Laboratory 8: Population Genetics and Evolution

*calculate allele and genotype frequencies using the Hardy-Weinberg theory
*discuss the effect of natural selection on allelic frequencies
*explain and predict the effect on allelic frequencies of selection against the homozygous recessive
*discuss the relationship between evolution and changes in allele frequencies, as measured by changes from the Hardy-Weinberg law of genetic equilibrium

In 1908 G. H Hardy and W. Weinberg independently suggested a scheme whereby evolution could be viewed as changes in the frequency of alleles in a population of organisms. Two formulas were used:

p (% dominant allele) + q (% recessive allele) = 1 referring to allele frequencies

p^2 (% homozygous dominant) + $2pq$ (hetero) + q^2 (% homo recessive) = 1 referring to genotype frequencies

These will remain constant from generation to generation only if five conditions are met:

1. the population is large
2. mating is random
3. no mutations occur
4. there is no immigration nor emigration
5. there is no natural selection

If any of these conditions are NOT met, then one should observe a change in the allele and genotype frequencies for the population....

****

The class is tested for ability to taste the chemical PTC (phenylthiocarbamide); a bitter-taste indicates presence of the dominant allele -- individuals will be homozygous dominant or heterozygous. Non-tasters are homozygous recessive! Once data is obtained, the frequency of each allele should be calculable......

	Phenotypes		Allele Frequency Based on the H-W Equation	
	% Tasters ($p^2 + 2pq$)	% Nontasters (q^2)	p	q
Class Population	.91	.09	.7	$\sqrt{.09}$ = .3
North American Population	0.55	0.45	.33	$\sqrt{.45}$ = .67

****

A simulation of Hardy-Weinberg can be made by having class members assume genotypes based on original allele frequencies and then "mating" with other class members to produce new combinations of genes....If an AA student mates with an aa student, all the offspring will be Aa; but if both parents are Aa, there are several possibilities for offspring. If the two offspring produced are then allowed to randomly produce two other offspring and so on, at the end of several generations, we should be able to see if frequencies remain stable.

32

Generation	Offspring's Genotype	Class Totals for Each Genotype		
	(AA, Aa, or aa)	AA	Aa	aa
1		8	17	7
2		9	18	5
3		9	19	4
4		9	18	5
5		7	18	7

A
16+17
33

a
17 + 14
31

****

It is possible to simulate selection by "killing off" one of the genotypes (for example, aa cannot survive) and noting the change in frequency of alleles after several generations.

Generation	Offspring's Genotype	Class Totals for Each Genotype	
	(AA or Aa)	AA	Aa
1		12	20
2		14	18
3		17	15
4		18	14
5		21	11

A
24+20

a
20

A
42+11

a
11

Free-Response Question 4

In a laboratory population of diploid, sexually reproducing organisms a certain trait is determined by a single autosomal gene and is expressed as two phenotypes. A new population was created by crossing 51 pure-breeding (homozygous) dominant individuals with 49 pure breeding (homozygous) recessive individuals. After four generations, the following results were obtained.

NUMBER OF INDIVIDUALS

Generation	*Dominant*	*Recessive*	*Total*
1	51	49	100
2	280	0	280
3	240	80	320
4	300	100	400
5	360	120	480

a. *Identify an organism that might have been used to perform this experiment, and explain why this organism is a good choice for conducting this experiment.*
b. *On the basis of the data, propose a hypothesis that explains the change in the phenotype frequency between generation 1 and generation 3.*
c. *Is there evidence indicating whether or not this population is in Hardy-Weinberg equilibrium? Explain.*

Question 4 Standards

Overall Commentary for Question 4

Question 4 is divided into three sections; addressing the subjects of appropriate choice of an experimental organism, Mendelian genetics, and the Hardy-Weinberg equilibrium. A perfect score of 10 could not be obtained unless a student included some correct response for each of the three parts (a maximum of 4 points can be earned for each section).

In part a, the students are asked to choose an appropriate organism which could have been used to provide the data and to justify their choice. A single point was awarded for naming an organism which might have had the given number of offspring in a reasonable amount of time. Appropriate organisms had to be diploid, sexually reproducing, and have a reasonably short life cycle. Acceptable answers therefore included: *Drosophila melanogaster* or fruit fly, housefly, mouse, rabbit, dog, cat, slug, a named diploid plant, e.g., maize, pea, or Brassica; or any other organism which meets the criteria stated. (*Note:* Peas are normally self-fertilizing plants and thus not ideal because their heterozygosity would decrease in each generation by a factor known as the inbreeding coefficient, or F; however, they were considered acceptable because they may have been crossbred by the experimenter.) Organisms not acceptable included: long-lived, prokaryotic, fungal (except diploid yeast), polyploid, protistan, or human organisms. One to three points could have been awarded for the students' justification of the organism

chosen. Such justification could include the ability of the organism to produce a large number of offspring per generation; having a reasonably short life cycle; being easy to maintain or easy to control in a laboratory; having clear, easily identified phenotypic traits or clear gender dimorphism; and interbreeding freely (without inbreeding). A maximum of 4 points was available for the first section.

In part b, students are asked to propose a hypothesis to account for the changes in phenotypic frequencies from the first to the third generation on the basis of the data. A point was given for a clear and logical hypothesis explaining all three generations; preferably in the typical if:then formulation, although any logical statement of a hypothesis was scoreable. One to two additional points were awarded for a correct explanation of these changes: one for the change from generation one to generation two because two homozygotes produce only heterozygotes; and another for the corresponding change from generation two to three, showing the genotypic ratios which would account for the data. A Punnett square could be used as an illustration in this context. Alternatively, a single point could be earned in the absence of either of the last two, for an overall explanation which reasoned from phenotype alone without allusion to genotype. One to two additional points were awarded if the student elaborated on the explanation of dominance and/or on the explanation of Mendel's law of segregation, which would account for the data. A maximum of 4 points was given for this section.

Part c asks the student whether or not there is evidence that the population is in Hardy-Weinberg equilibrium and further asks for an explanation for the answer given. The student received a point for the correct "yes" answer, but only if this was accompanied by at least some correct explanation. The next point was given if the student recognized that, at equilibrium, allele and genotype frequencies do not change. A further point was awarded if there was a definition or description of the equilibrium, usually in terms of the formula for distribution of genotypes after one generation: $p^2 + 2pq + q^2 = 1$. Another point was awarded if there was a calculation of q ($q^2 = 0.25$; $q = 0.5$). A final point was given if the student recognized three of the five conditions required for the equilibrium to occur: random mating, a large population, no (net) migration, no mutation, or no selection. A maximum of 4 points was possible in this section.

Part a: Choice of organism

Maximum 4 points

1 point — Name of organism that could be used to produce the kind of data shown:

> *Drosophila melanogaster* or fruit fly, housefly, mouse, dog, cat, rabbit, slug, named diploid plant, e.g., maize, pea, or Brassica; any other organism which reproduces sexually, is diploid, reproduces often, and has a reasonably short life cycle. (Peas accepted only because they may have been crossbred by the experimenter.) *(Not accepted were long-lived, prokaryotic, fungal [except diploid yeast], polyploid, protistan, or human organisms.)*

1-3 points — Reasons for choice:

- large number of offspring/generation
- reasonably short life cycle/generation
- easily maintained organisms OR easily controlled conditions
- clear, easily identified phenotypic traits/clear gender dimorphism
- interbreed freely (without inbreeding)

Part b: on the basis of data, hypothesis to explain change from generation 1 – 3; Mendelian genetics

Maximum 4 points

1 point — Correct formulation of a hypothesis; (if:then) logical statement
1 point — Explanation of genotypic change from generation 1 to 2 (AA × aa → Aa)
1 point — Explanation of genotypic change from generation 2 to 3 (Aa × Aa → ¼ AA, ½ Aa, ¼ aa) or Punnett square

OR

1 point — For only description of phenotypic change if neither of the above two points are given
1 point — Explanation of dominance (not just use of the word)/explanation of Heterozygosity
1 point — Explanation of Mendel's law of segregation

Part c: evidence for Hardy-Weinberg equilibrium

Maximum 4 points

1 point — Yes, with some correct explanation
1 point — Recognition that, at equilibrium, allele and genotype frequencies do not change
1 point — Describes Hardy-Weinberg equilibrium ($p^2 + 2pq + q^2 = 1$ after 1 generation)
1 point — Calculation of p and/or q ($q^2 = 0.25$; $q = 0.5$)
1 point — Elaboration: H-W only maintained if population is large, randomly mating, has No(net) mutation, no migration, or no selection for alleles in question (minimum of 3 stated)

4 a) A good organism that would be good for conducting such a study would be fruit flies. *Drosophila* have a short life-cycle and they reproduce in only a few days. Their phenotypes can clearly be observed with the aid of a disecting microscope. Also, *Drosophila* tend to follow basic Mendelian genetics and they also have a small number of chromosomes.

b) A large phenotypic change is seen between generation 1 and generation 3. ~~This change due~~ In generation 1 there is almost a 1:1 ratio of the dominant phenotype to the recessive phenotype. This ratio changes to a 3:1 ratio in the 3rd generation. This change can be explained by using simple Mendelian genetics. When an organism possesses the dominant allele (A), this allele masks the presence of the recessive allele (a). Thus when the homozygous dominant individuals ~~that~~ were crossed with the homozygous recessive individuals, they resulted in heterozygous individuals. As the punnet square shows

	A	a
A	AA	Aa
a	Aa	aa

~~these~~, when 2 heterozygous individuals are crossed the expected phenotypic ratio is 3 dominant : 1 recessive. This is the observed ratio.

c) The population is in Hardy Weinberg equilibrium. this can be seen because 360 of the 480 individuals exhibits the dominant phenotype. this is 75% of the individuals. That means that 25% (q^2) of the individuals exhibit the homozygous recessive. $p^2 + q^2 + 2pq = (p + q)^2 = 1$. q, which equals the abundance of the recessive allele must then equal $\sqrt{.25}$ which equals .5 This means that the population is 50% the recessive allele; and 50% the dominant all This ratio is simaler to the initial ratio of 49:51. Because there was very little change in allele frequencies over several gene the population must be in Hardy-Weinberg equilibrium. This means that no excessive migration, no natural selective random mating, a large sample size, and no mutations existed to large degree as condition on. The population.

Question 4 Overview

How prepared were the students?

The distribution of scores shows that nearly all students clustered tightly around the 4.57 mean which was maintained with no variation throughout the reading. Only 2% scored 0 and 15% less than 3; only 2% scored 10 as well, and 12% above 7. From the Readers, we know that the typical student, earning a score of 4 or 5, gained 3-4 points in part a, on the choice of a research organism, and one or two points on either part b or c.

In part b, students rarely wrote a plausible hypothesis and most typically only explained the Mendelian basis for the data. When hypotheses were attempted, they most often included only the first generation; students therefore rarely gained this point.

With regard to the Hardy-Weinberg equilibrium, students seem to describe it most often only in terms of its exceptions, i.e., the criteria under which it may obtain. Most often, in answering the question, students remarked on the stability of a 3:1 ratio rather than on the stability of alleles or genotypes.

How were the standards applied to the sample student responses?

FAIR ESSAY (5 points)

This response demonstrates an intermediate level of proficiency. The student chose a reasonable organism, the mouse, and justified the choice because of litter size and ease of handling. In part b, the student gave a correct explanation for the change to generation 2. In the final section, the student correctly answered "yes" with some correct information, however vague.

GOOD ESSAY (7 points)

This response is moderately proficient and receives a score of 7 points. The student chooses mice as an experimental organism and sees that they reproduce quickly and have clear characteristics. The response explains the changes from generation one to two and from two to three and explains dominance. In part c, the student only recognizes that equilibrium does exist.

EXCELLENT ESSAY (10 points)

This response demonstrates true competency and earns 10 points. The student correctly chooses an organism and gives two good justifications for the choice. In part b, both generations are explained and the student receives a point for explaining dominance. Part c is especially impressive, since the student correctly describes the equilibrium, correctly explains the formula, calculates the allele frequency, and understands that the relationship remains constant. Although the student has already received the maximum points allowed, the response goes on to explain the criteria for the equilibrium to be maintained.