General Genetic

A gene is a heritable factor that controls the expression of some trait, which may be morphological, behavioural, molecular, etc. Each such gene occupies a specific physical locus (pl. loci) on a particular chromosome. Alternative forms of these loci are termed alleles. Gene, locus, and allele are often used more or less interchangeably, and this can lead to confusion.

Gene is the popular and most general term, and is most appropriate when the inherited basis of a trait is emphasized, e.g., a “gene” for eye colour. Locus is most appropriate when the physical nature or position of a gene, especially with respect to other genes, is emphasized, as for example in gene mapping and linkage studies. Allele is most appropriate when the particular form(s) of a gene found in any particular individual or chromosome is (are) emphasized: e.g., there are “brown” and “blue” alleles of the eye colour gene. It is therefore inaccurate to say, for Example, “He has the gene for sickle-cell anaemia,” and more accurate to say “He has two HbS alleles at the beta-globin locus on Chromosome 6.” We all have the “gene” for every genetic condition; some of us have the particular allele(s) that result in the condition being expressed.

Drosophila Genetics

*Drosophila* are diploid, with two sets of chromosomes and therefore two alleles at each autosomal locus. If both alleles are identical, the individual is a homozygote and is described as homozygous. If the alleles differ from each other, the individual is a heterozygote and is described as heterozygous. If the gene occurs on a sex chromosome, females may be either homozygous or heterozygous, but a male fly with only one allele at a locus will be a hemizygote and would be described as hemizygous.

*Drosophila* of typical appearance are said to show the “wild-type” forms (phenotypes) of genetically-controlled traits for body colour, eye colour, wing shape, etc. Naturally-occurring or artificially-induced genetic variants (mutations) of the alleles that control these traits produce flies with different morphologies, according to the dominant or recessive nature of the alleles involved in the genotype. Such mutant alleles are designated by symbols that are typically abbreviations of the mutant name. For example, the typical body colour phenotype is grey. One mutant produces an ebony (shiny black) body colour. Because this allele is recessive, it is symbolized by a lower-case letter, e. Dominate traits are symbolised by capital letters e.g. E.

Genetic Crosses

An “X” is used to indicate that two individuals have been mated together. The parents are designated as P (for parental) and the offspring as F (for filial). When several generations are involved, subscripts are added to designate the generations.

P₁ give rise to F₁ (first filial) progeny. If the F₁ are crossed together they become P₂ and their progeny F₂.

A cross between members of the F₁ and members of the P₁ is a backcross.

A cross between members of the F₁ and the true breeding recessive P₁ is a test cross.
**Monohybrid Cross**

The simplest form of a cross is a **monohybrid cross**, which analyses a single trait and its associated variations. The diagram below shows the progression of a pair of alternative alleles for a single gene through two generations.

P1 - homozygous parents

All offspring (F₁) are heterozygous (Ee) and possess a wild-type body.

F1 – heterozygous parents

Phenotype ratio 3 wild-type body : 1 ebony body
Genotype ratio                 1 EE: 2 Ee: 1 ee

During gamete formation, the members of a pair of alleles are duplicated and then segregated from one cell into four separate gametes so that each contains only one member of the pair (Law of Segregation).

In cases such as the above example, the F2 phenotype ratio of 3:1 indicates a case of complete dominance. That is, one allele completely masks the expression of the other (recessive) allele.

In cases of incomplete dominance, neither allele masks the other, and heterozygous individuals express new phenotypes that are intermediate between the homozygous parents. This may arise for example if the dominant homozygous phenotype results from the expression of a double-dose of gene product, and the heterozygous phenotype from a single dose. The F2 phenotype ratio of 1:2:1 is characteristic. A non-Drosophila example of this is seen in red- and white-flowered snap dragons:

P1

All offspring (F1) are heterozygous Rr and have a pink flower, an intermediate between red and white.

When both alleles are expressed the effect is known as co-dominance. Heterozygous individuals express gene products from both alleles: unlike incomplete dominance, the phenotype is not an intermediate. This sort of interaction is seen in the ABO blood group system of humans. One allele controls the production of A antigen while the other controls the B antigen (a third allele O produces no antigen). Heterozygotes carrying the allele for
antigen A and the allele for antigen B have blood type AB in which both proteins are present in equal quantities. The F2 shows a ratio of 1:2:1, as in the case of incomplete dominance. shows a ratio of 1:2:1, as in the case of incomplete dominance.

Dihybrid Cross
Dihybrid crosses involve manipulation and analysis of two traits controlled by pairs of alleles at different loci. For example, in the cross body colour x wing shape:

\[ e \text{ is ebony body colour} \]
\[ E \text{ is wild-type body colour} \]
\[ v \text{ is vestigial wing shape} \]
\[ V \text{ is wild-type wing shape} \]

where the loci for ebony body colour and vestigial wing are on separate autosomes. Therefore the genotypes and gametes are the same for male and female.

CROSS DIAGRAM Autosomal Independent

P1: true breeding ebony fly with vestigial wings is mated with a true breeding wild-type body and wings. The genotype of the parents will be ee vv and EE VV. If these flies are mated all their offspring will be heterozygous for each gene (EeVv) and possess a wild-type body and wings. However, if the offspring from the F1 generation are crossed the phenotype ratio is 9:3:3:1.
In a dihybrid cross, each of the F1 parents can produce four different gamete types, so there are 16 (= $4 \times 4$) possible offspring combinations. Because the two traits show complete dominance and separate independently of each other (Law of Independent Assortment), the expected genotypic and phenotypic ratios from an analysis of these 16 possibilities can be calculated.

<table>
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<th>Phenotype</th>
<th>Genotype</th>
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These ratios can be derived from the results of a monohybrid ratio. A basic principle of probability theory is that the probability of two independent events occurring together is equal to the product of the two independent probabilities. For example, the expected proportions of flies with wild-type and ebony body colours in a monohybrid cross are $3/4$ and $1/4$, respectively. Likewise, in a monohybrid cross involving vestigial wings, the proportions are $3/4$ wild-type and $1/4$ vestigial-winged. In a dihybrid cross, the proportions of flies with various combinations of both characters can be calculated as:

- wild-type & wild-type = $3/4 \times 3/4 = 9/16$
- ebony = $1/4 \times 3/4 = 3/16$
- wild & vestigial = $3/4 \times 1/4 = 3/16$
- ebony & vestigial = $1/4 \times 1/4 = 1/16$

This produces the familiar 9:3:3:1 ratio.

**Autosomal Linkage**

Mendel’s work on peas was done before the discovery of chromosomes, and his Law of Independent Assortment postulated that each trait would segregate independently of every other. We know now that loci are arranged in linear fashion on chromosomes, and that loci that are physically close to each other will not segregate completely independently of each other. This phenomenon is called genetic linkage. Linkage may be complete (loci are so close that crossing over rarely if ever occurs between them, and only the parental type gametes are produced) or incomplete (where crossing over occurs between the two loci and produces some recombinant type gametes).

The chance that a cross-over will occur between the loci depends on the genetic distance between them. Loci located far enough apart on the same chromosome act as though they are unlinked and produce equal proportions of parental and recombinant gametes.

When the loci in a dihybrid cross are linked, it is necessary to indicate clearly the specific allelic combinations that are present on the two chromosomes in each of the parents, because these alleles will tend to stay together and not assort independently. In the case of a double heterozygote, a chromosome in which the two linked loci show alternately the recessive and dominant alleles is called the *trans* (repulsion) arrangement (a-b/ab-). A chromosome in which the two linked loci show either both recessive or both dominant alleles is called the *cis* (coupling) arrangement (a-b/-ab). The phenotypic expressions of *cis* and *trans* arrangements of heterozygous dihybrids are typically identical, but will produce different arrangements of alleles in their respective offspring.
Example of *Cis* arrangement

\[ P_1 \quad EEVV \text{ (wild type)} \times eevv \text{ (ebony body and vestigial wings)} \]

\[ F_1 \quad EeVv \quad (\text{all wild-type, } c\text{i} \text{s arrangement}) \]

\[ \begin{array}{c}
\text{E} \\
\text{e} \\
\text{V} \\
\text{v} \\
\end{array} \]

Gametes that are produced are EV and ev

\[ F_2 \quad 1 \text{ EEVV} : 2 \text{ EeVv} : 1 \text{ eevv} \]
3 wild-type : 1 ebony vestigial

Example of *Trans* arrangement

\[ P_1 \quad EEvv \text{ (normal body & vestigial wing)} \times eeVV \text{ (ebony body and normal wing)} \]

Gametes produced Ev and eV

\[ F_1 \quad EeVv \quad (\text{all wild-type, } t\text{ran} \text{s arrangement}) \]

\[ \begin{array}{c}
\text{E} \\
\text{e} \\
\text{v} \\
\text{V} \\
\end{array} \]

Gametes that are produced are Ev and eV

\[ F_2 \quad 1 \text{ EEvv} : 2 \text{ EeVv} : 1 \text{ eeVV} \]
1 vestigial wing normal body : 2 wild-type : 1 ebony body normal wing

The simplest mechanism for assessing linkage is a *test cross* (a mating in which one of the individuals is homozygous recessive for all traits considered). A non-linked dihybrid test cross will give a 1:1:1:1 ratio.

Reference
http://www.mun.ca/biology/dinnes/B2250/dros_intro.pdf