



Genetics made simple: I

It is widely recognised that individuals of the same species have similar characteristic features. This similarity is called *heredity*. However, individuals within the species will also show minor differences between each other. This is known as *variation*. Genetics is the study of how characteristics such as plant height or animal eye colour, are passed on from one generation to another (inherited) and how variation occurs.

DNA & chromosomes

The hereditary material, *i.e.* the material which carries the information to produce the specific characteristics of an organism or species is contained within the nucleus and is made up of the organic molecule **DNA** (deoxyribonucleic acid). DNA is an extremely long molecule consisting of two strands of nucleotides (see Factsheet 22 - Protein Synthesis) wound around each other into a double helix.

In the nucleus of eukaryotic cells DNA forms **chromosomes**. In chromosomes, long, tightly coiled strands of DNA are wound around globular proteins called **histones**. This arrangement acts as an efficient way of packaging the DNA molecule. Other proteins present in the chromosome are involved in regulating DNA activity.

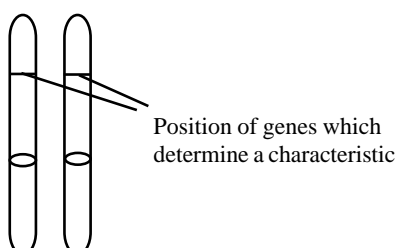
In humans, there are forty-six chromosomes in most cells of the body. This number is made up of twenty-two pairs of non-sex determining chromosomes (**autosomes**) and one pair of chromosomes which determine the individual's gender (**sex chromosomes**).

In most plants and animals the cells which make up the body are **diploid**, *i.e.* they contain a complete set of paired chromosomes. In humans, the diploid number is **46**, since most cells contain **23 pairs** of chromosomes. Gamete cells produced for sexual reproduction, *eg.* ovum and sperm cells, are **haploid**, *i.e.* they contain **half** the normal number - 23 single chromosomes.

During fertilisation, the combination of two haploid gametes produces a diploid cell (a zygote). This zygote will develop into an individual containing diploid cells. The production of haploid gametes for sexual reproduction has three important consequences:

1. The diploid number of a species stays the same in each generation. If gametes had the same number of chromosomes as body cells, the diploid number would double in each generation.
2. In the zygote the unpaired chromosomes from the two gametes pair up. This means that diploid cells contain pairs of chromosomes, with one member of each pair of chromosomes donated by the male parent (paternal) and the other member donated by the female parent (maternal). The pairs of chromosomes are known as homologous (*same-shape*) pairs, as they are similar in appearance (Fig 1).

Fig 1. Homologous pair of chromosomes

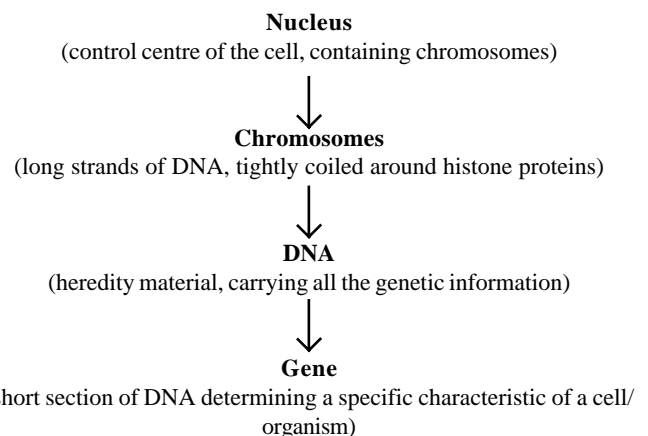


3. All offspring from sexual reproduction have approximately 50% of their heredity material from their mother and 50% from their father.

Genes

Thus, DNA is the heredity material, and carries all of the information for the characteristics of an individual. **Genes** are short sections of DNA (in other words, a sequence of nucleotides) which determine specific characteristics in cells and organisms. Fig 2 summarises the structural relationship between genes, DNA and chromosomes.

Fig 2. A summary of the structural relationship between genes, DNA and chromosomes.



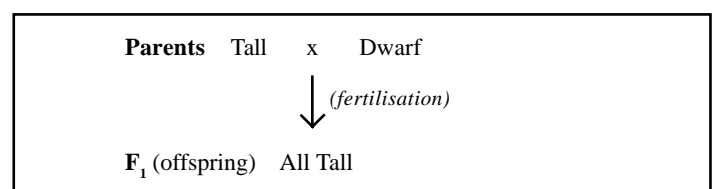
Mendelian Genetics

Gregor Mendel (1822-84) was an Austrian monk and teacher, who studied the inheritance of distinct characteristics, using **pure-breeding** individuals of the garden pea (*Pisum sativum*). These are individuals that, when bred with each other, produce the same characteristics over many generations. For example, pure-breeding parents with a tall trait, would produce all tall offspring.

Monohybrid inheritance

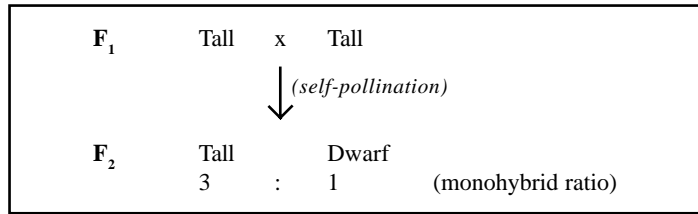
Mono = 1, so monohybrid inheritance is the inheritance of a **single** characteristic, such as height **or** shape of seed. Fig 3 shows the cross between two pure-breeding parents, one tall and one dwarf.

Fig 3. A monohybrid cross



The first generation is called the F_1 (first filial) generation. Mendel found that all the F_1 generation produced were tall. Individuals from the F_1 generation were then crossed with each other i.e. self-pollinated.

Fig 4. Self pollination of the F_1 generation



Mendel made the following deductions from these crosses:

- F_1 plants did not show characteristic **intermediate** between the parental characteristics, i.e. medium height. This showed that there was no **blending** or mixing of traits for a characteristic.
- Only one of the traits of the characteristic appeared in the F_1 , i.e. tall. This trait was **dominant** over the other, which was **recessive**.
- Since the recessive trait (dwarf) **reappeared** in the F_2 , the F_1 plants must contain one factor for tallness and one for dwarfness.

From these results, Mendel concluded that characteristics were passed from one generation to the next via gametes, and that parents must possess two pieces of information for each characteristic. This was the basis of Mendel's first law, the **Law of Segregation**:

The characteristics of an organism are determined by internal factors which occur in pairs. Only one of the pair of such factors can be represented in a single gamete.

Since Mendel devised this law, these 'factors' have been identified as the portions of the chromosome called **alleles**.

Genes and alleles

Mendel's experiments showed that, for any characteristic, there are alternative genes which produce different traits of that characteristic, eg. the trait for height can be either tall or dwarf. The name **allele** is given to the alternative forms of a gene. Every individual receives one allele from each parent, so that for most characteristics they have two alleles; one on each chromosome in a homologous pair.

The specific site at which an allele is found on a chromosome is called the **locus**. If the two alleles for a characteristic on a homologous pair of chromosomes are identical, e.g. they both produce a tall trait, they are said to be **homozygous**. When the two alleles are different, eg. one produces tall and one produces dwarf, they are **heterozygous**.

Genetic crosses

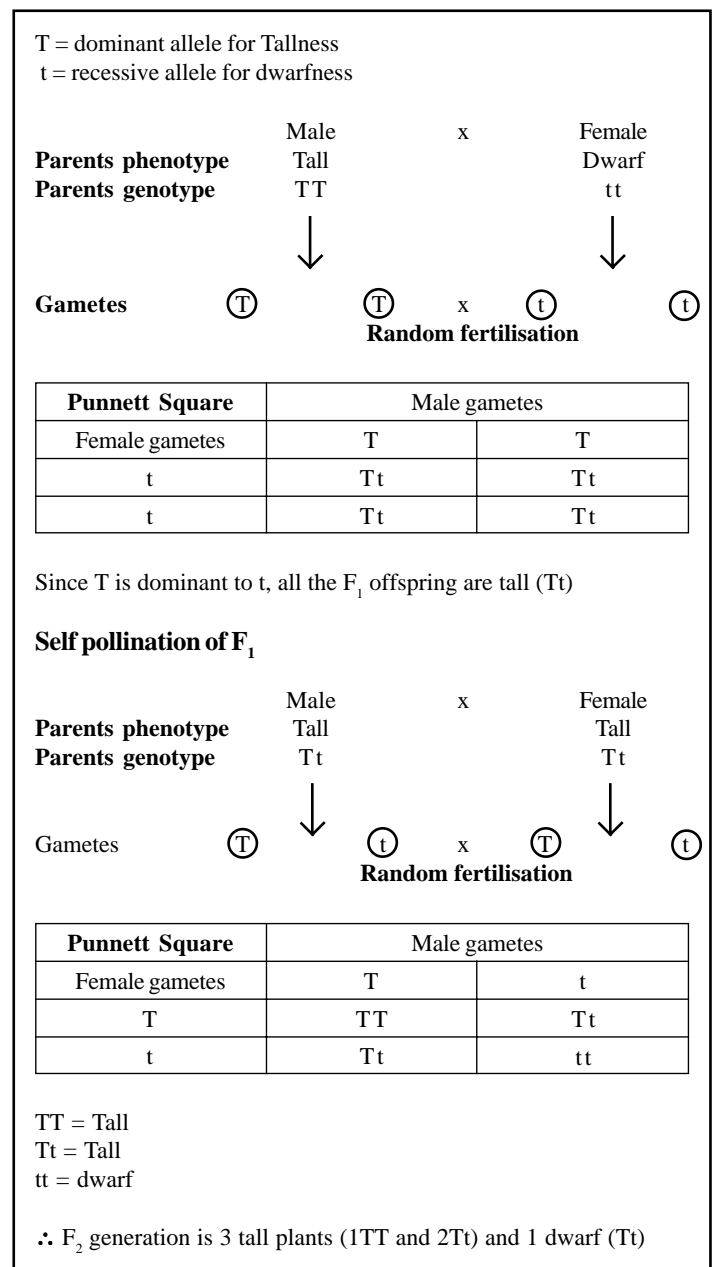
The inheritance of characteristics is represented using genetic diagrams. The following rules should be closely followed when producing these diagrams:

1. A single letter is used for each characteristic (generally the first letter of one of the traits).
2. A capital letter is used to represent the allele for the dominant trait and the lower case of the same letter to represent the recessive trait, eg. **T** for tall and **t** for dwarf (**always state clearly which trait each symbol represents**).

3. Parents are represented by an appropriate pair of letters for the pair of alleles in the diploid cells. This is the parents **genotype**. For example, a tall individual can be either TT, or Tt. A dwarf individual must be tt, because an individual with the recessive phenotype can only have recessive alleles.
4. Gametes are represented with only one letter, since gametes only possess one of the parental homologous chromosomes (since they are haploid). Genotype TT produces gametes which are all T; tt produces gametes all t; and Tt produces equal amounts of T gametes and t gametes.
5. All the possible F_1 genotypes are calculated using a system called a Punnett square (see the genetic diagram).
6. The **phenotype** (physical appearance due to the genotype) of each offspring, showing the number of each type is stated at the end of each diagram.

Fig 5 shows the genetic diagram for Mendel's cross in Fig 4 and Fig 5.

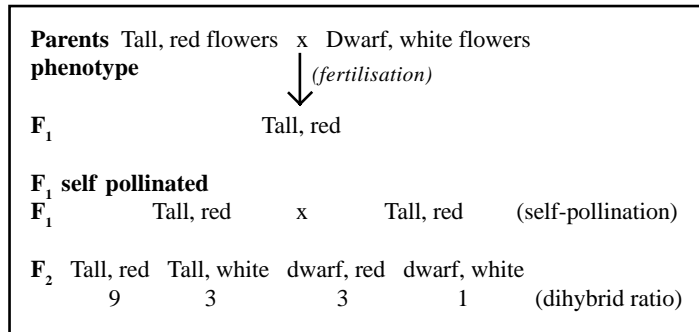
Fig 5.



Dihybrid crosses

A dihybrid cross is the inheritance of **two** characteristics. Mendel examined crosses between parents pure breeding for two characteristics, for example, height and flower colour (Fig 6). In the F₂, four phenotypes resulted in the ratio of 9:3:3:1.

Fig 6. A dihybrid cross

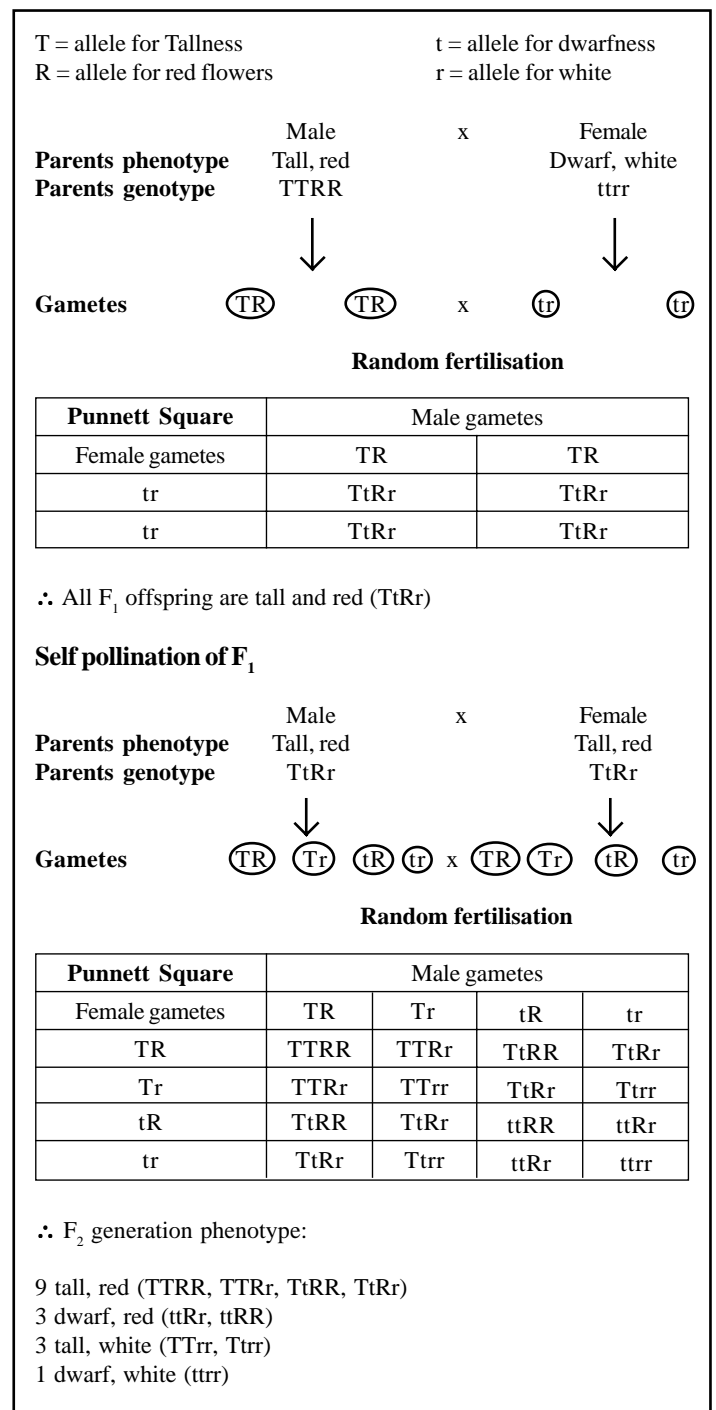


Mendel made the following deductions from these crosses:

- Since the F₁ generation are all tall and red, tall must be dominant over dwarf and red must be dominant over white.
- The factors (genes) for height and colour can be separated, because two **new combinations** of the traits appear; tall with white; and dwarf with red.
- In the F₂ generation, the ratios of tall:dwarf and red:white are both 3:1. Therefore, the factors (alleles) behave independently of each other.

These results were the basis of Mendel's second law, the **Law of Independent Assortment: Either of the pair of alleles may be combined in a gamete with either of another pair.** In other words, in a heterozygous individual (TtRr), either of the alleles for height can combine in a gamete with either of the alleles for flower colour. Fig 7 shows the genetic diagram for this cross:

Fig 7.



Exam hint - When the gametes are produced in dihybrid individuals, the separation of homologous pairs of chromosomes means that every possible genotype of gametes must be represented.

For example, a TTRR (homozygous) individual will produce gametes which are all TR. A TtRr (heterozygous) individual will produce equal amounts of the gametes: TR, Tr, tR, tr, i.e. each T/t goes with each R/r



Glossary

- Alleles** - Alternative forms of a particular gene
- Characteristics** - Specific physical features of cell or chromosome
- Chromosome** - A long strand of coiled DNA and associated proteins.
- Diploid** - Cells containing a complete set of paired homologous chromosomes.
- DNA (Deoxyribonucleic acid)**. Organic molecule containing the heredity information for organisms.
- Eukaryote** - A cell that possess a membrane-enclosed nucleus eg. plant or animal cells.

- Gametes** - Cells involved in sexual reproduction, eg. sperm cell.
- Genes** - A section of DNA (a sequence of nucleotides) that produces a particular characteristic in a cell or organism.
- Genotype** - Genetic composition of an individual
- Haploid** - Cells containing only one set of parental chromosomes
- Heterozygous** - Having two different alleles for a particular characteristic.
- Homozygous** - Having two identical alleles for a particular characteristic.
- Phenotype** - The physical appearance or outward expression of the genotype in an individual.
- Pure-breeding** - Individuals which are homozygous for a particular characteristic.

