

## 156. KEY TERMS: Did You Get It? (page 217)

1. Producer → 1st order consumer → 2nd order consumer → 3rd order consumer
2. carrying capacity (C), competition (F), consumer (E), food chain (A), food web (G), keystone species (J), mutualism (B), parasitism (H), random sampling (I), trophic level (D)
3. (a) Approximately 6000 individuals.  
(b) The population was entering an exponential phase of growth.  
(c) The population had crashed to a low point.  
(d) The population had exceeded carrying capacity by a large amount and would have run out of resources (food, space). Many individuals would have died and population growth would have fallen dramatically.

## 157. Chapter Review: Unit 1 Area of Study 2 (page 218)

No model answer. Summary is the student's own.

## 158. Synoptic Question: Unit 1 Area of Study 2 (page 220)

1. Animals need to maintain a stable internal environment so that the metabolic reactions essential to life can be carried out. To maintain homeostasis, the body must detect changes in the environment (through receptors), process this sensory information (brain and CNS), and respond to it appropriately via effectors (muscles and glands). Glucose is needed by cells for cellular respiration (to produce ATP, which provides the cell's energy), so providing a constant supply of glucose is essential to maintaining metabolism. In producers, glucose comes from photosynthesis. Heterotrophs obtain their glucose from other organisms (e.g. by eating).

In mammals, glucose is transported in the blood and must get into the body's cells to be available for metabolism. The process of regulating blood glucose levels and making it available to cells occurs by negative feedback involving two hormones, insulin and glucagon. Negative feedback has a stabilising effect and acts to discourage variations from a set point. It works by returning internal conditions back to a steady state when variations are detected.

Insulin and glucagon, secreted by the pancreatic islet cells, work together to control BG levels. Insulin is secreted by beta cells in the pancreas and decreases BG levels by causing cellular uptake of glucose. Glucagon is secreted by alpha cells and increases BG levels by causing mobilisation of glucose from stores in the liver. When BG levels rise (e.g. after eating a meal) insulin is secreted, causing glucose uptake by cells and glucose storage (as glycogen) in the liver. BG falls and insulin secretion stops (negative feedback). Low BG levels (e.g. from fasting or exercising) causes the secretion of glucagon. Glucagon stimulates the conversion of glycogen in the liver to glucose, which is released into the blood to raise BG levels. When BG is normalised again, glucagon secretion stops (negative feedback).

Several factors can disrupt BG homeostasis. Diabetes mellitus is a disease in which blood glucose is too high because the body's cells cannot take up glucose in the normal way. If glucose cannot get into the cells, the glucose levels in the blood remain high and glucose is excreted in the urine, leaving the cells 'starving' and leading to weakness, fatigue, and a host of other metabolic problems. In type 1 diabetes, the insulin producing cells of the pancreas are damaged and no longer produce insulin. In type 2 diabetes, the pancreas produces insulin, but the body does not respond to it. Type 1 diabetics require regular insulin injections to control BG levels. Type 2 diabetics can often control BG by lifestyle factors (e.g. losing weight, and exercising) but in some cases, insulin therapy may be required. Common recreational drugs such as alcohol and nicotine can disrupt BG homeostasis. Alcohol lowers BG by stimulating insulin production and inhibiting glucagon secretion. Nicotine increases BG indirectly (through adrenaline) causing inhibition of insulin production and stimulating glucose production by the liver. In both cases, BG levels do not return to normal until the body has finished metabolising the drug.

2. Hydrophytes are plants that are adapted to living in water (e.g. water lilies). Xerophytes (e.g. cacti) live in dry, arid regions and are adapted to conserve water. The adaptations of hydrophytes include a reduced root system, which provides anchorage but little water or nutrient absorption (nutrients are absorbed through the epidermis), air spaces in the stems and leaves, which help support the plant in the water, and stomata on the surface of the leaf to facilitate gas exchange. Hydrophytes are very susceptible to drying out, e.g. when water bodies shrink during summer. Xerophytes live in deserts. They generally have succulent stems and leaves that are reduced to spines. Spines provide protection from browsers, reduce surface area for water loss, and reduce air movement around the plant, increasing the relative humidity and further reducing water loss. Some xerophytes have leaves that curl up, keeping the stomata away from air currents and creating a moist microenvironment that reduces the gradient for diffusion of water vapour from the leaf.
3. (a) Bioprospecting is the search for and development of useful products derived from nature.  
(b) Areas of high biodiversity are likely to be very important for bioprospecting. The high diversity means that there are a large number of potentially useful species to investigate, some undiscovered, and it is likely that many unique chemicals have evolved to deal with a high diversity, competitive environment. This includes chemicals that reduce infection or parasitism, or are used as signal to communicate between different organisms.  
(c) Bioprospecting of low diversity areas is unlikely to yield large numbers of new chemicals or materials because the number of species is lower and the systems are simpler. However, it may produce ways to deal with pollution or low nutrient environments, depending on the reason for the low biodiversity.
4. (a) There is a mutualistic relationship.  
(b) The growth rate of both microbes A and B increases when they are together.

## 159. Why Cells Need to Divide (page 223)

1. (a) Cell division is responsible for growth in an embryo by increasing the number of cells. After birth, cell division continues to be important in growth until the adult size is reached.  
(b) Cell division replaces old or damaged cells with new ones (e.g. grazed skin). Other examples include the production of new cells to repair a broken bone or regeneration of a new limb in some organisms.  
(c) Some unicellular eukaryotes reproduce asexually using cell division. For example, baker's yeast reproduces by budding, producing a genetically identical daughter cell.

## 160. Binary Fission in Prokaryotes (page 224)

1. Binary fission is a form of asexual reproduction resulting in the division of a cell into two identical cells.
2. The cross wall is an in-growth of the cell wall and cell membrane of a cell dividing by binary fission. When its growth is completed, the cross wall completely divides the cell, resulting in the formation of two cells.
3. The generation time is the time taken for a population of bacterial cells to double.
4. Completed table below:
 

Min	No.	Min	No.	Min	No.
0	1	140	128	260	8,192
20	2	160	256	280	16,384
40	4	180	512	300	32,768
60	8	200	1,024	320	65,536
80	16	220	2,048	340	131,072
100	32	240	4,096	360	262,144
120	64				
5. (a) 8                      (b) 512                      (c) 262,144





The sport may be planted directly into the ground or micro-propagation can be used to produce many clones all at once.

## 167. Grafting (page 234)

- Grafting involves joining a scion (cutting) from one individual to another growing plant (the rootstock). A graft incorporates the (favourable) features of two individual plants.
- The scion is the part grafted onto the root stock - i.e. it is taken from a donor plant and attached to a new plant. The root stock is the plant to which the scion is added. It may be just a short part of the stem and the roots of a larger plant.
- Two (or more) plants can be joined together. One will be the root stock which will have its own desirable traits, e.g. resistance to root diseases. The other will be the scion, which will have different desirable qualities such as high fruit yield and quality. It is possible to graft multiple similar fruits together e.g. multiple types of pear on the same plant.
- Grafting of multiple plant types together can be useful as it means that two plant varieties bred specifically for different properties can be brought together without having to produce a hybrid by conventional means (which might lose aspects of the desirable properties and may not even be successful).

## 168. Micropropagation (page 235)

- The purpose of micropropagation is to produce many new plants from a small amount of original plant material.
- A callus is a mass of undifferentiated cells.
  - Several plant hormones are added to the culture in sequence. These will stimulate each phase of plant development.
- Compared with traditional propagation methods, micropropagation has a number of advantages. It enables the production of many clones from a single seed/explant and allows the selection of desirable traits directly from culture. It facilitates rapid propagation, with no wait for seed production and is ideal for plants with long generation times, low seed production, or seeds that are difficult to germinate. Micropropagation also facilitates the international exchange of plants without quarantine and allows researchers to eliminate plant diseases from propagation lines. It is also space saving and overcomes seasonal restrictions to propagation. However, there are several disadvantages to this technology. It is very labour intensive and trial and error is necessary to determine the ideal culturing conditions. Cultured plants may be genetically unstable/infertile and, over time, there may be a loss of genetic diversity.

## 169. Asexual Reproduction by Spores (page 236)

- A spore is an unicellular, haploid reproductive unit produced for dispersal.
- The purpose of spores in fungi is for dispersal and reproduction of genetically identical fungi.
- Spores are formed in sporangia that develop on upright hyphae.
- Endospores develop to survive unfavourable conditions, such as heat or drought.

## 170. Case Study: Cloning in Horticulture (page 237)

- Advantages** of cloning include:
  - Ability to produce large numbers of a particular variety quickly while preserving the genetic identity of that variety.
  - Cloning produces plants that yield predictable results. Barring mutation and environmental effects, there is no chance of unwanted variation.
  - Cloning is very important for the continuation of plants that are infertile or produce seedlings that differ from the specifically bred parent.
- Disadvantages** include:
  - Being identical there is no variation in immunity (except by chance mutation). In the case of disease (e.g. Panama disease in bananas and blight in potatoes) all plants will

- be affected in the same way.
- If all plants are clones breeding new varieties of the crop can be extremely difficult.
- Cloning is often labour intensive and costs can be high.

## 171. KEY TERMS: Did You Get It? (page 238)

- asexual reproduction (D), clone (E), cutting (A), grafting (F), micropropagation (C), scion (H), spore (G), vegetative propagation (B)
- Fission involves mitosis and cytokinesis and the cell splitting in two.
  - Fragmentation involves an organism splitting into two or more fragments, each of which develops into a new organism.
  - During budding, a small part of a parent organism separates as a bud, which then develops as a new individual (either remaining attached to the parent or separating as an independent organism).
- Cuttings are produced by taking a part of the plant and growing it into a new plant. Grafting involves taking a part of one plant and attaching it to a second plant (the root stock) where it will grow as part of that second plant.
- Haploid spores in plants are produced by meiosis as part of the sexual life cycle. Haploid spores in fungi are produced by mitosis as part of asexual reproduction (although fungi may also produce sexual spores). Bacterial endospores are produced by copying the original DNA and enclosing in a resistant casing. The parent cell then breaks down. Spores in plants and fungi are for dispersal whereas bacterial endospores are primarily for surviving unfavourable conditions.
- Micropropagation involves producing many clones of a small amount of plant material. It is possible because of the totipotent nature of plant tissue. It allows a large number of identical high value plants to be produced quickly and ensures a known outcome, so it is often used in plantation species or for propagating endangered plant species. However, as all the plants are clones, there is a risk of loss of genetic diversity and susceptibility to new diseases, and the set up and production costs can be high.

## 172. Cell Division (page 240)

- Mitosis occurs in body cells (somatic cells) in animals (and in meristematic tissue in plants).
  - Mitosis is responsible for growth of an organism, and repair and replacement of damaged cells.
  - False
- Meiosis occurs in sex organs in animals (ovaries and testes).
  - It produces sex cells (gametes or eggs and sperm) for the purposes of sexual reproduction.
  - True

## 173. Meiosis (page 241)

- In the first division of meiosis, homologous pairs of chromosomes pair to form bivalents. Segments of chromosome may be exchanged in crossing over and the homologues then separate (are pulled apart). This division reduces the number of chromosomes in the intermediate cells, so that only one chromosome from each homologous pair is present.
- In the second division of meiosis, chromatids separate (are pulled apart), but the number of chromosomes stays the same. This is more or less a 'mitotic' division.

## 174. Meiosis and Variation (page 242)

- Independent assortment refers to the random distribution of maternal and paternal homologues to the gametes. This results in  $2^n$  possible combinations of maternal and paternal chromosomes in gametes, where  $n$  is the haploid number, i.e. a large number of possible combinations.
- Crossing over is the mutual exchange of pieces of



chromosome (alleles) between non-sister chromatids of homologous chromosome pairs.

- (b) Crossing over increases variation by creating new combinations of alleles on the chromosomes involved in the crossing over. The more crossing over incidents, the greater the variation.

**175. Modelling Meiosis (page 243)**

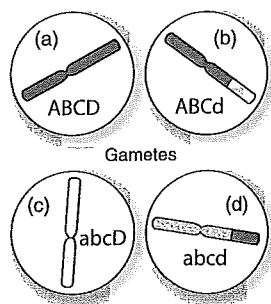
The genotype/phenotype of the offspring students obtain will depend on their own phenotypes.

**176. The Advantages of Sexual Reproduction (page 245)**

1. Sexual reproduction produces genetic variation in the offspring by the union of genetically variable gametes from different individuals.
2. Variation allows populations to adapt to a changing environment. While some individuals may be adversely affected by change others may not, or may be advantaged, allowing the population to continue.
3. Sexual reproduction produces variation in phenotypes, including variation in immune responses and susceptibility to disease. In any sexually reproducing population, individuals will have different susceptibility to different pathogens. Constant recombining of the genes associated with successful immune responses (i.e. genes for resistance) through sexual reproduction means that the population is able to adapt to the challenges presented by pathogens.

**177. KEY TERMS: Did You Get It? (page 246)**

1. Meiosis
2. Meiosis produces gametes (in animals) for sexual reproduction (in plants, meiosis produces haploid spores).
3. (a) Crossing over, in which genetic material is exchanged between non-sister chromatids of homologous chromosomes, and independent assortment, in which homologous chromosomes align randomly at the equator of the cell before being separated.  
(b) Crossing over allows chromosomes to exchange genes and produce more variation (recombination). Independent assortment means that each chromosome pair aligns at the equator (and then separates) independently of all other pairs so there are many possible combinations of chromosomes possible.
- 4.



5. B
6. B

**178. What Are Stem Cells? (page 248)**

1. (a) Potency - ability to differentiate into other cell types.  
(b) Self renewal - ability to maintain an unspecialised state.
2. (a) Totipotency: The ability to differentiate into any cell in the organism.  
(b) Pluripotency: The ability to differentiate into any cell except extra-embryonic cells e.g. the placenta.  
(c) Multipotency: Ability to differentiate into a limited number of cells related to the tissue of origin (e.g. blood cells).
3. Embryonic stem cells are pluripotent and can form over

220 cell types in the three primary germ layers (ectoderm, endoderm, mesoderm). Adult stem cells are multipotent and can divide only into a limited number of cell types, e.g. those of the blood, bone, epithelium.

4. Embryonic stem cells have vast potential for medical application as they can differentiate into many cell types and therefore, theoretically, be used to replace most damaged cell and tissue types. The medical use of adult stem cells is more limited than ESC but still valuable for repair or replacement of certain cell types (e.g. blood).
5. Embryonic stem cells require the creation of an embryo. This creates ethical problems associated with the creation and planned sacrifice of a life (the embryo) for medical research or therapeutic applications.
6. The iPSCs behave in the same way as embryonic stem cells. Induced pluripotent stem cells (iPSCs) have fewer ethical issues because they can be obtained without the need to create and then destroy embryos.

**179. Applications of Stem Cells (page 250)**

1. The stem cells from the donor may not be immunologically compatible and may be rejected by the recipient's immune system.
2. If stem cells that still carry a defective gene are placed into the patient, there will be no benefit. The disease will not be corrected because the stem cells carry the same genetic defect.
3. If the disease is the result of a genetic defect, the defective gene will also be present in the umbilical cord blood. If the genetic defect is uncorrected, the treatment will not be effective.
4. (a) Stem cells can be cultured to develop into retinal pigment epithelium cells, which can be injected into the eye to replace damaged cells.  
(b) The patient's cells will carry the defect which will reappear in the new cells, so the treatment will not be effective.  
(c) Using a patient's own cells means that there is no chance of immune rejection of the tissue. This is useful when replacing an organ damaged by a non-genetic disease.
5. (a) Type 1 diabetes results from the body's immune system destroying the insulin-producing cells of the pancreas so that no insulin is produced and, as a consequence, the cells cannot take up glucose.  
(b) Stem cells taken from donors who do not have diabetes are induced to develop into insulin producing cells. They are then transplanted into the patient.

**180. Stem Cells Give Rise to Other Cells (page 252)**

1. (a) A neutrophil develops from a multipotent red bone marrow stem cell into a myeloid precursor cell, and finally into a neutrophil.  
(b) A T lymphocyte develops from a multipotent red bone marrow stem cell, into a lymphoid stem cell. This stem cell then migrates to the thymus where it develops into a T lymphocyte.
2. 7
3. Differentiation is controlled by growth factors.
4. The daughter cells may remain as stem cells (self renewal) or become committed to developing into a new cell type.
5. The zygote is the ultimate stem cell because it is not only the first cell in a developing organism, it is able to develop into all cell types of the body.

**181. Stem Cells and Prenatal Development (page 253)**

1. Four days
2. The gastrula forms from the infolding (invagination) of the blastula.
3. Triploblastic means there are three layers.
4. (a) Endoderm gives rise to the embryonic gut (and subsequently the digestive system), glands, and part of



the respiratory system.

- (b) Mesoderm gives rise to the muscles, circulatory system, excretory system, dermis, skeleton, and connective tissues.
  - (c) Ectoderm gives rise to the epidermis, the nervous system, tooth enamel, some endocrine glands, and the cornea and lens of the eye.
5. (a) The development of the organs and limb buds.  
(b) Growth of the body's organ systems in preparation for birth.
  6. The cells of the gastrula can only develop into specific cells related the germ layer from which they arose, e.g. mesoderm cells can develop into muscle cells but not skin cells.
  7. At the early stages of development, tissues and organs are still developing (differentiation of cells) rather than just growing larger. Exposure to harmful substances can cause errors in the developmental pathways so that the tissues and organs do not form in the normal way.

### 182. Regulation of the Cell Cycle (page 255)

1. Cell checkpoints regulate the progression of the cell to make sure it is ready to divide, and to ensure that the cell divides correctly and in step with the rest of the tissue around the cell.
2. (a) The metaphase checkpoint makes sure the chromosomes are correctly attached to the spindle fibres.  
(b) If the chromosomes are not correctly attached then the chromosome may not separate correctly.
3. If the cell cycle was not regulated cells would go into uncontrolled cell division, resulting in the formation of tumours. Cycle regulation also ensures that the cell reaches the appropriate size and volume to divide successfully.

### 183. Disrupting the Cell Cycle (page 256)

1. (a) A mutagen is any substance that can cause a mutation (change in the base sequence) in the DNA.  
(b) Some common mutagens include ionising radiation, benzene, viruses, and tobacco smoke.  
(c) Carcinogens cause cancer. They cause mutations in proto-oncogenes, forming oncogenes - genes that cause cancer.
2. A tumour forms when a cells DNA mutates, causing the cell to ignore its checkpoints and divide in an uncontrolled manner. Some tumours may be benign, but if they invade surrounding tissue they become malignant (cancer).
3. (a) Genetic predisposition can increase the chances (risk) of a disease forming.  
(b) The risk could be offset by following a lifestyle that reduces exposure to the things that might trigger the disease e.g. not smoking if there a greater risk of a cancer - especially lung cancer.
4. Cancerous cells have lost control of the genetic mechanisms regulating the cell cycle so that the cells become immortal. They also lose their specialised functions and are unable to perform their roles.
5. The cell cycle is normally controlled by two types of gene: proto-oncogenes, which start cell division and are required for normal cell development, and tumour-suppressor genes, which switch cell division off. Tumour suppressor genes will also halt cell division if the DNA is damaged and, if the damage is not repairable, will bring about a programmed cell suicide (apoptosis).
6. Controls over the cell cycle can be lost if the proto-oncogenes or the tumour suppressor genes acquire mutations. Mutations to the proto-oncogenes (and formation of oncogenes), results in uncontrolled cell division. Mutations to tumour-suppressor genes result in a failure to regulate the cell repair processes and a failure of the cell to stop dividing when damaged.

### 184. KEY TERMS: Did You Get It? (page 258)

1. cancer (D), cell cycle (E), multipotent (F), pluripotent (C), stem cell (A), totipotent (B), zygote (G)
2. (a) Adult stem cell, e.g. cells from the bone marrow or gastrula germ layers.

- (b) Embryonic stem cells - cells from the blastula.
- (c) The zygote.

3. Stem cells can give rise to many cell types that make tissues and organs.

Self renewal is the ability to divide many times while maintaining an undifferentiated state.

Embryonic stem cells are derived from the inner cell mass of the blastocyst.

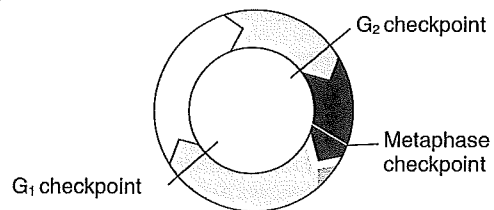
Gastrulation occurs when the blastula develops an infolding after about 16 days.

The cell cycle is controlled by three checkpoints, the G<sub>1</sub>, G<sub>2</sub>, and metaphase checkpoints

A mutation is any change in the DNA sequence, e.g. changing G to C.

A mutagen is any substance that causes a mutation in the DNA.

- 4 (a)



- (b) **G1 checkpoint:** checking the cell is large enough, that there are sufficient nutrients available, and that the correct signals from other cells have been received.

**G2 checkpoint:** checking that the cell is large enough, that replication of the chromosomes has been completed correctly, and that the proteins required for mitosis have been synthesised.

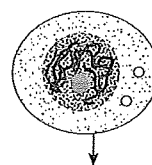
**Metaphase checkpoint:** checking that the chromosomes are correctly attached to the spindle fibres.

### 185. Review: Unit 2 Area of Study 1 (page 259)

No model answer. Summary is the student's own.

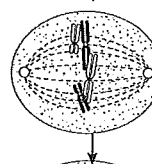
### 186. Synoptic Question: Unit 2 Area of Study 1 (page 261)

1. In prokaryotic binary fission, the circular chromosome is replicated and the cell elongates. The cell membrane grows inward and a cross wall forms. The cell then splits in two forming two new cells.
2. (a) Mitosis divides replicated genetic material into two identical cells for the purposes of growth, repair, or reproduction.  
(b) Presented as portrait view for reasons of space.



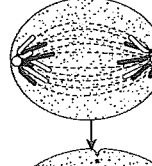
#### Prophase

- Nuclear membrane breaks down.
- Chromosomes condense.
- Centriosomes move to opposite poles.
- In late prophase, spindle forms and chromosomes attach to the spindle.



#### Metaphase

- Chromosomes (appearing as joined chromatids) align at the equator of the cell, attached to the spindle fibres by their centromeres.
- Some spindle fibres span the cell.



#### Anaphase

- Spindle fibres attached to chromatids shorten and chromatids are pulled apart.



#### Telophase

- Two nuclei form, a furrow forms across the middle of the parent cell.
- Chromosomes unwind.



3. (a) Asexual reproduction is the production of offspring without recombining chromosomes in meiosis. It produces genetically identical offspring.  
 (b) There are some simple animals that can carry out asexual reproduction, e.g. flatworms. Flatworms (planarians) carry out asexual reproduction by fragmentation. The animal may spontaneously fragment to produce new animals. The advantage of this is that the population can quickly increase to take advantage of favourable environmental conditions (e.g. a good food source).
4. (a) Vegetative propagation is the production of new individuals in plants without the need for seeds or spores.  
 (b) Vegetative propagation through cuttings can prevent plants from becoming extinct by providing a way to rapidly increase the number of plants. Although this does not increase genetic variation, it increases population numbers so that the risk of species loss is reduced and gives time for (1) reproduction by seeds if possible, or (2) favourable mutations to arise.
5. (a) The purpose of meiosis is to halve the chromosome number in sex cells and allow recombination of genetic material during the formation of gametes (or haploid spores in plants).  
 (b) During meiosis the cell undergoes two divisions to produce a cell with half the diploid chromosome number. These haploid cells will have new combinations of genes. Meiosis involves one cell division and produces cells genetically identical to the parent cell.  
 (c) Meiosis increases variation by crossing over between homologous chromosomes in late prophase I and independent assortment of chromosomes during metaphase I. Crossing over is the process in which homologous chromosomes may exchange genetic material. Independent assortment increases variation by allowing homologous pairs to line up independently of each other before separating into new cells.
6. Stem cells have the potential to be important medical tools because they can differentiate into multiple cell types. Adult stem cells can only differentiate into a limited number of cell types based on the tissue of origin. Embryonic stem cells (ESC) can differentiate into any type of somatic cell and so have a much wider range of possible applications. Applications of stem cells include replacement of damaged or diseased tissue and organs. This can be done by inducing the stem cells to develop into a certain cell type. These cells are then transplanted into a patient to correct malfunctioning cells or repair diseased tissue. Apart from technical difficulties associated with stem cell culture, there are several ethical issues associated with the use of stem cells, including their long term stability in the body (will they cause a worse kind of disease?). The use of ESC in particular is contentious because the cells are typically harvested from embryos, either donated or created for the express purpose of providing stem cells for harvest. Many people see this as creating a human (or other animal) while all the time intending to kill it, which they find morally objectionable regardless of the medical value of the ESC obtained.

**187. Genomes, Genes, and Alleles (page 264)**

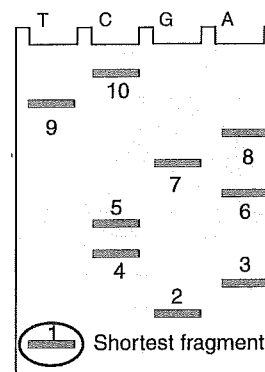
1. (a) All the genetic material in one haploid set of chromosomes.  
 (b) Genes are sections of DNA that code for proteins.  
 (c) An allele is a version of a gene. The allele will cause the production of a protein that varies slightly from one produced by a different allele.
2. In general the genome size increase from viruses to bacteria to eukaryotes,
3. Viruses and bacteria are haploid, with only one chromosome (excluding plasmids). Therefore they can only have one allele per gene. Because eukaryotes are diploid (two sets of chromosomes) they can have two different alleles for a gene. Teacher's note: some eukaryotes, especially plants and fish, can have more than two chromosome sets, e.g. triploid.

**188. Genome Sizes (page 265)**

- 1 (a) 49.67 times bigger  
 (b) 652.2 times smaller  
 (c) 18,750 times smaller
2. Large genomes require a lot of resources to replicate, thus reproduction and growth rates are low and this increases the risk of extinction.
3. Sequencing the mouse genome has given many useful insights in how genes behave and has advanced the development of medicines and treatments for genetic disease.
4. Sequencing the DNA of major crop plants gives information on the genetic basis of important characteristics, such as growth, yield, and disease resistance. This information can be used to improve productivity and breeding programmes.

**189. What is DNA Sequencing? (page 266)**

1. The purpose of DNA sequencing is to find out the sequences of bases in the DNA fragment.
2. (a) and (b)



(c) T G A C C A G A T C

3. (a) The modified nucleotides stop the growth of the DNA fragment.  
 (b) 1% modified DNA is enough to produce terminated DNA fragments but is small enough to allow other non-modified DNA to be incorporated in the DNA fragment. Too much modified DNA would cause termination of DNA replication too early in too many cases.
4. A reaction vessel is needed for each modified base. If all the modified bases were put in the same vessel there would be no way of distinguishing between them once placed on an electrophoresis gel.

**190. What was the Human Genome Project? (page 267)**

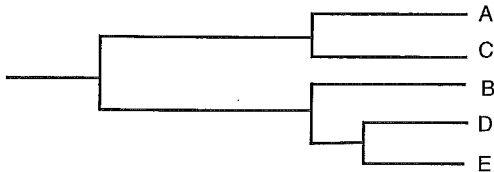
1. The HGP aimed to map the entire base sequence of every chromosome in the human cell (the human genome) to identify all genes in the sequence, determine what the gene products were (proteins produced), and determine the precise role of every gene on the chromosomes.
2. (a) Gene mapping is finding the location and role of specific genes on chromosomes.  
 (b) Gene mapping can help in understanding how heritable diseases are passed on. This helps in gene counselling and possible treatment of disease.  
 (c) Gene mapping can help locate genes that may play a role in cancer. It also helps in understanding mutations to those genes and the effect of these on the development and progression of cancer in different people.

## 191. DNA Differences Between Species (page 268)

- (a) Cow synthesised DNA:  
TGATTGTAAGCTTTTCAGGGTGGGTGATTA  
Sheep synthesised DNA:  
TAGTTGTAGGCTTTTTGGGTGGGTGATTA  
Goat synthesised DNA:  
TGGTTGTAGGCTTTCTGGGTGGGTAATTA  
Horse synthesised DNA:  
TGTTTGTAGGCCTTTAGAGTGGGTGATTA

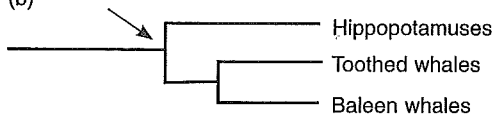
- Sheep and goat (3 differences)
- Goat and horse (6 differences)

2.



## 192. Using DNA to Determine Species Relatedness (page 269)

- Bioinformatics has allowed scientists to store and quickly access and analyse many different DNA sequences. This has led to greater understanding of evolutionary relationships.
- (a) Hippopotamuses  
(b)



- Evidence shows pigs branched off much earlier and are related to peccaries

## 193. Determining Gene Function Using Knockouts (page 270)

- Gene knockout produces a mouse with a non-functional gene. The effect of this can be examined and so used to determine what the gene does.
- Applications include using mice as models for genetic disease e.g. cystic fibrosis and diabetes, and to develop and screen drug treatments for human disease.
- Mice have many of the same genes as humans. They can be bred quickly and do not require large amounts of space.

## 194. Hunting for a Gene (page 271)

- The physical effects of Huntington's disease are shaking of hands and/or limbs and an awkward gait. More severe effects include the loss of muscle control and mental function leading to dementia.
- The mHTT gene was discovered using information from the family history of 10,000 people. Using a probe called G8, a map of the 4th chromosome was built up and each gene sequenced. The mHTT gene was shown to be one with a trinucleotide repeat expansion.
- HD is caused by a trinucleotide repeat expansion of the sequence CAG on the 4th chromosome. Repeats of over 35 cause the disease and the greater the number of repeats the more severe the disease. Because of the instability of the mHTT gene the number of repeats and severity of the disease tends to increase over generations.

## 195. Screening for Genes (page 272)

- (a) A positive test for a disease-causing gene can help early detection because it alerts the patient to their increased risk of developing the disease. This means the patient and doctors can look out for the signs of the disease and, if it arises, take early action to reduce its effects.  
(b) Advantages include being able to take steps to prevent or reduce the effect of the disease. This includes changing lifestyle or beginning preventative drug therapies.
- The family members may want to be screened to see if they have the gene associated with the disease and, if so, the risk of them passing it on to their offspring. This will help them to decide when or if to start a family.

## 196. Genomic Analysis and Disease Risk (page 273)

- (a) Sequence variations fall into distinct types that can be matched to a person's risk of disease. Different sequence variations may be associated with greater risk of disease or effect of a drug.  
(b) The frequency of DNA variations in a group of people with and without a disease are analysed to see if there is any significant differences in variations between the groups.
- Genomic analyses can provide probabilities for successful treatment outcomes, i.e. for a given genetic profile, a particular treatment will have a certain probability of being successful. This helps match drugs to a person's personal genetic profile based on their sequence variations.

## 197. KEY TERMS: Did You Get it? (page 274)

- allele (C), DNA sequencing (H), gene (D), gene mapping (E), genetic screening (A), gene therapy (G), genome (F), Human Genome Project (B).
- (a) See diagram below:  
(b) 6  
(c) 8  
(d) The ostrich and the emu  
(e) They have the least number of differences between their DNA sequences.  
(f) The photographs support the evidence. Emus and ostriches are morphologically similar (tall, long legs and necks, large body and similar feet).

## 198. Eukaryote Chromosome Structure (page 276)

- In the nucleus.
- DNA is a very long molecule. If it was not packaged up it could not fit into the nucleus.
- DNA wraps around the histone proteins, taking up less space that if it was spread out.
- Coiling the chromosome into tight structure helps the cell maintain order and ensures proper segregation of the chromosomes during mitosis and cell division.

## 199. Species Have Different Chromosome Numbers (page 277)

- (a) 2 (humans 46, chimpanzees 48)  
(b) Horse roundworm  
(c) Potato
- Many eukaryotes that are closer related have very different chromosome numbers. For example donkeys have 62 chromosomes while zebras have 44, even though they are in the same genus *Equus*.

Ostrich	ATGGC	C	CCCAAC	C	ATTC	GAAATC	G	CACCC	C	CTGCT	C	AAAATT	TATCAAC
Emu	ATGGC	C	CCTAAC	C	ATTC	GAAATC	C	CACCC	T	CTACT	C	AAAATT	CATCAAC
Turkey	ATGGC	A	CCCAAT	A	TATC	GAAATC	A	CACCC	C	TATTA	A	AAAACA	ATCAAC

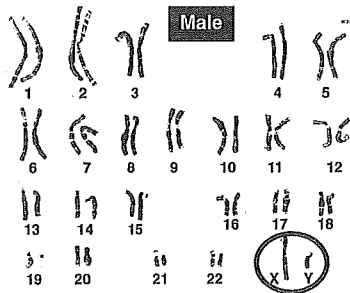
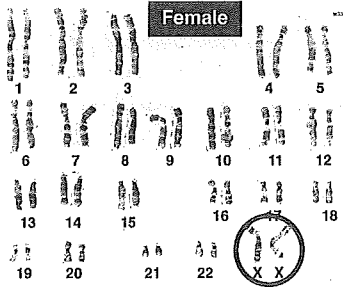


- The presence or absence of the Y chromosome. XX = female  
XY = male.
- The X chromosome has many genes that are not present on the Y chromosome, therefore an allele on the X chromosome will have no corresponding allele on the Y to override it.

**200. Karyotyping (page 279)**

- (a) A karyogram is the physically laid out image (i.e. a picture) of the karyotype. The karyotype is the complement of a cell or organism, characterised by the number, size, shape, and centromere position of the chromosomes.  
(b) It can provide information on sex and chromosomal abnormalities.

2. and 3:



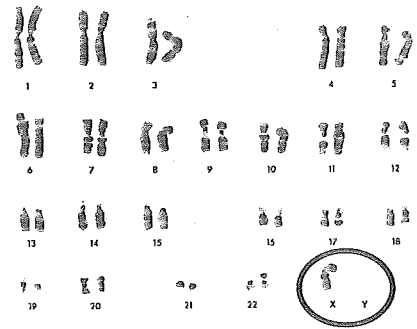
- (a) Female autosomes: 44 Sex chromosomes: XX  
(b) Male autosomes: 44 Sex chromosomes: XY
- (a) Chromosomes in human somatic cell: 46  
(b) Chromosomes in human sperm or egg cell: 23
- Autosomes are non-sex chromosomes that occur as 'matched' homologous pairs. Sex chromosomes (also called heterosomes) are the pair of chromosomes (XX in female humans and XY in male humans). Autosomes are not involved in determining sex, whereas the sex chromosomes are.
- Features include shape, size, centromere position, length of the arms, and banding pattern.
- They are of different size, shape, and banding pattern (and gene arrangement).

**201. Abnormal Chromosome Numbers (page 280)**

- The failure of chromosome to separate correctly during meiosis.
- (a) Klinefelter syndrome:



Turner syndrome:



- (b) 2N -1 Female  
(c) 2n+1 Male

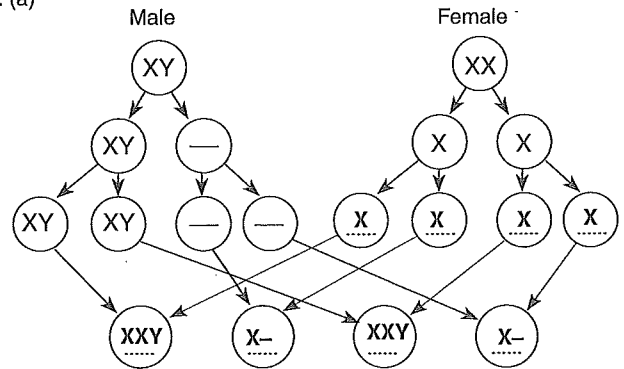
- The extra chromosome is tolerated because it only carries a few genes. Although extra copies cause abnormalities they are usually not serious enough to be lethal.
- Assume fertilisation with a normal (n = 2) gamete
  - 2 + 2 = 4 normal
  - 2 + 2 = 4 normal
  - 1 + 2 = 3 abnormal
  - 3 + 2 = 5 abnormal

**202. Making a Karyogram (page 281)**

- The karyotype should be organised as for the typical layout of a human karyotype on page 279, but it has one extra chromosome 21 (trisomy 21).
- (a) Sex: Male  
(b) Abnormal  
(c) 45 + XY (trisomy 21 or Down syndrome).

**203. KEY TERMS: Did You Get It? (page 284)**

- aneuploidy (H), autosome, (I), chromatid (B), chromosome (A), karyogram (G), karyotype (C), non-disjunction (E), polysomy (D), sex chromosome (F).
- (a) Sex (i) female (ii) female  
(b) (i) abnormal (trisomy 18) (ii) abnormal (trisomy 13)
- (a)



- (b) 0  
(c) (i) 2 n + 1, XXY  
(ii) 2 n - 1, X- (also written as XO)

**204. Alleles (page 286)**

- (a) Heterozygous: Each of the homologous chromosomes contains a different allele for the gene (one dominant and one recessive).  
(b) Homozygous dominant: Each of the homologous chromosomes contains an identical dominant allele.  
(c) Homozygous recessive: Each of the homologous chromosomes contains an identical recessive allele.
- (a) Aa (b) AA (c) aa
- Each chromosome of a homologous pair comes from a



different parent: one of maternal origin, one of paternal origin (they originated from the egg and the sperm that formed the zygote). They contain the same sequence of genes for the same traits, but the versions of the genes (alleles) on each chromosome may differ.

- Alleles are different versions of the same gene that code for the same trait. Different alleles provide phenotypic variation for the expression of a gene. There are often two alleles for a gene, one dominant and one recessive. In this case, the dominant allele will be expressed in the phenotype. Sometimes alleles for a gene can be equally dominant, in which case, both alleles will be expressed in the phenotype. Where three or more alleles for a gene exist (multiple alleles), there is more phenotypic variation in the population (for that trait) than would be the case with just two alleles.

## 205. Dominant and Recessive Traits (page 287)

- A trait is a variant of a phenotypic character e.g. brown eye colour, blonde hair colour.
  - True breeding (usually for a particular trait) refers to organisms that, when bred together (or self-crossed), always produce offspring with the same phenotype (with respect to that trait).
- 3:1
  - The wrinkled seed trait must have been masked by the smooth seed trait (i.e. it was recessive).

## 206. Influences on Phenotype (page 288)

- Sources include different alleles, sexual reproduction (recombination, independent assortment, random fertilisation), single nucleotide variations, and mutations.
  - Sources include nutrition, physical environment (e.g. temperature), drugs etc.
- Because the environment also plays a role in the phenotype identical twins will always be slightly different. This includes fingerprints, height, and weight. Depending on the extent of the environment's influence, these differences may range from very slight or more obvious.
- Behaviour (or components of it) can often be modified based on experience (learning). This means that the behavioural response can be dependent on the situation and the environment. For example, an animal can learn utilise new food sources in different environments.
- There are many examples of this. One is the growth of the queen bee. A honey bee larva becomes a queen if fed royal jelly. If not it becomes a worker. Other examples include temperature dependent sex determination in reptiles.

## 207. Mutations can Alter Phenotype (page 289)

- A mutation is a change in the DNA base sequence caused by a DNA copying error or by a mutagen.
- A person heterozygous for the mutation would not be affected (only people with the two recessive alleles are deaf).
- Being able to digest lactose in milk gave another energy source for ancient humans and allowed access to the extra nutrients in milk e.g. calcium.

## 208. Environment and Phenotype (page 290)

- Examples will vary based upon student's own choice:

The phenotype is the product of an organism's genotype (genetic make up) and the environmental influences to which it is subjected. Sometimes, the influence of the genotype predominates, e.g. in albinism, lack of pigment caused by a recessive mutation results in the albino phenotype. Some characteristics, skin colour, are determined by many genes (polygeny) and are further influenced by environment, e.g. degree of sun exposure. In both plants and animals, the full expression of the genotype (the genetic potential) is often constrained by growing conditions or nutrition.

Examples include:

- Trees that are grown at high altitude are stunted in growth.
  - Trees exposed to a strong prevailing wind appear distorted in shape (windswept).
  - Poor nutrition in infancy can retard brain and bone development.
  - Comb shape in chickens depends on what combination of alleles they inherit.
  - The colour of many flowers is determined by the dominance of one allele over another.
  - Skin colour in humans is the result of at least six allele combinations and the influence of environment.
- The helmet and spines develop as a response to chemicals released by the *Daphnia's* predator, the phantom midge larva.
    - The helmet and spines make the *Daphnia* more difficult to attack (handle and consume).
  - These are the cooler parts of the body. Body heat is lost from these areas which make it cool enough for the enzyme responsible for colour-pointing to remain active.
  - In hydrangeas, soil pH alters flower colour. In alkaline soils they are pink or red-purple, in acid soil the flowers are blue. The colour is due to the presence or absence of aluminium compounds in the flowers, and aluminium in the soil is only accessible to plants when soil pH is low. When aluminium is present within the plant, the flowers are blue. When the aluminium is absent, the flowers are pink. Note: Growers may apply aluminium sulfate to the soil to lower the pH and produce blue flowers. Adding lime produces pink flowers.
  - To ensure genetic potential is reached, provide the optimum growth conditions for that plant, e.g. suitable water availability (water regularly), adequate nutrient supply (fertilizer application), sufficient sunlight (keep out of shade), equable temperature (warmth, protect from frost and wind), protection from pests and diseases (companion plant or spray for pests).
  - A cline is a continuous, or nearly continuous, gradation in a phenotypic character within a species, associated with a change in an environmental variable such as temperature or wind.
    - Physical factors include: temperature, wind speed (faster at higher altitude), soil depth and organic content (less at higher altitude), nutrient levels (likely to be less at higher altitude) humidity and air pressure (both less at altitude).
    - Plant A: The observed phenotype (prostrate) of this species is not due to genetic factors, but to the effect of climate on growth. In the absence of a harsh environment, the plant reverts to its normal growing habit.  
Plant B: The low growing phenotype of this species is controlled by genes (not environmental factors). Species B continued to be low growing.
    - Plant A is most likely to show clinal variation.

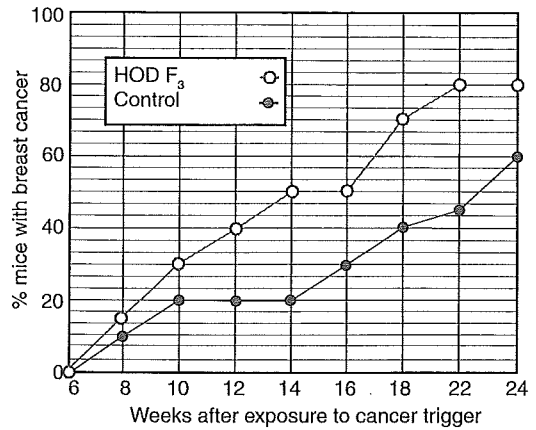
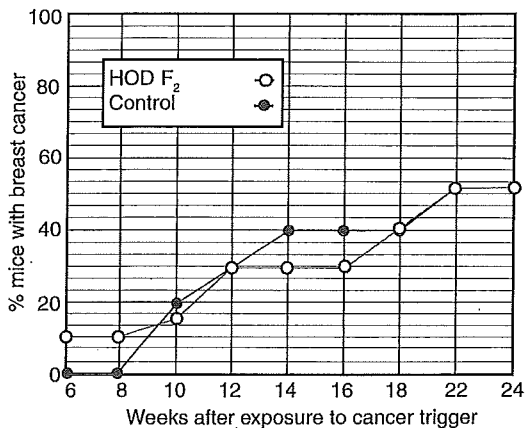
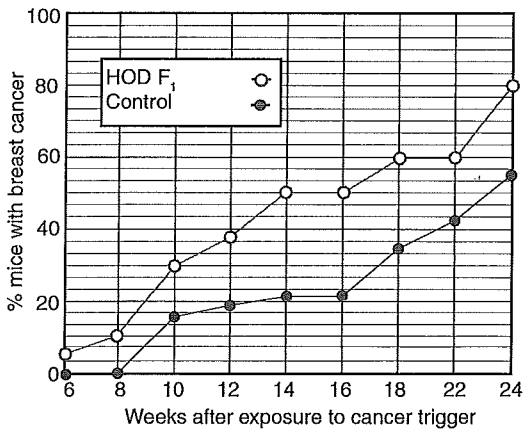
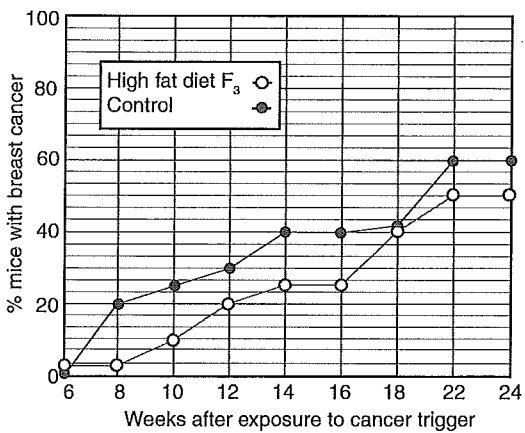
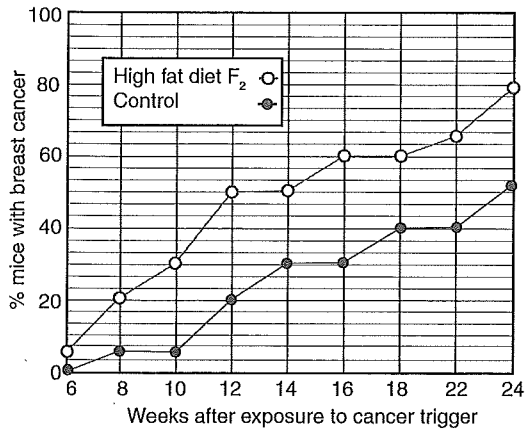
## 209. Gene Expression and Phenotype (page 292)

- DNA methylation is the adding of a methyl group (-CH<sub>3</sub>) to a DNA base (usually cytosine).
  - Methylation of cytosine turns off gene expression, so that the gene is not transcribed.
  - Methylation allows the developing embryo a way to rapidly respond to environmental changes (both inside and outside the uterus). Methylation also makes sure that each generation of cells develops into the correct cell type (by inheriting methylated DNA).
- The pregnant mother and the embryo are in constant close contact, primarily at the placenta but also in the amniotic fluid. Chemicals ingested or absorbed by the mother can make their way through the blood and cross the placenta to the embryo. These chemicals can then affect the developing embryo by affecting gene expression. The embryo is also exposed to any environment the mother is exposed to.



210. Epigenetic Factors and Phenotype  
(page 293)

1.



2. (a) F<sub>1</sub> and F<sub>2</sub>  
(b) F<sub>1</sub>, F<sub>2</sub>, and F<sub>3</sub>  
(c) The high oestrogen diet.
3. Experiments show that the effects of a parent's diet can be passed on to many following generations. Because the changes last beyond the F<sub>2</sub> generation these is evidence that the epigenetic effect is inherited.

211. Gene, Environment, and Continuous Variation  
(page 295)

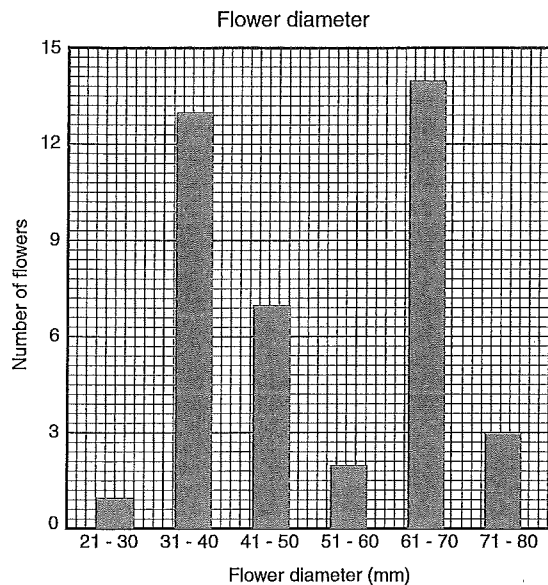
1. (a) Polygeny is the situation in which a single phenotype is controlled by more than one gene.  
(b) Many genes allows for many combinations of alleles and a greater chance for environmental affect on the genes. This will in turn lead to a wide range of variation.
2. Two phenotypes with continuous variation include skin colour and height. Environmental influences will alter the colour of a person's skin (such as tanning) and height (e.g. not enough calcium for bone development).
3. Frequency of the table is 1:6:15:20:15:6:1. Black and light/pale have the lowest frequencies (1) while medium skin colours have the highest frequency.
4. Traits with continuous variation show a normal distribution curve when sampled and a graded variation in phenotype in the population. Such phenotypes are usually determined by a large number of genes and/or environmental influence. Examples include height, weight, hand span, foot size. In contrast, traits with discontinuous variation fall into one of a limited number of phenotypic variants and do not show a normal distribution curve when sampled. Differences in the phenotypes of individuals in a population are marked and do not grade into each other. Such phenotypes are controlled by a few different alleles at a few genes, e.g. chin cleft.
5. Student's own plot. Shape of the distribution depends on the data collected. The plot should show a statistically normal distribution if sample is representative of the population and large enough.
  - (a) Calculations based on the student's own data.
  - (b) Continuous distribution, normal distribution, or bell shaped curve are all acceptable answers if the data conform to this pattern.
  - (c) Polygenic inheritance: Several (two or more) genes are involved in determining the phenotypic trait. Environment may also have an influence, especially if traits such as weight are chosen.
  - (d) A large enough sample size (30+) provides sufficient data to indicate the distribution. The larger the sample size, the more closely one would expect the data plot to approximate the normal curve (assuming the sample was drawn from a population with a normal distribution for that attribute).



## 212. KEY TERMS: Did You Get It? (page 297)

- allele (D), continuous variation (I), dominant (H), epigenetics (K), heterozygous (A), homozygous (C), homologous chromosomes (B), mutation (J), phenotypes (F), recessive (G), trait (E).
- (a) Fur colour  
(b) Black fur
- (a) Numbers may vary depending on the categories used by students to tally the flower diameter:  
21-30: 1  
31-40: 13  
41-50: 7  
51-60: 2  
61 - 70: 14  
71 - 80: 3

(b)



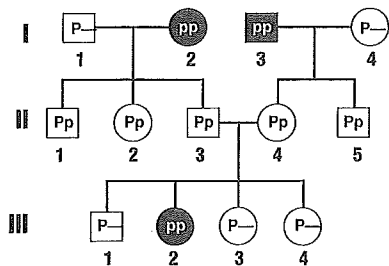
- (c) The graph has two peaks (it is bimodal). This may be because there are two different genotypes producing two ranges of flower diameter (i.e. smaller and larger), it may be due to not sampling randomly or not sampling enough flowers, or it may be due to environmental factors (e.g. more sun in one area).

## 213. Pedigree Charts (page 299)

Note: Strictly speaking, the allele for lactose tolerance is the faulty allele as this mutation occurred about 10,000 BC and causes the gene to fail to switch off. However, because it is so widespread, lactose intolerance is now often considered the 'faulty' state.

## 1. Autosomal recessive traits

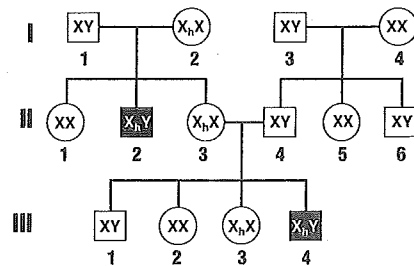
(a) Genotypes of individuals on the chart:



- (b) The genotypes of parents II-3 and II-4 have to be carriers (heterozygous) because they produced an affected offspring (homozygous recessive). Alternatively, we know that they must be carriers because each had an affected parent (I-2 and I-3).

## 2. Sex-linked recessive traits

(a) Genotypes of individuals on the chart:

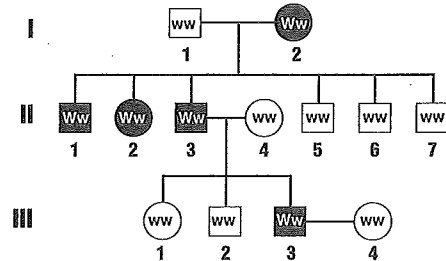


- (b) Males have only one X chromosome. If that single chromosome carries the affected allele, then it will be expressed. Males cannot be heterozygous, since they can only carry one copy of the gene.

## 3. Autosomal dominant traits

(a) Genotypes of individuals on the chart:

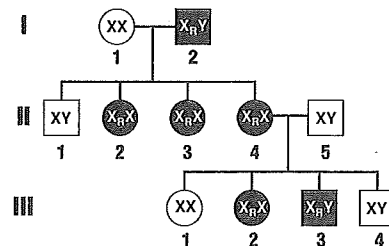
Teacher's note: Parent I-2 has to be heterozygous (Ww) since some of the offspring are normal (ww). This could not occur if I-2 was homozygous dominant (WW).



- (b) Each affected individual has an affected parent.

## 4. Sex-linked dominant traits

(a) Genotypes of individuals on the chart:



- (b) Females have two X chromosomes, and so have a greater probability that one of them will carry the affected gene.

## 5. Suggestions for interpreting a pedigree chart. Students can come up with their own list of guidelines:

If most of the males in the pedigree are affected, then the disorder is X-linked.

If it is a 50/50 ratio between men and women the disorder is autosomal.

If the disorder is dominant, one of the parents must have the disorder.

If the disorder is recessive, neither parent has to have the disorder because they can be heterozygous (carriers).

## Autosomal recessive

- Appears in both sexes with equal frequency.
- Trait tends to skip generations.
- Affected offspring are usually born to unaffected parents.
- When both parents are heterozygous, approximately 1/4 of the progeny will be affected.



**Autosomal dominant**

- Appears in both sexes with equal frequency
- Both sexes transmit the trait to their offspring
- Does not skip generations
- Affected offspring must have an affected parent unless they possess a new mutation

**X-linked dominant**

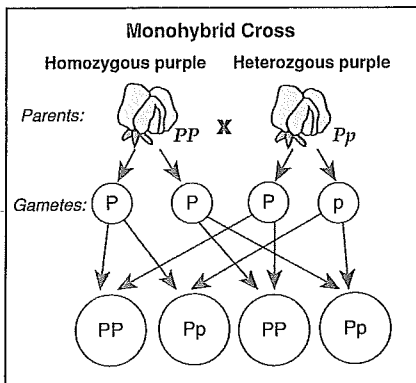
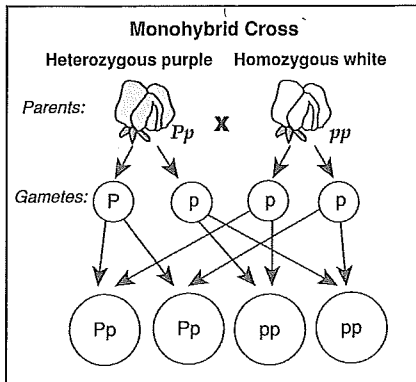
- Both males and females are affected; often more females than males are affected.
- Affected sons must have an affected mother.
- Affected daughters must have either an affected mother or an affected father.
- Affected fathers will pass the trait on to all their daughters.

**X-linked recessive**

- More males than females are affected.
- Affected sons are usually born to unaffected (carriers) mothers, thus the trait skips generations.

**214. The Monohybrid Cross (page 301)**

- All the F<sub>1</sub> generation are heterozygous and thus display the dominant flower colour. When these are crossed together a 3:1 phenotypic ratio appears so that the homozygous recessive feature will be seen again.
- 



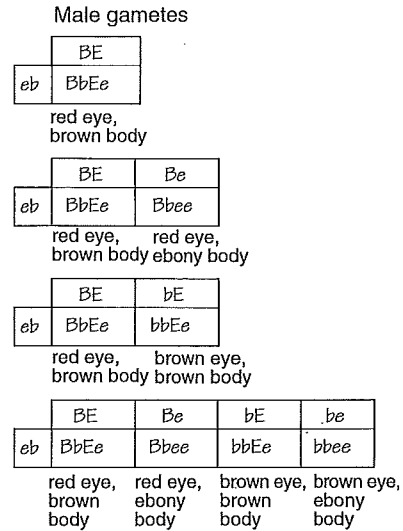
**215. The Test Cross (page 302)**

- To perform the test cross here you would have to cross the male with a female (brown eye, ebony body) homozygous recessive for both genes (bbee).

**Note:** Depending on the male's genotype the outcomes of the test cross would produce offspring that were:

- All wild type (brown body, red eye)
- Half wild type and half red eye, ebony body
- Half wild type and half brown eye, brown body
- 25% wild type, 25% red eye ebony, 25% brown eye, brown body, and 25% brown eye, ebony.

Students could present this in Punnett squares.



- A wild type male (brown or normal body and red eyes) could have one of four genotypes: BEBE, BEbE, BEbE, or BEbE
- (a) 50% are wild type and 50% are red eye, ebony so the male must BEbE.  
(b) This is the only genotype that produces this ratio of phenotypes in the offspring (as per the second Punnett square above)

**216. Sex Linkage (page 303)**

- Parental genotypes:  
Maternal X<sup>H</sup>X<sup>h</sup>      Maternal gametes X<sup>H</sup>, X<sup>h</sup>  
Paternal X<sup>H</sup>Y      Paternal gametes X<sup>H</sup>, Y  
Possible fertilisations: X<sup>H</sup>X<sup>H</sup>, X<sup>H</sup>X<sup>h</sup>, X<sup>h</sup>Y, X<sup>h</sup>Y

Male children	Genotypes	Phenotypes
	X <sup>H</sup> Y	Normal
	X <sup>h</sup> Y	Haemophilic
Female children	X <sup>H</sup> X <sup>h</sup>	Carrier
	X <sup>H</sup> X <sup>H</sup>	Normal

  - (a) Possible genotypes for the mother: X<sup>H</sup>X<sup>H</sup>, X<sup>H</sup>X<sup>h</sup>  
Possible genotypes for the father: X<sup>H</sup>Y  
Probability that she is a carrier is 25% or 1/4  
(b) If the mother is has a normal genotype (X<sup>H</sup>X<sup>H</sup>) the chance of the offspring being affected is 0%. If the mother is a carrier (X<sup>H</sup>X<sup>h</sup>) the chance of the offspring being affected is 50%. Because the genotype of the mother is unknown, the overall probability of the offspring being affected is 25%.
  - (a) Genotype X<sup>B</sup>X<sup>b</sup>  
(b) Phenotype Carrier (normal)  
(c) X<sup>b</sup>X<sup>b</sup>
  - Parent: Normal wife      Affected husband  
Parent genotype: XX      X<sup>R</sup>Y  
Gametes: X, X      X<sup>R</sup>, Y  
Children's genotypes: X<sup>R</sup>X, XY      X<sup>R</sup>X, XY  
Phenotypes: Affected girl, Normal boy      Affected girl, Normal boy
  - (a) Probability of having affected children = 50% or 0.5  
(b) Probability of having an affected girl = 50% or 0.5  
However, all girls born will be affected = 100%  
(c) Probability of having an affected boy = 0% or none
  - Parent: Affected wife      Normal husband  
Parent genotype: X<sup>R</sup>X      XY  
Gametes: X<sup>R</sup>, X      X, Y  
Children's genotype: X<sup>R</sup>X, X<sup>R</sup>Y, XX, XY  
Phenotypes: Affected girl, Normal girl, Affected boy      Normal girl, Normal boy
- Note:** Because the wife had a normal father, she must be heterozygous since her father was able to donate only an X-chromosome with the normal condition.

- (a) Probability of having affected children = 50% or 0.5  
 (b) Probability of having an affected girl = 25% or 0.25  
 However, half of all girls born may be affected.  
 (c) Probability of having an affected boy = 25% or 0.25  
 However, half of all boys born may be affected.

Background for question 6: Sex linkage refers to the location of genes on one or other of the sex chromosomes (usually the X, but a few are carried on the Y). Such genes produce an inheritance pattern which is different from that shown by autosomes:

- Reciprocal crosses produce different results (unlike autosomal genes that produce the same results).
  - Males carry only one allele of each gene.
  - Dominance operates in females only.
  - A 'cross-cross' inheritance pattern is produced: father to daughter to grandson, etc.
6. X-linked disorders are more commonly seen in males, because they have only one locus for the gene (and must express the trait). If the sex linked trait is due to a recessive allele, females will express the phenotype only when homozygous recessive. It is possible for females to inherit a double dose of the recessive allele (e.g. a colour blind daughter can be born to a colour blind father and mother who is a carrier), but this is much less likely than in males because sex linked traits are relatively uncommon.
- 7 (a) and (b)  
 - A Y-linked disorder must be expressed in the male offspring  
 - The male parent must be affected.

## 217. Inheritance Patterns (page 305)

### 1. Autosomal recessive:

(a) Punnett square:

**Male parent phenotype:**

Normal, carrier

**Female parent phenotype:**

Normal, carrier

	(P)	(p)
(P)	PP	Pp
(p)	Pp	pp

(b) Phenotype ratio:

Normal 3      Albino 1

### 2. Autosomal dominant:

(a) Punnett square:

**Male parent phenotype:**

Woolly hair

**Female parent phenotype:**

Woolly hair

	(W)	(w)
(W)	WW	Ww
(w)	Ww	ww

(b) Phenotype ratio:

Normal 1      Woolly 3

### 3. Sex linked recessive:

(a) Punnett square:

**Male parent phenotype:**

Normal

**Female parent phenotype:**

Normal, carrier

(b) Phenotype ratio:

**Females:**

Normal 2      Haemophilic 0

**Males:**

Normal 1      Haemophilic 1

	(X)	(X <sup>h</sup> )
(X)	XX	XX <sup>h</sup>
(Y)	XY	X <sup>h</sup> Y

### 4. Sex linked dominant:

(a) Punnett square:

**Male parent phenotype:**

Affected (with rickets)

**Female parent phenotype:**

Affected (with rickets)

(b) Phenotype ratio:

**Females:**

Normal 0      Rickets 2

**Males:**

Normal 1      Rickets 1

	(X <sup>R</sup> )	(X)
(X <sup>R</sup> )	X <sup>R</sup> X <sup>R</sup>	X <sup>R</sup> X
(Y)	X <sup>R</sup> Y	XY

## 218. Problems Involving Monohybrid Crosses

(page 306)

1.  $1/2 Ww$  and  $1/2 ww$ .

Ratio: 1 wire-haired : 1 smooth haired

Working: Parental genotypes are  $Ww \times Ww$ . The test cross of the F<sub>1</sub> (to the homozygous recessive by definition) is to a smooth haired dog ( $ww$ ).

$1/4$  of the F<sub>1</sub> will be wire-haired ( $WW$ ). When crossed with  $ww$  the result will be all wire-haired dogs ( $Ww$ ).

$1/2$  the F<sub>1</sub> will be wire-haired ( $Ww$ ). When crossed with  $ww$ , the result is  $1/2$  wire-haired and  $1/2$  smooth-haired.

$1/4$  of the F<sub>1</sub> will be smooth-haired ( $ww$ ). When crossed with  $ww$ , all offspring will be smooth-haired ( $ww$ ). Across all progeny, half will be  $Ww$  and half will be  $ww$ .

2. Probability of black offspring:  $(2/3 \times 1/4 =) 1/6$  or 0.16

Working: The parents genotypes are  $Bb \times Bb$ , and  $1/3$  of the white offspring ( $BB$ ) crossed with  $Bb$  will result in no black lambs while  $2/3$  of the white offspring ( $Bb$ ) crossed with  $Bb$  will result in  $1/4$  black lambs.

3. (a) They have an albino child ( $aa$ ) as well as unaffected ones ( $AA$  or  $Aa$ ), so the parents must both be  $Aa$ . Note: There is a 25% chance that any child of theirs will be albino.  
 (b) The family are all  $aa$ .  
 (c) The albino father must be  $aa$ . The mother must be  $Aa$ . The three unaffected children are  $Aa$ .  
 Note: There is a 50% chance that any child of theirs will be albino. The observed 3:1 ratio is not surprising, given the small number of offspring.

4. - **Couple #1** genotypes must be  $X^{H}X^{-}$  and  $X^{H}Y$  because neither is affected. Their son is affected  $X^{h}Y$ . If the mother is  $X^{H}X^{H}$  they could not have an affected son. If she is  $X^{H}X^{h}$ , there is a 50% chance that her son will be  $X^{h}Y$ .  
 - **Couple #2** genotypes must be  $X^{H}X^{-}$  and  $X^{h}Y$  and their son is  $X^{h}Y$ . The father did not pass an X chromosome to his son, so his genotype is irrelevant. If the mother is  $X^{H}X^{H}$ , all of her sons will be  $X^{H}Y$ , but if she is a carrier  $X^{H}X^{h}$ , there is a 50% chance that her son will be  $X^{h}Y$ .  
 - Either the hospital or the parents could be correct. The answer depends on the genotype of the mothers.

## 219. Dihybrid Inheritance (page 307)

1

	BL	Bl	bL	bl
BL	BBLL	BBLl	BbLL	BbLl
Bl	BbLL	BbLl	bbLL	bbLl
bL	BbLl	Bbll	bbLl	bbll
bl	Bbll	Bbll	bbll	bbll

1 BBLL      2 BbLL      2 BbLl      4 BbLl  
 1 BBll      2 Bbll  
 1 bbLL      2 bbLl  
 1 bbll

## 220. Inheritance of Linked Genes (page 308)

1. Linkage refers to the situation where genes are located on the same chromosome. As a result, the genes tend to be inherited together as a unit.  
 2. Gene linkage reduces the amount of variation because the linked genes are inherited together and fewer genetic combinations of their alleles are possible.

## 221. Recombination and Dihybrid Inheritance

(page 309)

1. It produces new associations of alleles in offspring.



**222. Problems Involving Dihybrid Crosses**  
(page 310)

1. (a) bbSS (brown/spotted) X BBss (solid/black)  
(which parent was male and which female is unknown. Parents must be homozygous since all the offspring are of one type: BbSs: black spotted).
- (b) F2 generation: BbSs X BbSs

	<b>BS</b>	<b>Bs</b>	<b>bS</b>	<b>bs</b>
<b>BS</b>	BBSS	BBSs	BbSS	BbSs
<b>Bs</b>	BBSs	BBss	BbSs	Bbss
<b>bS</b>	BbSS	BbSs	bbSS	bbSs
<b>bs</b>	BbSs	Bbss	bbSs	bbss

- (c) Spotted/black 9/16  
Spotted/brown 3/16  
Solid/black 3/16  
Solid/brown 1/16  
Ratio: 9:3:3:1 (described as above)

2. Note: Persian and Siamese parents are pedigrees (truebreeding) and homozygous for the genes involved.
  - (a) Persian: UUss, Siamese: uuSS, Himalayan: uuss
  - (b) F1: Genotype: all heterozygotes UuSs.
  - (c) F1: Phenotype: all uniform colour, short haired.
  - (d) F2 generation: UuSs X UuSs

	<b>US</b>	<b>Us</b>	<b>uS</b>	<b>us</b>
<b>US</b>	UUSS	UUSs	UuSS	UuSs
<b>Us</b>	UUSs	UUss	UuSs	Uuss
<b>uS</b>	UuSS	UuSs	uuSS	uuSs
<b>us</b>	UuSs	Uuss	uuSs	uuss

- (e) 1:15 or 1/16 uuss: Himalayan
- (f) Yes (only one type of allele combination is possible)
- (g) 3:13 or 3/16 (2 uuSS, 1 uuSS)
- (h) All of the following have different genotypes but produce a uniform colour-short hair cat:

UUSS, UuSS, UuSs, UUSs, because they all have at least one dominant allele for each gene. Similarly uuSs and uuSS both produce a colour pointed short hair cat, and UUss and Uuss both produce a uniform coloured, long hair cat.

3. (a) Yes
- (b) Four phenotypes were produced. If there was no crossing over there would only be two phenotypes (parental types).
- (c) CucuEbeb, cucuebeb, Cucuebeb, cucuEbeb.

**223. Genetic Screening** (page 311)

1. Benefits of genetic screening include:

Allows potential carriers to decide whether or not to have child and risk passing a genetic disease on to them.

Allows researchers to study how a genetic disease is inherited and if a treatment is possible.

Allows doctors to optimise a patient's drug therapy and make decisions about other medical options (e.g. surgery).

Allows parents with a possibly affected fetus to prepare for the consequences of that affect.

2. Discussion could include points on:

The rights of the affected child to a life free from pain or trauma.

The effect on the parents of supporting an offspring with a genetic defect, malfunctioning organ, or debilitating condition.

The effect on the younger sibling of knowing they were born as a solution to the older sibling's medical problem.

**224. KEY TERMS: Did You Get It?** (page 313)

1. (a) Recessive inheritance
- (b) The trait is present in the first generation, absent in the second, and present again in the third. Also it appears in the offspring of two unaffected parents.
- (c) Dominant inheritance
- (d) The trait appears in offspring when only one parent is affected. It does not appear in offspring when both parents are unaffected.

2. Punnett square:

	<b>YR</b>	<b>Yr</b>	<b>yR</b>	<b>yr</b>
<b>YR</b>	YYRR	YYRr	YyRR	YyRr
<b>Yr</b>	YYRr	YYrr	YyRr	Yyrr
<b>yR</b>	YyRR	YyRr	yyRR	yyRr
<b>yr</b>	YyRr	Yyrr	yyRr	yyrr

Yellow-round: 9/16  
Green-round: 3/16

Yellow-wrinkled: 3/16  
Green-wrinkled: 1/16

**225. Review: Unit 2 Area of Study 2** (page 314)

No model answer. Summary is the student's own.

**226. Synoptic Question: Unit 2 Area of Study 2**  
(page 316)

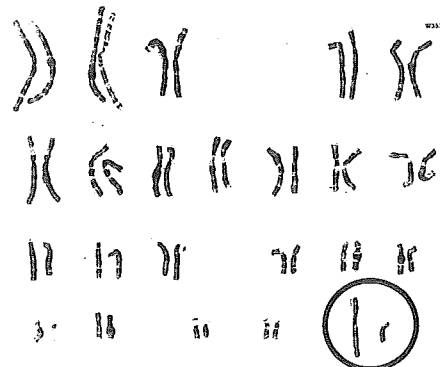
1. Genome sequencing refers to finding the DNA sequence of an organism. It has applications in understanding how species are related, determining the function of genes, and detecting and diagnosing human disease.

Sequencing the genome can be useful in studying relatedness between species. It allows the DNA sequences between different species to be compared and the sequence differences quantified. How many differences there are can be an indication of how closely related the species are. Fewer differences indicates species are closely related, whereas a large number of sequence differences indicates the species are more distantly related or unrelated.

Genome sequencing can also be useful in determining gene function. Finding differences between correctly working and defective genes can help to understand how genes function.

Finally genome sequencing can be useful in detecting human disease. Mutations to certain genes may increase the risk of a person developing a disease (e.g. BRCA1 mutations causing cancer). Genome sequencing can identify the mutations and help people make informed decisions about lifestyles or a possible course of treatment if a disease develops.

2. (a) Applications include finding out the sex of the individual. In some species it is very difficult to identify sex on appearance alone. Karyograms are also useful for finding any chromosome abnormalities.
- (b)



- (c) Male
- (d) Normal
- (e) There are two of every chromosome (except X and Y).

The person is therefore  $2n$  as would be expected.

3. (a) There would be three copies of chromosome 21.  
(b) It is an aneuploidy ( $2n + 1$ )  
(c) Non-disjunction of chromosome 21 resulting in a gamete with two no. 21 chromosome instead of one. The union of this gamete with a normal gamete produces a zygote with three no. 21 chromosomes.
4. Phenotype is the observable characteristic, e.g. hair colour. The genotype is the allele combination, e.g. Bb
5. (a) The genotype affects the phenotype by determining the variant of the phenotype, e.g. BB will determine a different phenotype from bb. The genotype defines the absolute genetic potential of an organism.  
(b) The environment will determine the extent to which the genetic potential of an organism will be reached and therefore the phenotype of the organism, e.g. a person's height may be determined by the genotype and the nutritional environment during their growth.  
(c) Phenotype can be influenced by epigenetics, i.e. modifications to how the DNA is packaged and read. These modifications can determine the level of gene expression and therefore how a cell or tissue develops.
6. The greater the number of genes associated with a phenotype the greater the potential variation in the phenotype. This is due to the greater number of allele combinations possible with greater numbers of contributing genes. For example, a monogenic trait may have two (possibly three) phenotypic traits caused by BB, Bb, or bb. A dihybrid trait may have four (or more) traits AABB, aaBB, AAbb or aabb. With more genes involved, there is also a greater opportunity for the environmental effects on the final phenotypic outcome.
7. The breeder would breed BBLL and bblL guinea pigs together. The gametes of the father would be BL and the mother bl. The offspring would all be BbLl heterozygotes. The breeder would then need to cross F<sub>1</sub> heterozygotes together as below:

		Female gametes			
		(BL)	(Bl)	(bL)	(bl)
Male gametes	(BL)	BBLL	BBLl	BbLL	BbLl
	(Bl)	BBLl	BBll	BbLl	Bbll
	(bL)	BbLL	BbLl	bbLL	bbLl
	(bl)	BbLl	Bbll	bbLl	bbll

The breeder then needs to look for all the guinea pigs that are white (bb) and have short hair (LL or Ll). This will be a small number as the cross ratio is 9:3:3:1, 9 black coat short hair, 3 white coat short hair, 3 black coat long hair, and 1 white coat long hair. From this cross it is already determined that the white coat, short hair guinea pigs will be pure breeding for white coats as this is recessive (bb). The breeder could then perform a test cross on the F<sub>1</sub> guinea pigs with white coats and short hair (i.e. a cross back to the homozygous recessive parent) to test which guinea pig is homozygous for the dominant allele (short coat). When crossed with the homozygous recessive parent, the heterozygotes will produce some long haired offspring but the homozygotes will not.

Test cross of F<sub>1</sub> generation

		Unknown gametes		Unknown gametes	
		(bl)	(bl)	(bl)	(bl)
Known gametes	(bl)	bbLl	bbll	bbLl	bbLl

Parents (of unknown genotype) that produced any long haired offspring can then be eliminated from the breeding group. The parent (of unknown genotype) that produced only white coated, short haired guinea pigs will be the true breeding guinea pig.

