characteristics could be heritable. In some cases (for example, scenario IV), however, it is not reasonable to assume the characteristic is heritable. That it is not heritable usually becomes obvious when you take the example to extremes. For example, if you take scenario IV to the extremes, then over time, the population would contain fewer and fewer very young and very old individuals and more and more middle-aged individuals. If you explain this to the class, some will recognize that there's something wrong with this idea. Others may have to be nudged further. Ask: “Which individuals in the population are most likely to breed? Given this, how would the number of newborns in the population be likely to change over time?”

Answers

Activity 23.1 A Quick Review of Hardy-Weinberg Population Genetics

Part A. Review Chapter 23 of Biology, 8th edition. Then complete the discussion by filling in the missing information.

If evolution can be defined as a change in gene (or more appropriately, allele) frequencies, is it conversely true that a population not undergoing evolution should maintain a stable gene frequency from generation to generation? This was the question that Hardy and Weinberg answered independently.

1. Definitions. Complete these definitions or ideas that are central to understanding the Hardy-Weinberg theorem.

   a. Population: An interbreeding group of individuals of the same species.
   b. Gene pool: All the alleles contained in the gametes of all the individuals in the population.
   c. Genetic drift: Evolution (defined as a change in allele frequencies) that occurs in small populations as a result of chance events.

2. The Hardy-Weinberg theorem. The Hardy-Weinberg theorem states that in a population that is not (is/is not) evolving, the allele frequencies and genotype frequencies remain constant from one generation to another.

3. Assumptions. The assumptions required for the theorem to be true are listed on page 472 of Biology, 8th edition, and are presented here in shortened form.

   a. The population is very large.
   b. There is no net migration of individuals into or out of the population.
   c. There is no net mutation; that is, the forward and backward mutation rates for alleles are the same. For example, A goes to a as often as a goes to A.
d. Mating is at random for the trait/gene(s) in question.
e. There is no selection. Offspring from all possible matings for the trait/gene are equally likely to survive.

4. The Hardy-Weinberg proof. Consider a gene that has only two alleles, $R$ (dominant) and $r$ (recessive). The sum total of all $R$ plus all $r$ alleles equals all the alleles at this gene locus or 100% of all the alleles for that gene.

Let $p =$ the percentage or probability of all the $R$ alleles in the population
Let $q =$ the percentage or probability of all the $r$ alleles in the population

If all $R + all \ r$ alleles = 100% of all the alleles, then

$$p + q = 1 \ (or \ p = 1 - q \ or \ q = 1 - p)$$

(Note: Frequencies are stated as percentages [e.g., 50%] and their associated probabilities are stated as decimal fractions [e.g., 0.5].)

Assume that 50% of the alleles for fur color in a population of mice are $B$ (black) and 50% are $b$ (brown). The fur color gene is autosomal.

a. What percentage of the gametes in the females (alone) carry the $B$ allele? 50%
b. What percentage of the gametes in the females (alone) carry the $b$ allele? 50%
c. What percentage of the gametes in the males carry the $B$ allele? 50%
d. What percentage of the gametes in the males carry the $b$ allele? 50%
e. Given the preceding case and all the Hardy-Weinberg assumptions, calculate the probabilities of the three possible genotypes ($RR$, $Rr$, and $rr$) occurring in all possible combinations of eggs and sperm for the population.

<table>
<thead>
<tr>
<th>Female gametes and probabilities</th>
</tr>
</thead>
<tbody>
<tr>
<td>$R(p)$</td>
</tr>
<tr>
<td>$r(q)$</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Male gametes and probabilities</th>
</tr>
</thead>
<tbody>
<tr>
<td>$R(p)$</td>
</tr>
<tr>
<td>$r(q)$</td>
</tr>
</tbody>
</table>

$RR$ (p²)  $Rr$ (pq)  $rr$ (q²)

Because the offspring types represent all possible genotypes for this gene, it follows that

$$p^2 + 2pq + q^2 = 1 \ or \ 100\% \ of \ all \ genotypes \ for \ this \ gene$$
Part B. Use your understanding of the Hardy-Weinberg proof and theorem to answer the questions.

1. According to the Hardy-Weinberg theorem, \( p + q = 1 \) and \( p^2 + 2pq + q^2 = 1 \). What does each of these formulas mean, and how are the formulas derived?

   \( p + q = 1 \): If you add all the dominant alleles for a gene to all the recessive alleles for the gene, you get all of the alleles for that gene, or 100% of the alleles for the gene.

   \( p^2 + 2pq + q^2 = 1 \): If you combine all the individuals that are homozygous dominant for a gene with all the heterozygotes and homozygous recessive individuals for that gene, you have counted or combined all the individuals in the population that carry that gene.

   (Note: This assumes the gene has only two alleles.)

2. Assume a population is in Hardy-Weinberg equilibrium for a given genetic autosomal trait. What proportion of individuals in the population are heterozygous for the gene if the frequency of the recessive allele is 1%?

   Assume that \( D \) is the dominant allele and \( d \) is the recessive allele. Because all the alleles are either \( d \) or \( D \), if the frequency of the \( d \) allele is 1% or \( 1/100 \) (= \( q \)), then the frequency of the \( D \) alleles must be \( 99/100 \) (= \( p \)). The frequency of heterozygous individuals in the population is \( 2pq \) or \( 2(99/100)(1/100) = 198/10,000 \).

3. About one child in 2,500 is born with phenylketonuria (an inability to metabolize the amino acid phenylalanine). This is known to be a recessive autosomal trait.

   a. If the population is in Hardy-Weinberg equilibrium for this trait, what is the frequency of the phenylketonuria allele?

      Assume \( P \) is the normal allele and \( p \) is the phenylketonuria allele. The frequency of homozygous \( pp \) individuals in the population is then equal to \( q^2 \), which is \( 1/2,500 \). The frequency of the \( p \) allele is the square root of \( 1/2,500 = 1/50 \) or 2%.

   b. What proportion of the population are carriers of the phenylketonuria allele (that is, what proportion are heterozygous)?

      Heterozygotes should occur in the frequency \( 2pq \): \( 2pq = 2(1/50)(49/50) = 98/2,500 \).

4. In purebred Holstein cattle, about 1 calf in 100 is spotted red rather than black. The trait is autosomal and red is a recessive to black.

   a. What is the frequency of the red alleles in the population?

      If 1 calf out of 100 is spotted red, then the frequency of the recessive red genotype is \( 1/100 = q^2 \). Therefore, \( q \) (the frequency of the red allele) = \( 1/10 \) or 10%.

   b. What is the frequency of black homozygous cattle in the population?

      \( p^2 = (1-q)^2 = (9/10)^2 = 81/100 \)
c. What is the frequency of black heterozygous cattle in the population?

\[ 2pq = 2(1/10)(9/10) = 18/100 \]

5. Assume that the probability of a sex-linked gene for color blindness is 0.09 = q and the probability of the normal allele is 0.91 = p. This means that the probability of X chromosomes carrying the color blindness allele is 0.09 and the probability of X chromosomes carrying the normal allele is 0.91.

a. What is the probability of having a color-blind male in the population?

Remember, males have only one X chromosome. Color blindness is a sex-linked gene and is found on the X chromosome and not on the Y. As a result, a male has a 0.09 probability of having the X with the color blindness allele and a 0.91 probability of having an X with the normal allele. For sex-linked genes, males display the allele frequency.

b. What is the probability of a color-blind female?

Unlike males, females have two copies of the X chromosome. As a result, they display the genotype frequency for genes on the X. A color-blind female is homozygous recessive for the color blindness allele. The frequency of the homozygous recessive genotype is \( q^2 \), or \( (0.09)^2 = 0.0081 \).

6. The ear tuft allele in chickens is autosomal and produces feathered skin projections near the ear on each side of the head. This gene is dominant and is lethal in the homozygous state. In other words, homozygous dominant embryos do not hatch from the egg. Assume that in a population of 6,000 chickens, 2,000 have no ear tufts and 4,000 have ear tufts. What are the frequencies of the normal versus ear tuft alleles in this population?

This gene is lethal in the homozygous condition. Therefore, homozygotes that are produced do not hatch and do not appear in the population. The population contains 4,000 heterozygous tufted chickens and 2,000 homozygous normal chickens. Because we don’t know how many eggs did not hatch (or how many of these contained homozygous tufted chickens), we need to calculate the allele frequencies by assigning alleles to the existing population.

<table>
<thead>
<tr>
<th>Number of</th>
<th>Number of</th>
</tr>
</thead>
<tbody>
<tr>
<td>T (tufted) alleles</td>
<td>t (normal) alleles</td>
</tr>
<tr>
<td>4,000 tufted chickens</td>
<td>4,000 T</td>
</tr>
<tr>
<td>2,000 normal chickens</td>
<td>4,000 t</td>
</tr>
<tr>
<td>Total alleles</td>
<td>8,000 T</td>
</tr>
<tr>
<td>Allele frequencies</td>
<td>8,000/12,000</td>
</tr>
</tbody>
</table>

0.33 0.66
If these are in Hardy-Weinberg equilibrium, we would expect the following offspring in the next generation:

<table>
<thead>
<tr>
<th>Frequency of alleles in eggs</th>
<th>$T$ ($p = 1/3$)</th>
<th>$t$ ($q = 2/3$)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Frequency of alleles in sperm</td>
<td></td>
<td></td>
</tr>
<tr>
<td>$T$ ($p = 1/3$)</td>
<td>$TT$ (1/9)</td>
<td>$Tt$ (2/9)</td>
</tr>
<tr>
<td>$t$ ($q = 2/3$)</td>
<td>$Tt$ (2/9)</td>
<td>$tt$ (4/9)</td>
</tr>
</tbody>
</table>

In the next generation, when you remove the homozygous lethals, the frequency of $TT$ and $tt$ genotypes would be equal. This indicates that the assumption is incorrect. In other words, the population is not in Hardy-Weinberg equilibrium.

7. **How can one determine whether or not a population is in Hardy-Weinberg equilibrium? What factors need to be considered?**

To determine whether a population is in Hardy-Weinberg equilibrium, you need to be able to calculate the numbers of individuals in the population that are homozygous versus heterozygous for the alleles. If you know the frequencies of each genotype, you can calculate the allele frequencies (as in question 6). Given the allele frequencies, you can calculate the genotype frequencies that would be expected if the population were in Hardy-Weinberg equilibrium. Then compare these values to the known values for the population. In reality, this is difficult to do because if alleles show dominance, it is hard to distinguish the homozygous dominants from the heterozygotes. As a result, we tend to look at the frequency of the homozygous recessive phenotype in a population. If this remains relatively constant from one generation to the next, we use it as evidence to assume that the population is in Hardy-Weinberg equilibrium.

8. **Is it possible for a population’s genotype frequencies to change from one generation to the next but for its gene (allele) frequencies to remain constant? Explain by providing an example.**

There are a number of ways that this is possible. Here is one example of how it could occur: Assume inbreeding has occurred in two populations of mice. In one population, the mice have become homozygous $AA$ at a particular gene locus. In the other population, the mice have become homozygous $aa$ at that gene locus. Equal numbers of $aa$ and $AA$ mice happen to migrate to a new habitat. The frequency of the $A$ allele is 0.5 and the frequency of the $a$ allele is 0.5 in this new population. All possible combinations of matings of these mice are listed in the following table.
<table>
<thead>
<tr>
<th></th>
<th>AA males</th>
<th>aa males</th>
</tr>
</thead>
<tbody>
<tr>
<td>AA females</td>
<td>$AA \times AA \rightarrow$ all $AA$ offspring</td>
<td>$AA \times aa \rightarrow$ all $Aa$ offspring</td>
</tr>
<tr>
<td>aa females</td>
<td>$AA \times aa \rightarrow$ all $Aa$ offspring</td>
<td>$aa \times aa \rightarrow$ all $aa$ offspring</td>
</tr>
</tbody>
</table>

Each of these matings should occur with equal probability. Therefore, each of the offspring types should occur with equal probability. As a result, $\frac{1}{4}$ of the offspring will be $AA$, $\frac{1}{2}$ will be $Aa$, and $\frac{1}{4}$ will be $aa$. In this case, within one generation the population has gone to Hardy-Weinberg equilibrium for genotypes; however, the allele frequency has remained the same.

23.1 Test Your Understanding

In each of the following scenarios, choose which assumption of the Hardy-Weinberg Law is being violated.

1. In a particular region of the coast, limpets (a type of mollusc) live on near shore habitats that are uniformly made up of brown sandstone rock. The principle predators of these limpets are shorebirds. The limpets occur in two morphs, one with a light-colored shell and one with a dark-colored shell. The shorebirds hunt by sight and are able to see the light ones on the dark sandstone easier than the dark ones. No selection—The shorebirds are selectively taking the most visible (light) limpets.

2. In *Chen caerulescens* (a species of goose), the white body form, the snow goose and the blue body form, the blue goose, occasionally coexist. In these areas of contact, white-by-white and blue-by-blue matings are much more common than white-by-blue matings.
   Random mating—Selective mating is occurring when white by white and blue by blue mating are more common than white by blue.

3. Prior to the Mongolian invasions which occurred between the 6th and 16th centuries, the frequency of blood type B across Europe was close to zero. The frequency of blood type B among the Mongols was relatively high. Today, it is possible to see fairly high frequencies of blood type B in the Eastern European countries and a gradual decrease in the frequency of blood type B as one moves from the Eastern European countries to the Western European countries, such as France and England. No migration—Mongolian invasions were more common in Eastern European countries than Western European countries. Offspring of Eastern Europeans and Mongols with blood type B led to the variability in distribution of the blood type in Europe.