

### **(Observed) Allele Frequencies**

#### What does it measure?

How many alleles of each type there are floating around in the population that you are looking at – relative to the total number.

#### Answers these kinds of questions:

“What are the frequencies of p, q, r (or A, B, C)” ; “What are the allele frequencies...” (pretty straightforward)

#### How do you do it?

If you are given the number of individuals with particular genotypes – you need to remember that there are two alleles per genotype.

First multiply the total number of individuals by 2 (as each individual has to alleles)

For every homozygote of the allele you are trying to count (e.g. AA), count it twice (because there are two A's!). For heterozygotes with that allele count them once (e.g. AB).

Divide this number over your doubled total number

Example:

Genotypes	AA	AB	BB	Total
No. individuals	20	30	10	60

**For Total:  $60 \times 2 = 120$**

**For A:  $(20 \times 2) + 30 / 120 = 70 / 120 = 0.583$  ( $p=0.583$ )**

**For B:  $(10 \times 2) + 30 / 120 = 50 / 120 = 0.417$  ( $q = 0.417$ )**

### **Genotype Frequencies**

#### What does it measure?

The proportion of each genotype as it occurs in the population you are looking at (how many of each type are there, relative to each other?)

#### Answers these kinds of questions:

“What is the *frequency* of AB individuals...”

“*If picking an individual at random, what is the probability you would get an AA individual from this population?*” (Remember relative frequencies and probabilities are the same thing!)

#### How do you do it?

Take the number of individuals of each genotype and divide them over the total number of individuals in the population. E.g.:

Genotypes	AA	AB	BB	Total
No. individuals	20	30	10	60
Freq genotypes	$20/60 = 1/3$ (0.33)	$30/60 = 1/2$ (0.5)	$10/60 = 1/6$ (0.17)	1

#### Extra Bits:

You may get a question like this: “If picking an individual at random, what is the probability you would get an AA **or** a BB? (remember for ‘OR’ we add them together. In this case it would be  $0.33 + 0.17 = 0.5$  or 50% chance (as these two genotypes make up 50% of the population)

N.B: The same rules apply for allele frequencies. If asked about selecting an allele at random, you are being asked about allele frequencies.

E.g. “If picking two alleles at random, what is the probability you would get an A **and** B?”. When we get and AND in the question we times the two together:

$\Pr(A) = 0.583$  (from above)

$\Pr(B) = 0.417$

$\Pr(A \& B) = 0.583 \times 0.417 = 0.243$

## **Hardy Weinberg Expected Numbers and Expected Frequencies**

### What does it measure?

Given the number of alleles you have floating around in your population, what arrangements (genotypes) would we expect them to be in, if the population was in H-W)?

### How do you do it?

The first thing you need to do is count the alleles you have in the population (see above). To get expected numbers (used in chi squared test) put the genotype numbers (the ones given – not their frequencies) in to the H-W formula:

#### **-2 alleles**

AA BB AB

$$p^2 + q^2 + 2pq$$

#### **-3 alleles**

AA BB CC AB AC BC

$$p^2 + q^2 + r^2 + 2pq + 2pr + 2qr$$

(where A = p, B = q, C = r etc)

#### **-Sex linked (X linked)**

Males: only one copy so

A B

p q

Females: have two copies so looks like normal 2 allele H-W

AA BB AB

$$p^2 + q^2 + 2pq$$

*If you are asked for expected frequencies put these numbers over the total number of genotypes.*

*Each sex separately should sum to 1 for frequencies in X linked HW.*

#### **-Dominance**

In this case we can only see two phenotypes. One is the heterozygous recessive ( $q^2$ ) and the other a combination of the homozygous dominant and heterozygote ( $p^2 + 2pq$ )

Start by getting q from  $q^2$

Once you have that, you can get p by  $1 - q$ . (See tutorial questions week 2 for an example)

## **Chi Squared ( $X^2$ )**

### What does it measure?

Measures how different a set of data is to what is expected of that set. Usually seeing if a population conforms to H-W.

### Answers these kinds of questions

“Check to see if the data conforms to... (H-W etc)”; “Do these meet the expectations of...”

### How do you do it?

$$X^2 = \sum (\text{Obs} - \text{Exp})^2 / \text{Exp}$$

*This can only be used on actual numbers – no good for frequencies.*

To get the numbers:

**Obs:** the numbers of individuals in each genotype. You are usually given these

**Exp:** get the allele frequencies (p and q) by counting. (See above). Multiply each frequency according to H-W (see above) to get  $p^2$ ,  $q^2$ ,  $2pq$  etc. (These should still all sum to 1). These are your expected genotype frequencies. We want numbers. So multiply your frequencies by the total number of individuals in the population. These are the expected values that you use

### Observed Heterozygosity ( $H_o$ )

What does it measure?

How many heterozygotes are there in the population? What percentage of individuals are heterozygotes?

How do you do it?

Count the number of heterozygotes you have (ABs, BCs etc) and then divide them over the total number of individuals

E.g.

Genotypes	AA	AB	BB	Total
No. individuals	20	30	10	60
Heterozygotes		30/60 = 1/2 (0.5)		

$H_o = 0.5$ . Half the individuals are heterozygotes

### Expected Heterozygosity ( $H_e$ )

What does it measure?

The proportion of heterozygotes for a **single loci** with multiple alleles, given H-W equilibrium

Answers these kinds of questions

“calculate heterozygosity ( $H_e$ ) for this locus”

How do you do it?

$$H = 1 - \sum p_i^2$$

This means 1- the sum of the alleles squared.

$H_e = 1 - (p^2 + q^2 + r^2 + \text{etc})$  E.g. (using the same data from above):

$p = 0.583$

$q = 0.417$

$H_e = 1 - (0.583^2 + 0.417^2)$

$H_e = 1 - 0.5138$

$H_e = 0.48622$

### Average Heterozygosity ( $H_e$ )

What does it measure?

The proportion of heterozygotes for a **multiple loci** with multiple alleles, given H-W equilibrium

How do you do it?

Sum of your heterozygosity measures ( $H_e$ 's) / Number of loci you measured them for.

I.e. take an average of all the  $H_e$  measures you have done. Easy! Deceptively complicated formula looks like this:

$$\frac{\sum H_i}{N}$$

## Polymorphism (P)

What does it measure?

How many *loci* are *polymorphic* out of **multiple loci**

How do you do it?

$P = \text{polymorphic loci} / \text{total loci}$

Count the number of loci

Count how many of these are polymorphic (have more than one allele)

## Allelic Diversity (A)

What does it measure?

The average number of alleles per loci (used for multiple loci)

How do you do it?

$$\sum(\text{_____})$$

Count how many alleles there are total (across all the loci)

Divide by number of loci you are looking at

E.g. (The numbers given are allele freqs)

Locus	Allele 1	Allele 2	Allele 3
1	0.60	0.24	0.16
2	0.63	0.17	0.10
3	0.67	0.33	0.00
4	0.76	0.14	0.00
5	1.00	0.00	0.00
6	1.00	0.00	0.00

Total number of alleles = 12 (Shaded cells)

Total number of loci = 6

$A = 12/6$

$A = 2$

## Effective Alleles ( $n_e$ )

What does it measure?

How evenly alleles are distributed. Use for a SINGLE loci. If there are multiple loci, calculate  $n_e$  for each one, and then take an average.

How do you do it?

$$n_e = \frac{1}{\sum P_i^2}$$

Where:

$P_i$  = frequency of each allele

1 divided by the sum of the frequencies of EACH ALLELE squared.

**E.g. If given allele frequencies:**

Locus	Allele 1	Allele 2	Allele 3
1	0.60	0.24	0.16
2	0.63	0.17	0.10

**ne for locus 1** =  $1 / (0.6)^2 + (0.24)^2 + (0.16)^2$

**n<sub>e</sub>** = 2.25

**ne for locus 2** =  $1 / (0.63)^2 + (0.17)^2 + (0.10)^2$

**n<sub>e</sub>** = 2.295

### Linkage Disequilibrium (D)

What does it measure?

Whether or not there is linkage happening in the population.

If there is no linkage – the two loci are assorting independently. The population is in *equilibrium*. If there is linkage between the two loci then there is *linkage disequilibrium*.

How do you do it?

$D = ru - st$  OR

$D = (\text{same} \times \text{same}) - (\text{different} \times \text{different})$

Where:

A1B1 and A2B2 are the same

A1B2 and A2B1 are different

E.g.

	A1B1	A1B2	A2B1	A2B2	Total
No. individuals	20	30	10	60	120
Frequencies	0.16	0.25	0.083	0.5	1
	r	s	t	u	

$D = (0.16 \times 0.5) - (0.25 \times 0.083)$

$D = 0.08 - 0.02075$

$D = 0.05925$

If  $D \neq 0$ , then there is linkage disequilibrium.

Chi-square degree of freedom in linkage disequilibrium is always 1

## **Response to Evolution / Adaptive Potential**

What does it measure?

The expected response to selection

Answers these kinds of questions:

“We conducted an *artificial selection experiment*...”;

“Say you wanted to breed bigger/smaller/faster/greener....” ; “A group of fish were *selectively bred*..”

How do you do it?

Using the breeders equation:  $R = S \times h^2$ ,

where:

$S$  = selection differential. This is the difference of the overall population mean and the mean of the individuals that you select.

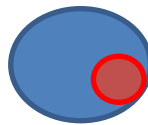
$h^2$  = heritability estimate (See above)

E.g. Imagine all the individuals are contained in the blue circle. This is your population.

Blue circle individuals have a mean height of 10. Within that circle are the very tall individuals. They have a mean of 15.

$S = 10 - 15 = 5$

$h^2$  for height is given as 0.5



$R = S \times h^2$

$R = 5 \times 0.5 = 2.5$

So if we breed only the red individuals, we expect their offspring to be on average 2.5 units taller. This is 2.5 units taller than the ORIGINAL mean (10 – from the blue circle) not the selected mean (15 – red circle).

$10 + 2.5 = 12.5$  is the expected mean height of the new individuals.

## **Heritability ( $h^2$ / $H^2$ )**

### **The Quick Guide**

Narrow Sense =  $V_A / V_P$ . Calculated with parents and offspring.

Broad Sense =  $V_G / V_P$ . Calculated with siblings. ( $V_G = V_A + V_I + V_{CE}$ )

$V_P = V_A + V_I + V_{CE} + V_E$

### **-Broad Sense**

What does it measure?

How much genetic variance ( $V_G$ ) can account for (causes) the variance you measure in the phenotype ( $V_P$ ).

The variance in genetics that you are looking at ( $V_G$ ) includes variance in genes which are additive, dominant, and epistatic, as well as the confound of shared environments. Broad sense heritability can be estimated by looking at similarities between SIBLINGS.

How do you do it?

$H^2 = V_G / V_P$

Where  $V_G = V_A + V_I + V_{CE}$

Where  $V_P = V_G + V_E$  (or  $V_P = V_A + V_I + V_{CE} + V_E$ )

### **-Narrow Sense**

What does it measure?

How much additive genetic variance ( $V_A$ ) can account for (causes) the variance that you measure in the phenotype ( $V_P$ ). The variance in genetics that you are looking at ( $V_A$ ) only includes variance in genes which quantifiably add to the phenotype. Narrow sense heritability can be estimated by looking at similarities between PARENTS AND OFFSPRING.

How do you do it?

$h^2 = V_A / V_P$

Where  $V_P = V_A + V_I + V_{CE} + V_E$