



# **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.

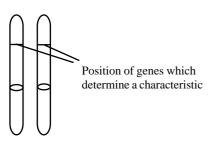
In humans, there are forty-six chromosomes in most cells of the body. This number is made up of twenty-two pairs of non-sex determining chromosomes (**autosomes**) and one pair of chromosomes which determine the individual's gender (**sex chromosomes**).

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. The 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

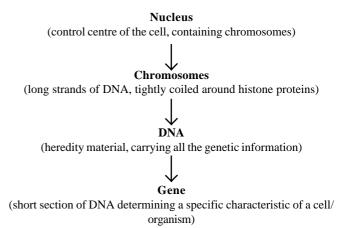


3. All offspring from sexual reproduction have approximately 50% of their heredity material from their mother and 50% from their father.

#### 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.



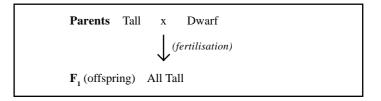
#### Mendelian Genetics

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

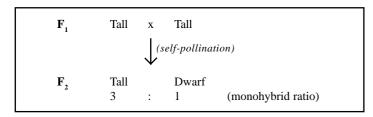
Mono = 1, so monohybrid inheritance is the inheritance of a **single** characteristic, such as height **or** shape of seed. Fig 3 shows the cross between two pure-breeding parents, one tall and one dwarf.

#### 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<sub>1</sub> 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<sub>1</sub>, *i.e.* tall. This trait was **dominant** over the other, which was **recessive**.
- Since the recessive trait (dwarf) **reappeared** in the F<sub>2</sub>, the F<sub>1</sub> 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

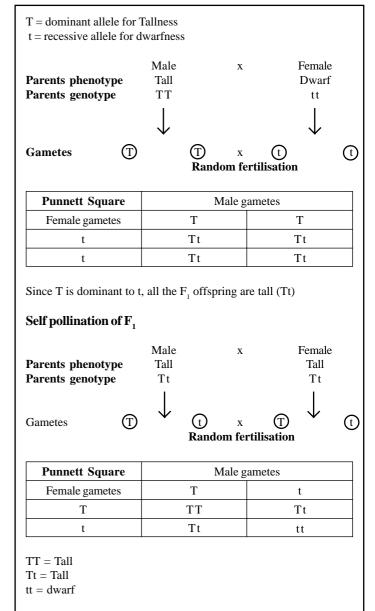
The inheritance of characteristics is represented using genetic diagrams. The following rules should be closely followed when producing these diagrams:

- 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.
- All the possible F<sub>1</sub> 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.

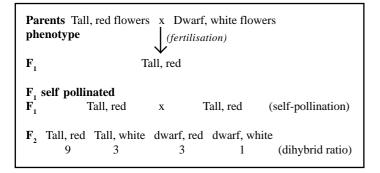


: F<sub>2</sub> generation is 3 tall plants (1TT and 2Tt) and 1 dwarf (Tt)

# **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  $F_2$ , four phenotypes resulted in the ratio of 9:3:3:1.

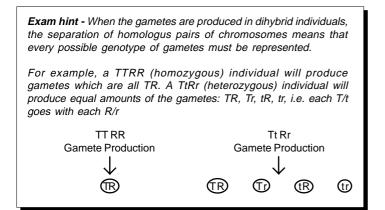
# Fig 6. A dihybrid cross



Mendel made the following deductions from these crosses:

- Since the F<sub>1</sub> 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 F<sub>2</sub> 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:



# Glossary

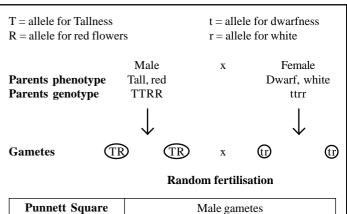
Alleles - Alternative forms of a particular gene

**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.

# Fig 7.



Punnett Square	Male gametes		
Female gametes	TR	TR	
tr	TtRr	TtRr	
tr	TtRr	TtRr	

: All F<sub>1</sub> offspring are tall and red (TtRr)

Self pollination of F<sub>1</sub>

Parents phenotype	Male e Tall, red	х	Female Tall, red
Parents genotype	TtRr		TtRr
	$\downarrow$		$\downarrow$
Gametes	(TR) (Tr) (TR)	tr x TR	(Tr) (TR) (Tr)

# **Random fertilisation**

Punnett Square	Male gametes			
Female gametes	TR	Tr	tR	tr
TR	TTRR	TTRr	TtRR	TtRr
Tr	TTRr	TTrr	TtRr	Ttrr
tR	TtRR	TtRr	ttRR	ttRr
tr	TtRr	Ttrr	ttRr	ttrr

 $\therefore$  F<sub>2</sub> generation phenotype:

9 tall, red (TTRR, TTRr, TtRR, TtRr)3 dwarf, red (ttRr, ttRR)3 tall, white (TTrr, Ttrr)1 dwarf, white (ttrr)

**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.

## **Practice questions**

1. Define each of the following terms:

(a)	
(i) gene	(3 marks)
(ii) genotype	(1 mark)
(iii) phenotype	(2 marks)

In many species of butterfly, the genotype of female is XY (heterogametic) and the genotype of male is XX (homogametic).

(b) Using a genetic diagram, show how reproduction in these species leads to approximately equal numbers of males and females in the next generation. (3 marks)

(c) Typical European Swallow-tail butterflies have yellow patches on their wings, but rare varieties have these areas shaded black. The allele for black patches is known to be recessive to the allele for yellow patches.

A butterfly breeder has an individual with yellow patches on the wings, but is unsure of the butterfly's genotype. Explain the simplest way by which the breeder could determine the genotype. (5 marks)

2. Some species of mice have characteristic fur colouring, with black fur on the top side of the body and tan coloured fur over the rest. The same species have either long or short ears.

The table below shows the result of interbreeding the  $F_1$  generation of such mice. The parents of the  $F_1$  generation were a pure-breeding black and tan male with long ears and a pure breeding black, short eared female.

F<sub>1</sub> interbred

	Black & tan Long ears	Black Long ears	Black & tan Short ears	Black Short ears
Totals in F <sub>2</sub>	43	15	13	5

 (a) Using suitable symbols to represent the traits, produce a genetic diagram to explain the result shown above. (5 marks)

(b) What can be deduced about the location of the genes for ear length and fur colour from the data? (2 marks)

#### Answers

Semicolons indicate marking points.

- 1. (i) sequence of nucleotides;
  - carried on a strand of DNA;
  - codes for a particular polypeptide.
  - (ii) Genetic/allelic composition of an individual/cell.

 (iii) Physical characteristics of an individual/cell; determined by genotype alone, or genotype and the environment.

<sup>(</sup>b)

PS	Male gametes		
Female gametes	Х	Х	
Х	XX	XX	
Y	XY	XY	

: Male:Female 1:1

- (c) Use Y for yellow patches allele and y for black patches; Individual of unknown genotype is either YY or Yy; Cross with individual with black patches (genotype yy); If offspring all have yellow patches, unknown genotype is YY; If offspring is 1:1, yellow patches : black patches, unknown genotype is Yy.
- 2. Ratio from table is approximately 9:3:3:1

$T = black & tan \qquad t = b \\ L = long ears \qquad l = s$						
Male x Female Parents phenotype Black & tan, long ears Black, short ears Parents genotype TTLL ttll ↓ ↓				nort ears		
Gametes	TL	X	tl			
: All $F_1$ offspring are b	: All $F_1$ offspring are black and tan and have long ears (TtLl)					
$\mathbf{F}_{1}$ generation						
F <sub>1</sub> genotype	Male TtLl					
Gametes $(T) (T) (T) (T) (T) (T) (T) (T) (T) (T) $						
Deres off Concerns	Random fertilisation					
Punnett Square	Male gametes					
Female gametes	TL	Tl	tL	tl		
TL	TTLL	TTLI	TtLL	TtLl		
Tl	TTLI	TTll	TtLl	Ttll		
tL	TtLL	TtLl	ttLL	ttLl		

 $\therefore$  F<sub>2</sub> generation phenotype:

t1

9 Black & tan, long ears (TTLL, TTLl, TtLL, TtLl) (43)

TtLl

Ttll

ttLl

ttll

- 3 Black & tan, short ears (TTll, Ttll) (13)
- 3 Black, long ears (ttLl, ttLL) (15)
- 1 Black short ears (ttll) (5)
- (b) Genes for fur colouration and genes for ear length must be carried on separate chromosomes; because new combinations of traits appear in  $F_2$  generation.

**Exam hint -** Candidates frequently lose easy marks by: (i) miscounting phenotypes (there are 16 individuals in the Punnet Square)

(ii) not telling the examiner which genotype gives which phenotype

# Acknowledgements;

This Factsheet was researched and written by Jim Sharpe

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ISSN 1351-5136