Evolution 4

- Population Genetics II
  - Hardy-Weinberg Equilibrium
    - Equilibrium
    - Human genetic traits
    - is a population at HWE?
    - examples
- iClicker Question #1

Yadyra C see me

Tim A see me

- Labs start this week; go to HMNH
  BE PREPARED
- no pre-lab this week; report due wk of 2/8
- go to HMNH anytime Mon-Sat 9-5 & Sun 1-5

* don't forget "Tree Building Survey" - see link at Evolution 6 (due Evolution 7)

from last time: if, in parental generation,

freq of R: $p = 0.2$ & freq of r: $q = 0.8$

& all 5 conditions hold

then the offspring will be:

× freq of RR kids $= p^2 = 0.04$

× freq of Rr kids $= 2pq = 0.32$

× freq of rr kids $= q^2 = 0.64$

if 100 kids ~~total~~

4 v. fast kids

32 fast kids

64 slow kids

if all these kids ~~have kids~~ (& all 5 conditions hold)

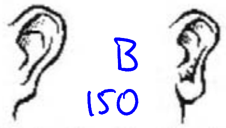
the allele & genotype frequencies will stay the same in all following generations

= Hardy-Weinberg equilibrium (HWE)

= NO EVOLUTION

Uses of HWE (1): Human Genetic Traits

Example: attached / detached earlobes (model for genetic disease)



Are your earlobes attached or unattached? A person with attached ear lobes will have the lowest point of the ear lobe attached to the face. A person with unattached ear lobes will not have the lowest point of the ear lobe attached to the face.

- large pop (yes)
- no mutation (low rate)
- no migration
- random mating } mating / survival do not depend on earlobes
- no selection }

allele	contribution to phenotype	frequency
E	unattached earlobe (dominant)	p
e	attached earlobe (recessive)	q

at HWE

	Genotype	Phenotype
p^2	EE	unattached
$2pq$	Ee	unattached
q^2	ee	attached

Q: How many people in class are carriers for the attached allele?

Assume that this trait is at HWE.

Problem: EE, Ee have same phenotype, so can't find p^2 directly

but $\text{freq (ee)} = q^2 = \dfrac{60 \text{ attached}}{210 \text{ total}} = 0.286$

$\therefore q = \sqrt{0.286} = 0.534$ & since $p + q = 1$ $p = 1 - q = 0.466$

$\therefore$ freq of carriers is $2pq = 2(.534)(.466) = 0.498$

$\therefore$ in class of 210 students, $210 \times 0.498 = 104$ should be carriers

Compare with reality - expect freq to be constant

uses (2) studying evolution

Q: is population X at HWE? (not evolving)

step 1: calculate allele frequencies of X

step 2: predict genotype freqs if X were at HWE

step 3: compare predicted and actual

example: given genotype #

example: given

genotype	#
AA	25
Aa	25
aa	50

is this pop. at HWE?

step 1: find allele freqs (since we know all genotype freqs)

$$\text{freq}(A) = p = \frac{\overset{2\times AA\ +\ Aa}{(2\times 25) + 25}}{200 \text{ total}} = 0.375$$

$$\text{freq}(a) = q = \frac{\overset{2\times aa\ +\ Aa}{(2\times 50) + 25}}{200 \text{ total}} = 0.625$$

check that $p+q=1$ Yes

∴ calculations are ok

- but not nec. at HWE

Step 2: if pop were at HWE (⟺ all 5 conditions hold) we'd expect:

genotype	predicted freq at HWE	Step 3 observed frequencies
AA	$p^2 = 0.140$	0.25
Aa	$2pq = 0.469$	0.25
aa	$q^2 = 0.391$	0.50

observed ≠ HWE predictions

∴ pop is not at HWE

∴ must be evolving

∴ one or more of HWE assumptions is being violated

Bio 112: Hardy-Weinberg Equilibrium Examples

General Info

- Allele frequencies: the frequency of each allele (R or r, for example) in the gene pool. The symbols p and q are used to represent these frequencies.
- Genotype frequencies: the frequency of each genotype (RR, Rr, rr for example) in the population. These are always equal to the number of individuals with a particular genotype divided by the total population size. They are sometimes equal to p^2, 2pq, and q^2 - only when the population is at HWE.

For a particular pair of allele frequencies (p = 0.2 and q = 0.8 for example), there are many possible sets of genotype frequencies that have the same allele frequencies (actually, infinitely many). This is illustrated by the 4 example populations below; all 4 of these populations have the same allele frequencies. However, given a pair of allele frequencies, there is only one set of these genotype frequencies that are at HWE. This is illustrated by the last two populations shown below; in addition to having the same allele frequencies as the other populations, their genotype frequencies match the predictions of HWE - that the frequency of RR = p^2 (0.04 in this example), the frequency of RR = 2pq (0.32 in this example), and the frequency of rr = q^2 (0.64 in this example).

Population 1

Genotype	#	Genotype frequency	#R's contributed to gene pool	#r's contributed to gene pool
RR	0	0	0	0
Rr	40	0.40	40	40
rr	60	0.60	0	120
		totals	40	160

Allele frequencies
Freq. of R = p = 40/200 = 0.2
Freq. of r = q = 160/200 = 0.8
Genotype frequencies
Freq. of RR = 0, not 0.04
Freq. of Rr = 0.4, not 0.32
Freq. of rr = 0.6, not 0.64
NOT AT HWE

different

same AF

Population 2

Genotype	#	Genotype frequency	#R's contributed to gene pool	#r's contributed to gene pool
RR	400	0.20	800	0
Rr	0	0.00	0	0
rr	1600	0.80	0	3200
		totals	800	3200

Allele frequencies
Freq. of R = p = 800/4000 = 0.2
Freq. of r = q = 3200/4000 = 0.8
Genotype frequencies
Freq. of RR = 0.2, not 0.04
Freq. of Rr = 0.0, not 0.32
Freq. of rr = 0.8, not 0.64
NOT AT HWE

Population 3

Genotype	#	Genotype frequency	#R's contributed to gene pool	#r's contributed to gene pool
RR	4	0.04	8	0
Rr	32	0.32	32	32
rr	64	0.64	0	128
		totals	40	160

Allele frequencies
Freq. of R = p = 40/200 = 0.2
Freq. of r = q = 160/200 = 0.8
Genotype frequencies
Freq. of RR = 0.04
Freq. of Rr = 0.32
Freq. of rr = 0.64
AT HWE

same AF

different

same

Population 4

Genotype	#	Genotype frequency	#R's contributed to gene pool	#r's contributed to gene pool
RR	12	0.04	24	0
Rr	96	0.32	96	96
rr	192	0.64	0	384
		totals	120	480

Allele frequencies
Freq. of R = p = 120/600 = 0.2
Freq. of r = q = 480/600 = 0.8
Genotype frequencies
Freq. of RR = 0.04
Freq. of Rr = 0.32
Freq. of rr = 0.64
AT HWE