By the end of this chapter you should be able to:

1. explain what is meant by the terms gene mutation and chromosome mutation;
2. describe the difference between continuous and discontinuous variation;
3. explain what is meant by variance;
4. explain the basis of continuous and discontinuous variation by reference to the number of genes which control the characteristic;
5. recognise that both genotype and environment contribute to phenotypic variation and describe examples of the effect of the environment on the phenotype;
6. describe the interaction between loci (epistasis) and predict phenotypic ratios involving epistasis;
7. explain the meaning of the terms linkage and crossing over, and explain the effect of linkage and crossing over on the phenotypic ratios from dihybrid crosses;
8. use the $\chi^2$ (chi-squared) test to test the significance of differences between observed and expected results.

Living organisms vary. The applied geneticist must be able to recognise the extent of this variation and to distinguish inherited variation from that caused by the environment. Only when the geneticist has that information can it be applied to such fields as selective breeding, genetic engineering and human genetics. The first steps, then, are to identify the source of variation, to describe it and measure its extent.

Mutation

The source of inherited variation is mutation, which may be defined as an unpredictable change in the genetic material of an organism. Such a change may be in the structure of a DNA molecule (a gene mutation), or in the structure or number of chromosomes in the cells of the organism (a chromosome mutation).

Gene mutations

Gene mutations are the source of the different alleles of a gene (see Biology 1 chapter 6). Each is a change in the sequence of base pairs in a part of a DNA molecule coding for a polypeptide. Such a change may be:
- substitution of one or more base pairs by others;
- addition of one or more base pairs;
- deletion of one or more base pairs.

These changes in the sequence of base pairs of DNA may, or may not affect the sequence of amino acids in the polypeptide coded by the gene. Any change in the sequence of amino acids may affect the three-dimensional structure of the polypeptide and hence alter its effect in the organism.

A substitution of one base pair for another does not always alter the amino acid sequence. For example, changing the triplet of bases on the sense strand of DNA from C–T–T to C–T–C does not alter...
2 Variation

the sense of the triplet. Both triplets code for the amino acid glutamic acid (glutamate). This is a silent mutation. However, changing the DNA triplet C–T–T to C–A–T alters the amino acid coded for from glutamic acid to valine. Just such a change in the gene for the β-globin chain of haemoglobin, substituting valine for glutamic acid, gives sickle cell haemoglobin and the symptoms of sickle cell anaemia (see Biology 2).

A substitution may have an even more drastic effect. Altering the DNA triplet A–T–G, which codes for the amino acid tyrosine, to A–T–T or to A–T–C produces a ‘stop’ triplet. During translation, polypeptide synthesis stops at that point and the full length polypeptide cannot be produced.

Either addition or deletion of a base pair alters the ‘reading frame’ of the triplet code of DNA, affecting all triplets downstream from the mutation. This usually changes the structure of the coded polypeptide significantly.

SAQ 1.1


After a mutation the DNA code is T–A–C–A–T–T–A–C–G.

a State what type of mutation has occurred.
b Explain the consequence of the mutation.
c Write out another change in the DNA sequence which would have the same effect.

When a whole triplet of base pairs is deleted the final polypeptide lacks one amino acid. An example of this sort of gene mutation is responsible for most cases of the genetic disease cystic fibrosis (CF), which you can read about in chapter 5. 70% of the sufferers of CF lack one amino acid, phenylalanine, in an ion channel in the plasma membranes of certain types of cells. The deletion removes just one amino acid from a chain of 1480 amino acids but as a result the polypeptide fails to reach its position in the plasma membrane of the cells concerned. More than 400 different mutations of the gene coding for this polypeptide have been identified, variously resulting in:

- no polypeptide synthesis;
- incomplete synthesis, leaving the polypeptide ‘stuck’ in the endoplasmic reticulum;
- the polypeptide reaching the plasma membrane but not functioning correctly.

SAQ 1.2

Suggest what types of gene mutation could account for each of these three different effects.

A DNA triplet of base pairs may be repeated many times, giving a string of the amino acid coded by that triplet in the final polypeptide. This sort of mutation is called a ‘stutter’ and is responsible for Huntington’s disease (see chapter 5) where the triplet repeat inserts a series of glutamines into the final polypeptide.

Chromosome mutations

Chromosome mutations include changes in chromosome structure or in the number of chromosomes.

Changes in chromosome structure can happen during division, when pieces of chromosome may:

- be duplicated;
- break off and be lost;
- break off and rejoin with the sequence of genes inverted;
- break off and attach to another chromosome.

The transfer of a portion of one chromosome to another is called translocation. The inherited form of Down’s syndrome (see chapter 5) results from the end of the long arm of chromosome 21 joining another chromosome.

Changes in chromosome number result from unequal separation of chromosomes between daughter cells during division. After meiosis, one daughter cell may lack one chromosome and the other have one chromosome too many. This is called non-disjunction. A fertilisation which joins a normal gamete with a gamete containing one chromosome too many results in three copies, or trisomy, of that chromosome in the zygote. Trisomy gives rise to various genetic diseases depending on which chromosome is involved. The usual form of Down’s syndrome is caused by trisomy 21 or three copies of chromosome 21 (see chapter 5).
**Discontinuous and continuous variation**

The total appearance of an organism is called its phenotype. Phenotypic differences between you and your friends include qualitative differences, such as different blood groups, and quantitative differences such as height and mass.

Qualitative characteristics fall into clearly distinguishable categories, with no intermediates. You are either male or female, and also have only one of four possible ABO blood groups: A, B, AB or O. This is discontinuous variation. In contrast, the quantitative differences between individual heights or masses may be small and difficult to distinguish. When the height of a large number of people is measured, there are no distinguishable height classes. Instead there is a range of heights between two extremes (figure 1.1). This is continuous variation.

**Variance**

The variation shown by a quantitative character can be given by the variance, which is a measure of how much spread there is about the mean (average) value for the character. Figure 1.2 shows two distribution curves with different variances. Different variances in phenotype result from differences in both an organism’s genotype (an organism’s genetic make-up) and the effects of its environment, as will be seen later.

A worked example showing how to calculate the mean and variance of a quantitative character is shown in the Box on page 4.

**The genetic basis of discontinuous and continuous variation**

Both qualitative and quantitative differences in phenotype may be inherited via genes. Both may involve several different gene loci. However, there are important differences between them.

In discontinuous (qualitative) variation:

- different alleles at a single gene locus have large effects;
- different gene loci have quite different effects on the character.

In continuous (quantitative) variation:

- different alleles at a gene locus have small effects;
different gene loci have the same, often additive, effect on the character; a large number of loci may have a combined effect on a particular phenotypic character. These are known as polygenes.

Discontinuous variation

Different alleles at a single locus

If possible, look again at chapter 4 of Biology 2 in this series. You will find there a number of examples of the inheritance of discontinuous variation showing the large effects of the different alleles of a single gene. The inheritance of the β-polypeptide gene of haemoglobin and of the gene responsible for red, pink and white flower colour in snapdragons (Antirrhinum) both show the effect of two codominant alleles, that is alleles which both have an effect on the phenotype in a heterozygote. In snapdragons, the two alleles of this flower colour gene are C<sup>R</sup> which gives red flowers, and C<sup>W</sup> which gives white flowers. The phenotypes produced by each genotype are:

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>C&lt;sup&gt;R&lt;/sup&gt;C&lt;sup&gt;R&lt;/sup&gt;</td>
<td>red flowers</td>
</tr>
<tr>
<td>C&lt;sup&gt;R&lt;/sup&gt;C&lt;sup&gt;W&lt;/sup&gt;</td>
<td>pink flowers</td>
</tr>
<tr>
<td>C&lt;sup&gt;W&lt;/sup&gt;C&lt;sup&gt;W&lt;/sup&gt;</td>
<td>white flowers</td>
</tr>
</tbody>
</table>

The inheritance of purple and green stem colour in tomato plants shows the inheritance of two alleles of a gene of which only one, the dominant allele, has an effect in the heterozygote. In a tomato plant which has one allele for purple stems and one allele for green stems, the stems are purple. The allele for green stems is said to be recessive.

Most genes have more than two alleles. The inheritance of the human ABO blood groups provides an example of this situation, known as multiple alleles. It also shows both dominance and codominance of the alleles concerned. The four blood groups, A, B, AB and O, are determined by three alleles of a single gene: I<sup>A</sup>, I<sup>B</sup> and I<sup>O</sup>. I<sup>A</sup> and I<sup>B</sup> are codominant, whilst I<sup>O</sup> is recessive to both I<sup>A</sup> and I<sup>B</sup>. The possible genotypes and phenotypes are:

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype (blood group)</th>
</tr>
</thead>
<tbody>
<tr>
<td>I&lt;sup&gt;A&lt;/sup&gt;I&lt;sup&gt;A&lt;/sup&gt;</td>
<td>A</td>
</tr>
<tr>
<td>I&lt;sup&gt;A&lt;/sup&gt;I&lt;sup&gt;B&lt;/sup&gt;</td>
<td>AB</td>
</tr>
<tr>
<td>I&lt;sup&gt;B&lt;/sup&gt;I&lt;sup&gt;B&lt;/sup&gt;</td>
<td>B</td>
</tr>
<tr>
<td>I&lt;sup&gt;O&lt;/sup&gt;I&lt;sup&gt;O&lt;/sup&gt;</td>
<td>O</td>
</tr>
</tbody>
</table>

Calculating mean and variance

A sample of maize cobs, from a variety known as Tom Thumb, was taken and the cobs measured to the nearest centimetre. The number of cobs in each centimetre length category was counted:

<table>
<thead>
<tr>
<th>Number of cobs in each length category (n)</th>
<th>5</th>
<th>6</th>
<th>7</th>
<th>8</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cob length (cm)</td>
<td>4</td>
<td>21</td>
<td>24</td>
<td>8</td>
</tr>
</tbody>
</table>

The mean cob length ($\bar{x}$) = sum ($\sum$) of all the cob lengths ($x$)/number (n) of cobs, or $= \sum x/n$

$\bar{x} = (5 \times 4)+(6 \times 21)+(7 \times 24)+(8 \times 8)/57 \text{ cm}$

$= 378/57 \text{ cm}$

$= 6.63 \text{ cm}$

Variance = $\sum \frac{n(x - \bar{x})^2}{\sum n - 1}$

$\sum n = 57$

$\sum (x - \bar{x})^2 = 37.44 \text{ cm}^2$

Variance = $37.44/57 - 1 = 0.67 \text{ cm}^2$

The mean cob length ($\bar{x}$) and variance for each length category are:

<table>
<thead>
<tr>
<th>Number of cobs in each length category (n)</th>
<th>x (cm)</th>
<th>(x - $\bar{x}$)</th>
<th>(x - $\bar{x}$)&lt;sup&gt;2&lt;/sup&gt; (cm)&lt;sup&gt;2&lt;/sup&gt;</th>
<th>n(x - $\bar{x}$)&lt;sup&gt;2&lt;/sup&gt; (cm)&lt;sup&gt;2&lt;/sup&gt;</th>
</tr>
</thead>
<tbody>
<tr>
<td>4</td>
<td>5</td>
<td>-1.63</td>
<td>2.66</td>
<td>10.64</td>
</tr>
<tr>
<td>21</td>
<td>6</td>
<td>-0.63</td>
<td>0.40</td>
<td>8.40</td>
</tr>
<tr>
<td>24</td>
<td>7</td>
<td>0.37</td>
<td>0.14</td>
<td>3.36</td>
</tr>
<tr>
<td>8</td>
<td>8</td>
<td>1.37</td>
<td>1.88</td>
<td>15.04</td>
</tr>
</tbody>
</table>

© Cambridge University Press
A woman with blood group A and a man with blood group B, both of whom were heterozygous at the ABO locus, could produce a child with any one of the ABO blood groups:

**SAQ 1.3**

Draw a genetic diagram to show the offspring expected from crossing two rats heterozygous at this locus.

**Different gene loci**

In discontinuous variation, different gene loci have different effects on a phenotypic character. For example, in tomato plants, a number of genes code for different features of the plants’ leaves. Among others, one gene codes for leaf shape, another for the presence or absence of hairs on the leaves and a third for the presence or absence of chlorophyll.

Genes at different loci may also interact to produce discontinuous variation, as in epistasis (see page 8).

**Continuous variation**

Two of the typical effects of the inheritance of continuous variation, namely the small effects of the different alleles of one gene on a phenotypic character, and the additive effect of different genes on the same character, may be seen in a hypothetical example of the inheritance of an organism’s height.

Suppose that the height of an organism is controlled by two, unlinked (that is, on different chromosomes) gene loci, $A/a$ and $B/b$, and that the recessive alleles of both loci ($a$ and $b$) each contribute $x$ cm to the height of the organism, whereas the dominant alleles ($A$ and $B$) each increase the height by $2x$ cm.

If the effect of each gene is additive, the homozygote recessive, $aabb$, therefore is potentially $4x$ cm tall and the homozygote dominant $AABB$ is potentially $8x$ cm tall. The other genotypes will fall between these extremes.

The inheritance of resistance to warfarin in rats is controlled at a single gene locus.

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
<th>Vitamin K requirement</th>
</tr>
</thead>
<tbody>
<tr>
<td>$Rw^R$</td>
<td>resistant</td>
<td>high</td>
</tr>
<tr>
<td>$Rw^S$</td>
<td>susceptible</td>
<td>normal</td>
</tr>
<tr>
<td>$Rw^R$</td>
<td>resistant</td>
<td>slightly increased</td>
</tr>
</tbody>
</table>

Three genotypes are possible, each giving a different phenotype as shown in **Table 1.1**.

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Resistance to Warfarin</th>
<th>Vitamin K Requirement</th>
</tr>
</thead>
<tbody>
<tr>
<td>$Rw^R$</td>
<td>susceptible</td>
<td>normal</td>
</tr>
<tr>
<td>$Rw^S$</td>
<td>resistant</td>
<td>slightly increased</td>
</tr>
<tr>
<td>$Rw^R$</td>
<td>resistant</td>
<td>high</td>
</tr>
</tbody>
</table>
6 Variation

Suppose now that the homozygotes, \textit{aabb} and \textit{AABB}, are interbred.

<table>
<thead>
<tr>
<th>Parental phenotypes</th>
<th>4x cm tall</th>
<th>8x cm tall</th>
</tr>
</thead>
<tbody>
<tr>
<td>Parental genotypes</td>
<td>\textit{aabb}</td>
<td>\textit{AABB}</td>
</tr>
<tr>
<td>Gametes</td>
<td>\textit{ab}</td>
<td>\textit{AB}</td>
</tr>
<tr>
<td>\textit{F}_1 \textit{genotypes}</td>
<td>\textit{AaBb}</td>
<td>all 6x cm tall</td>
</tr>
</tbody>
</table>

Interbreeding the \textit{F}_1 generation (see Box) gives all possible genotypes amongst the 16 possibilities:

![Gametes Table]

The number of offspring and their potential heights according to their genotypes are summarised in figure 1.3. These results fall approximately on a normal distribution curve.

<table>
<thead>
<tr>
<th>Potential height from genotype</th>
<th>4x cm</th>
<th>5x cm</th>
<th>6x cm</th>
<th>7x cm</th>
<th>8x cm</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of offspring with that genotype/16</td>
<td>1</td>
<td>4</td>
<td>6</td>
<td>4</td>
<td>1</td>
</tr>
</tbody>
</table>

\[\text{Figure 1.3} \text{ The additive effect of alleles.}\]
These imaginary results come from assuming that two gene loci on different chromosomes contribute to the height of the organism. Think about what will happen to a quantitative character if more gene loci, each with an additive effect, are involved (polygenes). Suppose that all the genes affecting height are on different chromosomes: the number of discrete height classes increases as more genes are involved, and the difference between these classes gets less.

Even if two or more of the loci are linked on the same chromosome, potentially reducing the number of classes of offspring and increasing the difference between them, crossing over in prophase I of meiosis will restore the variation. The differences between different classes will be further smoothed out by environmental effects, as discussed in the next section.

Both genotype and environment contribute to phenotypic variance

In the imaginary example of continuous variation just given, the heights shown are those expected from the genotype alone. If you were able to take a number of individuals, all with the same genotypic contribution to height, it would be most unlikely that their height would be exactly the same when measured. Environmental effects may allow the full genetic potential height to be reached or may stunt it in some way.

One individual animal might have less food, or less nutritious food, than another with the same genetic contribution. A plant may be in a lower light intensity or in soil with fewer nutrients than another with the same genetic potential height. Other examples of the effect of environment include the development of dark tips to ears, nose, paws and tail in the Himalayan colouring of rabbits and in Siamese cats. This colouring is caused by an allele which allows the formation of the dark pigment only at low temperature. The extremities are the coldest parts of the animals, so the colour is produced there.

When the number of gene loci controlling a quantitative character is large, it is not possible to identify them and assess the individual effects of their various alleles.

In selective breeding (chapter 2), it is important to know how much of the phenotypic variation is genetic, and how much is environmental in origin. There is no point in selecting parents for a breeding programme on the basis of environmental variation!

The genetic and environmental contributions to phenotypic variation are written simply:

\[ V_P = V_G + V_E \]

where \( V_P \) is the phenotypic variation, \( V_G \) is the genetic component, and \( V_E \) is the environmental component of the variation. The proportion of the phenotypic variation that is genetic is referred to as the heritability of the character. Successful selective breeding requires a high heritability (see chapter 2).

In a classic experiment the American geneticists Ralph Emerson and Edward East crossed two varieties of maize which differed markedly in cob length. Both of the parental varieties (Black Mexican and Tom Thumb) were pure-bred lines. The cob lengths of the plants used as parents and the first and second generations of offspring resulting from the cross were measured to the nearest centimetre. The number of cobs in each length category was counted. The results are shown in table 1.2.

<table>
<thead>
<tr>
<th>Cob length (cm)</th>
<th>5</th>
<th>6</th>
<th>7</th>
<th>8</th>
<th>9</th>
<th>10</th>
<th>11</th>
<th>12</th>
<th>13</th>
<th>14</th>
<th>15</th>
<th>16</th>
<th>17</th>
<th>18</th>
<th>19</th>
<th>20</th>
<th>21</th>
</tr>
</thead>
<tbody>
<tr>
<td>Black Mexican parents</td>
<td>3</td>
<td>11</td>
<td>12</td>
<td>14</td>
<td>26</td>
<td>15</td>
<td>10</td>
<td>7</td>
<td>2</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Tom Thumb parents</td>
<td>4</td>
<td>21</td>
<td>24</td>
<td>8</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Offspring 1</td>
<td>1</td>
<td>12</td>
<td>12</td>
<td>14</td>
<td>17</td>
<td>9</td>
<td>4</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Offspring 2</td>
<td>1</td>
<td>10</td>
<td>19</td>
<td>26</td>
<td>47</td>
<td>73</td>
<td>68</td>
<td>68</td>
<td>39</td>
<td>25</td>
<td>15</td>
<td>9</td>
<td>1</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

- Table 1.2 Variation in cob length of two parental varieties of maize and of the first and second generations of a cross between them.
Both parental varieties were pure-bred and therefore homozygous at a large number of loci. The first generation of offspring were genetically different from the parents, but were genetically the same as one another. The phenotypic variation that we see within the two parental lines and within the first generation is, therefore, environmental.

The average variance of the two parental varieties and of the first generation of offspring provides a measure of the environmental variance ($V_E$).

The second generation of offspring shows a much wider range of variation in cob length. This is both genetic and environmental.

**SAQ 1.4**

a From table 1.2, calculate the variance in cob length ($V_P$) shown by the Black Mexican parental variety and by the first generation of offspring. The variance of the Tom Thumb parental variety has already been calculated in box 1A.

b Calculate the environmental variance, $V_E$.

c Calculate the phenotypic variance ($V_P$) of the second generation of offspring, and from this, and your answer to b, find the genetic component of the variance ($V_G$).

---

### Interactions at one locus and between loci

The interactions between alleles at the same locus, of codominant alleles (as in flower colour in *Antirrhinum*), dominant and recessive alleles (as in tomato plant stem colour) and multiple alleles (as in the inheritance of ABO blood groups in humans), have already been discussed on page 4.

However, there are cases where different loci interact to affect one phenotypic character. When one gene locus (the epistatic gene) affects or inhibits the effect of another locus (the hypostatic gene), this is known as epistasis.

### Epistasis

In the inheritance of feather colour in chickens there is an interaction between two gene loci, $I$ and $C$. Individuals carrying the dominant allele, $I$, have white feathers even if they also carry the dominant allele, $C$, for coloured feathers. Birds that are homozygous recessive, $cc$, are also white. This is an example of dominant epistasis.

**SAQ 1.5**

List the genotypes that will result in coloured feathers.

---

**Figure 1.4**

a White Leghorn and b white Wyandotte chickens.

White Leghorn chickens (figure 1.4) have the genotype $IICC$, whilst white Wyandotte chickens have the genotype $iicc$. A white Leghorn is crossed with a white Wyandotte.
Now the F1 offspring are interbred to give an F2 generation.

<table>
<thead>
<tr>
<th>Parental genotypes</th>
<th>IiCc</th>
<th>IiCc</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gametes</td>
<td>IC or IC or IC or IC</td>
<td>IC or IC or IC or IC</td>
</tr>
<tr>
<td>Gametes from one parent</td>
<td>IC or IC or IC</td>
<td>IC or IC or IC</td>
</tr>
<tr>
<td>Gametes from the other parent</td>
<td>IC or IC or IC</td>
<td>IC or IC or IC</td>
</tr>
</tbody>
</table>

The usual Mendelian 9:3:3:1 ratio expected in the F2 generation has been modified by epistasis to (9+3+1):3 = 13 white : 3 coloured.

A different type of epistasis, recessive epistasis, is shown by the inheritance of flower colour in Salvia. A pure-breeding, pink-flowered variety of Salvia was crossed with a pure-breeding, white-flowered variety. The F1 generation had purple flowers. Interbreeding the F1 to give an F2 generation resulted in purple, pink and white-flowered plants, in a ratio of 9:3:4. Two loci on different chromosomes, A/a and B/b, are involved:

The homoyzgote recessive aa is epistatic to the B/b locus (the hypostatic gene). Neither the dominant allele, B, for purple flower colour, nor the recessive allele, b, for pink flower colour can be expressed in the absence of a dominant A allele.

**SAQ 1.6**

Draw a genetic diagram of the Salvia cross described above to show the 9:3:4 ratio in the F2 generation.

A different modification of the F2 dihybrid ratio involves complementary genes, where enzymes coded for by two genes act sequentially in a metabolic pathway. Only if a dominant allele of the gene for the first enzyme to act is present will a suitable substrate be formed to be further acted upon by the enzyme coded for by a dominant allele of the second locus. For example, a cross between two pure-breeding varieties of white-flowered sweet pea gave offspring which all had purple flowers. Interbreeding these gave plants with purple or with white flowers in a 9:7 ratio = 9:(3+3+1). Two gene loci, A/a and B/b, on different chromosomes, are involved, and pigment production depends on their combined action. The dominant alleles, A and B, each code for an enzyme in the metabolic pathway of pigment production (figure 1.5). The recessive alleles do not code for functioning enzymes. Purple pigment can be produced only when both dominant alleles are present in the genotype.
variation

10  Variation

Figure 1.5 Pigment production pathway of flower colour in sweet pea.

SAQ 1.7

a State the genotypes of the two pure-breeding varieties of white-flowered sweet pea used in the cross on page 9.
b Draw a genetic diagram of the cross to show the 9:7 ratio of the F2 generation.

Epistasis is not inherited. It results from the interaction of the gene loci of a particular genotype. Look back to the section on continuous variation. The different alleles had an additive effect on height. If we were to introduce epistasis as well as dominance, the number of possible phenotypes would be much reduced. Dominance and epistasis reduce phenotypic variation.

Linkage

The number of phenotypic classes resulting from a cross is also reduced by the phenomenon known as linkage.

When two or more gene loci are on the same chromosome, they do not assort independently in meiosis as they would if they were on different chromosomes. The loci are said to be linked.

Drosophila normally has a striped body, and antennae ending in a much branched spike (figure 1.6). The gene for body colour and the gene for antennal shape are close together on the same chromosome (chromosome 3) and so are linked.

A black body, with no stripes, results from a recessive allele called ebony. A recessive allele for antennal shape, aristopedia, gives an antenna looking rather like a Drosophila leg, with two claws on the end (figure 1.7). A homozygous fly with a striped body and normal antennae was crossed with a homozygous ebony-bodied fly with aristopedia antennae. All the offspring had striped bodies and normal antennae. The male F1 flies were then crossed with the parental type homozygous for ebony body and aristopedia antennae. This kind of cross, between the F1 and the double recessive parental type, is known as a test cross and allows us to work out the genotype of the F1. In this case the test cross produced the two original parental types in equal numbers in the offspring. This can be explained by the diagram on page 11.

G Figure 1.5 Pigment production pathway of flower colour in sweet pea.

G Figure 1.6 Drosophila melanogaster (x 20).

G Figure 1.7 Normal and aristopedia Drosophila antennae.