EXPERIMENT 17 DETERMINATION OF ALLELIC AND GENOTYPIC FREQUENCIES

Structure

17.1 INTRODUCTION

In this lab exercise you will learn certain simple calculations relating to the frcqucncics of alleles and genotypes in populations. The term frequency refers to the number of times an allele or a gcnotypc occurs in a given total population For instance, in a population consisting of 500 people, if 180 of them carry the gcnotypc AA, then the frequency of AA in the given population is 180/500 or 36% or 0.36. You have learnt that Mendelian monohybrid ratio for a given pair of allclcs (A and a) is 3:1 for phenotypes and 1:2:1 for genotypes. Similarly if two pairs of alleles are involved (A and a, B and b) the phenotypic ratio is 9:3:3:1 and the genotypic ratio is 1:2:1:2:4:2:1:2:1. These ratios refer to the number of times a phenotype or a genotype appears relative to olhcr phenotypes or genotypes in specific crosses. They do not give the picture of frequencies of gcnolypcs occurring in a givcn populalion. You will now learn the process of calculaling the frequencies of dominant and rcccssivc allcles and those of the genotypes that these alleles producc using some simple algebraic expressions.

Objectives

At the end of this excrcise, you should bc able lo

- calculate the frequencies of dominant and rcccssive allclcs based on the population data available for the phenotypes,
- arrive at the froqucncics of gcnolypcs bascd on allclic frequencies.
- cxtcnd the concept to the multiple allclic systems and calculalc the frequencies of blood group allclcs and the genotypes lhcy producc.

17.2 DETERMINATION OF FREQUENCY OF DOMINANT AND RECESSIVE ALLELES AND GENOTYPES

For purposes of calculation, let us assume that the total frequency of a givcn pair of alleles, A and a is cqual to 1. Let the frequency of allele A bc equal to p and that of the recessive allelc bc equal lo q. Then,

Frequency of A t [illegible] $p + q = 1$

In other words, frequency of A $= p = (1-q)$ and that of a $= q = (1-p)$

The two alleles, as you are aware, form three genotypes, AA, Aa and aa and the three genotypes are in the ratio of 1:2:1

(1AA : 2Aa : laa).

The frequencies of the genotypes could then be.

$1AA = p \times p = p^2$

$2Aa = p \times q = pq$

$q \times p = pq$

$laa = q \times q = q^2$

Assuming the total frequency of the three genotypes = 1,

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

Let us now look into an example. In a population of 304 students in a college, 214 were able to taste the chemical phenylthiocarbamide (PTC) bitter and the rest 90 did not taste it. **PTC** tasters have the dominant allele T in their genotype and the non-tasters are recessive homozygotes (tt). Based on the data provided, let us calculate the frequency of alleles T and t in the given student population as well as the frequency of genotypes (TT, Tt and tt).

Totalnumber of students = 304

Tasters = 214

non-tasters = 90

Frequency of taster phenotypes = 214/304 = 0.7

Frequency of non-taster phenotypes = 90/304 =0.3

Frequency of tt = $q^2 = 0.3$

Frequency of the allele $t = \sqrt{q^2} = \sqrt{0.3} = 0.55$

Frequency of the allele T = (1-q) = (1-0.55) = 0.45

Based on the allelic frequencies (T = 0.45 and t = 0.55), we can predict the genotype frequencies of the population

Frequency of TT = p^2	= 0.45 x 0.45 =	0.2025
Frequency of Tt	= pq = 0.45 x 0.55 =	0.2475
Frequency of tT	= qp = 0.55 x 0.45 =	0.2475
Frequency of tt	= q^2 = 0.55 x 0.55 =	0.3025
		1.0000

Thus, from the frequencies of alleles, those of genotypes are calculated. The data on total population number and the number of persons possessing the recessive trait are sufficient to arrive at the frequencies of alleles and the genotypes, provided that the two alleles have simple dominant—recessive relationship. You attempt the following problem to test your understanding of determining allelic and genotypic frequencies in a population.

A survey conducted in a population of 930 individuals showed that 325 of them are non-tasters of PTC. What is the frequency of T and t alleles in the population? Also, calculate the frequency of TT, Tt and tt genotypes.

17.3 DETERMINATION OF FREQUENCIES OF MULTIPLE ALLELES

In the earlier seclion you have learnt the application of equations ($p + q = 1$) and $(p+q)^2 = 1$, in which the alleles exhibiting simple dominant-recessive relationship are involved. When more than two allcles occupy a locus, then the frequcncy of additional alleles also should bc included in the cquation. For instance, in ABO system of blood group inheritance, thcrc arc three allcles involved. The blood groups A, B, AB and O are controlled by a multiple allclic system of I^A, I^B and I^O. I'' and I^B arc dominant to I^O but codominant themselves. The genotypes of the iour blood groups arc

Blood group	Genotype
A	I^AI^A, I^AI^O
B	I^BI^B, I^BI^O
AB	I^AI^B,
O	I^OI^O

Let the frequency of allclc I'' = p, I^D =q and I^O = r

Then p + q + r = 1

Let us now analyse the blood group allclic frcqucncies in a population.
Blood group determination of a population of 1000 individuals revealed that 328 of them belonged to **A** group, 122 of them to B group, 32 of them to AB group and 518 to O group. Calculate the allclic and genotypic frequencies in the population.

Type **A** persons — 328
Type B persons — **122**
Type AB persons — 32
Type O persons — 518

Assuming the frequency of I^A, I^B and I^O to bc p, q and r rcspcctivcly, the frequcncy of the genotypes formed by the three allcles would bc

$$(p+q+r)^2 = p^2 + 2pq + q^2 + 2qr + r^2 + 2rp$$

Let us summarise the frequencies of phcnolypcs and probable genotypes in the form of a table.

Phenotype	Phenotypic Frequency	Genotype	Genotype Frequency	Sum of the frequencies of genotypes with similar phenotypes
A	$\frac{328}{1000} = 0.328$	I^AI^A I^AI^O	p^2 2pr	$p^2 + 2pr$
B	$\frac{122}{1000} = 0.122$	I^BI^B I^BI^O	q^2 2qr	$q^2 + 2qr$
AB	$\frac{32}{1000} = 0.032$	I^AI^B	2pq	2pq
O	$\frac{518}{1000} = 0.518$	I^OI^O	r^2	r^2

$r^2 = 0.518$

$r = \sqrt{0.518} = 0.72$

The frcquency of I^O = r = 0.72

Frcqucncy of I^A = p = 1 – Frequency of B + frcquency of O

Frcqucncy of B = $q^2 + 2qr$

and the frequency of O = r^2

Frcqucncy of B + frequency of O = $q^2 + 2qr + r^2 = (q+r)^2$ And the frcqucncy of

$I^A = 1 - \sqrt{(q + r)^2}$

$= 1 - \sqrt{(q^2 + 2qr + r^2)}$

$= 1 - \sqrt{0.122 + 0.518}$

$= 1 - \sqrt{0.640}$

$= 1 - 0.8$

$= 0.2$

Frequency of I^A = 0.2

Since the frcquency of I^A and I^O are known, the frequency or I^B can be calculated using the expression.

Frequency of I^B = 1 – (frequency of I^A + frequency of I^O)

= 1 – (0.72 + 0.2)

= 1 – 0.92 = 0.08

Frequency of I^R = 0.08

The allelic frequencies are : I^A = 0.200

I^B = 0.080

I^O = 0.720

1.000

The genotype frequencies would then be

I^AI^A	$= p^2$	$= 0.2 \times 0.2$	= 0.0400
I^AI^A	$= 2pq$	$= 2 \times 0.2 \times 0.72$	= 0.2880
$I^BI^$	$= q^2$	$= 0.08 \times 0.08$	= 0.0064
I^BI^O	$= 2qr$	$= 2 \times 0.08 \times 0.72$	= 0.1152
I^AI^B	$= 2pq$	$= 2 \times 0.2 \times 0.08$	= 0.0320
I^OI^O	$= r^2$	$= 0.72 \times 0.72$	= 0.5184
		$(p+q+r)^2$	= 1.0000

The above calculations show that the sum of the genotype frequencies is also equal to 1.

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

Since alleles I^A and I^B exhibit codominance and I^O is recessive to both alleles, the distribution of alleles and gcnotypcs differs from the distribution of phcnotypcs. You will study more about the distribution of allelic and genotypic frequcncics and the changes which they undergo in the population from your LSE-03 Genetics course in the Unit titled "Bchnviour of Genes in Populations".