Continuous and discontinuous variation

Variation, the small differences that exist between individuals, can be described as being either **discontinuous** or **continuous**.

Discontinuous variation
This is where individuals fall into a number of distinct **classes** or categories, and is based on features that cannot be measured across a complete range. You either **have** the characteristic or you **don't**. Blood groups are a good example: you are either one blood group or another - you can't be in between. Such data is called **discrete** (or **categorical**) **data**. Chi-squared statistical calculations work well in this case.
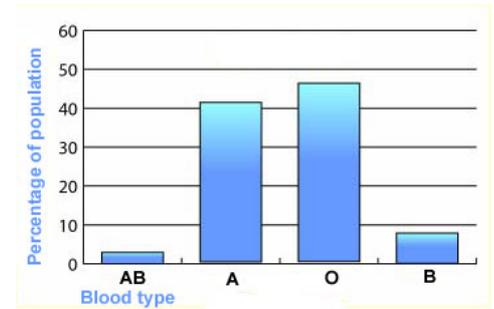
Discontinuous variation is controlled by alleles of a single gene or a small number of genes. The environment has little effect on this type of variation.

Continuous variation
In continuous variation there is a complete range of measurements from one extreme to the other. **Height** is an example of continuous variation - individuals can have a complete range of heights, for example, 1.6, 1.61, 1.62, 1.625 etc metres high.
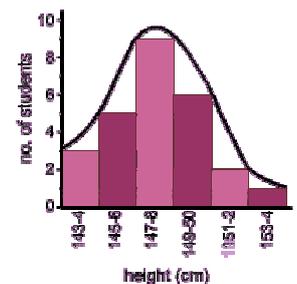
Other examples of continuous variation include:

- Weight; Hand span; Shoe size; Milk yield in cows

Continuous variation is the combined effect of many genes (known as **polygenic inheritance**) and is often significantly affected by environmental influences. Milk yield in cows, for example, is determined not only by their genetic make-up but is also **significantly affected by environmental factors** such as pasture quality and diet, weather, and the comfort of their surroundings.
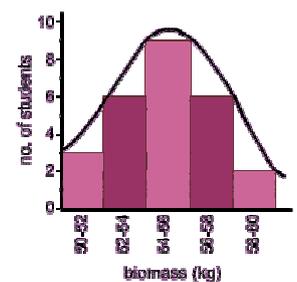
When plotted as a histogram, these data show a typical bell-shaped **normal distribution curve**, with the **mean** (= average), **mode** (= biggest value) and **median** (= central value) all being the same.

Standard Deviation (σ)

This is a measure of the variability of a data set. Put simply, the taller and narrower the histogram, the **lower** the SD (σ) and the **less variation** there is. For a low, wide, histogram, the opposite applies:
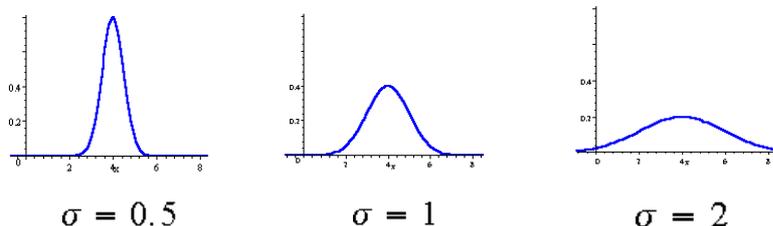
Standard Error is a refinement of this, which takes into account sample size (n). The formula is:

$$\sigma = 0.5 \qquad \sigma = 1 \qquad \sigma = 2$$

$$s = \frac{\sigma}{\sqrt{n}}$$

Sampling Error

When collecting data, it is vital that the data is **reliable** and **reflects real differences** in the population. This can be ensured by having a **large data size**, but it is also necessary to ensure that the **data collected is typical** of the whole population – in other words, that it is **randomly collected**. In the laboratory, we repeat experiments 3 times and/or pool class data. In field-work, we use **random number tables**; grids laid out with tape-measures, and large numbers of quadrats to ensure that our data are reliable and randomly collected.

N.B. It is easy to (unwittingly) introduce bias into the collection of results, and **'to avoid bias' is an important point to make in your answers** to questions on this topic

Causes of Variation

Variation in the phenotype is caused either by the environment, by genetics, or by a combination of the two. Meiosis and sexual reproduction introduces variation (see Ch 1), through **Independent assortment** of the parental chromosomes**;** through **Crossing-over** during Prophase I; and through the **random fertilisation** that forms the zygote.

Mutations

These changes to the genetic make-up of an individual which **cannot** be accounted for by the normal processes (*above*) may (rarely) involve **chromosomal mutations** (e.g. *Down's Syndrome*, where an individual has **trisomy (= 3 copies) of chromosome 21;** they therefore have 47 chromosomes), but more commonly, these mutations are confined to a **single gene** and are known as **gene mutations**. Since DNA replication is **not** perfect, with an error rate of about 1 in 10^{12} bases, we gradually acquire more of these during our lives. Certain chemicals and radiation increase this error rate, but note that 99% of our DNA is 'junk DNA' and so the vast majority of mutations are unlikely to have any effect whatsoever.
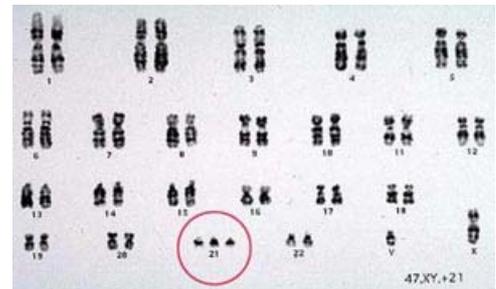
Base deletion means a single base is omitted. This will **affect all subsequent amino-acids** in that protein and the effects are therefore likely to be severe.

Base substitution means that one base is replaced by another. This will **only affect a single amino-acid,** and normally this has **little effect unless the active site of an enzyme is affected**. One example where the effects **are** dramatic is **sickle-cell anaemia**, which is caused by a single amino-acid substitution (*see later notes*)

Gene pool

This is the total number of **alleles** (note, **not** genes!) in a breeding population. A human population will have 3 alleles for blood groups (A, B, O). Since everyone **must** have one of each of these alleles, the **total frequency must be 1.0** and the frequency of each allele can then be expressed as a decimal. In the above case, the frequencies vary greatly amongst different ethnic groups (*see table*)

Population	I^A	I^B	I^O
African	15	10	75
Caucasian	30	15	55
Orientals	20	22	58
Amer. Indians	0-55	0	45-100
Australasian	50	0	50
Worldwide	22	16	62
Europe	22	11	67

Hardy – Weinberg

This principle states that the proportion of the different alleles in a gene pool (breeding population) only changes as a result of an external factor. Thus providing the following assumptions are met, each generation will be the same as the present one:

- **Large population**
- **No migration** – either in (**immigration**) or out (**emigration**)
- There is **random mating**
- **No mutations** occur
- All **genotypes are equally fertile**

Worked Example:

In a population containing 2 alleles, **T** and **t**, three genotypes will exist – **TT**, **Tt** and **tt**. If **T** is a **dominant allele**, then it is impossible to tell which individuals are **Tt** and which are **Tt**. It will also be impossible to tell **the relative proportions** of these two genotypes. The Hardy-Weinberg equation allows us to calculate that.

Stages in the calculation:

Let the proportion of **T** alleles = p and the proportion of **t** alleles = q. Then:

$$p + q = 1$$

Both p and q are decimal fractions, and therefore both will be less than 1.0.

That means that the proportion of the different genotypes is:

TT individuals are p^2;
Tt individuals are $2pq$; and
Tt individuals are q^2
Since that is the whole population, it follows that:

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

Now, we **can** see, and count, those individuals that are **tt** (double recessive), so:

1. **Work out the proportion (as a decimal fraction, i.e. 5% = 0.05) of tt individuals in the whole population.** The question will give you this data, so just use your calculator!
2. **Take the square root** of that number. That gives you q
3. Since $p + q = 1$, you can now calculate p (it is $1 - q$).
4. **Substitute the values you now** have in the equation $1 = p^2 + 2pq + q^2$ to give you the decimal fraction of each of the genotypes.
5. **If the question asks you** to calculate 'how many individuals in then population are….', then **multiply the number you have (for the required genotype) by the total population**. Remember that **you cannot have a fraction of an individual**, so round up/down to the nearest whole number.
6. Easy, isn't it!
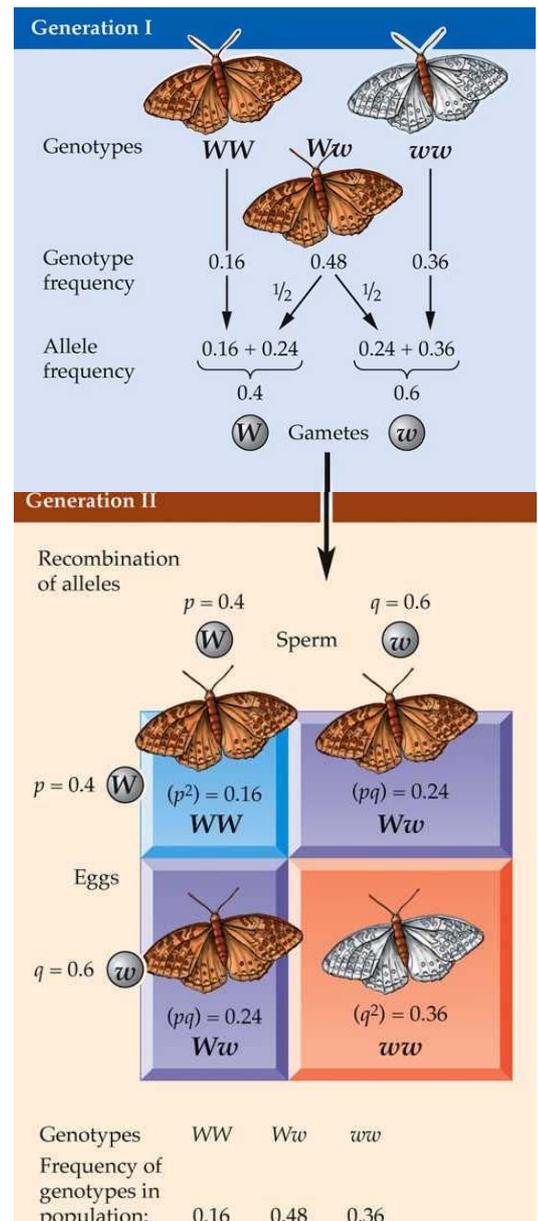
Natural selection

The Hardy-Weinberg equation requires such stringent conditions to be met, that, in the real world, it is seldom, if ever, true. Thus, the equation is used to show that **populations change over time**, something we normally call **evolution.**

The driving force behind evolution is **natural selection**, and this can occur in three main ways:

Stabilising selection:

This is **the most common**, and is a **response to a stable environment.** A good example is birth weight, since under-weight babies are clearly less likely to survive, and over-weight babies are likely to get stuck during birth, killing not only themselves, but also their mothers! The result is that **the mode stays the same, but the SD of the population falls**, i.e. the **population graph gets narrower and taller**, as **selection against mutations** takes place. This is most apparent in small, isolated populations, or when a **bottleneck event** takes place.
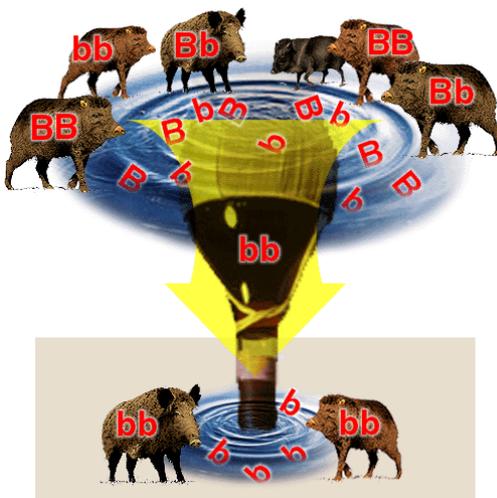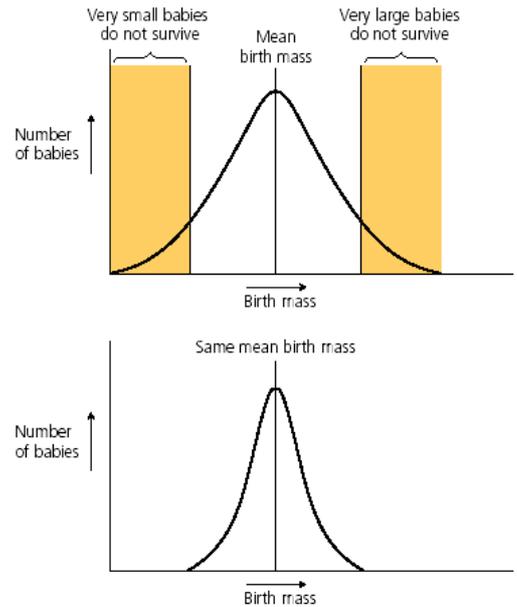
A **bottleneck event** occurs when a population is reduced to just a few breeding individuals (e.g. cheetahs). Though the total population may later recover, they will all be descendants of the few originals (Adam and Eve?) and so will have a **much narrower gene pool** than the original population.

There have been a number of events in geological history ('**mass extinctions'**) when all forms of life on Earth were reduced to around 1% of the previous community. These events are **followed by rapid evolution and an increase in diversity**, as species evolve to fill every available **niche**. The last such event was 65 million years ago, when the Dinosaurs became extinct, allowing mammals to replace reptiles as the dominant life-forms on Earth.

Directional selection:

This is the most common form of selection that results in a population with a new trait. It takes place **whenever change occurs in the environment**, such as when a poison is used and resistant individuals begin to occur. These soon become the dominant type within the population. It does not matter if the example is Warfarin resistance in rats, DDT resistance in mosquitoes, or antibiotic resistance in bacteria. In Nature, such selection can also occur, as we saw, in spine selection in Holly, or when cacti are grazed (*see left*):
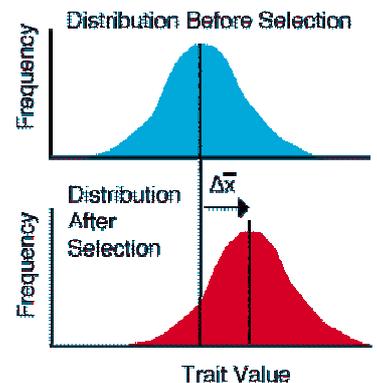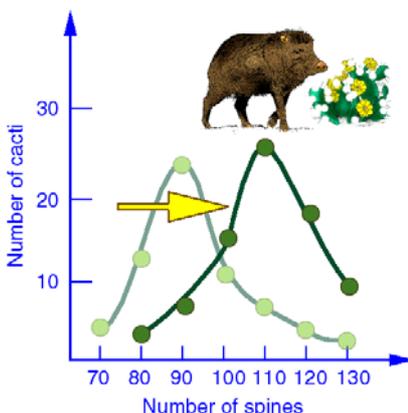
Disruptive selection:

This is much less common and results in two distinct populations, or **morphs**. Eventually, these two forms may become so distinct that they become new populations.
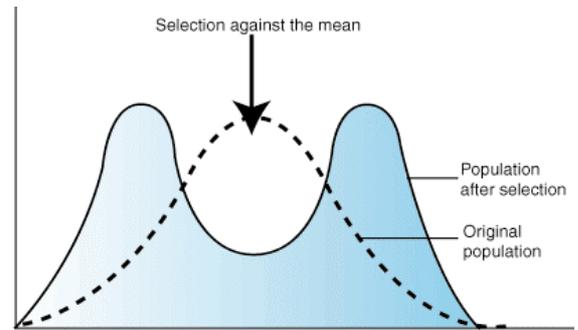

The example usually used is *Biston betularia,* the **Peppered Moth** (*left*). This moth hides on birch-trees during the day, and two forms are favoured – like (clean air, pale tree-trunks) and dark (sooty air, dark tree-trunks). Any intermediate forms are obvious on **any** tree, and so are selected against. The dark (or **melanic)** form became more common in polluted city air after the Industrial Revolution, but has now become rare again, following the Clean Air Acts of the last 50 years. This is usually used as an example of **Natural Selection** in action.

Sickle-Cell Anaemia

Sickle-cell is an inherited condition in which two **co-dominant alleles** exist – Hb^S and Hb^A. This condition is caused by a single point substitution mutation, resulting in a single amino-acid being changed. With 2 alleles, three phenotypes therefore exist:
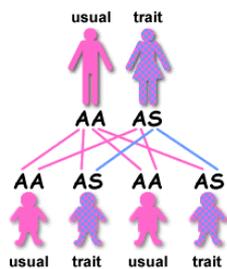
1. Hb^SHb^S. These individuals have **sickle-cell anaemia**, and normally die young.
2. $Hb^S Hb^A$. These individuals have **sickle-cell trait**, have only a few sickle-cells **and are resistant to malaria**
3. $Hb^A Hb^A$. These individuals are **normal**, and are **susceptible to malaria.**
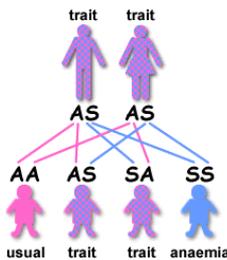
In most parts of the world, **stabilising selection** selects against this mutation, so it is very rare. However, **in parts of the world where malaria is common, there is a selective advantage in having sickle-cell trait**. These individuals are resistant to malaria but suffer few side-effects, and can live healthy and productive lives. Normal people may well be infected with malaria and die, whilst those with sickle-cell anaemia die young. That means the population is selected for this mutation (*left*):

Note that, with modern medicine, sickle-cell individuals can survive and reproduce, so we can now **also** have:

Frequency of individuals (y-axis)

Phenotypes (shell color) (x-axis)

Original population

Evolved population / Original population
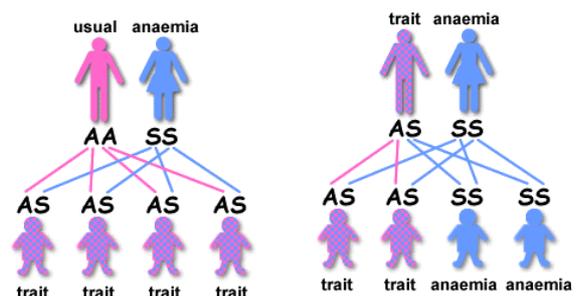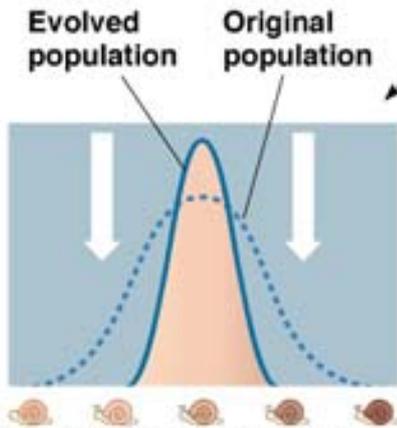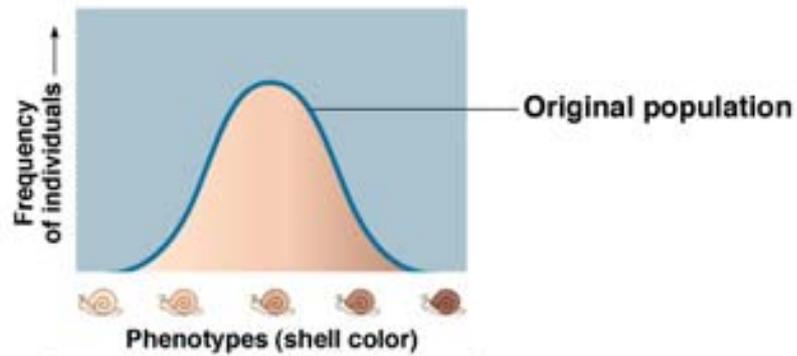
Stabilizing selection culls extreme variants from the population, in this case eliminating individuals that are unusually light or dark. The trend is toward reduced phenotypic variation and maintenance of the status quo.
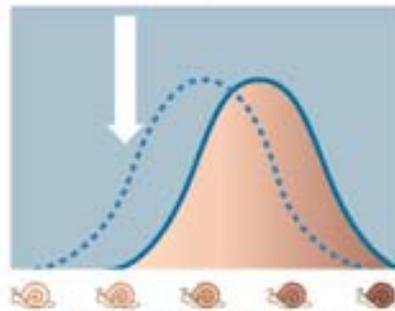
Directional selection shifts the overall makeup of the population by favoring variants of one extreme. In this case, the trend is toward darker color, perhaps because the landscape has been blackened by lava.
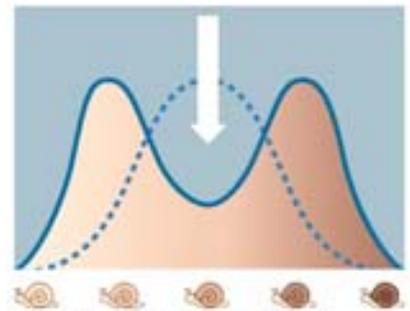
Diversifying selection favors variants of opposite extremes over intermediate individuals. Here, the relative frequencies of very light and very dark snails have increased. Perhaps the snails have colonized a patchy habitat where a background of white sand is studded with lava rocks.