Population Genetics

Hardy-Weinberg Law

Assumptions

1. Autosomal Locus

2. No Mutation

3. No Migration

4. No Selection

5. Random Mating (*i.e.*, Random Pairing of Gametes)

6. Population is **Very Large**.

After a single generation, population will be in **Hardy-Weinberg Equilibrium** with genotypic array:

$$p^2 AA + 2pq Aa + q^2 aa$$

Determining if Population Has Hardy-Weinberg Proportions

Example:

MN Blood group in Eskimos.

MM	MN	NN	Total
2,087	390	23	2,500

Test for Hardy-Weinberg Proportions

1. Compute the Gene Frequencies

$$M: \quad (2087 + 390/2)/2500 = 0.9128$$
$$N: \quad (23 + 390/2)/2500 = 0.0872$$

2. Compute the Expected Hardy-Weinberg Frequencies

MM:	$(0.9128)^2$	=	0.8332
MN:	$2(0.9128)(0.0872)$	=	0.1592
NN:	$(0.0872)^2$	=	0.0076

3. Use the Expected Frequencies to Create a Chi-Square Table

Genotype	Observed	Expected	O-E	Chi-Square
MM	2,087	2,083	4	0.0076
MN	390	398	-8	0.1608
NN	23	19	4	0.8421
Total	2,500	2,500	0	1.0106

{ Degrees of Freedom

3 Genotypes - 1 for Total - 1 to Estimate p = 1

{ Statistical Conclusion

Fail to Reject the Hypothesis

{ Genetic Conclusion

The data are consistent with Hardy-Weinberg proportions for the MN locus in Eskimos.

Exceptions to Hardy-Weinberg Assumptions

Sex-Linked Genes (X-Linked)

	Female			Male		
Genotypes	AA	Aa	aa	AY	aY	
Gametes	A		a	A	a	Y

Crosses:

Start with a Female from a pure breeding A-Strain and a male from a pure breeding a-Strain.

Females		Males
AA	×	aY
Aa	×	AY
½ AA + ½ Aa	×	½ AY + ½ aY
3/8 AA + ½ Aa + 1/8 aa	×	¾ AY + ¼ aY

Several Generations

4/9 AA + 4/9 Aa + 1/9 aa   2/3 AY + 1/3 aY

Mutation

Parameters

μ = Probability[Mutation from **A** to **a**]

ν = Probability[Mutation from **a** to **A**]

Recurrence Equation

$p_1 = \nu\,(\,1 - p_0) + p_0\,(\,1 - \mu\,)$

Migration

Mainland / Island Model

Parameters

m = migration rate from Mainland

P = Frequency of A on Mainland

p_0 = Frequency of A on Island

Recurrence Equation

$p_1 = (1 - m) p_0 + mP$

Selection

Parameters

Selection Coefficients:

Genotype	AA	Aa	aa
Relative Fitnesses (W)	W_{AA}	W_{Aa}	W_{aa}

Examples

Gene Action	AA	Aa	aa
Recessive Lethal	1.00	1.00	0.00
Dominant Lethal	1.00	0.00	0.00
Overdominant (Stabilizing Selection)	1.00	1.30	0.20
Underdominant	1.00	0.30	0.70

Summary Value

$\overline{W} = p^2 W_{AA} + 2pq W_{Aa} + q^2 W_{aa}$ = Mean Population Fitness

Selection Over One Generation

Compute Starting Genotypic Frequencies & List Fitnesses

Genotypes	AA	Aa	aa	Gene Frequencies	
				A	a
Frequency	0.36	0.48	0.16	0.6	0.4
Fitness	1	0.95	0.3		

Note: Save some space here for the table to exand

Selection Over One Generation

Compute the Survivors (the parents after selection)

Genotypes	AA	Aa	aa	Gene Frequencies	
				A	a
Frequency	0.36	0.48	0.16	0.6	0.4
Fitness	1	0.95	0.3	Total	
Survivors				This is the	
(Fitness × Frequency)	0.36	0.456	0.048	0.864	Popula-tion Fitness

Selection Over One Generation

Adjust the Genotypic Frequencies (This is the Offspring Generation)

Genotypes	AA	Aa	aa	Gene Frequencies	
				A	a
Frequency	0.36	0.48	0.16	0.6	0.4
Fitness Survivors	1	0.95	0.3	Total	
(Fitness × Frequency)	0.36	0.456	0.048	0.864	
Adjusted	0.4167	0.5278	0.0555	1	

The Gene Frequencies for the next generation are:

$p_1 = \frac{1}{2}(0.5278) + 0.4167 = 0.6806$

$q_1 = \frac{1}{2}(0.5278) + 0.0555 = 0.3194$

NonRandom Mating

Inbreeding

Inbreeding is the result of mating between individuals who are related.

Definition

Identical by Descent. Two genes (alleles) are Indentical by Descent if they are derived from the same ancestral gene at some time in the past.

Parameters

F = Inbreeding Coefficient

= Probability that the two alleles from an individual at any gene are Identical by Descent.

Inbreeding

Effect on the Population

In a population undergoing **systematic inbreeding**

    wGene frequencies will not change from generation to generation;

    wThe genotypic array is:

$$(p^2 + Fpq)AA + 2(1 - F)pqAa + (q^2 + Fpq)aa$$

for every gene (although p & q will be different for each gene, F will remain the same).

Assortative Mating

Definition

Individuals choose mates based on phenotypic similarity. For "pure" assortative mating, *all* individuals have the same chance of mating. The only change from random mating is in their choice of mates.

Results (Qualitative)

wGene Frequencies do not change

wIncrease in homozygosity for

{ Traits selected

{ Genes linked to the traits selected

Small Population Size

Concepts:

Random Drift

Gene frequencies (and genotypic frequencies) change due to *random chance* (no selection). Eventually, all alleles except one will be lost from the population.

Founder Effect

{ Random Drift fixes genes that don't necessarily have selective advantage

{ Founder Population may not have **started** with genes in the same frequencies as the population as a whole.

Example of a More

Complex System

Mutation & Selection

There is recurrent mutation to lethal genes.

Loss due to selection = increase due to mutation

Genetic Load: Reduction in population fitness due to the number of lethal genes.

Small Population Size

Population Bottleneck: Catastrophic decrease in population size. Could be caused by natural disaster, human influences, *etc*.

Inbreeding

Appearance of lethals (in homozygous form) due to inbreeding. This may lead to extinction.

Population Dynamics

Population Size after Bottleneck