PTC Genetics

Introduction

Different alleles produce variation in inherited characteristics such as hair colour or blood type. In an individual, one form of the allele (the dominant form) may be expressed more than another form (the recessive form).



Concepts

• Allele frequency

• Evolution

• Hardy-Weinberg equilibrium model

Background

Allele frequency is a measure of the relative frequency of an allele in a population. Usually allele frequency is expressed as a proportion or a percentage. In population genetics, allele frequencies show the genetic diversity of a species population. Four evolutionary forces affect the frequency of an allele within a population—natural selection, genetic drift, mutation, and migration.

Individuals feel the effects of evolution, but it is the population as a whole that actually evolves. Godfrey Hardy (1877–1947), an English mathematician, and Wilhelm Weinberg (1862–1937), a German physician, developed a definition of evolution independently in 1908. They used mathematical modeling of probability to predict that gene pool frequencies are inherently stable but that continual evolution must be expected in all populations. Hardy and Weinberg concluded that evolution would *not* occur in a population if all of the following conditions were met:

- 1. Mutation is not occurring.
- 2. Natural selection is not occurring.
- 3. The population is infinitely large.
- 4. All members of the population breed.
- 5. All mating is totally random.
- 6. Everyone produces the same number of offspring.
- 7. There is no migration in or out of the population.

In other words, 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 occur in the real world, evolution is the inevitable result.

Hardy and Weinberg developed a simple equation, now called the *Hardy-Weinberg equilibrium equation* or the *Hardy-Weinberg principle or law* (Equation 1). Scientists use the equation to determine the probable gene frequencies in a population and to track the changes in gene frequencies from one generation to another.

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

where

p is the frequency of the dominant allele (A) for a trait

q is the frequency of the recessive allele (a) for a trait

In the case where there are only two alleles for a trait (e.g., A and a), the frequency of p plus the frequency of q equals 100%, which is stated mathematically as Equation 2.

$$p + q = 1$$
 Equation 2

The only observable genotype is that of a homozygous recessive (aa) organism because its phenotype is different from that seen in a homozygous dominant (AA) or a heterozygous (Aa) organism. Assuming that a population is in Hardy-Weinberg equilibrium, the number of dominant alleles and the number of recessive alleles in a population can be calculated by counting the number of homozygous recessive organisms (aa = q^2) and doing some math.

One way to test the Hardy-Weinberg Equilibrium Model is to use a simple, easy-to-test, human trait. Using the class as a

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sample population, the allele frequency of a gene controlling the ability to taste phenyl thiocarbamide (PTC) will be determined. A bitter-taste reaction to PTC (called "tasters") is evidence for the presence of the dominant allele in either the homozygous (AA) or heterozygous (Aa) genotype. The inability to taste the chemical (called "non-tasters") depends on the presence of homozygous recessive alleles (aa). In order to estimate the frequency of the PTC-tasting allele in the population, the number of non-tasters, who are homozygous recessive (aa), must be counted. This is q^2 . After determining q^2 , p^2 can be calculated and finally the frequency of dominant alleles and the number of recessive alleles can be determined.

Materials

Phenyl thiocarbamide (PTC) taste test paper

Calculator

Safety Precautions

Although the materials in this lab activity are considered nonhazardous at low concentrations, follow normal safety precautions. Handle the taste test papers as little as possible before use. Once any taste has been detected, immediately remove the test paper from the mouth and discard. Do not reuse any test papers; always use a fresh test paper for each individual. Wash hands after completing this laboratory. Please review current Safety Data Sheets for additional safety, handling and disposal information.

Control taste test paper

PTC paper is soaked with phenyl thiocarbamide (PTC). The LD_{50} (Rat) for PTC is 3 mg/kg. Such a low LD_{50} suggests that this is a very toxic substance. However, the solution used to make the taste test papers contains approximately 50 mg of PTC per litre of water. Using crude arithmetic, we calculate that each strip of PTC paper would contain approximately 0.03 mg of PTC, and that an average person would have to lick 5000 strips of PTC paper to reach the LD_{50} for a body weight of about 50 kilograms (110 lbs).

Procedure

- 1. Record the number of students in the class.
- 2. Have each student place a piece of control taste test paper on his or her tongue and note the taste of the control paper.
- 3. Have each student place a piece of PTC taste test paper on his or her tongue. If he or she senses a bitter taste (it will be obvious), then he or she is a taster of PTC. If the test paper tastes like the control paper, then he or she is a non-taster.
- 4. Record the number of non-tasters in the class. These students are homozygous recessive (aa) for the PTC tasting gene.
- 5. Calculate q^2 by dividing the number of non-tasters by the total number of students. Record q^2 .
- 6. Calculate q by taking the square root of q^2 .
- 7. Calculate p by subtracting q from 1.
- 8. Determine the frequency of homozygous dominant (AA) genotypes in the class by calculating p^2 .
- 9. Determine the frequency of heterozygous genotypes in the class by calculating 2pq.

Disposal

It is recommended that you consult your local school board and/or municipal regulations for proper disposal methods that may apply before proceeding. PTC and Control taste test papers may be thrown in the regular trash.

Tips

- The gene that primarily affects the ability to taste PTC is called *TAS2R38*. It is part of a family of genes called *TAS2R* all of which are related to the ability to taste bitter substances. *TAS2R38* is located on chromesome 7, location q35–q36. Three amino acid substitutions are the typical differences between tasters and non-tasters. Other *TAS2R* genes play a minor role in the ability to taste PTC. Certain *TAS2R* genes contribute to those individuals called "super-tasters."
- One study found that many people who react strongly to PTC do not like to eat foods such as broccoli or coriander nor are they likely to smoke cigarettes or drink coffee or tea.
- The ability to taste PTC is present in about 70% of the overall human population, varying from 58% for Australians to 98% for Native American populations.
- Test the frequency of thiourea tasters (Flinn Catalogue No. AP7892) and sodium benzoate tasters (Flinn Catalogue No. AP7891) and compare the data to that of the PTC tasters.
- See the Flinn Scientific Canada kit—"Genetics of Taste" (Flinn Catalogue No. FB1431) that discusses the genetics of PTC paper.

Materials for PTC Genetics are available from Flinn Scientific Canada Inc.

Catalogue No.	Description
<u>AP7891</u>	Sodium Benzoate Taste Test Paper
<u>AP7892</u>	Thiourea Taste Test Paper
<u>AP7989</u>	PTC Paper
<u>AP7990</u>	Control Test Paper
<u>FB1431</u>	Genetics of Taste

Consult www.flinnsci.ca or your Flinn Scientific Canada Catalogue/Reference Manual for current prices.

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