Surname	StemUp Model Solution	Centre Number	Candidate Number
Other Names			2

GCE A LEVEL

A400U20-1

S18-A400U20-1

BIOLOGY – A level component 2
Continuity of Life

MONDAY, 11 JUNE 2018 – AFTERNOON

2 hours

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Question	**Maximum Mark**	**Mark Awarded**
1.	**23**	
2.	**15**	
3.	**17**	
4.	**19**	
5.	**17**	
6.	**9**	
Total	**100**	

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01

ADDITIONAL MATERIALS

In addition to this examination paper, you will need a calculator and a ruler.

INSTRUCTIONS TO CANDIDATES

Use black ink or black ball-point pen. Do not use gel pen. Do not use correction fluid.

Write your name, centre number and candidate number in the spaces at the top of this page.

Answer **all** questions.

Write your answers in the spaces provided in this booklet. If you run out of space, use the additional pages at the back of the booklet, taking care to number the question(s) correctly.

INFORMATION FOR CANDIDATES

The number of marks is given in brackets at the end of each question or part-question.

The assessment of the quality of extended response (QER) will take place in question **6**.

The quality of written communication will affect the awarding of marks.

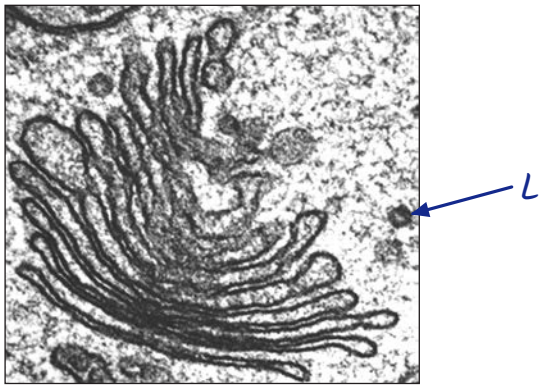
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Answer **all** *questions.*

1. Lysosomal storage disorders are a group of about 50 diseases that are characterised by an accumulation of waste products in the lysosomes. Two examples are Fabry disease and Tay-Sachs disease. Sufferers of Tay-Sachs disease die in childhood.

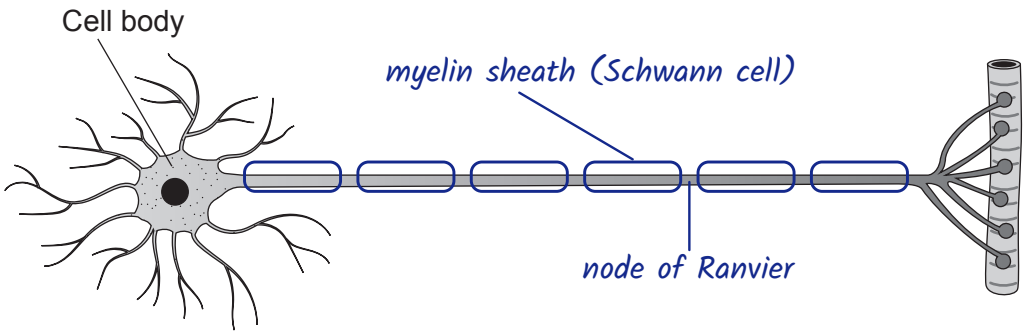


(a) The electron micrograph above shows the cell organelle responsible for producing lysosomes. Name the organelle, **draw an arrow labelled L** on the micrograph to identify a lysosome and describe a general function of lysosomes in normal cells. [3]

Golgi apparatus. Lysosomes contain hydrolytic (digestive) enzymes that break down (recycle) worn-out organelles and macromolecules.

(b) The symptoms of Tay-Sachs disease are a consequence of abnormal accumulation of fatty substances in neurones.

(i) **Complete the diagram** of a neurone below to show how this fatty substance is usually distributed, your labels should include the name of the fatty substance. [2]



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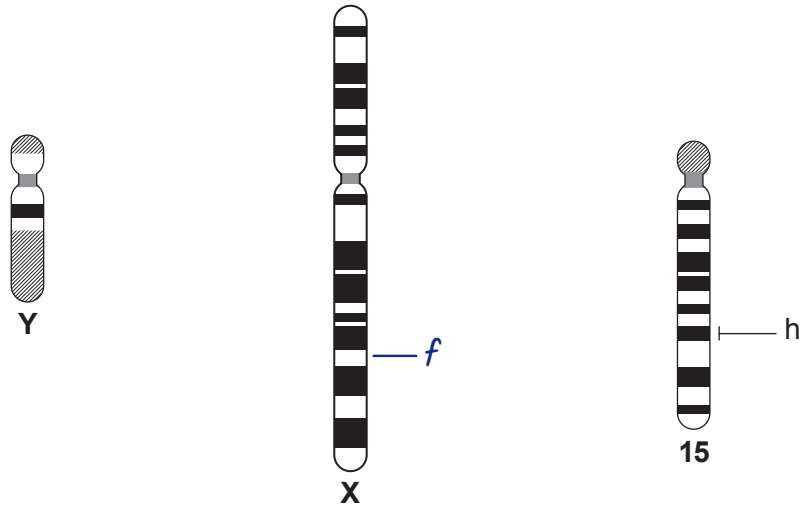
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(ii) Explain how the arrangement of the fatty substance affects the transmission of nerve impulses. [3]

Myelin is an electrical insulator, so it prevents the movement of ions across the axon membrane where it is present. Local currents therefore extend further, so action potentials are only generated at the nodes of Ranvier. The impulse leaps from node to node (saltatory conduction), which speeds up transmission.

(c) The diagrams below show gene maps of human sex chromosomes and chromosome **15**. The recessive allele **(f)** that causes Fabry disease is carried on the **X** chromosome. The recessive allele **(h)** that causes Tay-Sachs is carried on chromosome **15**, in the locus labelled h.



(i) **Mark, by placing the letter f on the diagram,** a possible locus for the gene that causes Fabry disease. [1]

(ii) With reference to the positions of the gene loci explain the following observations: [2]

I. Males with the **f** allele always suffer Fabry disease but females can have the **f** allele without suffering the disease.

Males have one X and no equivalent locus on the Y, so one f allele causes it. Females have two X so can be carriers (X^FX^f).

II. Males and females can have the **h** allele without suffering Tay-Sachs disease.

The h locus is on chromosome 15, an autosome, so both sexes have two alleles and can be heterozygous carriers.

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(d) Complete the genetic diagram below to show how Fabry disease might be inherited from parents neither of whom suffers from the condition and state the probability of this couple producing a child with Fabry disease. [4]

	male		female
Parental phenotype:	unaffected		unaffected (carrier)
Parental genotypes:	X^FY	x	X^FX^f
Gametes:	X^F and Y	x	X^F and X^f

gametes	X^F	X^f
X^F	X^FX^F	X^FX^f
Y	X^FY	X^fY

X^FX^F unaffected female; X^FX^f carrier female

X^FY unaffected male; X^fY male with Fabry disease

Key: F = normal allele, f = Fabry allele (on the X chromosome)

Probability of this couple producing a child with Fabry disease = 0.25 (¼)

(e) A genetic counsellor advised a woman with Fabry disease in her family to have an amniotic fluid test, ten weeks into her pregnancy, in order to determine the sex of the embryo.

(i) Explain the function of amniotic fluid. [1]

It acts as a shock absorber, protecting the embryo from physical damage.

(ii) Why might it be important for the woman with Fabry disease in her family to know the sex of the embryo and what ethical issues might this cause? [2]

Only males suffer Fabry: a male has a 50% chance of being affected.

Ethical issue: whether to terminate the pregnancy if it is male.

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The Hardy-Weinberg Principle states that the frequency of alleles for a given gene remains constant from generation to generation, providing the population is large and no selection takes place.

The frequency of Tay-Sachs disease is 1 in 360 000 births in the general population of the USA. However, the frequency is 1 in 40 000 in certain populations, which isolate themselves culturally. One such population is the Old Order Amish of the Kishacoquillas Valley, Pennsylvania, USA. Recent estimates put the size of this population at 40 000.

(f) (i) Use the Hardy Weinberg equations, given below, to calculate the number of people in the Amish community in the Kishacoquillas Valley that carry the Tay-Sachs allele without suffering the disease. [3]

$p + q = 1.0$ $\quad\quad$ $p^2 + 2pq + q^2 = 1.0$

$q^2 = 1 \div 40\,000 = 0.000025$

$q = \sqrt{0.000025} = 0.005$

$p = 1 - 0.005 = 0.995$

carriers: $2pq = 2 \times 0.995 \times 0.005 = 0.00995$

number $= 0.00995 \times 40\,000 = 398$

Number of people who are carriers of Tay-Sachs allele = 398

(ii) Explain why the frequency of Tay-Sachs is higher in isolated populations and predict, with a reason, what is likely to happen to the frequency of the Tay-Sachs allele in the general population. [2]

Small isolated population (small gene pool, no immigration): genetic drift (founder effect), and two carriers are more likely to have children. In the general population the h allele frequency will decrease, as sufferers die in childhood before reproducing, so it is selected against.

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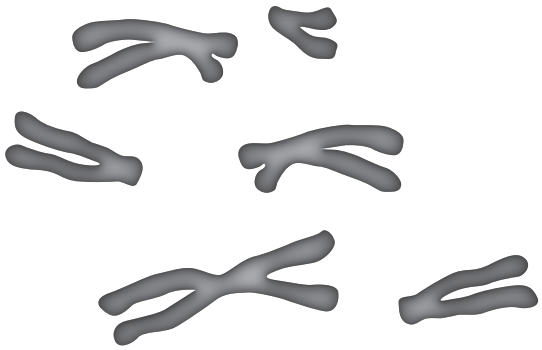
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2. The photograph shows all the chromosomes from a blood cell of a mammal.



(a) (i) Explain why red blood cells could not have been used to produce the photograph of the chromosomes. [1]

Red blood cells have no nucleus, so they contain no chromosomes.

(ii) Use the photograph to deduce the sex of the mammal and explain your choice. [1]

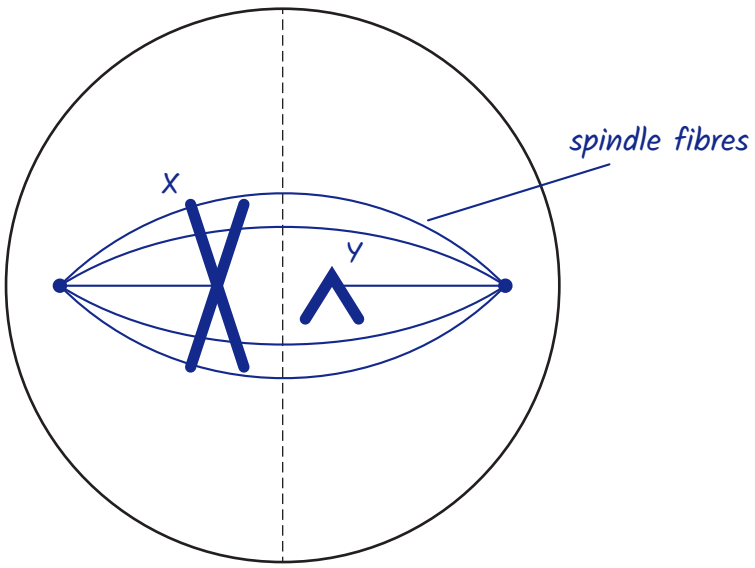
Male: one pair is not homologous (one large X and one small Y).

(iii) In this mammal, how many chromosomes would be present in [1]

I. a kidney cell; 6

II. gametes? 3

(iv) The circle below represents the outline of the mammal cell at metaphase I of meiosis, and the dotted line the equator of the cell.
Complete the drawing to show the spindle and how **the sex chromosomes** as shown above would be arranged. [2]



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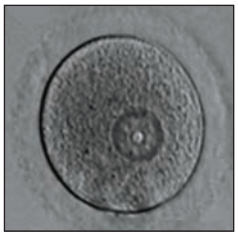
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(b) Starfish, *Asterias rubens*, reproduce sexually by spawning. When starfish spawn, the males release spermatozoa and the females release oocytes in large numbers.



The oocytes are suspended at early prophase of the first meiotic division.



The nuclei of early oocytes are called germinal vesicles.



Breakdown of the germinal vesicle indicates resumption of meiosis (re-initiation).

Re-initiation is naturally stimulated by contact with spermatozoa. Scientists have shown that it is possible to stimulate re-initiation by exposing the oocytes to suitable concentrations of calcium ions (Ca^{2+}).

Sea water has a typical calcium concentration of 400 mg dm^{-3}.

Suggest a suitable hypothesis for this investigation and design an experiment to investigate how calcium ion concentration affects meiosis in starfish. Make sure you suggest suitable values for the independent variable as well as identifying the dependent variable and **two** controlled variables. [6]

Hypothesis: the greater the Ca^{2+} concentration, the greater the percentage of oocytes in which meiosis re-initiates (germinal vesicle breaks down).

Place 50 oocytes from the same starfish into equal volumes of artificial sea water with Ca^{2+} at 0, 100, 200, 300, 400, 500 and 600 mg dm^{-3}.

Independent variable: Ca^{2+} concentration. Dependent variable: percentage of oocytes whose germinal vesicle has disappeared after 30 minutes, counted under a microscope.

Controlled variables: temperature (water bath at 15 °C), pH, volume of solution, concentration of other ions and number of oocytes. Repeat three times and find the mean.

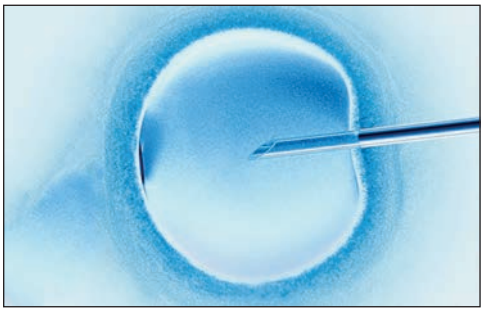
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(c) Some human fertility problems are due to poor morphology (abnormal shape) or poor motility (not moving normally) of spermatozoa. These causes of infertility can be treated by a special type of In Vitro Fertilisation (IVF) called Intra-Cytoplasmic Sperm Injection (ICSI). It differs from conventional IVF in that a single spermatozoon is injected directly into a secondary oocyte, instead of fertilisation taking place in a dish where many spermatozoa are placed near a secondary oocyte.



Intra-Cytoplasmic Sperm Injection

However, fertilisation does not always proceed to completion. A solution of a 'calcium ionophore' may be used to stimulate gated calcium ion channels in the oocyte plasma membrane. Ionophores are molecules that facilitate ion passage in or out of cell membranes.

(i) Use your knowledge of the structure of the plasma membrane to state what is meant by the term *'gated calcium ion channel'*. [2]

A transmembrane (intrinsic) protein with a channel that can be opened or closed to allow or prevent Ca^{2+} ions passing through.

(ii) In humans, before fertilisation the oocytes are suspended in metaphase II. Explain how the ionophore molecules might improve the success rate of ICSI. [2]

The ionophore causes Ca^{2+} ions to enter the oocyte through the gated channels.

The rise in Ca^{2+} stimulates re-initiation of meiosis (meiosis II is completed), so fertilisation proceeds to completion.

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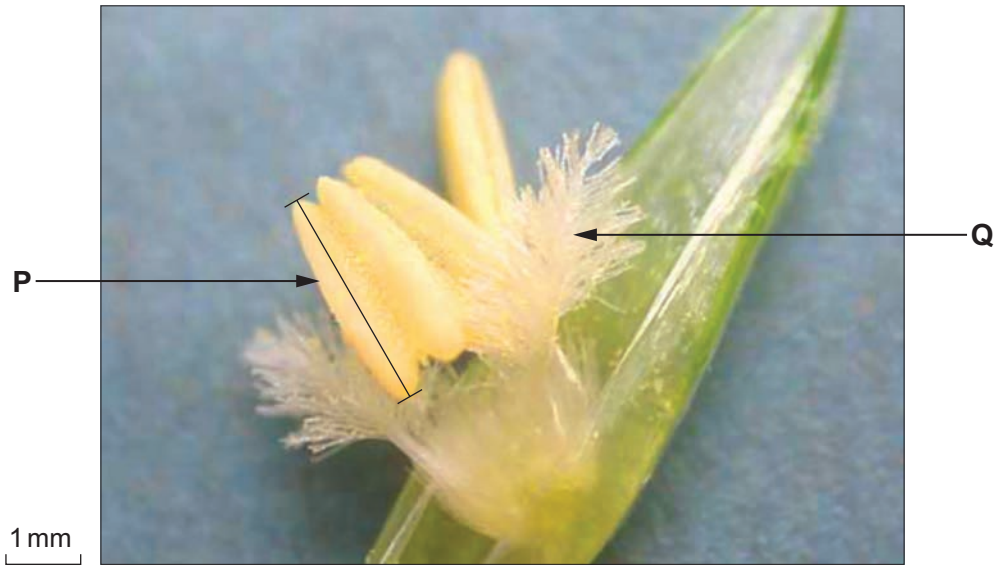
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3. *(a)* The photograph below shows a flower of the wheat plant, *Triticum aestivum*.



(i) Name the parts labelled **P** and **Q**. [1]

P anther

Q stigma

(ii) Calculate the actual length of structure **P**. [2]

length of P on photo = 30 mm

scale bar 1 mm = 10 mm, so magnification = × 10

actual length = 30 ÷ 10

Size = 3 mm

(iii) Describe **two** features of the flower shown in the photograph which suggest it is wind pollinated. [1]

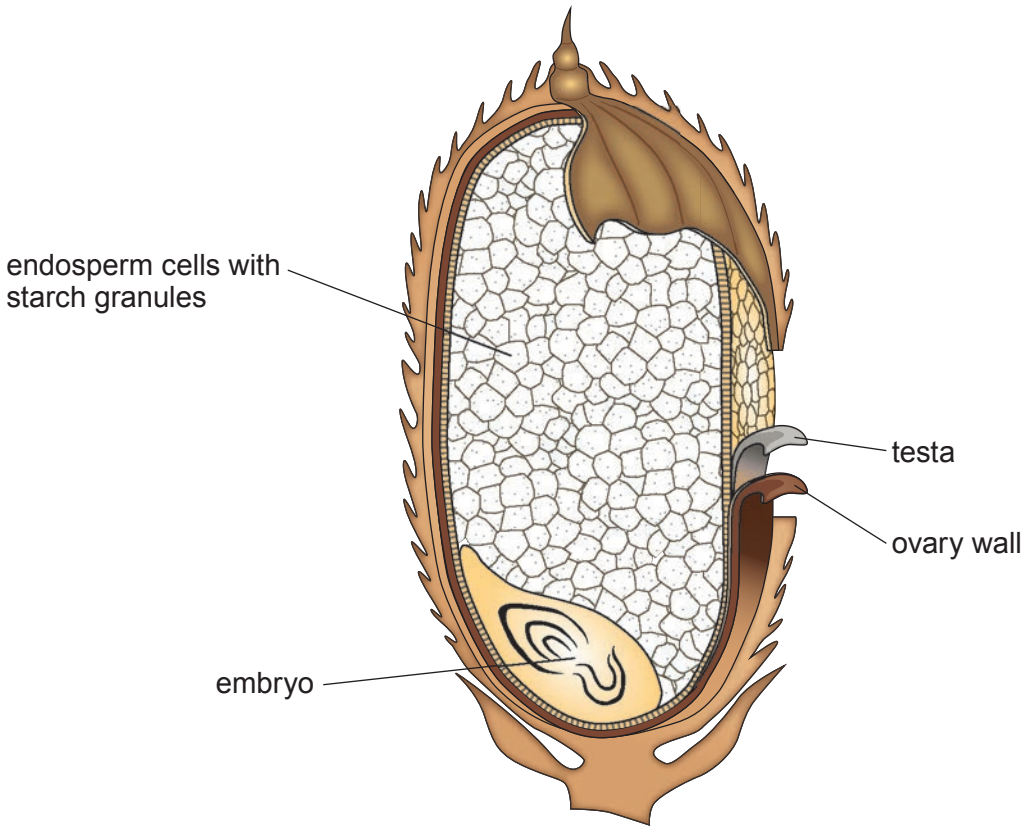
Large anthers hanging outside the flower and feathery stigmas outside the flower; no coloured petals.

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(b) The diagram below shows a wheat grain.



(i) Use information shown in the diagram to explain why the grain is more correctly described as a fruit rather than a seed. [2]

The grain retains the ovary wall, which is fused to the testa, so it is a fruit. A seed would only have a testa as its outer layer.

(ii) Explain why the events taking place in the embryo sac of a wheat flower are described as a double fertilisation. [2]

Two male nuclei are involved: one fuses with the egg cell (female gamete) to form the zygote, and the other fuses with the two polar nuclei to form the (triploid) endosperm nucleus.

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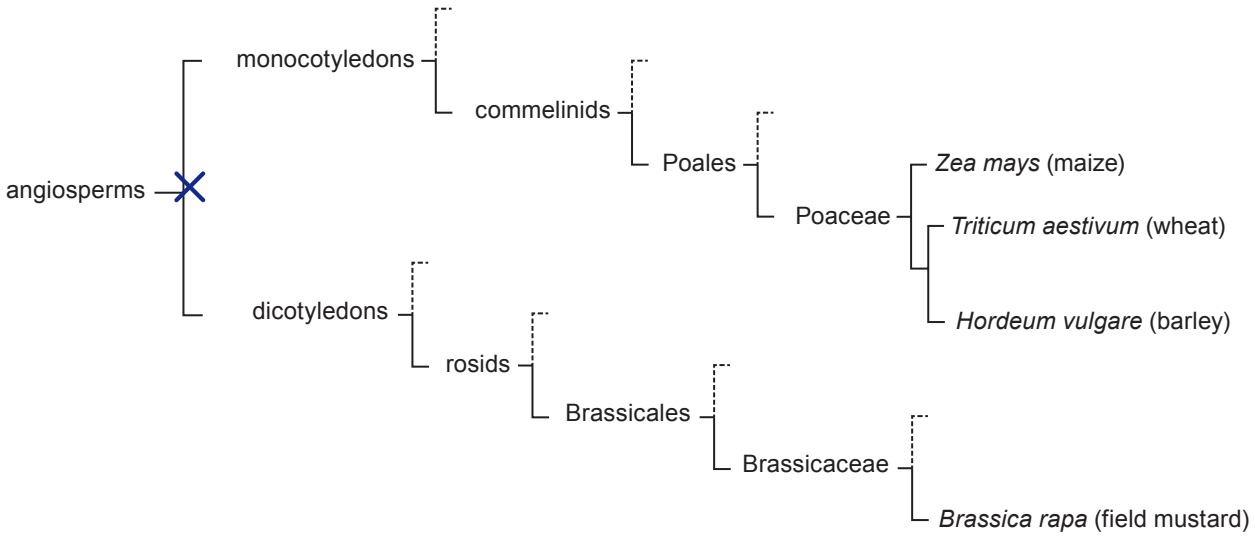
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(c) The photograph below shows a flower of the field mustard plant, *Brassica rapa*.



Diagram **1** below shows the classification of wheat and field mustard and their evolutionary relationship.

Diagram 1



Each branch on the diagram represents a clade, i.e. an ancestor together with all its descendants.

(i) **Mark with an X**, the position on the diagram where wheat and field mustard share a common ancestor. [1]

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(ii) In modern plant classification systems hierarchical groups like Divisions and Classes have been replaced with unranked clades, but orders and families have been retained and have names ending in **~ ales** and **~ aceae** respectively.

Use the diagram opposite to complete the classification of field mustard and wheat. [2]

Kingdom:	Plantae	Plantae
(unranked):	angiosperms	angiosperms
(unranked):	Dicotyledons	monocotyledons
(unranked):	Rosids	Commelinids
Order:	Brassicales	Poales
Family:	Brassicaceae	Poaceae
Genus:	*Brassica*	*Triticum*
Species:	***B. rapa***	***T. aestivum***

(iii) Using the information given and your own knowledge, describe how the seed of field mustard differs from the wheat grain in terms of where it stores nutrients. [2]

Wheat stores nutrients (starch) in the endosperm. Field mustard is a dicotyledon, so its seed stores nutrients in the two cotyledons.

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(d) The amino acid sequence of the protein cytochrome C has been used to investigate the evolutionary relationship between organisms. The table below shows the amino acid sequences of parts of the protein from three sources, wheat, barley and maize. The amino acids are represented by using a single letter code, e.g. G = glycine.

SOURCE	Position of amino acid in polypeptide chain																									
	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15-60	61	62	63	64	65	66	67	68	69	70	71	72-112
1	M	D	S	F	A	E	A	P	A	G	N	P	T	T		V	I	W	E	E	N	T	L	Y	D	Y	
2	M	A	P	F	D	E	A	P	P	G	K	S	K	A		V	E	W	E	E	K	T	L	T	D	Y	
3	M	A	S	F	D	E	A	P	P	G	K	P	K	A		V	E	W	E	E	K	T	L	Y	E	Y	

(i) Explain why determining the amino acid sequence of proteins from different organisms can be used to show evolutionary relationships. [1]

The more closely related the organisms, the fewer differences in the amino acid sequence, as they share a more recent common ancestor.

(ii) Count the differences between the amino acid sequences from sources **2** and **3**. Enter your answer in the table below. [1]

Between sources	No. of differences
1 and 2	11
2 and 3	4
1 and 3	9

(iii) Use the table above and diagram **1** on page **12** to conclude which source came from which species. [2]

Species	Source
Maize (*Zea mays)*	1
Wheat *(Triticum aestivum)*	3
Barley *(Hordeum vulgare)*	2

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4. Wheat grain size is determined by the plants' genetics (i.e. variety), and the length of the grain filling period (time between fertilisation and harvesting). Plant breeders cross varieties of wheat in order to increase grain size. The photograph below shows the results of one such cross.



(a) Distinguish between **continuous** and **discontinuous** variation. You should give an example, which is visible in the photograph, for each one. [3]

Continuous variation shows a gradation from one extreme to the other (many genes), e.g. grain length. Discontinuous variation has clear-cut categories (single gene), e.g. coat colour or a hairy or smooth tip.

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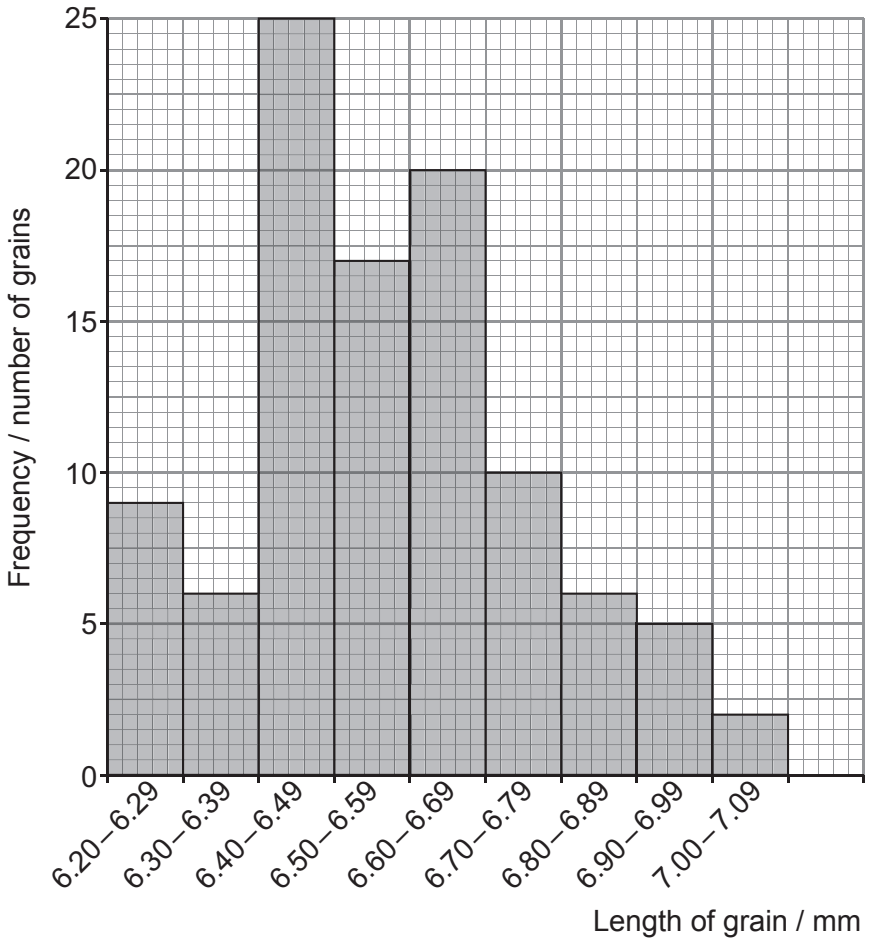
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The graph below shows the distribution of grain length in a sample of the hybrid wheat variety. The grains were measured to the nearest 0.01 mm.



(b) (i) What is the total number of grains in the sample? [1]

100 grains

(ii) The student calculated three different averages for the length of hybrid grains. The mean was 6.57 mm. The other two values were 6.45 mm and 6.52 mm. With reference to the graph, state which of these values is more likely to be the mode and give a reason for your choice. [2]

6.45 mm, because it lies in the 6.40 to 6.49 mm class, which is the tallest bar (the most common class, 25 grains).

(iii) What evidence is there from the graph that the length of hybrid grains is not normally distributed? [1]

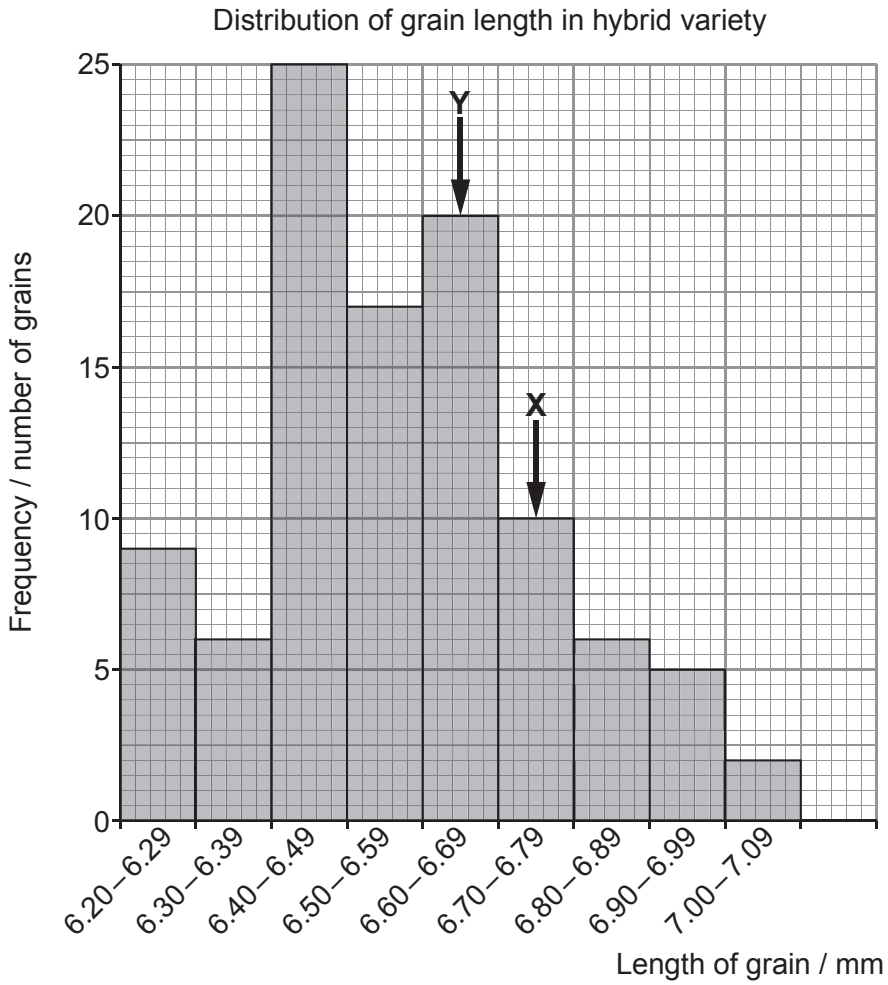
It is not symmetrical (not bell shaped); it is skewed to the left.

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For each parental variety the mode and median were the same. Arrow **X** shows the mode and median for parental variety A and Arrow **Y** shows the mode and median for parental variety B.



(iv) Consider the following statement:

'There is no significant difference in the length of the hybrid grains and the length of the grains of parental type B.'

Describe any evidence from the graph above to support the statement and name the terms used to describe; the type of hypothesis represented by the statement and the statistical test that could be used to test the hypothesis. [3]

Evidence *the hybrid mode and median are closer to Y (parent B) than to X (parent A), so hybrids have a similar average length to parent B.*

Type of hypothesis *null hypothesis*

Statistical test *Student's t-test*

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As soon as fertilisation has occurred, the embryo and endosperm begin to develop with the plant redirecting products of photosynthesis to the developing grains. The longer the period of grain fill the larger wheat grain size. Grain size can be influenced by the availability of water and nutrients, and disease management.

(c) With reference to the information provided above, explain the following:

(i) there is a saying amongst farmers, "When you think the crop is ready to harvest, take a holiday"; [1]

Leaving the crop longer increases the grain fill time, so more photosynthetic product goes into the grains and they grow larger.

(ii) the use of nitrate fertiliser to increase grain size; [3]

Plants absorb nitrates to make amino acids, proteins, chlorophyll and nucleic acids.

These are used for the growth of leaves, which increases photosynthesis, so more photosynthetic product is redirected to fill the grains.

The grain also contains more protein, so it is larger.

(iii) the use of fungicide to increase grain size. [2]

Fungicide kills the fungus and prevents disease.

With less disease the leaves carry out more photosynthesis, so more photosynthetic product reaches the grain, and fewer nutrients are taken from the grain by the fungus.

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(d) In the USA wheat prices are quoted per bushel, (a unit of volume). In Europe wheat prices are quoted per kg. Rex Ryder is a Kansas farmer who harvests his wheat into a truck that holds 998 bushels. During the harvest in 2015 he filled the truck 22 times. His farm covers 492 acres.

Estimate how many bushels of wheat he produced per acre to the nearest bushel and suggest **one** advantage and **one** disadvantage of using prices per kg rather than per bushel. [3]

Estimate

998 × 22 = 21 956 bushels; 21 956 ÷ 492 = 44.6, so about 45 bushels per acre.

Advantage

Mass is more accurate: it takes account of grain quality, and grains settle over time, decreasing their volume. Kg is an internationally understood unit.

Disadvantage

The grain must be weighed, and grains dry out and lose mass over time.

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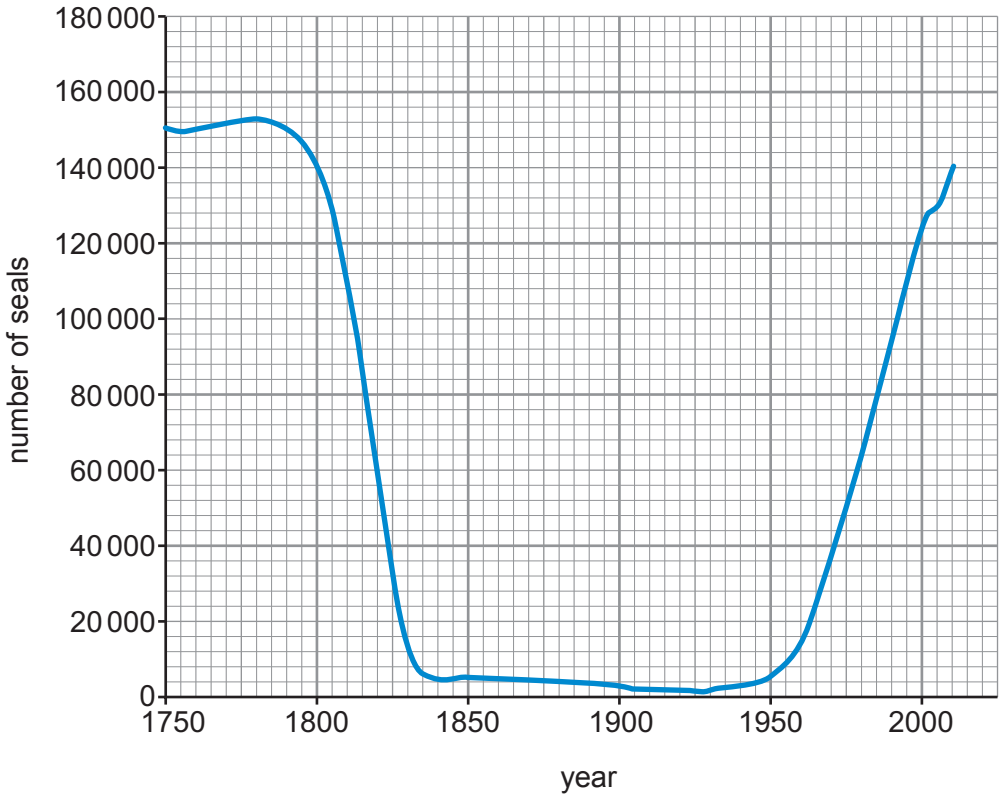
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5. The northern elephant seal, *Mirounga angustirostris*, is named because the male has a large nose that resembles an elephant's trunk. They feed on squid, octopus and fish. They have a thick layer of blubber which was used by humans in the nineteenth century to produce lamp oil.



The population curve below shows estimates of the population of northern elephant seals from 1750 to 2010.



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(a) Factors affecting population size are described as density dependent or density independent. Use the graph to identify when density dependent and density independent factors would have been acting, between the years 1750 and 1830, and suggest what these factors may have been. [4]

From 1750 to 1800 the population was stable at about 150 000, so it was regulated by density dependent factors, e.g. competition for food, mating sites or mates, disease and predation.

From about 1800 to 1830 the population crashed because of density independent factors, e.g. hunting by humans for blubber.

(b) An event in which the size of an existing population is drastically reduced is called a **population bottleneck**.

By 1910, Northern Elephant Seals were only found on Guadalupe Island off Baja California, Mexico. The species was protected under U.S. and Mexican law early in the twentieth century.

Explain how the graph illustrates an example of a *'population bottleneck'* and describe evidence from the graph which suggests that conservation measures were successful. [2]

The population crashed to a very small number (about 4000) by 1900, a bottleneck. After protection the numbers recovered to about 140 000, almost the level before hunting, so conservation was successful.

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(c) Today, all Northern Elephant Seals are descended from the 50 or so Guadalupe seals.

In order to assess the impact of the population bottleneck on the genetic diversity of northern elephant seals a group of scientists compared nucleotide sequences of DNA. Samples were taken from 185 present day northern elephant seals and compared with 22 museum samples of northern elephant seals collected in the nineteenth century.

Five different base sequences have been identified at the same point in the same gene locus. The results table below shows the number of specimens showing each of these alternative base sequences.

Base sequence at a particular point in the gene locus.	Number of specimens showing this sequence	
	present day	nineteenth century
GTA	50	8
GAA	0	8
GAG	0	4
AAG	135	1
AAA	0	1

[A. Rus Hoelzel *et al*; J. Evol. Biol. **15** (2002) 567-575)]

(i) Name a technique that the scientists could have used to amplify the DNA available from the museum specimens. [1]

Polymerase chain reaction (PCR)

(ii) Give **one** limitation of the experiment that produced the results in the table. [1]

Small 19th century sample (only 22); only one gene locus was sampled.

(iii) With reference to the data, draw a conclusion about the impact of population bottlenecks on genetic diversity of northern elephant seals. [2]

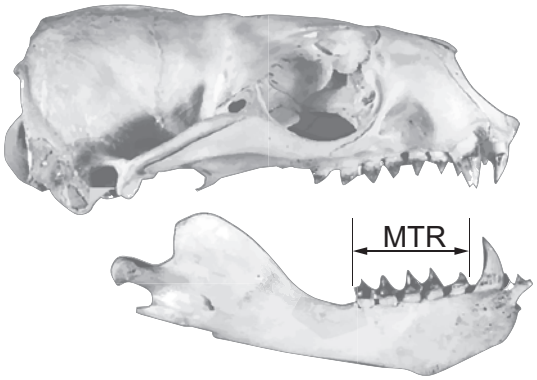
The bottleneck has decreased genetic diversity: all 5 base sequences were found in the nineteenth century samples but only 2 (GTA and AAG) in present day seals, so GAA, GAG and AAA have been lost from the gene pool.

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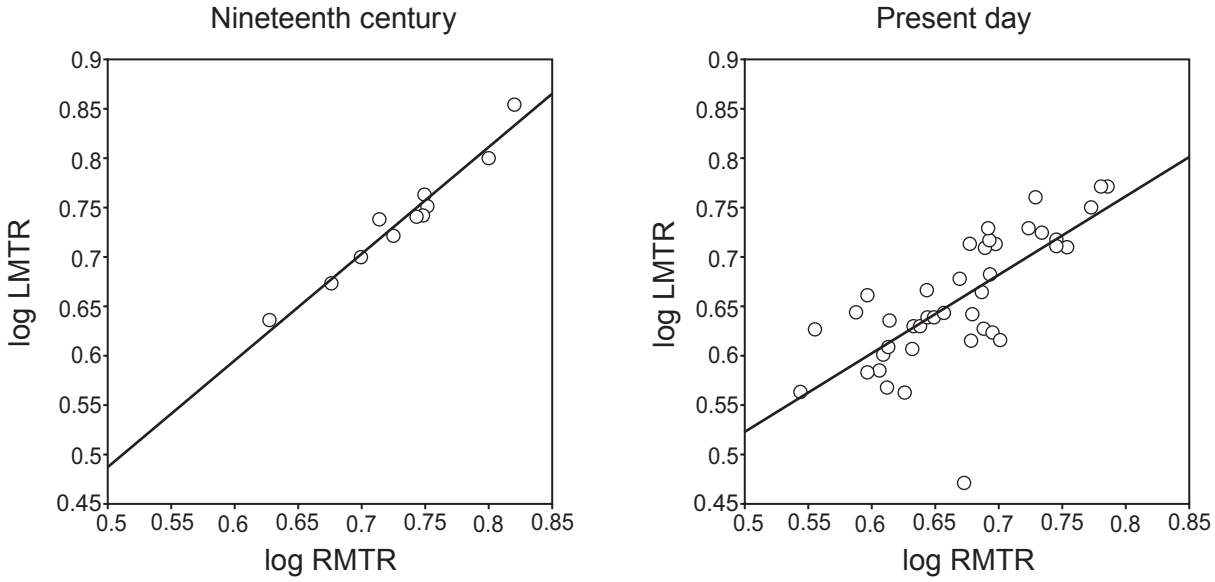
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In order to investigate the impact of the population events on development in the northern elephant seals, the same group of scientists also compared the skulls of present day seals with the museum specimens. They measured the length of the Mandibular Tooth Row (MTR) shown in the diagram below. By comparing the lengths from the right and left sides of each skull they were able to assess their symmetry.



The graphs below show the results of plotting logs of values for the Left Mandibular Tooth Row (LMTR) against logs of values for the Right Mandibular Tooth Row (RMTR).



(d) Use the graphs above to draw a conclusion about the impact of population bottleneck of the nineteenth century on the symmetry of skulls in northern elephants seals. [2]

Present day skulls are less symmetrical: their points show greater scatter (more deviation from the line, lower correlation).

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The scientists also measured the skulls of present day southern elephant seals, *Mirounga leonina*, which did not suffer as badly from the population bottleneck of the nineteenth century.

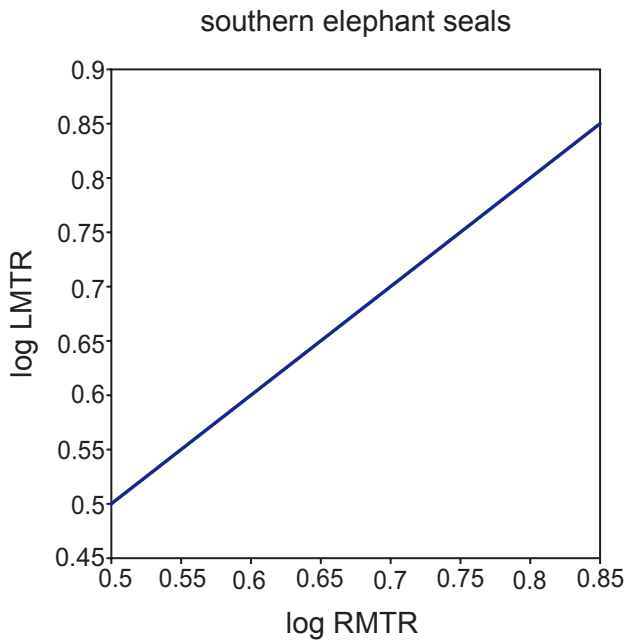
(e) (i) Using the information, what is the evidence that the northern elephant seal is closely related to the southern elephant seal? [1]

Both belong to the same genus, Mirounga.

(ii) How does measuring southern elephant seals improve the validity of the conclusion? [1]

A control: a similar species that did not suffer the bottleneck.

(iii) **Draw a line on the grid below** to represent the expected relationship between the LMTR and RMTR for the southern elephant seals. [1]



(f) How might the effects of the population bottleneck of the nineteenth century on the DNA described in part *(c)* and the development described in part *(d)* affect the survival of northern elephant seals in the future? [2]

Low genetic diversity: they may not adapt to environmental change;

asymmetrical jaws may make them less able to catch food or win mates.

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6. In April 2003 one of the most significant scientific breakthroughs of modern times was announced. After years of painstaking research carried out by thousands of dedicated scientists across the world, the complete genetic code of a human being – their genome – could now be made freely available on-line.

The Human Genome Project, as this work was known, was the largest international collaboration ever undertaken in biology with British scientists leading the global race to read the human genome using a technique called sequencing.

To bring the predicted benefits of genomics to NHS patients the 100 000 Genomes Project was launched in late 2012 and by 2017 had sequenced the genomes of 100 000 NHS patients. The project focussed on patients with a rare disease and their families, and on patients with cancer.

Scenario:

It is 2025 and Sharon has a painful skin infection that she just can't get rid of. Her doctor would like to prescribe an antibiotic called phenyloxacillin, since it is especially effective against the bacteria *(Staphylococcus aureus)* that are causing the infection. However, her doctor knows that in a small number of cases phenyloxacillin can cause serious liver damage so suggests a genome test. She tells Sharon that there is a law giving people the right not to disclose the results of genetic tests to insurers.

Explain what is meant by 'sequencing' the human genome and describe the type of data that might be made available on-line.
Explain how the extra information provided by the '100 000 Genomes Project' might be used in medicine, and describe how the scenario above illustrates one possible beneficial application and an ethical dilemma of genome sequencing. [9 QER]

Sequencing the human genome means determining the order of the nucleotide bases (A, T, C and G) in all of the DNA of a human. The Human Genome Project used Sanger sequencing: DNA fragments were copied using fluorescently labelled terminator nucleotides, separated by length by electrophoresis, and the base sequence was read and assembled by computer.

The data made available on-line include the base sequences and loci of individual genes, showing which sequences code for specific amino acids and therefore proteins, the positions of exons (coding regions) and introns (non-coding regions removed before translation), and variations between individuals.

The 100 000 Genomes Project used Next Generation Sequencing (NGS), which is much faster and cheaper. By comparing the genomes of patients who have a rare genetic disorder or cancer with the normal genome of a large number of individuals, scientists can locate the genes and mutated sequences

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responsible. This helps with diagnosis and understanding the causes of disease, identifying a predisposition to disease, prenatal diagnosis and developing new treatments such as gene therapy and targeted drugs.

In the scenario the genome test is a beneficial application, because it allows personalised medicine: the doctor can match the drug to Sharon's genetic make-up and find out whether she has alleles that make phenyloxacillin likely to cause liver damage before prescribing it. If she is at risk, a different antibiotic can be used, so she is treated safely and effectively.

However, it also raises an ethical dilemma over the rights of access to genomic information. Her genome could reveal a predisposition to other diseases, which could be misused by insurers or employers and lead to social discrimination; this is why the law gives her the right not to disclose results to insurers. The knowledge could also cause anxiety and distress, and it has wider implications for her family's health, since relatives share many of her alleles and may not wish to know.

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