A) DISCONTINUOUS GENETIC VARIATION

Example: shell colouration in the land snail, *Cepaea nemoralis*. Shell is highly polymorphic in appearance. **Background colour**: highly variable – yellow, pink or brown (or others). **Banding**: shells vary in number of bands superimposed on background - zero, one, three or five bands.

Breeding tests - background colour and band number are determined by alleles at a series of independent loci, with clear dominance relations. Background colour - one locus and alleles giving darker colours are dominant. Banding - under the control of several different loci.

Data is available on the evolution of these colour variants. Natural selection plays role because variation in gene frequencies correlated with variation habitat. But very little known about the molecular structure or the mode of action of these genes. For the **background** genes we can presume that they are responsible for pigment production. For the **band** genes, a more complicated mechanism is required which controls the location of pigment.

B) DISCONTINUOUS ENVIRONMENTAL VARIATION

Discontinuous variation is **not** always caused by genetic variation; it can be purely environmental in origin - i.e. a consequence of the conditions encountered during development or subsequently. Often found where organisms receive reliable cues of their future environment and where it therefore pays to adopt a particular phenotype. In such a situation, the **same genotype can give rise to different phenotypes**.

Example: Shell morphs in the ACORN BARNACLE living in the gulf of California. The ‘**conic**’ form is more common and widespread. A ‘**bent**’ form is found in the northern gulf. Distribution coincides with the distribution of one of the barnacle’s main predators (snail, *Acanthina angelica*). ‘**Bent**’ form is adapted to the presence of the predator because the snail has reduced feeding success on the bent form. ‘**Bent**’ morphs are the consequence of a developmental shift, triggered by the presence of the snail.

In experiments, snails were either added or not added to cultures of developing barnacles that had settled. ‘**Bent**’ forms were produced where the snail was added; presumably due to a chemical cue. These morphs are stable but are the result of a environmental switch. Why does the ‘**conic**’ form persist? The reason is that, in the absence of the predator, it is fitter with higher feeding efficiency, growth rate and reproductive success.

Such environmentally-based discontinuous variation also called **ADAPTIVE PHENOTYPIC PLASTICITY**. Common in plants - can be radically different forms within a single population of a single species. Note that phenotypic plasticity isn’t always adaptive. Environment can come in the form of poor nutrition and disease causing dwarfism = maladaptive phenotypic plasticity.

C) CONTINUOUS VARIATION

Most evolutionarily interesting characters are not discontinuously variable but show continuous variation. The individuals in a population cannot be allocated to discrete categories. Classic example is body size. If you measure a linear dimension e.g. human height, flower length, then what you typically find is a bell-shaped normal frequency distribution. The majority of measures fall in the middle of the distribution with a range of observations tailing away on either side. This is CONTINUOUS VARIATION of **QUANTITATIVE CHARACTERS**.

How is this sort of variation caused? At DNA level, genetic variation is discontinuous. So do continuously variable characters have a genetic basis? In reality it is almost always the case that quantitative characters are caused to vary by a combination of underlying genetic variation and by the environment (e.g. nutrition levels, temperature during development, disease etc.).
Typically the genetic effects underlying quantitative characters come from many gene loci with each individual gene having such a small effect that we do not pick it up as discontinuous variation. The analyses of these genes require the techniques of quantitative genetics.

**How do we know that such characters have any genetic basis at all?**

Ask whether, when we control for the effects of environment, do offspring resemble their parents (or other relatives) for the character of interest? If all the variation in the character in the parents is caused by variation in the environment, then it should not be transmitted to the offspring. Numerous demonstrations that offspring do resemble their parents for quantitative characters in controlled environments. Example (HANDOUT) of *Drosophila* wing length.

The mid-parent is the mean of the character in the parents. Offspring value is the mean trait value of several progeny from that pair of parents. Here, the axes are drawn at the mean for the parents (X-axis) or the mean for the progeny (Y-axis) and they cross at the grand mean. There is a strong correlation - parents with short wings tend to produce offspring with short wings.

A key assumption is that environment is standardised for both generations. If, for example, the nutrition levels in offspring generation were reduced then all offspring could be smaller and this could obscure the underlying relationship.

In general, this approach is useful but this example was done in the laboratory where the environment is benign and constant. So could ensure that parents and offspring grow up in more or less the same environment. Perhaps under these conditions genes might play a large part in producing the character? What about the real world? In the field, life is harsher and the environment is a lot more variable. So can this genetic resemblance between parents and offspring be detected for characters of evolutionary interest under field conditions?

Field measures are still rare, because difficult to do but some good examples: e.g. an island population of song sparrows, *Melospiza melodia*. Examined beak depth (which affects ability to feed on different sizes of seed). Measured trait for breeding pairs of adults, banded all their nestlings in the nest. Recaptured nestlings as adults and measured bill-depth when fully grown.

Working with an island population meant that recapture rates were high. HANDOUT shows the results. Top graph shows the relationship between parent and offspring and there is a clear strongly positive correlation. But there are potential problems with this under field conditions. There could be a common environment in terms of territory quality, or food supply or parental size. So big parents might be better providers of food and so increase their progeny's size.

However the nice thing about the song sparrow data is that they are from young that were cross-fostered either as eggs or as very young nestlings. So any correlations with parental ability or territory quality should be broken. And indeed these factors seem to be of little importance because in lower graph, we have the same data for the progeny now plotted against their foster parents. There is no significant correlation. There is a genetic basis to this typical quantitative trait. Also checked for any maternal effects such as variation in mother's deposition of food (egg yolk). Did separate regressions for mother and father on the progeny values - these plots were not significantly different from each other.

**How do we know that several genes are involved in these characters?**

Why for instance couldn't it be one gene with many alleles? Example: classic work with tobacco plants. Tobacco habitually self-fertilises. You know already that the genetic consequence of inbreeding is a loss of genetic variability, that is, homozygosity.

Tobacco breeders have produced lines that are completely homozygous at all their loci. These true breeding lines have no variability except through new mutations. These lines are used to investigate the genetic basis a typical quantitative character in tobacco, the corolla length of the tobacco flower (HANDOUT). Let’s examine the pattern of results from some breeding tests.
and compare two rival explanations of the genetic basis of corolla length to see which one fits the facts better.

Step 1 - Start with 2 parental lines with different corolla lengths (~40mm versus ~90mm). Note variation within a parental line. Must be entirely environmental variation because within a line they are genetically identical with same alleles as each other for each locus. So between them the parental lines cannot have more than two alleles at any locus.

Step 2 - Make the F1. TOBACCO can cross breed as well as self (with aid of a person using a paintbrush to transfer pollen between flowers). The F1 distribution of corolla length is intermediate with about the same spread as the parents. Note that again this must be all environmental variation – all the F1 plants are the same as each other in genotypic terms.

**Hypothesis 1**: If only 1 gene responsible - then the selfed F1 should give only 3 classes because there are a maximum of 2 alleles at the critical locus that have come from the parents. Expect Parent 1, Parent 2, and F1 phenotypes; 3 classes with double the area under the middle peak.

**Hypothesis 2**: If more than 1 gene is responsible, then we will see more phenotypic classes. This is what is observed.

Step 3: Make the F2. The F2 distribution has a large spread of values. The F2 distribution has the same corolla length, on average, as in the F1. But the variance of the distribution is much larger. In the F2 we have 2 sources of variation: a) the segregation and independent assortment of the different pairs of alleles controlling corolla length and b) environmental factors.

Step 4: Make the F3. This is the critical step. The F3 results prove that must be dealing with the effects of many gene loci. Four different F3 populations were produced by inbreeding particular classes of plants from different parts of the “range” of F2 corolla length values. There was substantial variation between and within the F3 populations (see picture). This pattern is true irrespective of where in the F2 population their parents were drawn from.

These F3 results show that a typical quantitative trait is controlled by several loci.

Most of the recommended texts for BIOL2007 describe extensions of this to a general picture. **HANDOUT** shows the changes in the phenotypic distribution of a quantitative trait under different genetic models ranging from a single locus with 2 alleles to many loci each with two alleles.

**Analysis of quantitative traits**

The fact that the variation is quantitative, not qualitative, poses potentially formidable problems for the geneticist. Segregation ratios are difficult to discern because the number of phenotypes is large and one phenotype blends imperceptibly into the next. The key is to think in terms of the distribution of phenotypes for a quantitative trait. We use a statistical approach to determine the properties of the distribution.

The thrust of the statistical analyses is to breakdown (‘partition’) the observed variation in a trait into components that are genetic and environmental. Then the genetic component can be used to make predictions about the phenotypes of the offspring of particular controlled crosses.

Before going onto discuss some of the standard terms in quantitative genetics, you need to remind yourselves about some basic statistical quantities and their properties. I’ve put these reminder definitions on 1 side of the **HANDOUT** – this shows how to calculate some relevant items.

**Standard ways of describing quantitative characters.**

First think of an individual: phenotype = genotype + environment

\[ P = G + E \]
Now think of a population of these individuals: we have a range of phenotypic scores which have an associated variance (defined on statistics page of HANDOUT) and so have a variance in genotypic effects and a variance in environmental effects.

Can rewrite equation as: \( V_p = V_g + V_e \)

Variance in genotypic effects can be divided into 3 components

**ADDITIVE (A)** – effects of individual alleles that are inherited.

**DOMINANT (D)** effects – due to the combinations of alleles (homozygotes versus heterozygotes) produced in any given generation but are not inherited by offspring.

**INTERACTION/EPISTASIS (I)** effects - due to epistatic interactions between alleles at different loci. Again not inherited by offspring - when the specific combinations are broken up (e.g. by recombination) the effect disappears.

So can rewrite equation as: \( V_p = V_a + V_d + V_i + V_e \)

A fundamental quantity in quantitative genetics is **HERITABILITY**. This is a description of the amount of the total phenotypic variance in a character that is genetic. Usually written \( V_a/V_p \).

The winglength picture (HANDOUT) is relevant. The idea is that relatives can have similar genotypes (because of the genes that they have in common) and this will produce a correlation between their phenotypic character values.

**What is the diagonal line?** It is a regression line which quantifies the resemblance between the 2 sets of relatives, plotted on the X and Y axes, as MID-PARENTAL and OFFSPRING values.

The slope of this line can vary from 0 to 1 and estimates the HERITABILITY of the character. If all the variation in the character is **environmental**, then the slope will be **ZERO**. If all of the variation was heritable **genetic** variation then the value would be **ONE**. The symbol for heritability is \( h^2 \). In the case of the winglength, it is 0.58 plus or minus 0.07 standard error, i.e. it is significantly positive.

A very important fact about these figures is that they are only true for the particular stock and the conditions under which the measurement was made. They could well be different for a different stock or a different environment.

**Quantitative genetic variation can produce discontinuous phenotypic variation**

**Note that although we’ve emphasised the notion that genetic variation at many loci can give rise to continuous phenotypic variation, it is possible for such variation can cause discontinuous phenotypic variation.** An example is where a character comes in whole numbers; bristles on the abdomen of *Drosophila* (we don’t see fly phenotypes with half a bristle). Whether or not to make a bristle is an all-or-none decision, but the number made is influenced by many genes.

Bristle number behaves like a standard quantitative trait. We can use the same type of analysis as described before. These types of character, (ones that you can count) are called **MERISTIC CHARACTERS**.

Sometimes, the trait can be more complex e.g. some human medical disorders such as **SCHIZOPHRENIA** appear to be “threshold” characters. These are explained as having an underlying variable called "LIABILITY" which is both genetic and environmental in origin, with a threshold dose above which the character appears. **Full details of such threshold traits is beyond the scope of BIOL2007.**