UNIT 2

# How is continuity of life maintained?

AREA OF STUDY 2

## How is inheritance explained?

### Outcome 2

Apply an understanding of genetics to describe patterns of inheritance, analyse pedigree charts, predict outcomes of genetic crosses and identify the implications of the uses of genetic screening and decision making related to inheritance.

### Key knowledge

**Genomes, genes and alleles**

- the distinction between a genome, gene and allele
- the genome as the sum total of an organism's DNA measured in the number of base pairs contained in a haploid set of chromosomes
- the role of genomic research since the Human Genome Project, with reference to the sequencing of the genes of many organisms, comparing relatedness between species, determining gene function and genomic applications for the early detection and diagnosis of human diseases.

**Chromosomes**

- the role of chromosomes as structures that package DNA, their variability in terms of size and the number of genes they carry in different organisms, the distinction between an autosome and a sex chromosome and the nature of a homologous pair of chromosomes (one maternal and one paternal) as carrying the same gene loci
- presentation of an organism's set of chromosomes as a karyotype that can be used to identify chromosome number abnormalities including Down, Klinefelter and Turner syndromes in humans.

**Genotypes and phenotypes**

- the use of symbols in the writing of the genotypes for the alleles present at a particular gene locus
- the distinction between a dominant and recessive phenotype
- the relative influences of genetic material, environmental factors and interactions of DNA with other molecules (epigenetic factors) on phenotypes
- qualitative treatment of polygenic inheritance as contributing to continuous variation in a population, illustrated by the determination of human skin colour through the genes involved in melanin production or by variation in height.

**Pedigree charts, genetic cross outcomes and genetic decision-making**

- pedigree charts and patterns of inheritance including autosomal dominant, autosomal recessive, X-linked and Y-linked traits
- the determination of genotypes and prediction of the outcomes of genetic crosses including monohybrid crosses, and monohybrid test crosses
- the inheritance of two characteristics as either independent or linked, and the biological consequence of crossing over for linked genes
- the nature and uses of genetic testing for screening of embryos and adults, and its social and ethical implications.

# Genes and DNA

The common thread weaving through all living organisms is the **DNA** (**deoxyribonucleic acid**) contained within their cells. This genetic material contains the genes that are responsible for the inherited features of organisms.

- **Gene:** unit of inheritance; composed of DNA
- **Genome:** the complete set of genes contained within an individual organism, measured in base pairs in a hapolid set of chromosomes.

Nuclear material is arranged in strands of DNA called **chromosomes**.

The DNA is composed of a double helical structure. This helix is like a rope ladder that has been twisted (Figure 2.16). Each strand of the DNA helix is made up of a series of linked subunits called **nucleotides**.

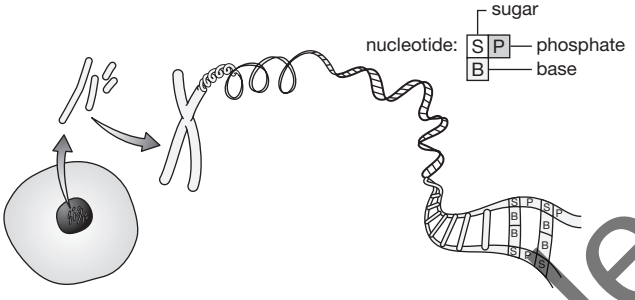



**Figure 2.16** The DNA contained in the nucleus unravels to reveal the double helix

There are four different kinds of nucleotides in DNA. Common to each is a **deoxyribose sugar** (5-carbon) molecule and a **phosphate** component. Nucleotides vary in the base unit they contain **adenine**, **thymine**, **guanine** or **cytosine**.

The vertical backbone of the DNA helix is composed of the sugar–phosphate groups. The rungs of the 'ladder' are represented by the bases. The bases occur in **complementary pairs**, with *adenine* (A) pairing with *thymine* (T) and *cytosine* (C) pairing with *guanine* (G) as in Figure 2.17.

The complementary strands in a molecule of DNA are referred to as **antiparallel** because one runs 5′→3′ while the other runs 3′→5′ (Figure 2.17).

The number of chromosomes in the **somatic cells** of organisms is characteristic of particular species. Somatic cells are body cells other than **sex cells**. Sex cells are called **gametes**. In animals, these are the ova and sperm.

Somatic cells are typically **diploid**. That is, they contain two sets of chromosomes.

**Table 2.3** Diploid numbers

| Species | Diploid number |
|---|---|
| **Humans** | 46 |
| **Chimpanzee** | 48 |
| **Cat** | 38 |
| **Blowfly** | 12 |
| **Eucalypt** | 22 |

Gametes are **haploid**. That is, they contain only one set of chromosomes—half the full set.

**Table 2.4** DNA features

| | Feature | Examples |
|---|---|---|
| **Purines** | Double ring structure | Adenine, guanine |
| **Pyrimidines** | Single ring structure | Thymine, cytosine |

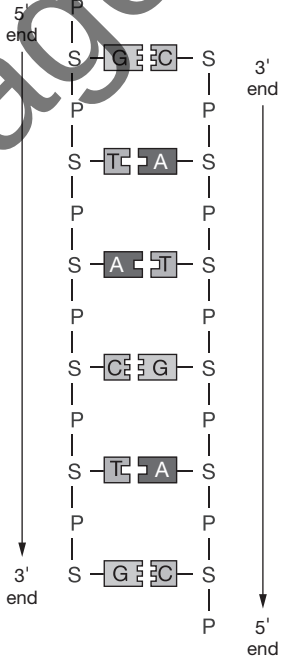



**Figure 2.17** Nucleotides arranged in complementary base pairs

# Genetic technologies

A range of technological advances in genetics allows scientists to investigate, measure and manipulate the genetic information of species. These include tools to sequence genomes, clone organisms, genetically transform or modify organisms, and diagnose and treat genetic conditions.

**DNA sequencing** is a process that is used to determine the order of nucleotide bases along a segment of DNA. Bases are 'tagged' so that each appears a different colour when viewed under fluorescent light. Chromatography is used to observe the tagged bases in a series of coloured peaks. The order of the coloured

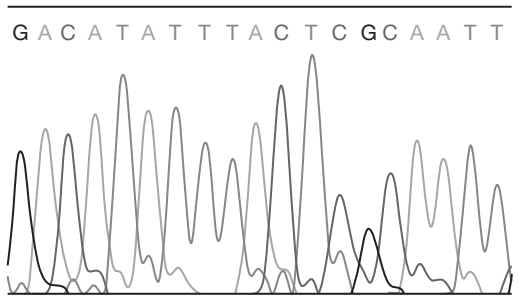



**Figure 2.18** In DNA sequencing, each peak reflects a particular nucleotide base.

peaks reflects the order of the bases in the DNA strand (Figure 2.18). Identifying the base sequence of the human genome in the Human Genome Project is a classic example of the use of this technology.

**Gene cloning** literally makes many copies of a particular gene. This technology involves the production of **transgenic** (or **recombinant**) bacteria. A foreign gene is inserted into a bacterial plasmid and the bacteria are allowed to reproduce, thereby making many copies or clones of the required gene. The cloned genes can then be delivered to the target tissue, e.g. using a disabled virus.

Gene cloning is used in the production of human insulin for the treatment of diabetes.

**Gene therapy** is a process used to treat patients with certain genetic diseases, such as cystic fibrosis. A normal gene from a healthy individual is inserted into the DNA of a vector, such as a disabled virus. The vector is then used to deliver the normal gene into the cells of the affected person. Transcription of the normal gene results.

**Cloning** technology allows the production of genetically identical individual organisms. Plant clones are easy to achieve using asexual reproduction. Animal cloning involves inserting the nucleus of a mature body cell into an emptied ovum that is ready for fertilisation. After a period of laboratory incubation, the new embryo is implanted into the uterus of an adult female where development proceeds in the usual way.

Cloning applications include the production of crops and stock with desirable characteristics, e.g. pest-resistant crops. Therapeutic cloning produces compatible tissue for transplanting in humans, e.g. to treat burns victims with new skin.

**Karyotyping** is a process of sorting chromosomes according to size (Figure 2.19). In a karyotype, chromosomes are arranged in **homologous** (similar) pairs and usually organised in order from largest to smallest. Karyotypes are used to determine gender and to diagnose chromosomal abnormalities, e.g. Down syndrome, in which there are three number 21 chromosomes instead of two (Figure 2.20).



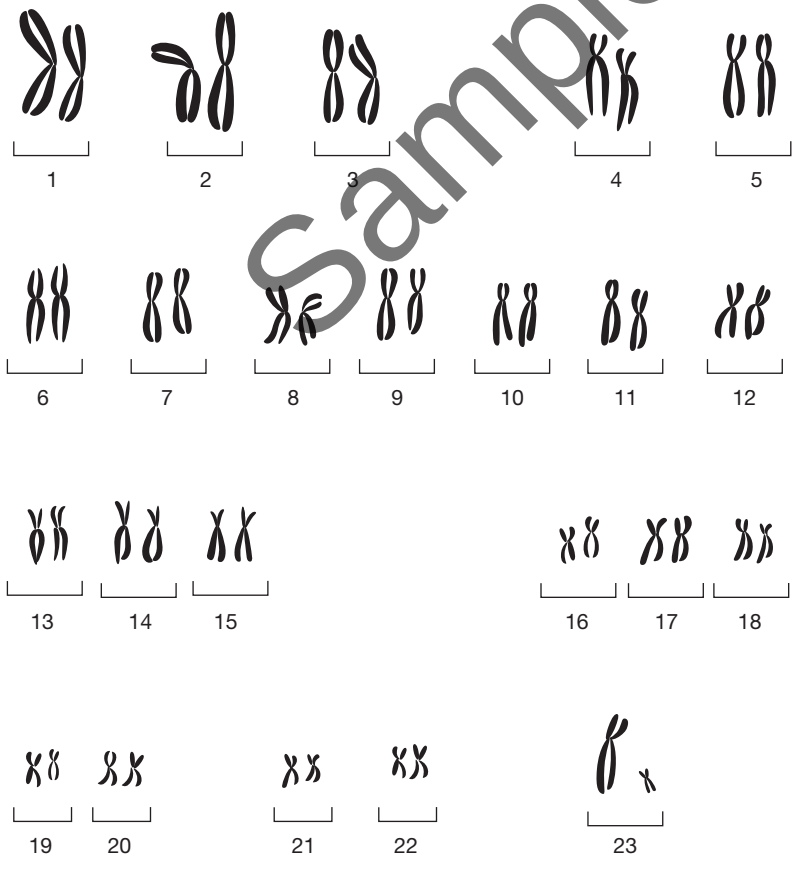



**Figure 2.19** Human karyotype

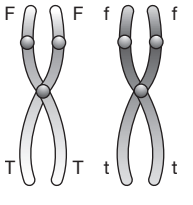



**Figure 2.20** Homologous chromosomes

**FISH** (**fluorescent in situ hybridisation**), also called chromosome painting, identifies particular entire chromosomes by applying a 'colour tag' that is specific for each homologous pair. Homologous pairs of chromosomes are easily identified according to colour using the fluorescent dyes. This technique also allows the order of homologous pairs to be established in karyotyping.

### Errors in meiosis

**Non-disjunction** occurs when chromosomes fail to separate during meiosis. This results in some gametes having two copies of a particular chromosome, and others missing that chromosome altogether.

When a chromosome with the incorrect chromosome number is involved in fertilisation, a zygote is formed that has either one too many or one too few chromosomes. This can have serious repercussions for the well-being of an individual.

**Down syndrome** is an example of **trisomy 21**, that is, the individual has three copies of chromosome 21 in each of the somatic cells. Individuals with Down syndrome have characteristic facial features, are relatively short in stature and display delayed physical development and intellectual impairment.

**Translocation Down syndrome** occurs when the number 21 chromosome in a cell is joined to the number 15 chromosome. During meiosis, gametes are formed that contain a normal number 21 chromosome as well as a chromosome 15–21, effectively two copies of chromosome 21. During fertilisation, trisomy 21 occurs.

## Chromosomes and sex determination

- **Sex chromosomes** are chromosomes that are involved in sex determination (see Figure 2.21). In humans, these are the X and Y chromosomes—XX: female; XY: male.
- **Autosomes** are chromosomes that are not involved in sex determination.
- Diploid cells in humans contain 46 chromosomes, arranged in 23 pairs.
- There are 22 homologous pairs—the autosomes—and 1 pair of sex chromosomes.
- Females are **homogametic**, that is, the sex chromosomes are homologous.
- Males are **heterogametic**, that is, the sex chromosomes are not a homologous pair.
- Unlike humans, female birds are heterogametic and males are homogametic.

## Genotype and phenotype

**Gregor Mendel** is credited with laying the foundations of our modern understanding of genetics. He carried out breeding experiments with various aspects of garden pea in a successful attempt to understand and interpret the patterns of inheritance that he observed.

- An organism's **genotype** is the combination of alleles that make up its genetic information.
- The **phenotype** of an organism is the observable expression of its genotype.
- An organism's phenotype is influenced by both its genotype and environmental factors.

**phenotype = genotype + environment**

- **Genes** are the units of heredity.
- **Alleles** are alternative forms of genes.

The chromosomes of an homologous pair may carry the same or different alleles for a given gene.

- **Homozygous** describes the state of an organism that carries the same alleles for a particular gene on both chromosomes of an homologous pair.
- **Heterozygous** refers to the state of an individual that carries alternative alleles for a given gene.

*Example*: In humans, 'handedness' is a genetic trait controlled by a gene with two alternative alleles. Right-handedness is dominant to left-handedness.



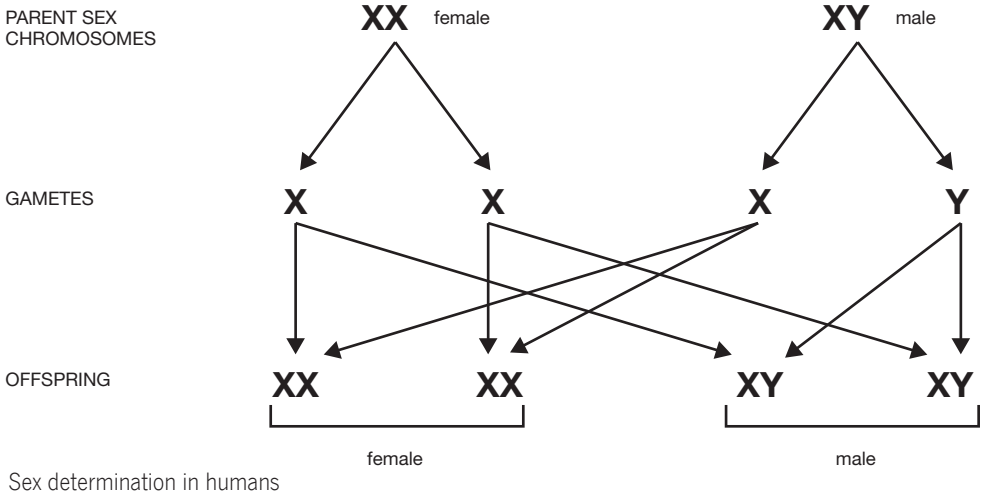



**Figure 2.21** Sex determination in humans

Notation for handedness: *R*: right-handed
*r*: left-handed
*RR*: homozygous right-handed individual
*Rr*: heterozygous right-handed individual
*rr*: homozygous left-handed individual

- Phenotypic traits can be described as **dominant** or **recessive**.

A trait is **dominant** when it appears in the phenotype of a heterozygote.

**Recessive traits** only appear in the phenotype of homozygotes; they do not appear in the phenotype of a heterozygote.

**Pedigree analysis** provides the opportunity of tracking the pattern of inheritance of particular traits from one generation to the next in families. This is a useful method of establishing the mode of inheritance for characteristics.

Using the information provided in the legend, together with appropriate allelic notation, allows genotypes to be assigned to at least some individuals in the pedigree. Such an approach is useful in determining the *mode of inheritance* of a particular characteristic.

The pedigree in Figure 2.22 illustrates that right-handedness is inherited as an **autosomal dominant trait**. Its inheritance pattern is not linked to gender (so the gene is carried on an autosome) and the trait appears in individuals who are heterozygous (making it fit the definition of dominance).

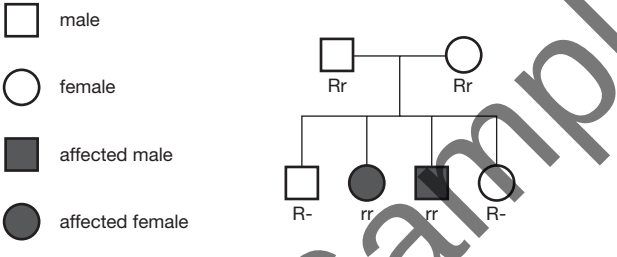



**Figure 2.22** Right-handedness is inherited as an autosomal dominant trait.

Some traits do not show simple dominance or recessiveness. There are instances in which both alleles are expressed to varying degrees in the phenotype. This is called co-dominance. The ABO blood grouping system is an example—a single gene locus features multiple alleles, $I^A$, $I^B$ and $i$. Individuals carrying alleles for both A antigens and B antigens express both in the phenotype with blood type AB.

| Genotype | Phenotype (blood type) |
|---|---|
| $I^AI^A$, $I^Ai$ | A |
| $I^BI^B$, $I^Bi$ | B |
| $I^AI^B$ | AB |
| $ii$ | O |

Flower color in snapdragons is another example. When red-flowered snapdragons (homozygous) are crossed with white-flowered snapdragons (homozygous), they produce pink-flowered offspring. In this instance both the alleles for red color and white color are partially expressed.

Genetic explanation: *R*: red
*W*: white
Parents: *RR* (red) × *WW* (white)
Offspring: all *RW* (pink)

When the pink-flowered snapdragons are crossed, they produce three different phenotypes. A **Punnett square** can be used to show this.

| gametes | R | W |
|---|---|---|
| R | RR | RW |
| W | RW | WW |

¼ red : ¾ pink : ¼ white

A **1 : 2 : 1 phenotypic ratio** is typical of the second generation in a cross involving traits that are co-dominant.

## ENVIRONMENTAL IMPACT ON GENOTYPE

Chocolate-point Siamese cats demonstrate the impact of the environment on phenotype. This breed of cat carries genetic information that results in the production of dark pigment in the extremities, i.e. the tips of the ears, snout, tail and paws. In temperate climates, the chocolate points are evident. However, when Siamese cats are raised in hot climates, the fur that grows at the extremities lacks the dark pigment.

Other examples of an interplay between genotype and environment can be observed in individuals diagnosed with phenylketonuria whose health is managed by diet; fur colour in Himalayan rabbits; and correction of tooth gap using braces in humans.

## MONOHYBRID CROSSES

A **monohybrid** cross is a cross that involves a single gene locus.

*Example*: Inheritance of colour in pea seeds
Yellow pea colour is dominant to green pea colour.
Notation: *Y*: yellow (dominant)
*y*: green (recessive)
$F_1$: first filial generation (offspring)
$F_2$: second generation
Two pure-breeding plants are crossed.
Parents: *YY* (yellow) × *yy* (green)
$F_1$: *Yy* (all yellow)

A **Punnett square** is used to calculate the ratio of genotypes and phenotypes in the $F_2$ generation.

| gametes | *Y* | *y* |
|---|---|---|
| *Y* | *YY* | *Yy* |
| *y* | *Yy* | *yy* |

¾ of the offspring will be yellow; ¼ of the offspring will be green.

A **3 : 1 phenotypic ratio** is typical of a cross between heterozygotes in a monohybrid cross where the gene under investigation has two allelic forms.

## DIHYBRID CROSSES

A **dihybrid cross** is a cross that involves two gene loci.

*Example*: Inheritance of colour and shape in pea seeds

Round pea shape is dominant to wrinkled pea shape.

Yellow pea colour is dominant to green pea colour.

Notation: *R*: round (dominant)
*r*: wrinkled (recessive)
*Y*: yellow (dominant)

When pure-breeding round, yellow pea producing plants are crossed with pure-breeding wrinkled, green pea producing plants, all the offspring produce round, yellow peas.

Parents: *RRYY* (round, yellow) × *rryy* (wrinkled, green)

$F_1$: *RrYy* (all round, yellow)

Punnett square to calculate the $F_2$ ratio:

| gametes | *RY* | *Ry* | *rY* | *ry* |
|---|---|---|---|---|
| *RY* | *RRYY* | *RRYy* | *RrYY* | *RrYy* |
| *Ry* | *RRYy* | *RRyy* | *RrYy* | *Rryy* |
| *rY* | *RrYY* | *RrYy* | *rrYY* | *rrYy* |
| *ry* | *RrYy* | *Rryy* | *rrYy* | *rryy* |

This reveals a phenotypic ratio of 9/16 round, yellow: 3/16 round, green: 3/16 wrinkled, yellow: 1/16 wrinkled, green.

A **9 : 3 : 3 : 1 phenotypic ratio** is typical of a dihybrid cross between heterozygotes where the traits under investigation are controlled by genes with two alleles.

## TEST CROSSES

A **test cross** is a cross between an individual displaying the dominant phenotype and a homozygous recessive individual. Test crosses are carried out to determine whether the individual with the dominant phenotype is homozygous or heterozygous. If offspring displaying the recessive phenotype are produced, the individual must be heterozygous. If all offspring show the dominant phenotype, this suggests the individual is probably homozygous. The larger the number of offspring, the more reliable the results.

Monohybrid test crosses reveal a phenotypic ratio of 1 : 1.

| gametes | *R* | *r* |
|---|---|---|
| *r* | *Rr* | *rr* |

Dihybrid test crosses reveal a phenotypic ratio of 1 : 1 : 1 : 1.

| gametes | *RY* | *Ry* | *rY* | *ry* |
|---|---|---|---|---|
| *ry* | *RrYy* | *Rryy* | *rrYy* | *rryy* |

## GENE LINKAGE

Genes located on the same chromosome and that are likely to be inherited together form a **linkage group**. **Linkage** refers to the tendency for alleles located on the same chromosome to be inherited together.

*Example*: Consider linked genes *P* and *Q*, represented by alleles *P*, *p* and *Q*, *q* respectively.

Notation: *PQ* denotes that alleles *P* and *Q* are located on one chromosome and *pq* denotes alleles *p* and *q* are located on the other

During meiosis, two kinds of gametes are expected to be produced, *PQ* and *pq*. These are called **parental types** (also called parental gametes).

The further apart the gene loci are located on the chromosome, the more likely that **crossing over** will occur between them. Crossing over will rearrange the genetic material, resulting in new combinations of alleles. Such gametes are called **recombinants**. Crossing over increases variation in the kinds of gametes produced.

Genes are considered to be linked if less than 50% of the gametes produced are recombinant. When a dihybrid test cross deviates from the expected 1 : 1 : 1 : 1 ratio, it indicates the gene loci in question are linked.

## SEX LINKAGE

Genes located on the sex chromosomes are said to be **sex-linked**. This is because the phenotype is linked to the gender of the individual. Tracking the pattern of inheritance of characteristics in pedigree analysis is a useful method of establishing whether or not genes are sex-linked.

Colour-blindness and haemophilia in humans are sex-linked characteristics; genes controlling both characteristics are located on the X-chromosome (Figure 2.23).

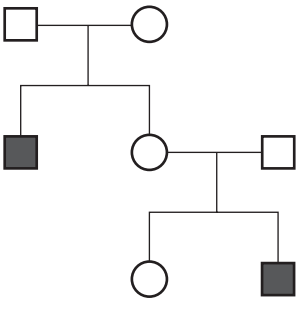


**Figure 2.23** Colour-blindness is inherited as a sex-linked recessive trait.

**X-linked dominant characteristics:** An affected male will pass the trait to all his daughters but not his sons.

**X-linked recessive characteristics:** An affected female will pass the trait to all her sons.

**Y-linked characteristics:** Pattern of inheritance is always father to son.

## CONTINUOUS AND DISCONTINUOUS VARIATION

Many characteristics come under the control of more than one gene. This is called **polygenic inheritance**.

**Continuous variation**: Traits are controlled by **polygenes** and characterised by a range of phenotypes; their distribution can be represented graphically by a typical bell curve.

Examples include the inheritance of height, eye colour and skin colour.

**Discontinuous variation**: Traits are typically controlled by a single gene, usually with two allelic forms and characterised by distinct phenotypes.

*Examples*:

- Handedness—individuals are either right-handed or left-handed.
- Flower colour in snapdragons—two alleles result in three distinct phenotypes.
- ABO blood grouping—three different alleles result in four distinct phenotypes.

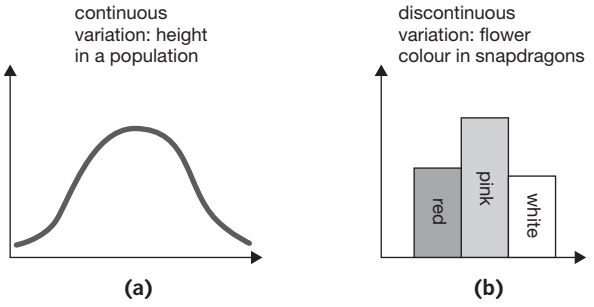



**Figure 2.24** **(a)** Continuous variation **(b)** Discontinuous variation

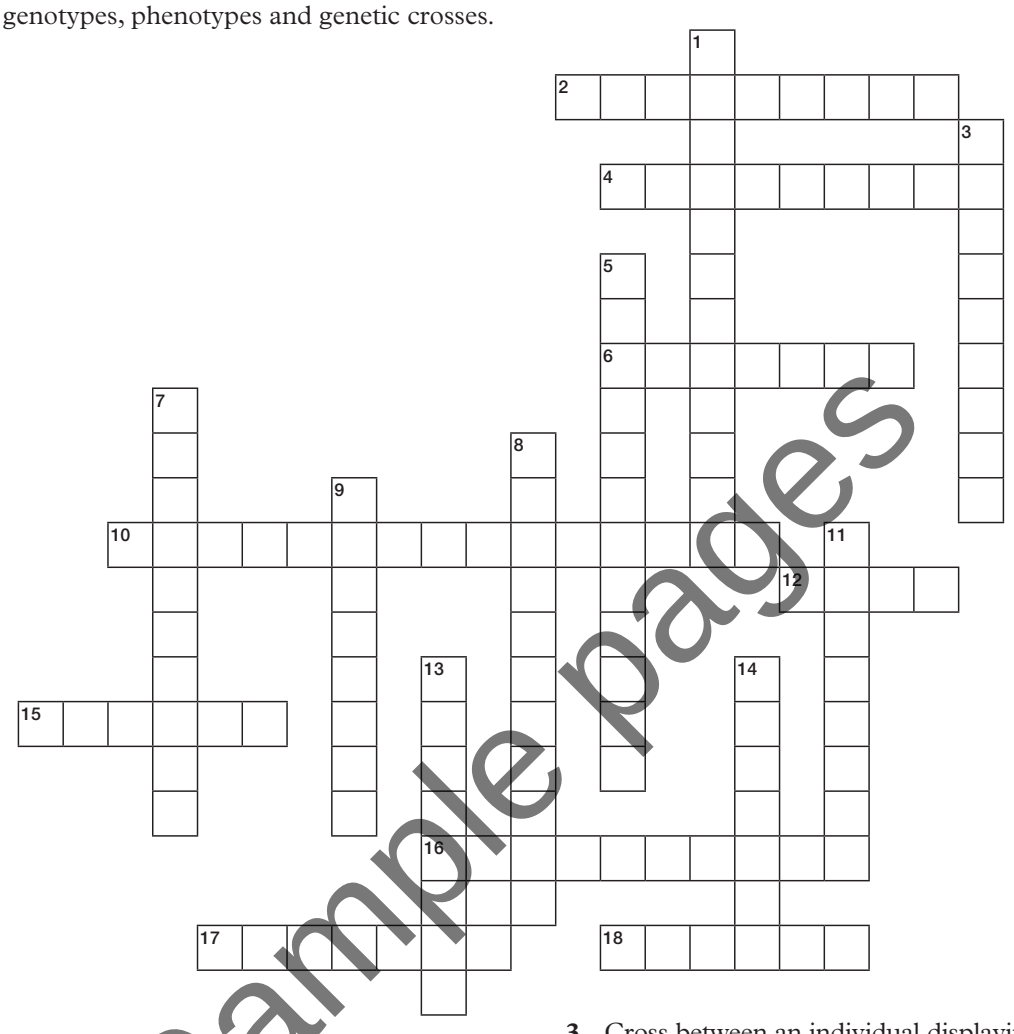
## TRACKING GENETIC PEDIGREE

Stud books are records outlining breeding relationships over a number of generations. They represent a useful way of tracking the individuals that have been bred, their characteristics and their relationships with one another. Stud books are kept for thoroughbred race horses, and for breeding cats and dogs. They are important in breeding livestock, particularly in the selection of animals identified with desirable characteristics. Stud books are also important record-keeping tools in breeding programs for wildlife management, especially in relation to endangered species, where maintaining genetic variation and avoiding inbreeding are paramount.

# WORKSHEET 31

## Crossword—genotype, phenotype and crosses

Complete the crossword puzzle to help you check your knowledge and understanding of key terms and processes related to genotypes, phenotypes and genetic crosses.





### Across

**2** Describes the characteristic that is observed in the phenotype of an individual homozygous for a particular allele, but not observed in the heterozygote. [9]

**4** Physical expression of genotype. [9]

**6** Nitrogenous base complementary to adenine in DNA. [7]

**10** Cross between individuals that takes into account one particular characteristic. [15]

**12** A unit of hereditary information that determines the characteristics of an organism. [4]

**15** The full complement of genes in an individual organism. [6]

**16** The structural unit of nucleic acids. [10]

**17** Nitrogenous base complementary to cytosine in DNA. [7]

**18** Alternative form of a gene. [6]

### Down

**1** Describes the status of an individual that carries two different forms of a gene in relation to a particular characteristic. [12]

**3** Cross between an individual displaying a dominant phenotype and an individual displaying a recessive phenotype (homozygous) for the purposes of determining the genotype of the individual with the dominant phenotype. [9]

**5** Describes the arrangement of the two complementary strands in a DNA molecule as they run in opposite directions. [12]

**7** Describes the status of an individual's genotype when identical alleles are present. [10]

**8** Describes the alternative forms of a characteristic that are either fully or partially expressed in the phenotype of an individual. [11]

**9** Nitrogenous base complementary to guanine in DNA. [8]

**11** Genetic make-up of an individual in relation to one or more genes. [8]

**13** Describes a characteristic that is observed in the phenotype of a heterozygote. [8]

**14** Nitrogenous base complementary to thymine in DNA. [7]

# WORKSHEET 32

## Nuclear puzzle—same pieces, different species

Millions of different species of organisms have evolved on Earth—plants, animals, algae, fungi, protists, bacteria and more. Within a single species there is also enormous diversity. And yet, we account for every individual using the same fundamental threads of genetic material—DNA. Not only this, the DNA that codes for the staggering number of different organisms and the features that make each one unique comes in only four different forms. The pieces of the DNA puzzle, the nucleotides, are characterised by a different base molecule—**adenine**, **thymine**, **cytosine** or **guanine**. It is the infinite number of combinations that gives us such a titanic degree of variety.

**1** Nucleotides are composed of the same three components. Name the molecule represented by

P: _____ S: _____

A, T, C, G: _____

**2** The nucleotide sequence in Figure 2.25 is part of the human β-haemoglobin gene.

Use coloured pencils to colour-code the nucleotide bases in the legend.

Follow your code to colour the different bases along the base sequence.

**3** Use appropriate symbols and colour-coding to draw the complementary DNA strand against this template strand.

**4** Look carefully at the details of your double-stranded DNA. Describe two features of DNA that ensure complementary base-pairing occurs.

Feature 1: _____

_____

_____

Feature 2: _____

_____

_____

**5** Add another symbol to the base legend in Figure 2.24 that will allow the construction of an RNA molecule.

**6** Use the space at the right of the DNA template strand in Figure 2.25 to draw in the complementary RNA strand.

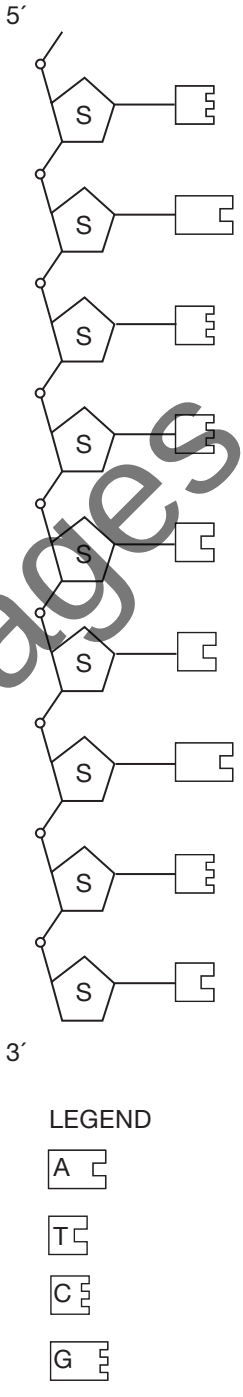
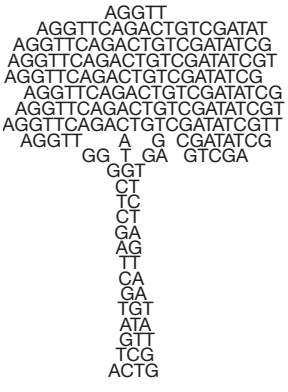
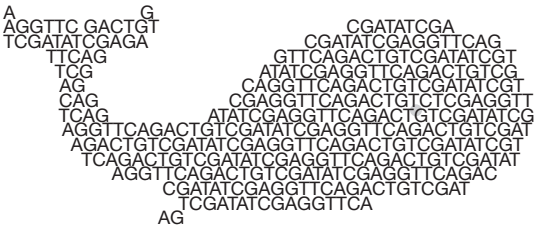



**Figure 2.25** Sequence of nucleotide bases in the human haemoglobin gene



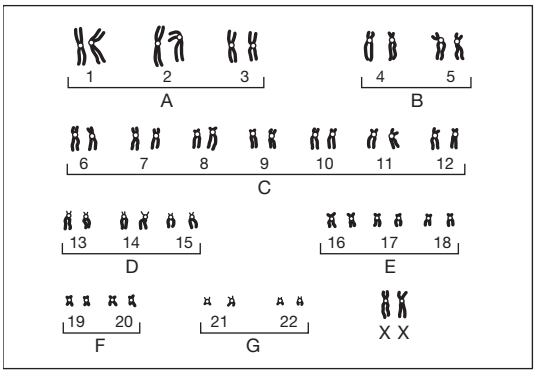
# WORKSHEET 33

## Counting on karyotypes—chromosomal diagnoses

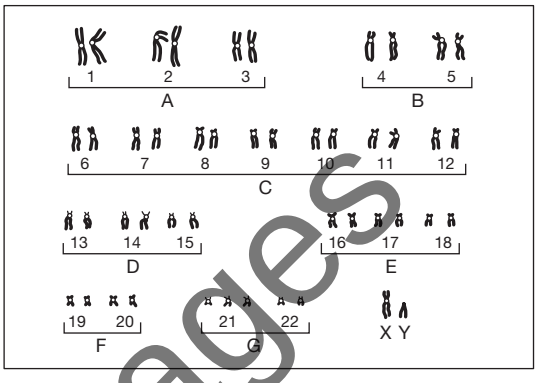
A karyotype is a visual display of an individual's chromosomes set out in homologous pairs—pairs are arranged in order of length, usually longest to shortest.

**1** Examine each of the karyotypes displayed. In the space below each, write information that can be determined about the individual from the chromosomal information provided. Include the general genotype in each case, e.g. 46XY.

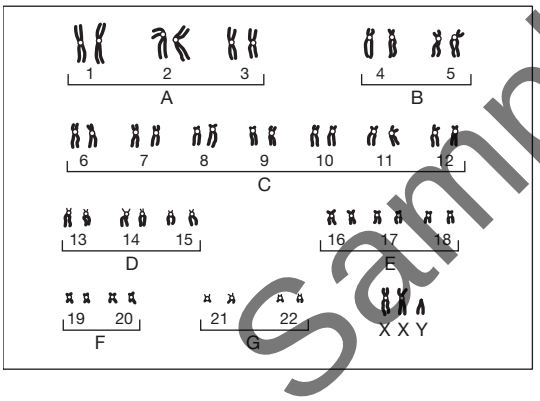
INDIVIDUAL A



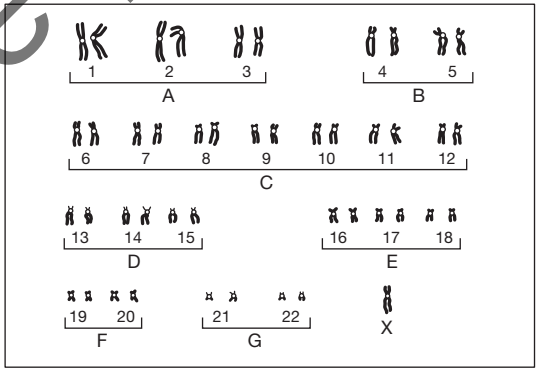

INDIVIDUAL B




INDIVIDUAL C




INDIVIDUAL D






**2** Describe the kind of information that karyotyping provides about an individual.

**3** What kind of genetic information is not available from karyotype analysis?

**4** Outline a technique that may be used in conjunction with karyotyping that allows geneticists to arrange and interpret chromosomes with greater accuracy.

WORKSHEET 34

# Counting characteristics—variations in a group

A glance around your classroom reveals an enormous degree of variation in a small group of humans. There is variation in the form of hair colour and texture, eye colour, skin colour and texture, gender and height. You may add many more just by brief observation.

In this exercise, you will collect data about the number of people with varying forms of some characteristics in your class.

**1** What is the sample size for your class? $n =$ _____

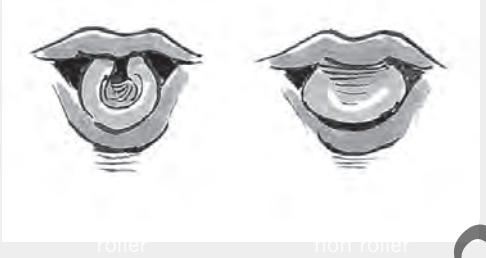
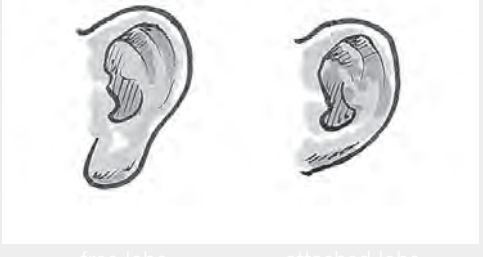
**2** Take each characteristic in turn. Count the number of individuals who display the different forms.

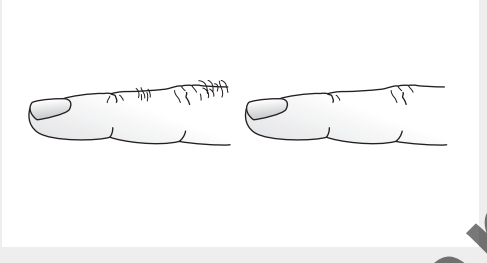
**a** Enter the data into the table.

**b** Use the data and the formula provided to calculate the frequency of each characteristic for your class.

Frequency F (%) $= \frac{x}{n} \times \frac{100}{1}$, where $x$ = number of individuals with a particular form of a characteristic, and $n$ = total number of individuals in sample.

**c** Convert the percentages of the alternative forms of characteristics into the simplest ratio. Enter this into the table, e.g. 77% : 23% approximates a 3 : 1 ratio.

| Trait | Frequency (%) | |
|---|---|---|
| Tongue rolling ability  roller non roller | Can roll tongue: $x =$<br>F = | Cannot roll tongue: $x =$<br>F = |
| | Ratio | |
| Hand clasping  | Left thumb on top: $x =$<br>F = | Right thumb on top: $x =$<br>F = |
| | Ratio | |
| Earlobe shape  free lobe attached lobe | Free lobes: $x =$<br>F = | Attached lobes: $x =$<br>F = |
| | Ratio | |

| Trait | Frequency (%) | |
|---|---|---|
| Hair line  | Widow's peak: $x$ =<br>F = | Straight: $x$ =<br>F = |
| | Ratio | |
| Second toe length  | Longer than first: $x$ =<br>F = | Shorter than first: $x$ =<br>F = |
| | Ratio | |
| Mid-digital hair  | Present: $x$ =<br>F = | Absent: $x$ =<br>F = |
| | Ratio | |



**3** Outline any consistencies that became apparent in the statistics for your class.

**4** Explain why a larger sample size is likely to provide more reliable results.

WORKSHEET 35

# Linkage and pedigrees—a summary

Make a selection from the list to fill in the missing words in each summary statement.

| | | | | |
|---|---|---|---|---|
| X-linked | gene complex | continuous | pedigree | sex linkage |
| polygenic | linkage | crossing over | stud books | Y-linked |
| discontinuous | recombinant | dominant | recessive | |

- When two or more genes are located on the same chromosome they are referred to as a _____ group. The more closely such genes are situated on a given chromosome, the greater the likelihood that they will be inherited together.
- _____ gametes are formed as a result of _____ at a chiasma during prophase I in meiosis.
- Genes that are so closely linked on a chromosome that crossing over between them is a rare event are referred to as _____ _____.
- _____ _____ refers to the presence of genes on either of the sex chromosomes. In this case, the inheritance of characteristics is linked to the sex of the individual.
- Colour-blindness is an example of an _____ characteristic. The mode of inheritance for this condition is X-linked recessive—transmission is typically from female parent to male offspring. X-linked characteristics appear less often in females because there are two X-chromosomes. When one X-chromosome is carrying the affected allele, it may be masked by a normal allele on the second X-chromosome. Males are more often affected because the allele is present on their only X-chromosome.
- _____ inheritance shows a pattern of transmission from father to son. Characteristics that follow this mode of inheritance are never observed in females.
- _____ analysis is a strategy that allows geneticists to track the pattern of inheritance of particular characteristics. This provides important information about the mode of inheritance of characteristics and can be useful in calculating the likelihood of genetic diseases occurring in families.
- Animal breeders looking to breed desirable characteristics avoid inbreeding and maintain genetic variation by keeping records called _____ _____ to track breeding stock over generations.
- When alternative forms of a particular characteristic can be clearly placed into non-overlapping groups, _____ variation is said to exist. Such characteristics are typically governed by single genes.
- _____ variation describes the kinds of characteristics that show wide variation across a range. Such characteristics are typically governed by a number of genes and are referred to as _____ traits.
- Characteristics that appear in the phenotype of a heterozygote are described as _____.
- Characteristics that do not appear in the phenotype of the heterozygote are described as _____.

WORKSHEET 36

# Puzzling pedigrees—analysing family histories

## BLOOD RELATIVES

The ABO blood group of an individual can be determined by identifying the kinds of proteins (antigens) that are present on the surfaces of red blood cells. The single gene locus that codes for the production of these antigens has three alleles ($I^A$, $I^B$ and $i$). The genotypes and phenotypes of respective individuals are shown in the Table 2.5.

**Table 2.5** ABO blood groups

| Genotype | Phenotype |
|---|---|
| $I^AI^A$ | A |
| $I^Ai$ | A |
| $I^BI^B$ | B |
| $I^Bi$ | B |
| $I^AI^B$ | AB |
| $ii$ | O |

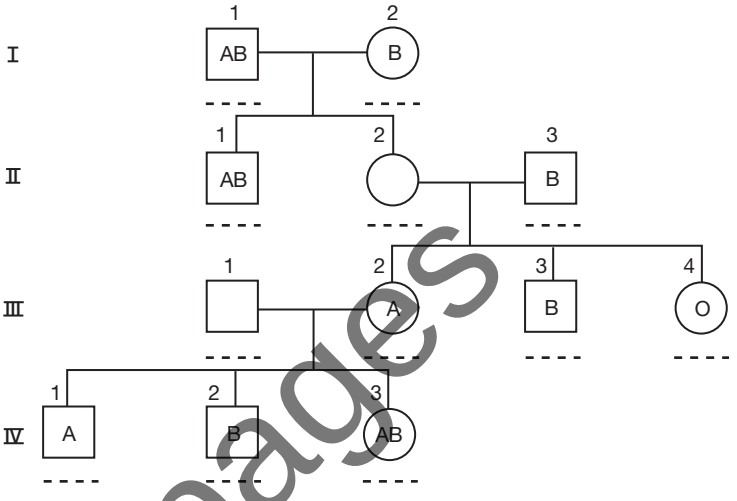



**Figure 2.26** ABO blood group pedigree

**1** The pedigree in Figure 2.26 on page 161 indicates the blood type for some individuals. Use your understanding of inheritance and the alleles above to assign genotypes and blood types to individuals II-2 and III-1.

**2** Outline the relationship between the phenotypic expression of the $I^A$, $I^B$ and $i$ alleles.

## ROYAL BLOOD

The pedigree in Figure 2.27 represents part of the family tree for a European royal family. It also tracks the inheritance of haemophilia, a blood disorder that leaves sufferers without an important clotting factor, leading to uncontrolled bleeding after even minor injury. Today, haemophiliacs are successfully treated with blood transfusions, but in the past individuals born with this disorder usually did not survive childhood.

# WORKSHEET 36

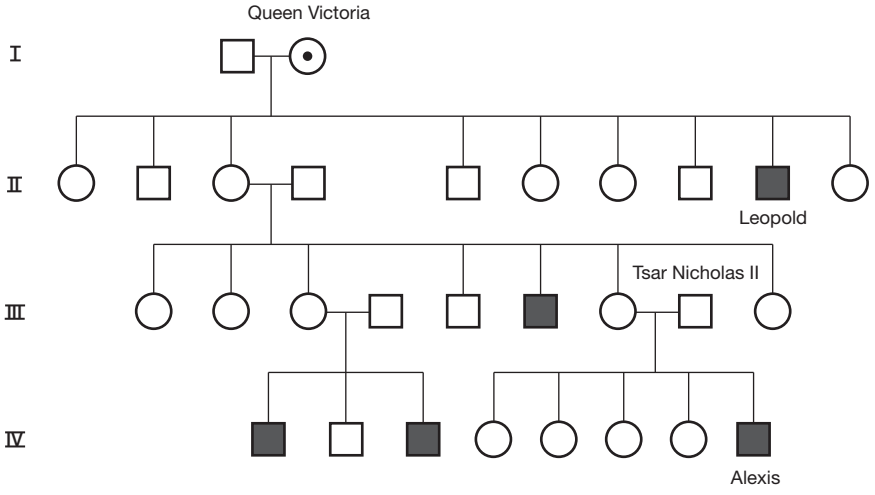



**Figure 2.27** Pattern of inheritance of the blood disorder haemophilia in a European royal family

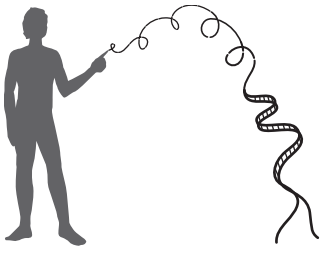
**3** Suggest why only males in this family tree are affected by haemophilia.

**4** Queen Victoria's son, Leopold, was the first person in the family's history to have been diagnosed with the condition. The cause of the disease in this family is attributed to a mutation that occurred early in the embryological development of Queen Victoria or in a germ-line cell from one of her parents. Describe the evidence from this family tree that points to Queen Victoria as the origin of haemophilia in the family, and not her son Leopold.

**5** In the pedigree, Queen Victoria's status as a carrier is denoted by the dot symbol. Use the same notation to identify all of the other carriers of haemophilia in this family.

# PRACTICAL ACTIVITY 17

## DNA distillery—extracting DNA



### MATERIALS

- 1 level teaspoon wheat germ
- 20 mL hot water (about 50°C)
- 1 mL dishwashing detergent
- meat tenderiser powder
- methylated spirits or ethanol
- 50 mL beaker
- 50 mL glass measuring cylinder
- glass stirring rod
- glass hook
- pipette

### PURPOSE

To extract DNA from the nuclei of wheat germ cells.

### PROCEDURE

1 Place the teaspoon of wheat germ in the beaker, add the hot water and stir for about 5 minutes (not too vigorously).
2 Add the detergent then stir VERY GENTLY every minute or so for the next 5 minutes. Be careful to avoid creating any foam. If any foam does form, use a pipette to remove it.
3 Add the meat tenderiser. Again, stir through VERY GENTLY every minute or so for the next 2 or 3 minutes.
4 Gently pour the mixture into the measuring cylinder.
5 Tilt the measuring cylinder a little and very carefully pour some methylated spirits down the inside of the cylinder until it forms a layer about 2 cm deep above the wheat germ–detergent mixture.
IMPORTANT NOTE: Slow and careful addition of the methylated spirits will help prevent the methylated spirits penetrating the wheat germ mixture.
6 Leave the preparation on the bench for 10 minutes. Check your preparation every few minutes.

### DISCUSSION

1 Describe your observations. Include a diagram of your complete preparation.

**2** The white material that has emerged at the top of the mixture is the DNA that has been extracted from the wheat germ cells. Use the glass hook to gently lift some of this from the surface of the mixture. Describe its appearance.

**3** Suggest the reason for adding:

**a** detergent:

**b** meat tenderiser:

(Hint: Think about the role of detergents in removing grease, and the role of meat tenderiser in meat preparations. How can these be related to the cells of the wheat germ?)

**4** Describe any limitations you encountered in this activity.

**5** What measures could you take to reduce these limitations next time?

# PRACTICAL ACTIVITY 18

## Modelling DNA—simulating the structure

### MATERIALS

- 2 different colours of plasticine or similar
- set of 2 different coloured beads (approximately 10 of each)
- set of nucleotides photocopied from template provided (at least 4 of each A, T, G, C)
- scissors
- material to fix nucleotide 'cut-outs' to paper

### INTRODUCTION

DNA, or deoxyribonucleic acid, is often referred to as the 'blueprint' for life; a universal code that provides the instructions for protein synthesis in all living organisms.

The building blocks of DNA are nucleotides, composed of three parts—a deoxyribose sugar, a phosphate component and a nitrogenous base (one of either adenine, thymine, guanine or cytosine). The nucleotides link together to form two strands running in an antiparallel arrangement with complementary base pairing between adenine and thymine and between guanine and cytosine.

A single DNA molecule may measure over a metre in length when fully unwound. In order to fit within the nucleus of a cell and maintain order to the code, the DNA coils tightly to form a chromosome.

### PURPOSE

To investigate the structure of DNA through a modelling activity.

### DURATION

40 minutes



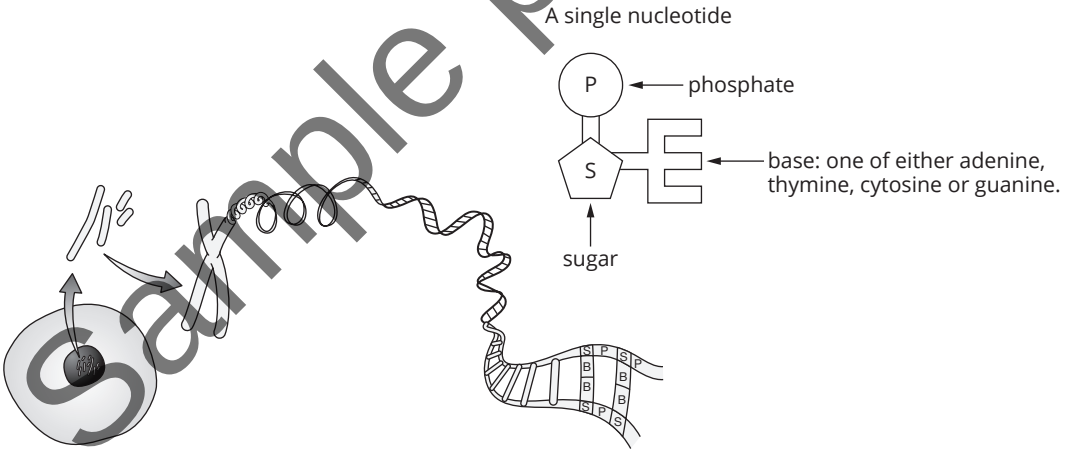



**Figure 2.28** The DNA contained in the nucleus unravels to reveal the double helix

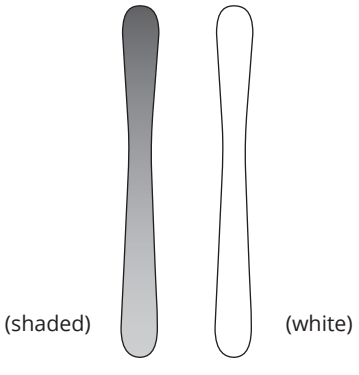



**Figure 2.29** Single-stranded chromosomes

### PROCEDURE

## PART A MODELLING DNA AT THE CHROMOSOMAL LEVEL

1 Using two different colours of plasticine, roll out two single-stranded chromosomes, one of each colour. Ensure that the size is similar. This represents a homologous pair. The different colours represent the maternal and paternal inheritance of each chromosome.

1 Describe how the chromosomes in the homologous pair are similar and different to each other.

**2** Use the coloured beads provided to mark two gene loci on your plasticine model. For one locus, show identical alleles and for the other show different alleles.

**2** Outline the difference between a 'gene' and an 'allele'.

**3** Explain what is meant by the terms 'homozygous' and 'heterozygous'.

**3** During DNA replication, the double helix produces an identical copy in preparation for cell division. At this point of the cell cycle, the duplicated chromosome is held together by a centromere. Add another plasticine roll to each single chromosome to model a duplicated homologous pair.

**4** Add further beads to your duplicated pair to show the alleles now present on each chromatid.

**4** Sketch a diagram of your duplicated pair of homologous chromosomes. Use colour coding and the following terms to add labels:

| | | |
|---|---|---|
| duplicated chromosome | chromatid | centromere |
| gene loci | homozygous | heterozygous |



**5** Identify and describe a feature of DNA which is clearly demonstrated by your chromosome model.

**6** Share your response to Question 5 with others. Are the responses of your classmates similar or different to your suggestion? Explain.

## PART B MODELLING DNA AT THE BIOMOLECULAR LEVEL

In part B we are going to zoom in on the DNA structure as the chromosome unwinds. When the double helix is completely unwound, the nucleotides appear in two complementary strands that resemble a ladder, with the ribose sugar and phosphate groups forming the uprights and the nitrogenous bases forming the rungs of the ladder.

**5** Allocate a colour for each of the four nucleotides and colour in each nucleotide accordingly. Note how each of the four types of nucleotides are similar and different from each other.

**6** Cut out the photocopied templates provided for each of the four types of nucleotides. You should have four of each type, so 16 individual nucleotides in total.

**7** Arrange a strand of nucleotides to resemble a single strand of DNA running in the 5' to 3' direction as per the image below. Consider the order of your nucleotides. What does the order that you selected represent?

**8** Fix the nucleotides in position downwards along the length of the page on the blank space provided on pages 168 and 169.

**9** Arrange corresponding nucleotides to demonstrate the complementary base pairing in the other strand. Fix the nucleotides in position.

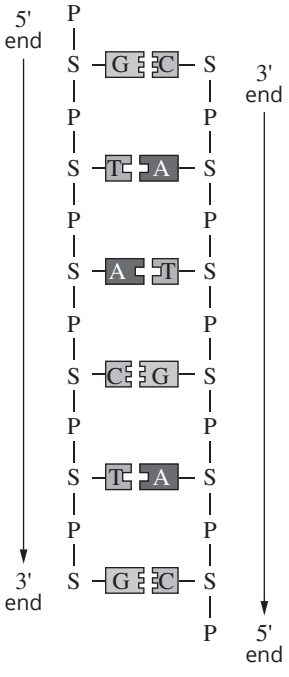



**Figure 2.30** Nucleotides arranged in complementary pairs

**7** Look carefully at the arrangement of the two complementary DNA strands you have constructed. Explain why the DNA molecule is described as 'antiparallel'.

**8** Describe two features of DNA that ensure complementary base pairing is maintained between the strands.

**9** One of the limitations of this modelling activity is that it does not accurately demonstrate the three-dimensional structure of the double helix. Suggest how you could modify your model to more accurately reflect the three-dimensional structure of the double helix.

**10** Identify and describe another limitation of your model in demonstrating the structure and/or function of the DNA molecule.



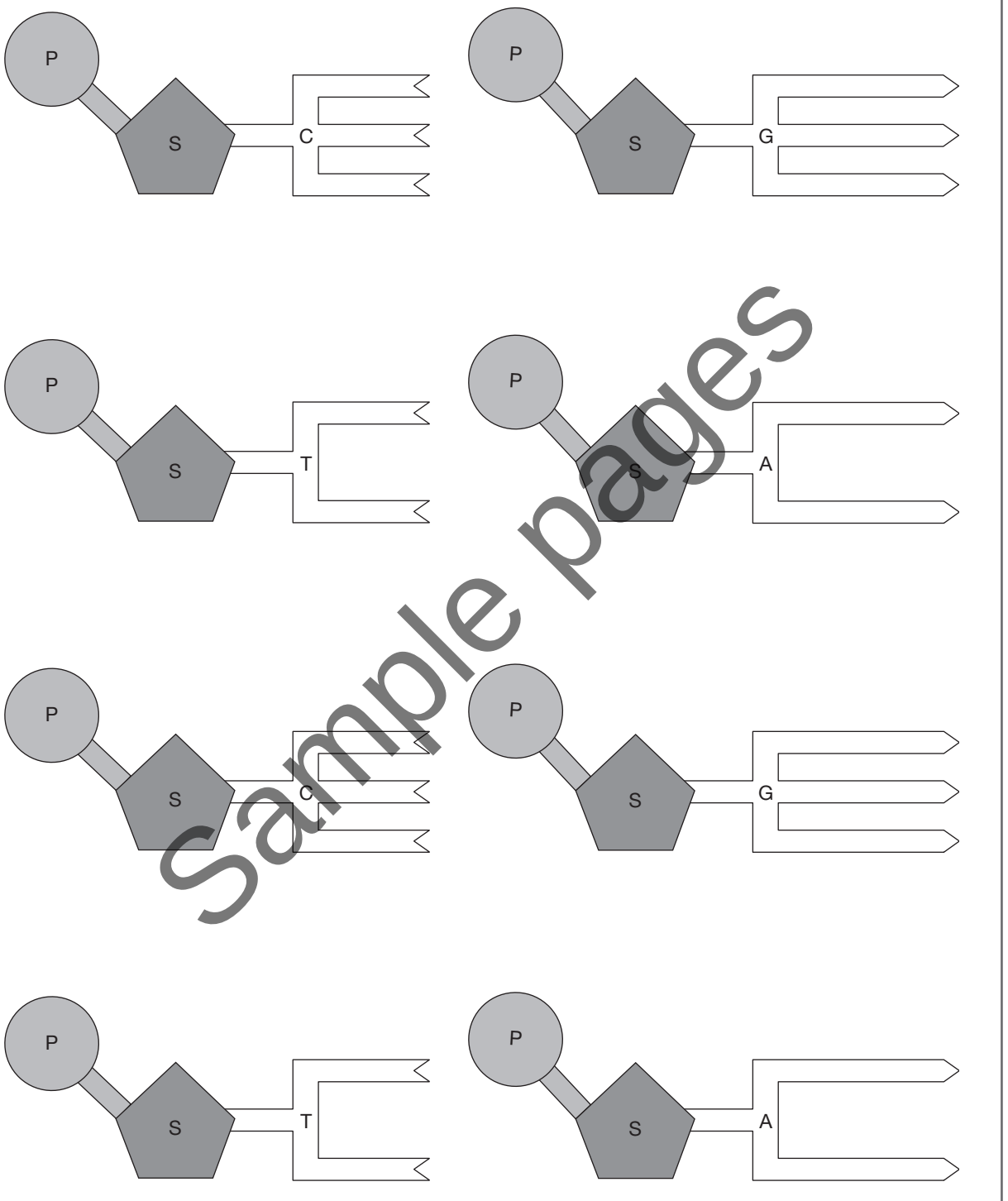
### CONCLUSIONS

**11** The DNA molecule is composed of threads of nucleotides. Name the three main components of a single nucleotide.

**12** Summarise the way nucleotides are arranged to form the double helix of the DNA molecule.

# PRACTICAL ACTIVITY 18

## Photocopiable templates



PRACTICAL ACTIVITY 19

# Genetic roulette—people and pedigrees

## INTRODUCTION

A look around your class illustrates many similarities between unrelated individuals—we share hair, eyes, ears, nose, arms, legs, and many other characteristics. Of course, there are many differences in the form of these characteristics between members of the group. For example, hair can be blond, brown, red or black; it can be straight, wavy or curly; it may be fine in some individuals and coarse in others. There are enormous variations in eye colour. But look within a single family—there are more similarities between related individuals within a family than there are between unrelated members of your class. For example, fair skin and reddish hair are likely to be shared by individuals within one family. These characteristics are also likely to be shared by other generations within the family. Why is this? Family trees or pedigrees provide a useful way of analysing information that is inherited from one generation to the next within a family. Analysis of pedigrees gives us a clue about the way characteristics are inherited, and their pattern or mode of inheritance. They can also be used to determine the genetic make-up, or genotype, of an individual, and to predict the chances of children having particular features.

## PURPOSE

- To analyse selected pedigrees to determine the mode of inheritance of genetic traits.
- To predict the possible outcomes in children born of particular partnerships in relation to inherited diseases.
- To construct pedigrees from family histories in order to determine modes of inheritance.



## PROCEDURE

Carefully read the foundation ideas in the background notes on page 171. This will familiarise you with the symbols used in pedigree analysis. The information in Table 2.6 outlines the key features that distinguish the modes of inheritance for different phenotypic characteristics. Use the information to help you answer the questions related to each of the pedigrees presented in part A and the problems raised in part B of this activity.

**Table 2.6** Observed patterns for different types of traits

| Pattern of inheritance | Key features |
|---|---|
| **Autosomal dominant** | Gene loci on chromosomes other than sex chromosomes; either sex can be affected. Characteristic appears in the phenotype of a heterozygote. Affected individuals must carry at least one allele for the dominant trait. Unaffected parents will not produce affected offspring (unless a new mutation occurs). |
| **Autosomal recessive** | Gene loci on chromosomes other than sex chromosomes; either sex can be affected. Characteristic does not appear in the phenotype of the heterozygote. Affected individuals are homozygous recessive. Unaffected parents can produce affected offspring. |
| **X-linked dominant** | Affected males pass trait to all their daughters and none of their sons. |
| **X-linked recessive** | Affected females produce only affected sons. Expect half the sons of unaffected female carriers to be affected. Affected males produce only unaffected sons. |

# PRACTICAL ACTIVITY 19

## BACKGROUND

In pedigree analysis, symbols are used to provide specific information about individuals in a clear and simple way. Use the following legend as a guide to interpreting the pedigrees in this activity.

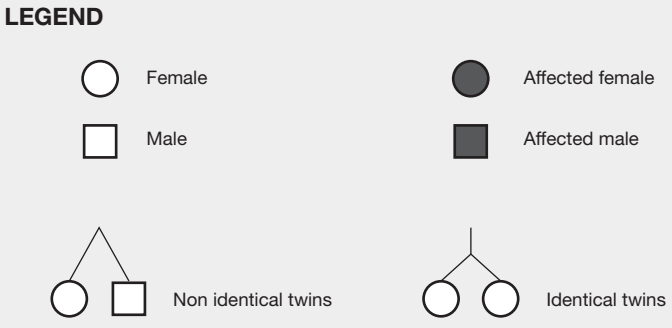



**Figure 2.31** Pedigree legend

## Scenario 1

The pedigrees in Figures 2.32 and 2.33 show the inheritance pattern of earlobe shape in two different families. 'Free lobes' are dominant to 'attached lobes', which are recessive. The gene responsible for earlobe shape has two alternative alleles represented by *E* (free lobes) and *e* (attached lobes).

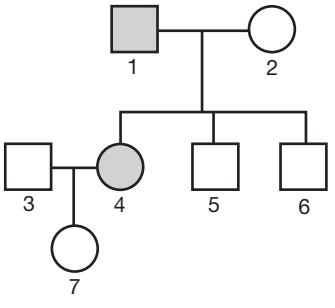



**Figure 2.32** Pedigree of earlobe inheritance

## PART A PEDIGREE ANALYSIS

**1** Assign genotypes to as many individuals as possible in Figure 2.32.

**2** Describe the pattern or mode of inheritance for earlobe shape in humans.

**3** Examine the pedigree in Figure 2.33. Assign genotypes to as many individuals as possible.

**4** Why is it difficult to do this with confidence for individuals 1 and 3?

**5** How can you be sure of the genotypes of individuals 6 and 7?

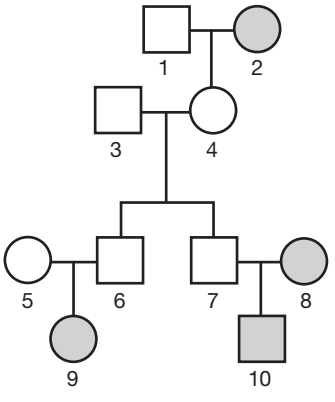



**Figure 2.33** Pedigree of free and attached earlobe inheritance in a family

## Scenario 2

Known family histories are also useful to geneticists in establishing the mode of inheritance for particular genetic diseases. Pedigree analysis for families that show such diseases is also important so that genetic counselling can be provided to families about the likelihood of future children being affected or carrying the allele in question. Figure 2.34 illustrates the inheritance of Huntington's disease in two unrelated families. Huntington's disease is a neurological disorder that leads to gradual, permanent deterioration of nerve and muscle control with eventual complete dependence on care. Death results after some years. The onset of the symptoms does not occur until at least the mid to late thirties.


**Figure 2.34** Two pedigrees of Huntington's disease

**6** Assign genotypes to each person in both pedigrees.

**7** Name the mode of inheritance for Huntington's disease. Explain your choice.

Individuals 7 and 8 are engaged to be married. Both individuals are keen to raise a family.

**8** **a** What are the chances of any children from this union developing Huntington's disease? Show your working.

**b** Suggest options that a genetic counsellor might discuss with such a couple.

## Scenario 3

People with galactosaemia are unable to digest milk sugar (galactose).

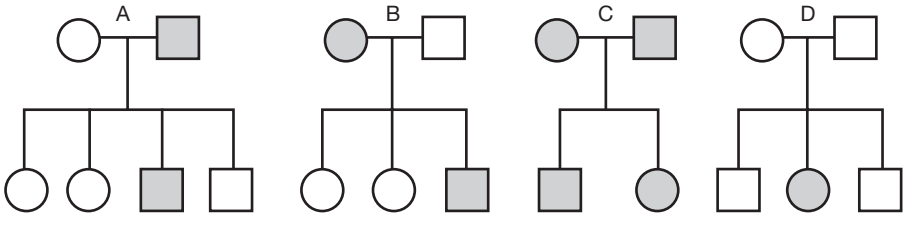



**Figure 2.35** Four families with galactosaemia

**9** From the evidence of the pedigrees shown in Figure 2.35, suggest which pedigree shows beyond doubt that galactosaemia is inherited as an autosomal recessive condition. Explain your reasoning.

# PRACTICAL ACTIVITY 19

## Scenario 4

Red–green colour blindness is a relatively common condition, inherited as an X-linked recessive trait. Figure 2.36 shows the pedigrees of three families in which this condition occurs.

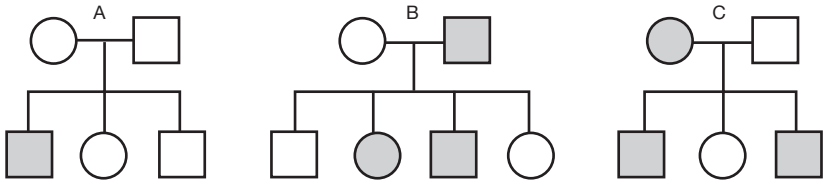



**Figure 2.36** Pedigrees of red–green colour blindness.

**10** Which of the three pedigrees best establishes the mode of inheritance for this trait? Explain your reasoning.

## PART B PICTURING PEDIGREES

Prepare a pedigree chart in the space provided for each of the scenarios described below.

**11** **a** An affected child is born to parents, neither of whom shows the characteristic.

**b** Determine the mode of inheritance. Explain.

**12** **a** A man displaying a characteristic inherited as an X-linked trait marries an unaffected female. They have two affected daughters and two unaffected sons.

**b** Explain whether the trait is inherited as dominant or recessive.

**13** **a** A woman showing a trait that has an X-linked recessive mode of inheritance has twins, a girl and a boy, followed by another girl and another two boys. Her partner is unaffected.

**b** Assign genotypes to all individuals in the pedigree.

## CONCLUSION

**14** Describe the feature of a pedigree that establishes the mode of inheritance for a particular characteristic as:

**a** autosomal dominant:

**b** autosomal resessive:

**c** X-linked dominant:

**d** X-linked recessive:

# PRACTICAL ACTIVITY 20

## Betting on barley—a monohybrid cross

### BACKGROUND

Pigmentation in barley is controlled by a single gene with two alternative alleles. In the heterozygote, expression of green pigment masks the effect of the allele coding for no pigment (albino). The genetic barley used in this experiment is the result of a cross between plants heterozygous for the gene locus in question.

This activity recommends that a total of around 200 barley seeds be grown by the class. Your teacher will assign a number of seeds to your group, depending on the number of groups in your class.

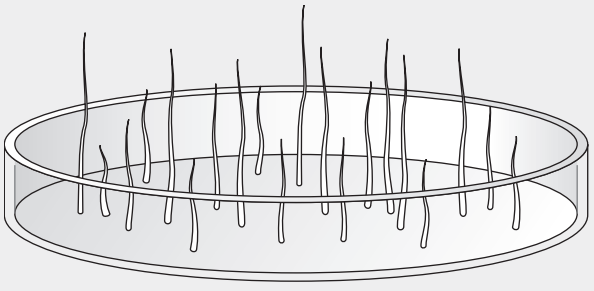


**Figure 2.37** Germinating barley seedlings

### MATERIALS

- 20 seeds genetic barley
- sheet of cotton wool
- large Petri dish
- water spray dispenser
- forceps

### PURPOSE

To investigate the mode of inheritance of a genetic trait in a monohybrid cross using genetic barley.



### PROCEDURE

1 Lay a sheet of cotton wool inside the Petri dish and spray generously with water until the cotton wool is quite damp.
2 Use the forceps to arrange the barley seeds on the cotton wool so that they are evenly spaced about 1 cm apart.
3 Spray a little more water to ensure the seeds are dampened.
4 Leave the Petri dish on a bench near a window.
5 Spray the seeds twice daily to ensure they do not dry out. You should continue to do this after initial germination until the barley seedlings are at least 2 cm tall. This is likely to take a couple of weeks.

### TWO WEEKS LATER

1 Count the number of different coloured seedlings and enter your results into the table below..

2 Collate the class data. A spreadsheet, whiteboard or overhead projector will be useful for this. Enter the class data into the table.

| | Number of seedlings of each colour | | Total |
|---|---|---|---|
| | Green | Albino | |
| **Own data** | | | |
| **Class data** | | | |

**3** Calculate the ratio of green seedlings to albino seedlings for:

**a** your own data: _____

**b** the class data: _____

**4** **a** How does the ratio for your own data compare with the ratio for the class data?

_____

_____

**b** Which set of data is likely to be more reliable? Explain.

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_____

**5** Use appropriate notation to assign genotypes to the different coloured seedlings.

_____

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_____

**6** Homozygous green barley plants are indistinguishable from heterozygotes. In ordinary circumstances, a geneticist would carry out a test cross to determine the genotype of an individual that shows the dominant characteristic.

**a** Describe a test cross.

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**b** Outline how a test cross is useful in determining the genotype of such an individual. Use a worked example to illustrate your answer.

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## SUMMARY REPORT

Summarise your findings in this activity. You should include the following items in your discussion:

- Identify the mode of inheritance for green pigmentation in barley.
- Use a model to explain how the data supports your theory.