

S1. The phenotypic frequency of individuals who cannot taste phenylthiocarbamide (PTC) is approximately 0.3. The inability to taste this bitter substance is due to a recessive allele. If we assume that there are only two alleles in the population (namely, tasters, T , and nontasters, t) and that the population is in Hardy-Weinberg equilibrium, calculate the frequencies of these two alleles.

Answer: Let p = allele frequency of the taster allele and q = the allele frequency of the nontaster allele. The frequency of nontasters is 0.3. This is the frequency of the genotype tt , which in this case is equal to q^2 .

$$q^2 = 0.3$$

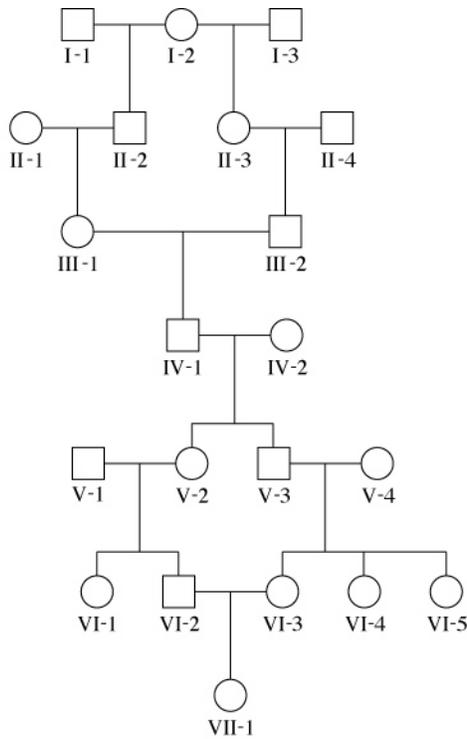
To determine the frequency q of the nontaster allele, we take the square root of both sides of this equation:

$$q = 0.55$$

With this value, we can calculate the frequency p of the taster allele.

$$\begin{aligned} p &= 1 - q \\ &= 1 - 0.55 = 0.45 \end{aligned}$$

S2. In the pedigree shown here, answer the following questions with regard to individual VII-1:



- Who are the common ancestors of her parents?
- What is the inbreeding coefficient?

Answer:

- The common ancestors are IV-1 and IV-2. They are the grandparents of VI-2 and VI-3, who are the parents of VII-1.
- The inbreeding coefficient is calculated using the formula

$$F = \sum (1/2)^n (1 + F_A)$$

In this case there are two common ancestors, IV-1 and IV-2. Also, IV-1 is inbred, because I-2 is a common ancestor to both of IV-1's parents. So, the first step is to calculate F_A , the inbreeding coefficient for this common ancestor. The inbreeding loop for IV-1 contains five people, III-1, II-2, I-2, II-3, and III-2. Therefore,

$$n = 5$$

$$F_A = (1/2)^5 = 0.03$$

Now we can calculate the inbreeding coefficient for VII-1. Each inbreeding loop contains five people: VI-2, V-2, IV-1, V-3, and VI-3; and VI-2, V-2, IV-2, V-3, and VI-3. Thus,

$$F = (1/2)^5 (1 + 0.03) + (1/2)^5 (1 + 0)$$

$$= 0.032 + 0.031 = 0.063$$

S3. As discussed in chapter 25, John Hubby and Richard Lewontin studied genetic variation in *Drosophila pseudoobscura*. They focused their attention on 18 different structural genes that were known to encode enzymes. The activity of these enzymes had already been well characterized by biochemists. Hubby and Lewontin wanted to know how much polypeptide sequence variation there was in these enzymes within different populations of *D. pseudoobscura*. At the molecular level, genetic variation in a structural gene may produce enzymes that have slight differences in their amino acid sequences. Based on previous work, Hubby and Lewontin knew that very small differences between the amino acid sequences of two enzymes could be detected by gel electrophoresis. For example, two alleles for a particular enzyme may differ only by a single amino acid. However, this amino acid difference may influence the mobility of the enzyme when it is subjected to gel electrophoresis. This is particularly evident when the amino acid alteration affects the overall charge on the protein. The 20 amino acids differ with regard to their side chain chemistry. The amino acid side chains are positively charged, negatively charged, or neutral. An amino acid substitution that changes a neutral amino acid to a charged one, or vice versa, will affect the net charge on the protein. The data from their study are shown here.

Population	Number of Monomorphic Genes	Number of Polymorphic Genes	Percentage of Polymorphic Genes
Strawberry Canyon	12	6	33
Wildrose	13	5	28
Cimarron	13	5	28
Mather	12	6	33
Flagstaff	13	5	<u>28</u>
			30 (average)

Discuss these data.

Answer: In this study, Hubby and Lewontin looked for genetic variation in 18 different enzymes in five different populations of fruit flies. On average, 30% of the enzymes were found as two or more allozymes. This means that the genes encoding these enzymes have DNA sequence differences resulting in alleles that cause the encoded proteins to have slightly different amino acid sequences. It should be pointed out that this study tends to underestimate genetic variability because some amino acid substitutions will not alter protein mobility during gel electrophoresis. Nevertheless, the conclusion from this work is that the genetic variability within these populations is quite high.

S4. The Hardy-Weinberg equilibrium provides a way to predict genotype frequency based on allele frequency. In the case of mammals, males are hemizygous for X-linked genes while females have two copies. Among males, the frequency of any X-linked trait will equal the frequency of males with the trait. For example, if an allele frequency for a X-linked disease-causing allele was 5%, then 5% of all males would be affected with the disorder. Female genotype frequencies are computed using the Hardy-Weinberg equation.

As a specific example, let's consider the human X-linked trait known as hemophilia A (see chapter 22 for a description of this disorder). In human populations, the allele frequency of the hemophilia A allele is approximately 1 in 10,000, or 0.0001. The other allele for this gene is the normal allele. Males can be affected or unaffected, whereas females can be affected, unaffected carriers, or unaffected noncarriers.

- What are the allele frequencies for the mutant and normal allele in the human population?
- Among males, what is the frequency of affected individuals?
- Among females, what is the frequency of affected individuals and heterozygous carriers?

D. Within a population of 100,000 people, what is the expected number of affected males? In this same population, what is the expected number of carrier females?

Answer: Let p represent the normal allele and q represent the allele that causes hemophilia.

A. X^H normal allele, frequency = $0.9999 = p$

X^h hemophilia allele, frequency = $0.0001 = q$

B. X^hY genotype frequency of affected males = $q = 0.0001$

C. X^hX^h genotype frequency of affected females = $q^2 = (0.0001)^2 = 0.00000001$

X^HX^h genotype frequency of carrier females = $2pq = 2(0.9999)(0.0001) = 0.0002$

D. We will assume that this population is composed of 50% males and 50% females.

$$\text{Number of affected males} = 50,000 \downarrow 0.0001 = 5$$

$$\text{Number of carrier females} = 50,000 \downarrow 0.0002 = 10$$

S5. The Hardy-Weinberg equation can be modified to include situations of three or more alleles. In its standard (two-allele) form, the Hardy-Weinberg equation reflects the Mendelian notion that each individual inherits two copies of each allele, one from both parents. For a two-allele situation, it is written as

$$(p + q)^2 = 1 \text{ (Note: The number 2 in this equation reflects the idea that the genotype is due to the inheritance of two alleles, one from each parent.)}$$

This equation can be expanded to include three or more alleles. For example, let's consider a situation in which a gene exists as three alleles: $A1$, $A2$, and $A3$. The allele frequency of $A1$ is designated by the letter p , $A2$ by the letter q , and $A3$ by the letter r . Under these circumstances, the Hardy-Weinberg equation becomes

$$(p + q + r)^2 = 1$$

$$p^2 + q^2 + r^2 + 2pq + 2pr + 2qr = 1$$

where

p^2 is the genotype frequency of $A1A1$

q^2 is the genotype frequency of $A2A2$

r^2 is the genotype frequency of $A3A3$

$2pq$ is the genotype frequency of $A1A2$

$2pr$ is the genotype frequency of $A1A3$

$2qr$ is the genotype frequency of $A2A3$

Now here is the question. As discussed in chapter 4, the gene that affects human blood type can exist in three alleles. In a Japanese population, the allele frequencies are

$$I^A = 0.28$$

$$I^B = 0.17$$

$$i = 0.55$$

Based on these allele frequencies, calculate the different possible genotype frequencies and blood type frequencies.

Answer: If we let p represent I^A , q represent I^B , and r represent i , then,

$$p^2 \text{ is the genotype frequency of } I^A I^A, \text{ which is type A blood} = (0.28)^2 = 0.08$$

$$q^2 \text{ is the genotype frequency of } I^B I^B, \text{ which is type B blood} = (0.17)^2 = 0.03$$

$$r^2 \text{ is the genotype frequency of } ii, \text{ which is type O blood} = (0.55)^2 = 0.30$$

$$2pq \text{ is the genotype frequency of } I^A I^B, \text{ which is type AB blood} = 2(0.28)(0.17) = 0.09$$

$$2pr \text{ is the genotype frequency of } I^A i, \text{ which is type A blood} = 2(0.28)(0.55) = 0.31$$

$$2qr \text{ is the genotype frequency of } I^B i, \text{ which is type B blood} = 2(0.17)(0.55) = 0.19$$

$$\text{Type A} = 0.08 + 0.31 = 0.39, \text{ or } 39\%$$

$$\text{Type B} = 0.03 + 0.19 = 0.22, \text{ or } 22\%$$

Type O = 0.30, or 30%

Type AB = 0.09, or 9%

S6. Let's suppose that pigmentation in a species of insect is controlled by a single gene existing in two alleles, D for dark and d for light. The heterozygote Dd is intermediate in color. In a heterogeneous environment, the allele frequencies are $D = 0.7$ and $d = 0.3$. This polymorphism is maintained because the environment contains some dimly lit forested areas and some sunny fields. During a hurricane, a group of 1,000 insects is blown to a completely sunny area. In this environment, the fitness values are $DD = 0.3$, $Dd = 0.7$, and $dd = 1.0$. Calculate the allele frequencies in the next generation.

Answer: The first step is to calculate the mean fitness of the population.

$$\begin{aligned} p^2W_{DD} + 2pqW_{Dd} + q^2W_{dd} &= \bar{W} \\ \bar{W} &= (0.7)^2(0.3) + 2(0.7)(0.3)(0.7) + (0.3)^2(1.0) \\ &= 0.15 + 0.29 + 0.09 = 0.53 \end{aligned}$$

After one generation of selection, we get

Allele frequency of D :

$$\begin{aligned} p_D &= \frac{p^2W_{DD}}{\bar{W}} + \frac{pqW_{Dd}}{\bar{W}} \\ &= \frac{(0.7)^2(0.3)}{0.53} + \frac{(0.7)(0.3)(0.7)}{0.53} \\ &= 0.55 \end{aligned}$$

Allele frequency of d :

$$\begin{aligned} q_d &= \frac{q^2W_{dd}}{\bar{W}} + \frac{pqW_{Dd}}{\bar{W}} \\ &= \frac{(0.3)^2(1.0)}{0.53} + \frac{(0.7)(0.3)(0.7)}{0.53} \\ &= 0.45 \end{aligned}$$

After one generation, the allele frequency of D has decreased from 0.7 to 0.55, while the frequency of d has increased from 0.3 to 0.45.