Biology 317

Hardy-Weinberg Equilibrium

The biological sciences now generally define **evolution** as being the sum total of the genetically inherited changes in the individuals who are the members of a population's gene pool. It is clear that the effects of evolution are felt by individuals, but it is the population as a whole that actually evolves. Evolution is simply a change in frequencies of **alleles** in the **gene pool** of a **population.** For instance, let us assume that there is a trait that is determined by the inheritance of a gene with two alleles--B and b. If the parent generation has 92% B and 8% b and their offspring collectively have 90% B and 10% b, evolution has occurred between the generations. The entire population's gene pool has evolved in the direction of a higher **frequency** of the b allele--it was not just those individuals who inherited the b allele who evolved.

In 1908 Hardy and Weinburg concluded that gene pool frequencies are inherently stable but that evolution should be expected in all populations virtually all of the time. They resolved this apparent paradox by analyzing the net effects of potential evolutionary mechanisms.

For a population to be in **Hardy-Weinberg equilibrium**, (where allele and genotypic frequencies will remain constant over time) it must satisfy five main conditions:

1. **The population is very large**
2. **There is no gene flow; that is, there is no migration of individuals or gametes into or out of the population.**
3. **Mutations (changes in genes) do not alter the gene pool.**
4. **Mating is random.**
5. **All individuals are equal in reproductive success; that is, natural selection does not occur.**

These five conditions are rarely met, therefore, we do not expect populations to be in Hardy-Weinberg equilibrium.

If no mechanisms of evolution are acting on a population, evolution will not occur--the gene pool frequencies will remain unchanged. However, since it is highly unlikely that any of these seven conditions, let alone all of them, will happen in the real world, evolution is the inevitable result. Hardy and Weinberg went on to develop a simple equation that can be used to discover the probable genotype frequencies in a population and to track their changes from one generation to another. This has become known as the **Hardy-Weinberg equilibrium equation**. In this equation ($p^2 + 2pq + q^2 = 1$), p is defined as the frequency of the dominant allele and q as the frequency of the recessive allele for a trait controlled by a pair of alleles (A and a). In other words, p equals all of the alleles in individuals who are homozygous dominant (AA) and half of the alleles in people who are heterozygous (Aa) for this trait in a population. In mathematical terms, this is

$$p^2 + 2pq + q^2 = 1$$

where, **p = frequency of the dominant allele in the population**

q = frequency of the recessive allele in the population

p^2 = percentage of homozygous dominant individuals

q^2 = percentage of homozygous recessive individuals

2pq = percentage of heterozygous individuals

Suggested Method:

1. Attempt to use the recessive trait to find allele frequency.
   (If q = 16, the allele freq. = square root which = 0.04)
2. The dominant + recessive frequencies = 1
   (p + q =1, therefore, p + 0.4 = 1, then p = 0.6)
3. Determine the individual frequencies using
   $p^2 + 2pq + q^2 = 1$

Sample Problem: Albinism

Albinism is a rare genetically inherited trait that is only expressed in the phenotype of homozygous recessive individuals (aa). The most characteristic symptom is a marked deficiency in the skin and hair pigment melanin. This condition can occur among any human group as well as among other animal species. The average human frequency of albinism in North America is only about 1 in 20,000. Referring to the Hardy-Weinberg equation $p^2 + 2pq + q^2 = 1$, the frequency of homozygous recessive individuals is q^2. Therefore:

$q^2 = 1/20{,}000 = 0.00005$

or, $q = 0.007$ (in other words, the frequency of the recessive albino allele (a) is 0.00707 or 1 in 140)

to solve for p: $p = 1 - q$

$p = 1 - 0.007$

$p = 0.993$

The frequency of the dominant, normal allele (A) is, therefore, 0.99292 or about 99 in 100. We can now plug the frequencies of p and q into the Hardy-Weinberg equation:

$$p^2 + 2pq + q^2 = 1$$
$$(0.993)^2 + 2\,(0.993)(0.007) + (0.007)^2 = 1$$
$$0.986 + 0.014 + 0.00005 = 1$$

This gives us the frequencies for each of the three genotypes for the trait in the population:

p^2 = predicted frequency of homozygous dominant individuals = 0.986 = 98.6%
2pq = predicted frequency of heterozygous individuals = 0.014 = 1.4%
q^2 = predicted frequency of homozygous recessive individuals = 0.00005 = 0.005%

With a frequency of 0.005% (about 1 in 20,0000), albinos are extremely rare. However, heterozygous carriers for this trait, with a predicted frequency of 1.4% (about 1 in 72), are far more common than most people imagine. There are roughly 278 times more carriers than albinos.

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Practice Problems:

PROBLEM #1

You have sampled a population in which you know that the percentage of the homozygous recessive genotype (aa) is 36%. Using that 36%, calculate the following:

i. The frequency of the "aa" genotype.

ii. The frequency of the "a" allele.

iii. The frequency of the "A" allele.

iv. The frequencies of the genotypes "AA" and "Aa."

v. The frequencies of the two possible phenotypes if "A" is completely dominant over "a."

PROBLEM #2

There are 100 students in a class. Ninety-six did well in the course whereas four blew it totally and received a grade of F. Sorry. In the highly unlikely event that these traits are genetic rather than environmental, if these traits involve dominant and recessive alleles, and if the four (4%) represent the frequency of the homozygous recessive condition, please calculate the following:

i. The frequency of the recessive allele.

ii. The frequency of the dominant allele.

iii. The frequency of heterozygous individuals.

PROBLEM #3

Within a population of butterflies, the color brown (B) is dominant over the color white (b). And, 40% of all butterflies are white. Given this simple information, which is something that is very likely to be on an exam, calculate the following:

i. The percentage of butterflies in the population that are heterozygous.

ii. The frequency of homozygous dominant individuals.

PROBLEM #4

Sickle-cell anemia is an interesting genetic disease. Normal homozygous individuals (SS) have normal blood cells that are easily infected with the malarial parasite. Thus, many of these individuals become very ill from the parasite and many die. Individuals homozygous for the sickle-cell trait (ss) have red blood cells that readily collapse when deoxygenated. Although malaria cannot grow in these red blood cells, individuals often die because of the genetic defect. However, individuals with the heterozygous condition (Ss) have some sickling of red blood cells, but generally not enough to cause mortality. In addition, malaria cannot survive well within these "partially defective" red blood cells. Thus, heterozygotes tend to survive better than either of the homozygous conditions. If 9% of an African population is born with a severe form of sickle-cell anemia (ss), what percentage of the population will be more resistant to malaria because they are heterozygous (Ss) for the sickle-cell gene