affect the H-W model
  - Incest taboos prohibit mating between closely related individuals, making inbreeding less common than simple random mating would predict
  - Incest taboos would have the effect of increasing heterozygotes

### Inbreeding

- Small isolated populations end up with high levels of inbreeding, even when incest taboos are followed
  - Neel estimated that the average relationship (based on shared genes) between individuals in a Yanomamo village is nearly the same as between brothers and sisters
  - The result is increased homozygotes

### Mutation

- Mutation, the alteration of genetic material, is the source of all truly new variability in the genome
- Mutation has a very small influence on changes in allele and genotype frequencies from one generation to the next
- Primarily important for the occurrence of new alleles

### Gene Flow

- Gene Flow, the intermarriage or mixing between populations, has the effect of altering allele and genotype frequencies so that the two (or more) populations involved come to resemble each other in terms of genetic frequencies

### Genetic Drift

- An infinite population size eliminates the chance or random influences on gene frequencies from one generation to the next which are especially significant in small populations
- One such example comes from the island of Tristan da Cunha, settled in 1816 by a group of 16 Scottish soldiers and their spouses

## Tristan da Cunha

Year	Event	Size
1816	Settlement of island	16
1855	Dispute, causing 33 (of 103) to leave	70
1885	Population back up to 106, boat wreck kills 15 males, families begin to leave	106
1891	Population starts growing again	59
1961	Continued growth from 1891	270

## Selection

- Selection causes changes in allele and genotype frequencies from one generation to the next due to differential net reproductive success of individuals with different genotypes
  - If individuals with genotype AA consistently have twice as many offspring as individuals with AB and BB genotypes, the frequency of the A allele will increase and eventually, everyone will have the AA genotype

## Selection

- There are two elements contributing to the differential reproductive success of individuals with differing genotypes
  - Viability or survival: individuals must survive to maturity in order to be able to reproduce
  - Fertility: individuals must produce offspring in order to pass on their genes
- The genotype producing the most offspring on average is the most fit

## Natural Selection

- Natural selection is a two-step process
  - Production of variation (mutation)
  - Differential reproduction of favorable variants (selection)