

# P8149 — Human Population Genetics: Homework 1

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## Question 1

Three allelic variants A, B, and C of the red cell acid phosphatase enzyme were found in a sample of 178 people. All genotypes were distinguishable by electrophoresis, and the frequencies in the sample were:

Genotype	AA	AB	BB	AC	BC	CC
Frequency (%)	9.6	48.3	34.3	2.8	5.0	0.0

What are the *gene (allele)* frequencies in the sample?

- $P(A) = P(AA) + P(AB) / 2 + P(AC) / 2 = 35.15\%$
- $P(B) = P(BB) + P(AB) / 2 + P(BC) / 2 = 60.95\%$
- $P(C) = P(CC) + P(AC) / 2 + P(BC) / 2 = 3.9\%$

## Question 2

What will be the genotypic composition of the next generation under random mating if the initial genotypic composition is  $P_{11} = 0.2$ ,  $P_{12} = 0.4$ ,  $P_{22} = 0.4$ ?

From the genotype frequency, we can calculate the allele frequency:

- $P(1) = P_{11} + \frac{1}{2}P_{12} = 0.4$
- $P(2) = P_{22} + \frac{1}{2}P_{12} = 0.6$

Since it's random mating, the genotype frequency of the next generation  $P'$  is only decided by allele frequency:

- $P'_{11} = P(1)^2 = 0.16$
- $P'_{22} = P(2)^2 = 0.36$
- $P'_{12} = 2 * P(1) * P(2) = 0.48$

## Question 3

About 30% of people do not recognize the bitter taste of phenyl-thiocarbamate (PTC). Inability to taste is due to a single autosomal recessive gene. Assuming HWE, what is the frequency of the non-tasting gene?

Let A/a be the tasting/non-tasting gene. From the question, we know  $P(aa) = 30\%$ .

Assuming HWE,  $P(a) = \sqrt{30\%} = 54.8\%$  and  $P(A) = 1 - P(a) = 45.2\%$ .

## Question 4

Albinism occurs with a frequency of about 1 in 20,000 in European populations. Assuming it is due to a single autosomal recessive gene and HWE, what is the approximate proportion of carriers?

Let A/a be the corresponding dominant/recessive gene.

From the question, we know the genotype frequency  $P(aa) = \frac{1}{20000}$ . So the allele frequency  $P(a) = \sqrt{P(aa)} = 0.7\%$ .

Since  $P(a)$  is very small, the approximate proportion of carriers is  $2P(a) = 1.4$ .

## Question 5

Observed counts:

Genotype	M	MN	N
Count	1787	3039	1303

- (a) What genotype frequencies are observed?

The total observed count is 6129.

- $P(MM) = 1787/6129 = 29.16\%$
- $P(MN) = 3039/6129 = 49.58\%$
- $P(NN) = 1303/6129 = 21.26\%$

- (b) What are the gene (allele) frequencies (M and N)?

- $P(M) = P(MM) + P(MN)/2 = 53.9\%$
- $P(N) = P(NN) + P(MN)/2 = 46.0\%$

- (c) Test whether the genotypes are in HWE.

From the calculation above, we can see that

- $P(N) + P(M) \approx 1$
- $2 * P(N) * P(M) = 2 * 53.9\% * 46.0\% = 59.59\% \approx P(MN)$

So the genotypes are in HWE.

## Question 6

A certain population is *triploid*. A locus has two alleles  $A_1$  and  $A_2$  with frequencies  $p$  and  $q = 1 - p$ . Calculate the genotypic composition after one generation of random mating (assume HWE conditions).

From the binomial expansion  $(pA_1 + qA_2)^3 = p^3 A_1^3 + 3pq^2 A_1 A_2^2 + 3p^2 q A_1^2 A_2 + q^3 A_2^3$ , we have:

- $P(A_1 A_1 A_1) = p^3$
- $P(A_1 A_1 A_2) = 3p^2 q$
- $P(A_1 A_2 A_2) = 3pq^2$
- $P(A_2 A_2 A_2) = q^3$