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POPULATION GENETICS & GENETIC DRIFT

Evolution is not only the development of new species from older ones, as most people assume. It is also the minor changes within a species from generation to generation over long periods of time that can result in the gradual transition to new species.

Population genetics

- Population genetics is a subfield of genetics that deals with genetic differences within and between populations, and is a part of evolutionary biology. Studies in this branch of biology examine phenomena such as adaptation, speciation, and population structure .Population genetics seeks to understand how and why the frequencies of alleles and genotypes change over time within and between populations.
- Population genetics provides the deepest and clearest understanding of how evolutionary change occurs. These changes in genetic composition that result from the operation of various factors, including natural selection. Population genetics is intimately bound up with the study of evolution and natural selection, and is often regarded as the theoretical foundation of modern Darwinism. This is because natural selection is one of the most important factors that can affect a population's genetic composition. Natural selection occurs when some variants in a population out-reproduce other variants as a result of being better adapted to the environment, or 'fitter'. Presuming the fitness differences are at least partly due to genetic differences, this will cause the population's genetic makeup to be altered over time. By studying formal models of gene frequency change, population geneticists therefore hope to shed light on the evolutionary process, and to permit the consequences of different evolutionary hypotheses to be explored in a quantitatively precise way.
- The biological sciences now generally define evolution as being the sum total of the genetically inherited changes in the individuals who are the members of a population's gene pool. It is clear that the effects of evolution are felt by individuals, but it is the population as a whole that actually evolves. Evolution is simply a change in frequencies of alleles in the gene pool of a population. For instance, let us assume that there is a trait that is determined by the inheritance of a gene with two alleles-B and b. If the parent generation has 92% B and 8% b and their offspring collectively have 90% B and 10% b, evolution has occurred between the generations. It was not just those individuals who inherited the b allele who evolved. Here the entire population's gene pool has evolved in the direction of a higher frequency of the b allele.

The Hardy-Weinberg Principle

The Hardy-Weinberg principle, discovered independently by G.H. Hardy and W. Weinberg in 1908, is one of the simplest and most important principles in population genetics. It is a fundamental principle of population genetics that is approximately true for small populations and holds with increasing exactness for larger and larger populations

Key points of Hardy-Weinberg principal

1. When a population is in Hardy-Weinberg equilibrium for a gene, then it is not evolving, and allele frequencies will stay the same across generations.

The principle states that the allelic frequency remains constant through generations and the gene pool remains constant if the following assumptions of hardy Weinberg law are fulfilled.

Assumption of Hardy Weinberg law

- No mutations must occur so that new alleles do not enter the population.
- No gene flow can occur (i.e. no migration of individuals into, or out of, the population).
- Random mating must occur (i.e. individuals must pair by chance)
- The population must be large so that no genetic drift (random chance) can cause the allele frequencies to change.
- No selection can occur so that certain alleles are not selected for, or against.

When these conditions meet for a population, then this population is called in **genetic equilibrium.**

2. The population should be a panmictic population.

A **panmictic population** is one where all individuals are potential partners. This assumes that there are no mating restrictions, neither genetic nor behavioural, upon the population, and that therefore all recombination is possible.

3. If these assumptions are not met for a gene, the population may evolve for that gene (the gene's allele frequencies may change).

The Hardy-Weinberg law can be used under some circumstances to calculate genotype frequencies from allele frequencies.

Let A1 and A2 be two alleles at the same locus,

p is the frequency of allele A1	0 =< p =< 1
q is the frequency of allele A2	0 =< q =< 1 and p + q =1
if they reproduce: p+q=1, then $(p + q)^2$ $= p^2 + 2pq + q^2 = 1$	

Where:

p^2 = frequency of the A1 A1 genotype <— HOMOZYGOTE

2pq = frequency of the A1 A2 genotype <— HETEROZYGOTE

q^2 = frequency of the A2 A2 genotype <— HOMOZYGOTE

These frequencies remain constant in successive generations.

Example : Autosomal recessive inheritance with alleles A and a, and allele frequencies p and q:

frequency of the genotypes: : HOMOZYGOTE A	AA = p^2	the phenotypes	(A) = p^2 + 2pq/2
HETEROZYGOTE AB	Aa = 2pq		[ab] = 2pq
HOMOZYGOTE B	aa = q^2		(B)=q^2 + 2pq/2

Example:

Phenyl ketonuria (recessive autosomal), of which the deleterious gene has a frequency of 1/100:

—> q = 1/100

therefore, the frequency of this disease is q^2 = 1/10 000, and the frequency of heterozygotes is 2pq = 2 x 99/100 x 1/100 = 2/100;

Note that there are a lot of heterozygotes: 1/50, two hundred times more than there are individuals suffering from the condition. .

There are two formulas that must be memorized:

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

PROBLEM #1.

- You have sampled a population in which you know that the percentage of the homozygous recessive genotype (aa) is 36%. Using that 36%, calculate the following:
- The frequency of the "aa" genotype.
- The frequency of the "a" allele.
- The frequency of the "A" allele.
- The frequencies of the genotypes "AA" and "Aa."
- The frequencies of the two possible phenotypes if "A" is completely dominant over "a."

Soln.

- The frequency of the "aa" genotype. Answer: 36%, as given in the problem itself.
- The frequency of the "a" allele. Answer: The frequency of aa is 36%, which means that q^2 = 0.36, by definition. If q^2 = 0.36, then q = 0.6, again by definition. Since q equals the frequency of the a allele, then the frequency is 60%.
- The frequency of the "A" allele. Answer: Since q = 0.6, and p + q = 1, then p = 0.4; the frequency of A is by definition equal to p, so the answer is 40%.
- The frequencies of the genotypes "AA" and "Aa." Answer: The frequency of AA is equal to p^2, and the frequency of Aa is equal to 2pq. So, using the information above, the frequency of AA is 16% (i.e. p^2 is 0.4 x 0.4 = 0.16) and Aa is 48% (2pq = 2 x 0.4 x 0.6 = 0.48).
- The frequencies of the two possible phenotypes if "A" is completely dominant over "a." Answers: Because "A" is totally dominate over "a", the dominant phenotype will show if either the homozygous "AA" or heterozygous "Aa" genotypes occur. The recessive phenotype is controlled by the homozygous aa genotype. Therefore, the frequency of the dominant phenotype equals the sum of the frequencies of AA and Aa, and the recessive phenotype is simply the frequency of aa. Therefore, the dominant frequency is 64% and, in the first part of this question above, you have already shown that the recessive frequency is 36%.

PROBLEM #2.

Within a population of butterflies, the colour brown (B) is dominant over the colour white (b). And, 40% of all butterflies are white. Given this simple information , calculate the following:

- The percentage of butterflies in the population that is heterozygous.
- Frequency of homozygous dominant individuals.

Soln. The first thing you'll need to do is obtain p and q. So, since white is recessive (i.e. bb), and 40% of the butterflies are white, then bb = q^2 = 0.4. To determine q, which is the frequency of the recessive allele in the population, simply take the square root of q^2 which works out to be 0.632 (i.e. 0.632 x 0.632 = 0.4). So, q = 0.63. Since p + q = 1, then p must be 1 - 0.63 = 0.37. Now, to answer our questions. First, what is the percentage of butterflies in the population that are heterozygous? Well, that would be 2pq so the answer is 2 (0.37) (0.63) = 0.47. Second, what is the frequency of homozygous dominant individuals?

That would be p2 or (0.37)2 = 0.14.

PROBLEM #3.

Cystic fibrosis is a recessive condition that affects about 1 in 2,500 babies in a population. Calculate the following.

1. The frequency of the recessive allele in the population.

Soln. We know from the above that q^2 is 1/2,500 or 0.0004. Therefore, q is the square root, or 0.02. That is the answer to first question: the frequency of the cystic fibrosis (recessive) allele in the population is 0.02 (or 2%).

2. The frequency of the dominant allele in the population.

Soln. The frequency of the dominant (normal) allele in the population (p) is simply 1 - 0.02 = 0.98 (or 98%).

3. The percentage of heterozygous individuals (carriers) in the population.

Soln. Since 2pq equals the frequency of heterozygotes or carriers, then the equation will be as follows: 2pq = (2)(.98)(.02) = 0.04 or 1 in 25 are carriers.

Mechanisms of evolution (the mechanism that disturb genetic equilibrium)

Different Hardy-Weinberg assumptions, when violated, correspond to different mechanisms of evolution.

Mutation:

Although mutation is the original source of all genetic variation, mutation rate for most organisms is pretty low. So, the impact of brand-new mutations on allele frequencies from one generation to the next is usually not large. (However, natural selection acting on the results of a mutation can be a powerful mechanism of evolution)

Non-random mating

In non-random mating, organisms may prefer to mate with others of the same genotype or of different genotypes. Non-random mating won't make allele frequencies in the population change by itself, though it can alter genotype frequencies. This keeps the population from being in Hardy-Weinberg equilibrium.

Gene flow

Gene flow involves the movement of genes into or out of a population, due to either the movement of individual organisms or their gametes (eggs and sperm, e.g., through pollen dispersal by a plant). Organisms and gametes that enter a population may have new alleles, or may bring in existing alleles but in different proportions than those already in the population. Gene flow can be a strong agent of evolution. It also means change in gene pool and frequencies of allele in it.

Small population size (genetic drift)

Genetic drift involves changes in allele frequency due to chance events – literally, "sampling error" in selecting alleles for the next generation. Drift can occur in any population of non-infinite size, but it has a stronger effect on small populations. We will look in detail at genetic drift and the effects of population size.

Natural Selection.

- The most famous mechanism of evolution natural selection occurs when one allele (or combination of alleles of different genes) makes an organism more or less fit, that is, able to survive and reproduce in a given environment. If an allele reduces fitness, its frequency will tend to drop from one generation to the next. We will look in detail at different forms of natural selection that occur in populations.
- All five of the above mechanisms of evolution may act to some extent in any natural population. In fact, the evolutionary trajectory of a given gene (that is, how its alleles change in frequency in the population across generations) may result from several evolutionary mechanisms acting at once. For instance, one gene's allele frequencies might be modified by both gene flow and genetic drift. For another gene, mutation may produce a new allele, which is then favoured (or disfavoured) by natural selection.