5.8 Population genetics and genetic variation

Genes and environment in evolution; and genetic drifting

Variation

There are two main categories of variation: **discontinuous variation** refers to qualitative differences between phenotypes (clearly distinguishable categories, rather than a spectrum, such as gender and blood type); and **continuous variation** which refers to quantitative differences between the phenotypes, that could be scaled and are measurable, such as height and body mass.

Both continuous and discontinuous variation might be the result of more than one gene, although if there is more than one gene involved in *discontinuous* variation, the genes will interact in an **epistatic** way where one gene masks or influences the expression of another. In discontinuous variation, different alleles at a single gene locus have significantly different effects on the phenotype, and different gene loci have quite different phenotypic effects – examples include codominance, dominance and recessive patterns of inheritance.

In continuous variation, traits are controlled by two or more genes. Each gene provides an additive component to the phenotype. Different alleles at each gene locus have a smaller effect on the phenotype. A large number of different genes may have one combined effect on the phenotype (these are known as **polygenic**, as opposed to discontinuous which is **monogenic**).

Environmental influences

It cannot be said that everything is down to genetics. The *nature-nurture debate* considers how far certain phenotypes are due to the genotype, the environment, or a mixture of the two. For example, intelligence in humans is partly determined by genes, and partly by environment. Children inherit many genes, with alleles from each parent, giving a genetic potential. However, that potential is only realised with the help of a stimulating learning environment both at home and at school. It is also aided by healthy diet for proper development of the brain and nervous system. These are all environmental influences on biological explanations, which suggests not everything is down to nature.

The role of genes and environment in evolution

Whether the environment (as in *natural selection*) or humans (as in *artificial selection*) are doing the selecting, genetic variety within the population is necessary. When the environment changes, those individuals that are well-adapted will survive and reproduce, passing on their advantageous alleles to their offspring.

Environmental factors which may limit the growth of a population include space (for plants to grow, or animals to defend a feeding territory), availability of food, light, minerals or water, predation and infection by pathogens. Some of these factors are **biotic** (living) and others are **abiotic** (non-living), but they all offer **environmental resistance**.

Due to the natural variation within the population, some members will be better adapted than others for survival, and have a better chance of out-competing the others as a result. Those with advantageous adaptations are much better equipped for survival in the struggle for existence. Environmental factors which limit the growth of a population are known as **selection pressures** (e.g. for prey animals, such as a rabbit, are more likely to survive predation if they are well camouflaged and quick to escape – so the selection pressure for them is predation).

How do small populations occur in nature?

A natural disaster, such as a large volcanic eruption, or a disease pandemic, may cause a **population bottleneck** (sudden shrinkage to a very small size). For example, in 1775 in the Pingelap atoll in the western Pacific Ocean, a storm and famine reduced the population to about 30 people. Today's 2,000 inhabitants are descended from those 30 survivors. About 5% of them have a form of eye defect caused by a recessive allele. This disorder is extremely rare in other human populations, but family pedigrees have shown that one of the original survivors was a chief who was a heterozygous carrier for the condition. If he was the only one, there would have been the allele frequency of 0.016 (or 1 out of 60) amongst those 30 people.

Biological and phylogenetic species concepts

A **species** is a starting point for the classification of living organisms, by our modern classification system. There are two main definitions of what a species is, from two different approaches to biology:

- the **biological species concept** suggests that a species is a 'group of organisms that can interbreed and produce fertile offspring, and are reproductively isolated from other such groups' although this proves problematic when trying to classify species which do not reproduce sexually; and a further weakness of this concept is that it does not take into account that some members of the same species are physically and physiologically different
- the **phylogenetic species concept** suggests that a species is a 'group of organisms which share characteristics of morphology, physiology, embryology and behaviour and occupy the same ecological niche'

Population genetics and genetic drift

A group of individuals carries a larger number of different alleles than an individual. This gives rise to a pool of genetic diversity that can be measured using the *Hardy-Weinberg principle* (below). Factors such as migration, selection, **genetic drift** and mutation can alter the amount of genetic variation within a population of a species.

Genetic drift occurs where there are changes in *allele frequency* amongst the population. In extreme cases, genetic drift may lead to the chance elimination of one allele from the entire population. It reduces genetic variation and may reduce the ability of a population to survive in a new environment. It could contribute to the extinction of a population or species, or could even lead to the production of a new species.

What we observe in members of a population is the phenotype, and not the genotype. To measure the frequency of an allele, we need to know firstly the mechanism of inheritance for the trait in question, and also how many different alleles of the gene for that trait exist in that population.

Calculating allele frequency

In a population of 100, 36 individuals are **MM**, 48 are **MN** and 16 are **NN**. The 36 MM individuals represent 72 M alleles, and the 48 MN individuals represent another 48 M alleles

Therefore out of 200 alleles in the population for this trait, 120 are M, so the frequency of the allele is 120/200 or 0.6

Therefore, the frequency of N is 0.4

For traits that show codominance, the frequency of the heterozygous phenotype is the same as the frequency of the heterozygous genotype.

The Hardy-Weinberg principle

The mathematician Hardy and the doctor Weinberg established a formula to calculate the allele frequency in a population. The **Hardy-Weinberg principle** states that allele and genotype frequency will remain the same when the population is very large, to eliminate sampling error; when mating in the population is random; when there is no selective advantage to certain genotypes and when there is no mutation, migration or genetic drift.

There are also some useful equations. If the frequency of dominant alleles is ***p*** and the frequency of recessive alleles is ***q***:

$$p + q = 1 \quad \text{and} \quad p^2 + 2pq + q^2 = 1$$

- p^2: frequency of homozygous dominant
- $2pq$: frequency of heterozygous dominant
- q^2: frequency of homozygous recessive

This has a number of uses, for example, the Hardy-Weinberg principle can be used to calculate the number of **albinism** carriers, based on knowing how many people have the condition. Albinism occurs in 1 of every 10,000 of the British population, and we can use this to work out what percentage of the population are carriers of the allele.

- we know that $q^2 = 0.0001$
- therefore $q = \sqrt{0.0001} = 0.01$
- and since $p + q = 1$ we can calculate that $p = 0.99$
- so the frequency of albinism allele carriers = $2pq = 2 \times 0.01 \times 0.99 = 0.0198$
- so the percentage of the population is 1.98% of Britons