

Introductory Biology -- Population Genetics Problem Set

1. Suppose that you study the genetics of a locus that controls flower color (C). There is no dominance and homozygous C^R give red flowers, homozygous C^W give white flowers and the heterozygotes are pink. You go out and write down the phenotypes of a number of plants and get the following:

red = 70
pink = 120
white = 110

(a) What are the allele frequencies in the study population? What are the genotype (phenotype) frequencies in this population?

Let the frequency of allele $C^R = p$ and
the frequency of allele $C^W = q$

$$\begin{aligned} p &= (\text{total num. of } C^R \text{ alleles}) / \text{total num. of alleles in pop.} \\ &= ((2 * \# \text{ homozygotes}) + \text{heterozygotes}) / 2 * (\text{total pop size}) \\ &= (2 * 70 + 120) / 2 * (70+120+110) = 260 / 600 = \mathbf{0.433} \end{aligned}$$

$$\text{and since } q = 1 - p = 1.0 - 0.433 = \mathbf{0.567}$$

Using the same concept to calculate phenotypes frequencies:

freq. = # of a certain phenotype / total number of individuals

$$\text{freq. (red)} = 70 / (70+120+110) = 0.233$$

$$\text{freq. (pink)} = 120 / 300 = 0.40$$

$$\begin{aligned} \text{freq. (white)} &= 1.0 - \text{freq. (red)} - \text{freq. (pink)} = 1.0 - 0.233 - 0.4 \\ &= 0.367 \end{aligned}$$

Check for freq.(white) --- $110/300 = 0.367$
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(b) Is this population at Hardy-Weinberg Equilibrium?

We know the actual allele frequencies in the entire population from the calculation we did in #1(a).

We need to use the Hardy-Weinberg expression to generate a series of expected genotypic frequencies. These are the frequencies that would exist given the allele frequencies we just worked out IF THE POPULATION IS AT H-W EQUILIBRIUM.

$$\text{freq. of red} = \text{freq. } C^R C^R = p^2 = 0.433 * 0.433$$

$$\text{freq. of white} = \text{freq. } C^W C^W = q^2 = 0.567 * 0.567$$

$$\text{freq. of pink} = \text{freq. } C^R C^W = 2pq = 2 * 0.433 * 0.567$$

NOTE THAT THESE MUST SUM TO 1.0 SINCE

$$p^2 + 2pq + q^2 = 1.0$$

THUS:

$$0.433^2 + 2 * 0.433 * 0.567 + 0.567^2 = \mathbf{0.1875 + 0.491 + 0.3215}$$

(Notice that these DO all add to 1.0, as they must regardless of whether or not the pop is at H-W equilibrium. IF THEY DON'T YOU HAVE MADE A MATH ERROR – NOTHING MORE, NOTHING LESS)

Now, when we compare them to the frequencies in the actual population (last page) note that:

$$\text{observed red flowers} = 0.233$$

$$\text{predicted reds (H-W)} = 0.433^2 = 0.1875$$

We need go no further because these are not equal. The observed does not come close to the expected and so the population is not at H-W equilibrium. For practice sake, let's go ahead anyway.

$$\text{observed white flowers} = 0.367$$

$$\text{predicted reds (H-W)} = 0.3215$$

(not very close to predicted by H-W)

observed pink flowers = 0.40

predicted pinks (H-W) = 0.491

(not even close)

The observed values do not reflect the values predicted from the allele frequencies in the observed population. Thus the population is not at equilibrium -- one of more of the H-W assumptions has been violated.

2. Suppose that the following frequencies are observed for blood type alleles in a human population:

$$I^A = 0.40$$

$$I^B = 0.05$$

$$i = 0.55$$

(a) Write and expand an equation to predict the frequency of each genotype at Hardy-Weinberg equilibrium.

$$\text{let freq.}(I^A) = p$$

$$\text{let freq.}(I^B) = q$$

$$\text{let freq.}(i) = r$$

$$\text{so: } p + q + r = 1.0$$

Since human folk, like most organisms, are diploid, all possible combinations of alleles are derived by squaring this binomial:

$$(p + q + r)^2 = 1.0^2$$

$$p^2 + 2pq + 2pr + q^2 + 2qr + r^2 = 1.0$$

(b) If the population is at Hardy-Weinberg equilibrium, give the frequency of each of the following:

(i) type "O" individuals

These are homozygous recessive (earlier in the course) and therefore if **the population is at H-W equilibrium**:

$$\text{freq. (type O)} = r^2 = 0.55^2 = \mathbf{0.3025}$$

(ii) A blood types who are homozygotes

$$I^A I^A = p^2 = 0.40^2 = \mathbf{0.16}$$

(iii) total proportion who have type A blood

$$\text{This is } I^A I^A + I^A i = p^2 + 2pr = 0.4^2 + 2 * 0.4 * 0.55 = \mathbf{0.60}$$

(iv) frequency of type AB

$$\text{This is } I^A I^B = 2pq = 2 * 0.4 * 0.05 = \mathbf{0.04}$$

(c) What will the frequencies be of alleles I^A , I^B , and i in the next generation? How about type O? What will be the frequency of each allele and type O in three more generations?

This is easy -- since the population is given as being at H-W equilibrium, there will be total stasis of all genotype and allele frequencies. Thus, no changes from above.

3. Suppose that you know a population is at or very close to H-W equilibrium with respect to a certain locus. Furthermore, assume that this locus, called A, shows complete dominance with only two alleles present (the dominant and recessive). Suppose that you count individuals with the two different phenotypes and find:

Phenotype A	125
Phenotype a	375

(a) What are the frequencies of the two phenotypes?

There are a total of 500 individuals in the pop.

0.25 show the dominant (A) phenotype ($125/500 = 0.25$) show the dominant (A)

therefore $1 - 0.25 = 0.75$ show the recessive (a) phenotype (since there are only two phenotypes present). (note – it is equally correct to compute this answer as $375/500 = 0.75$)

(b) What genotypes make up each phenotype?

A phenotype = AA + Aa

a phenotype = aa

(c) Assuming that the freq. (A) = p and freq. (a) = q, give the expressions from the H-W equation used to calculate the frequencies of each genotype.

$$AA = p^2 ; Aa = 2pq ; aa = q^2$$

(d) Calculate the frequencies of each allele. Note -- you will not be able to use the equation we learned in class that works for situations where there is lack of dominance. Why? Hint: Look at your answer to part (c) and see if any of H-W terms for genotypes corresponds to a group that we did actually measure.

The obvious problem is that you can't tell whether A phenotype individuals are homozygous or heterozygous and therefore you cannot use the equation we learned to find the frequencies for any population where you can tell them apart (example -- where there is no dominance). However, since :

$$q^2 = \text{freq. (a phenotype - which we can measure)}$$

AND since we know that this is a H-W population then:

$$q = \sqrt{q^2} = \sqrt{\text{freq. ("a" phenotype)}}$$

$$\text{then } q = 0.75^{0.5} = 0.866$$

-- we need not know the proportion of alleles in the heterozygotes in this case!

$$\text{So, the } p = \text{freq. (A allele)} = 1.0 - 0.866 = 0.134$$

(e) Find the frequencies of each homozygote and the heterozygotes.

Since the population is given as a H-W population, then:

freq.(AA genotype) = $0.134^2 \approx 0.0179$ (*i.e.*, about 9 individuals -- see why the large pop matters!)

freq.(heterozygote) = $2 \cdot 0.134 \cdot 0.866 \approx 0.232$

NOTE THAT THE FREQUENCY OF THE DOMINANT PHENOTYPES IS $0.232 + 0.0179 \approx 0.25$ (-- THE SAME AS 125, AS GIVEN)

The last step is trivial since we just reverse what we had already done to find "q"

freq.(aa genotype) = $q^2 = 0.866^2 = 0.75$ or the same as 375 individuals in the population of 500

(f) Would this technique work if the pop was not at H-W equilibrium?

Absolutely NOT -- it is based on the equilibrium between the heterozygote and homozygous recessives that only occurs under H-W conditions - any other use is fallacious. Other techniques must be used when the pop is not at H-W equilibrium.

(g) If a pop. is at H-W equilibrium for one locus, must it also be for others? Explain.

Absolutely not. Although selection operates on the individual, selection still affects different loci differentially. Here's an easy way to remember this. The locus you have for blood type matters very little at all and is probably close to H-W equilibrium within most human populations. On the other hand, imagine another locus in your body that in most people does a certain set of things that results in normal function but in a few homozygous recessive individuals produces disease. Obviously, the recessive will be slowly removed in this case and therefore the allele

frequencies will change from one generation to the next with the recessive decreasing.

(h) If a population is at H-W equilibrium for a number of generations and suddenly there is selection for one of the homozygotes over the other phenotypes, what will happen to the other phenotypes and to the allele frequencies? (give a qualitative answer).

See the previous answer -- once selection starts, the allele that produces the favored homozygote will increase. The other allele must then decrease. The favored homozygotes will gradually become more frequent and the less favored homozygotes increasingly rare; as will the heterozygotes.