Section 12: Population genetics

1. Introduction

Population genetics:

• Study of gene frequency and genotypes

• Study of the factors likely to modify these frequencies over successive generations, such as mutations, migrations, etc.

Population genetics is a branch of genetics that studies the distribution and changes in allele frequency in populations over time, under the influence of evolutionary forces such as natural selection, genetic drift, mutation, migration and non-random mating.

The genetic structure of a population: is both the genotypic and allelic structure.

2. Concepts of genetics of populations

a) Population

- A population is a group of individuals of the same species living in a given area and reproducing amongst themselves.

b) Gene and allele

- A gene is a unit of hereditary information.
- An allele is a specific version or variant of a gene.

c) Genetic Pool

- All the alleles present in a population at a given time.

d) Allelic frequency

- Proportion of a particular allele in relation to all the alleles for a given gene in a population.

e) Biological evolution

Over time, a species accumulates variations; as a result, its descendants differ from their ancestors. In this way, a new species develops from those that already exist. Evolution can result from anything that causes a change in the genetic composition of a population. Many processes can lead to evolutionary change:

● According to Darwin, the evolution of species occurred through the process of natural selection. Individuals in a population acquire certain heritable traits that ensure their offspring survive longer than individuals without these traits. As a result, the population will contain more and more individuals with the advantageous traits. In this way, the population evolves and adapts better to local contingencies.

● Lamarck contributed a rival theory which is that of the hereditary transmission of acquired traits, according to which individuals pass on to their descendants the physical and behavioural changes acquired during their lives.

The Hardy-Weinberg Principle

The Hardy-Weinberg principle is used to predict genotype frequencies. Hardy-Weinberg equilibrium is reached when the observed genotype frequencies match the predicted frequencies calculated. This is only possible when evolutionary processes do not modify the distribution of alleles or genotypes in the population. The original proportions of genotypes in a population will remain constant from generation to generation as long as the following conditions are met:

- No mutations occur

- No gene comes from other sources, i.e. no immigration has taken place

- Fertilisation is random.

- The population is very large.

- There is no selection.

Example: Consider a population of 100 cats: - 84 have the (black) phenotype with a frequency of 0.84 (84%) - 16 have the (white) phenotype with a frequency ofv0.16 (16%).

If the black cats are homozygous dominant B/B or heterozygous B/b, we can calculate the frequency of the two alleles from the proportion of black and white individuals, assuming that the population is in Hardy-Weinberg equilibrium.

p = frequency of allele B; q = frequency of allele b. The sum of p and q must always equal 1 (total population).

The sum of the three genotype frequencies must also equal 1.

If the frequency of the B allele is p, the probability that an individual has two B alleles is simply the probability that each of its alleles is a B.

The probability that the individual received a B allele from his father is p and the probability that he received a B allele from his mother is also p, while the probability that he received both is $p \times p = p^2$.

By the same reasoning, the probability of an individual having two b alleles is q^2.

The individual could receive a B from his father and b from his mother, or vice versa. The probability of the first case is p x q; and the probability of the second is q x p. Since in both cases the result is the heterozygosity of the individual, its probability is the sum of the two probabilities: 2pq.

To summarise:

If a population is in Hardy-Weinberg equilibrium with allele frequencies, p and q, the probability that an individual has one of the three possible genotypes is: p 2 + 2pq + q 2

If the probability of any individual being heterozygous is 2pq, the proportion of heterozygous individuals in the population is 2pq; the frequency of homozygotes BB and bb should be p^2 and q^2.

In our example:

If white coat is a recessive trait, these individuals must have the b/b genotype. If the frequency of this genotype is $q^2 = 0.16$ (the frequency of white cats), then q (the frequency of the b allele) = 0.4.

Since p + q = 1, then p, the frequency of the B allele, would be 1 - 0.4 = 0.6.

Homozygous dominant BB cats would make up the p^2 group, and the value of $p^2 = (0.6)^2 = 0.36$, or 36 homozygous dominant B/B individuals in a population of 100 cats.

Heterozygous cats have the B/b genotype and would have the corresponding frequency of 2pq or 2 x 0.6 x 0.4 = 0.48, or 48 heterozygous B/b individuals.

Thus, we assumed that the union of sperm and egg in these cats was random, so that any combination of B and b alleles could occur. The alleles are mixed randomly and are represented in the next generation in proportion to their original representation. Each individual egg or

sperm cell has a 6 in 10 chance of receiving a B allele in each generation (p=0.6) and a 4 in 10 chance of receiving a b allele (q=0.4).

At the next generation, the chance of combining two B alleles is p^2 or 0.36, i.e. 0.6 x 0.6, and approximately 36% of individuals in the population will continue to have the B/B genotype. The frequency of b/b individuals is q^2 (0.4 x 0.4), so it will remain around 16%, and the frequency of B/b individuals will be 2pq (2 x 0.6 x 0.4), on average 48%.

Phenotypically, out of 100 cats, we will still have around 84 black cats (with B/B or B/b genotypes) and 16 white cats (with b/b genotype).

5- Adaptive value :

Adaptive value is defined by the reproductive success of an organism compared with that of other members of its population. Its success depends on its survival, the frequency with which it mates and the number of offspring it produces each time. The relative adaptive value assigns numerical values to the different phenotypes relative to those of the best adapted phenotype.

Exercise 1 of the application

The following table shows the results of a study of the MN blood group in a human population made up of 730 Australian aborigines:

Groupe sanguin	Génotype	Nombre
[M]	MM	22
[MN]	MN	216
[N]	NN	492

Question: Calculate the genotypes and all types of frequences

Answer:

Genotype frequencies::

f(MM) = 22/730 = 0.03

f(MN) = 216/730= 0.30

f(NN) = 492/730 = 0.67

Allelic frequencies:

f(M)= f(MM)+ f(MN)/2= 0.03 + 0.15 = 0.18

f(N)=f(NN)+ f(MN)/2= 0.67 + 0.15 = 0.82

Exercise 2

A study of the ABO blood group system in a sample of 5,000 individuals produced the results shown in the table below.

Génotype	AO	BB	AB	OO
Nombre	1600	700	200	2500

Calculate the frequencies of all A, B and O

Answer:

F(A)= ½ (1600 /5000) + ½ (200/5000)

F(B)= 700/5000 + ½ (200/5000)

F(O)= 2500/5000 + ½ (1600/5000)

Exercise 3

In one butterfly species, the gene coding for colour is carried by an autosome. The allele responsible for the dark colour (C) dominates the allele responsible for the light colour (c).

Assume that the population is in equilibrium and that the frequency of the dark phenotype is 0.98.

1 - Calculate the frequencies of the C and c alleles in this population.

2 - Calculate the genotypic frequencies.

Answer:

At the same time: The frequency of the dominant phenotype + the frequency of the dominant phenotype = 1

We have :

The frequency of the dominant phenotype = 0.98.

So the frequency of the recessive phenotype = 1- 0.98 = 0.02

The frequency of the recessive phenotype = the frequency of the recessive genotype.

Therefore: F(cc)= 0.02.

F(cc)= q^2 => q=$\sqrt{0.02}$ => q= 0.14 => F(c)= 0.14

We know that: p+q = 1. Therefore: p= 1-q = 1- 0.14 => p= 0.86 => F(C) = 0.86

F(CC)= p^2 = $(0.86)^2$ => F(CC)= 0.74

F(Cc) = 2pq = 2x 0.86 x 0.14 => F(Cc) = 0.24