Surname	Centre Number	Candidate Number
Other Names		2

GCE A LEVEL – NEW

1400U40-1

S17-1400U40-1

BIOLOGY – A2 unit 4
Variation, Inheritance and Options

TUESDAY, 20 JUNE 2017 – MORNING

2 hours

	For Examiner's use only		
	Question	**Maximum Mark**	**Mark Awarded**
Section A	**1.**	**13**	
	2.	**20**	
	3.	**12**	
	4.	**10**	
	5.	**6**	
	6.	**9**	
Section B	**Option**	**20**	
	Total	**90**	

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ADDITIONAL MATERIALS

In addition to this paper, you will require 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.

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

INFORMATION FOR CANDIDATES

This paper is in 2 sections, **A** and **B**.

Section **A**: 70 marks. Answer **all** questions. You are advised to spend about 1 hour 35 minutes on this section.

Section **B**: Options; 20 marks. Answer **one option only**. You are advised to spend about 25 minutes on this section.

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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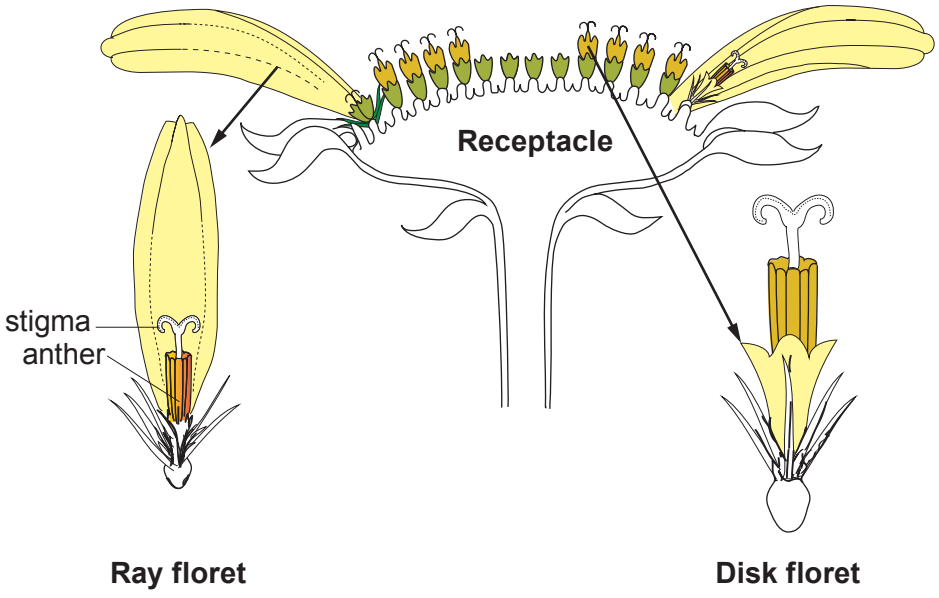
Answer **all** *questions.*

Section A

1. The photograph below shows one species of the genus *Senecio*, commonly known as groundsel. It is a plant that colonises open ground, crevices in walls and between paving slabs.

The flower head consists of many small modified flowers called florets which are cross pollinated. The structure of the flower and florets is shown.



Examiner only

(a) (i) State whether the florets shown in the diagram are genetically identical or genetically different. Explain your answer. [2]

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(ii) State **one** way shown in the diagram in which the structure of a floret reduces the chance of self-pollination. [1]

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(iii) Explain the advantage of preventing self-pollination. [2]

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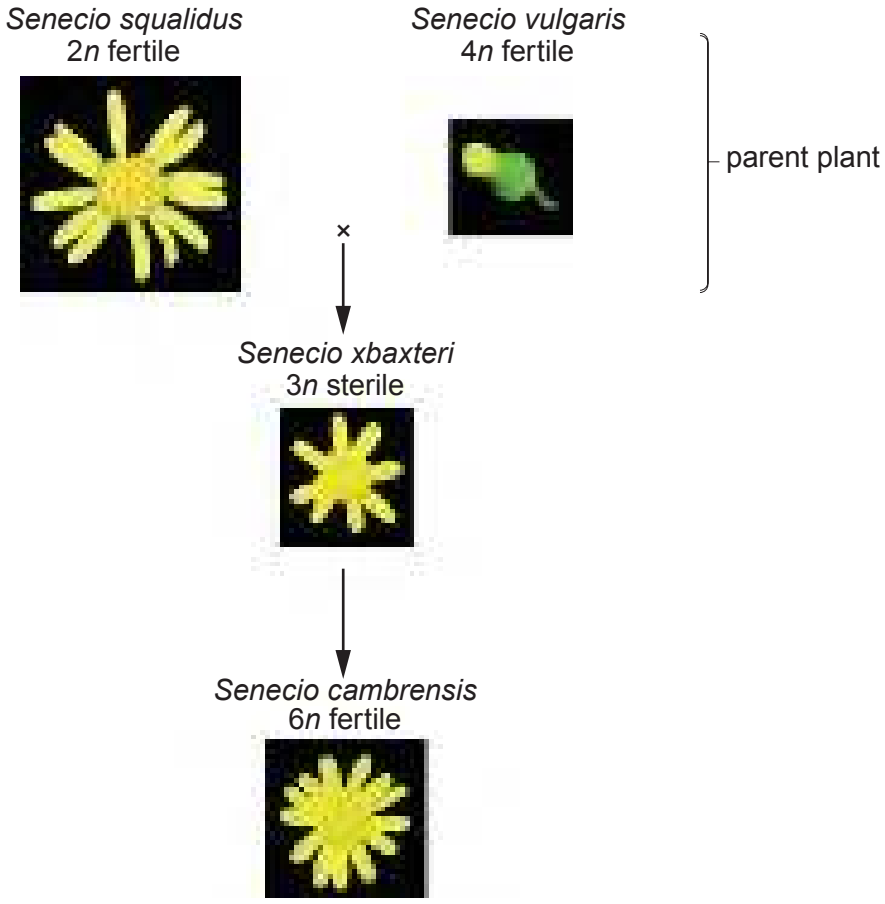
(b) In 1690, *Senecio squalidus* was introduced to the Oxford Botanic Garden from Italy. It escaped into the wild and became an invasive species. In North Wales, it hybridised with a native species, *Senecio vulgaris*, and gave rise to a new species, *Senecio cambrensis*, as shown below.



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(i) Leaf cells of *S.squalidus* contain 20 chromosomes. For this plant, state the number of chromosomes found in the following: [2]

I. Female gamete .....

II. Primary endosperm nucleus .....

III. Petal cells .....

(ii) *S.xbaxteri* has three copies of each chromosome. Describe the change that has resulted in the formation of the hybrid *S.cambrensis* and explain why it is fertile. [5]

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(iii) *S.cambrensis* appeared independently in Edinburgh in 1974. Originally there were 102 plants in three populations surrounded by parental plants growing on derelict land. Suggest **one** reason why the species became extinct in Edinburgh by 1993. [1]

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Examiner only

2. Duchenne muscular dystrophy (DMD) is a recessive sex-linked condition which affects 1 in 3500 boys. It is caused by a mutation of a gene which codes for the protein dystrophin, which gives structural stability to all membranes within muscle cells. As a result, calcium ions enter the mitochondria and they burst. This results in the death of the muscle cell. The mutation involves a deletion involving an intron and one or more exons.

(a) (i) Explain why a mutation involving an intron would not affect the primary structure of a polypeptide but a mutation in an exon might. [2]

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(ii) Explain why the mitochondria burst after calcium ions enter them. [2]

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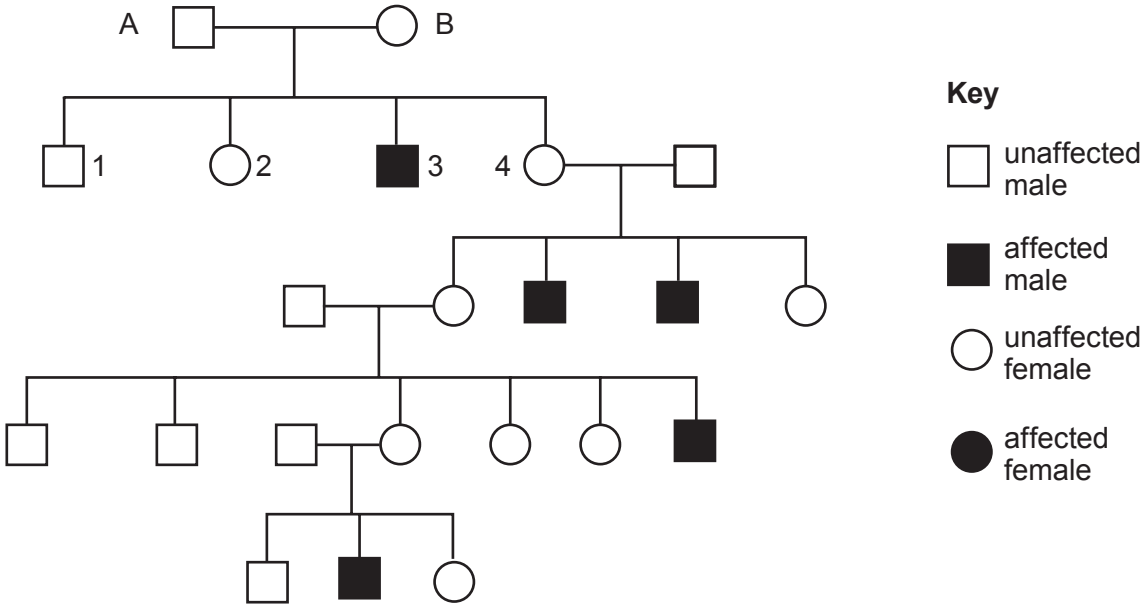
(b) The pedigree chart below shows the transmission of DMD in humans from one generation to another.



Examiner only

(i) Using the symbols $\mathbf{X^D}$ for unaffected and $\mathbf{X^d}$ for affected by DMD to represent alleles, construct a genetic diagram to show the genotypes and phenotypes of the parents A and B and the possible genotypes of their offspring. [4]

Parental phenotypes ....

....

Parental genotypes ....

Gametes ....

Offspring genotypes ....

(ii) Explain why it is not possible to determine the genotype of **child 2**. [1]

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(c) Through gene therapy, it is hoped that the functional version of the gene can be isolated and introduced into muscle cells. Historically, a virus has been used as the vector.

Suggest **two** potential problems of using a virus to treat DMD. [2]

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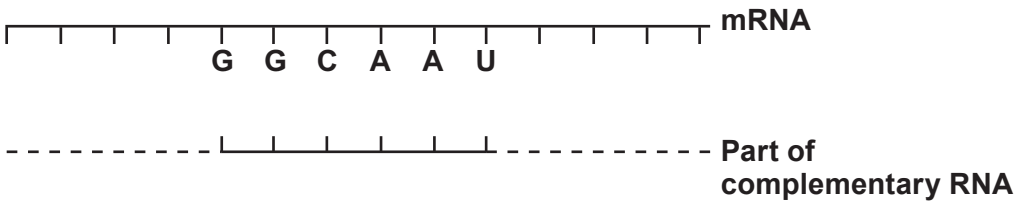
Examiner only

(d) DMD can be treated using a drug which acts as a 'molecular patch' on an mRNA molecule transcribed from the mutated DNA. The drug contains a short RNA molecule which is complementary to a specific sequence of bases on the mRNA. It prevents translation of the covered mRNA but allows ribosomes to continue translating the strand following the patch.

(i) **Complete the diagram** below to show the complementary base pairing between the mRNA and the molecular patch. [1]



(ii) State **one** difference between the structure of the protein dystrophin synthesised after the use of the molecular patch and normal dystrophin. [1]

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(iii) There is another drug available which is used to remove mutated exons from the dystrophin gene. State the advantage of using this drug to treat DMD rather than the molecular patch on mRNA. [1]

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(iv) Scientists are also investigating a means of using a molecular patch for germ line gene therapy. State **one** ethical issue of using germ line gene therapy in the treatment of DMD.

[1]

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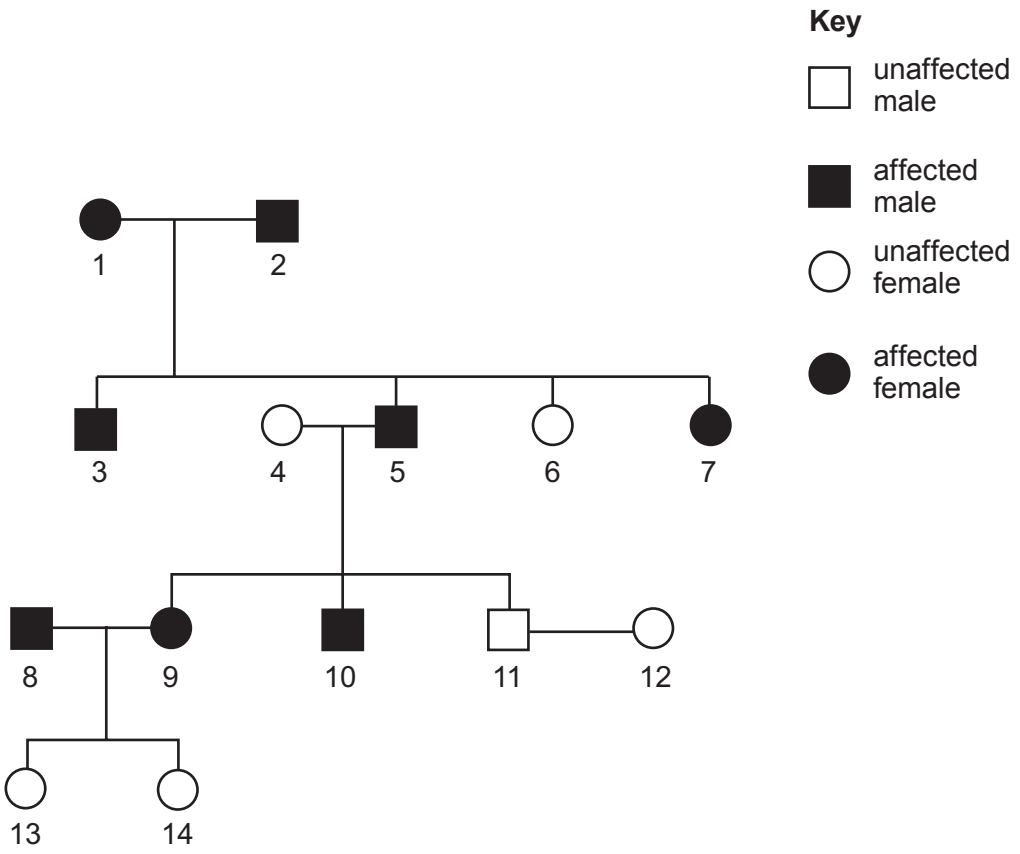
Examiner only

(e) The pedigree chart below shows the transmission of another form of muscular dystrophy caused by a gene mutation.



What conclusions can be made about the inheritance of this form of muscular dystrophy? **Giving 3 specific examples** from the pedigree chart, explain how you arrived at your conclusions. [5]

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3. *(a)* The micrograph below shows a human sperm cell.

5μm

(i) Calculate the actual length of the sperm cell. [2]

..... μm

(ii) The mitochondria in the sperm do not enter the secondary oocyte at fertilisation. What does this suggest about the origin of all mitochondria in an organism? [1]

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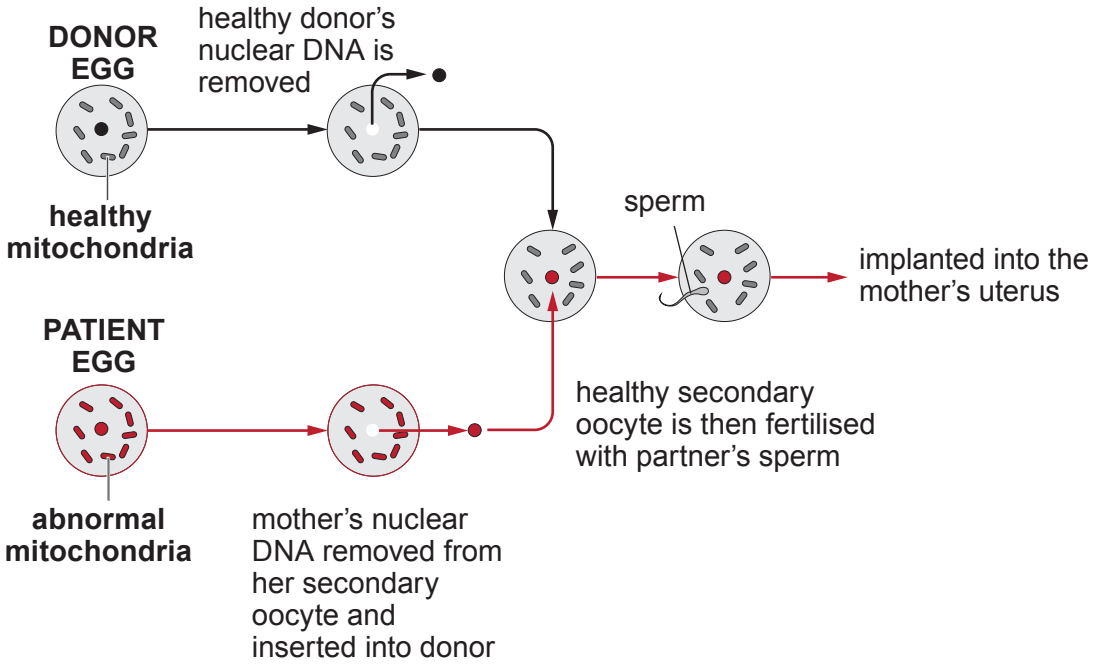
(iii) A woman has inherited a mutation in her mitochondrial DNA resulting in mitochondrial disease. Her husband and father do not have this disease. Complete the table below to indicate which of her relatives will suffer from the **same** mitochondrial disease. [2]

Relative	Mitochondrial disease (✓ or x)
Son	
Daughter	
Daughter's children	
Son's children	

Examiner only

(iv) The inheritance of mitochondrial diseases can be prevented using the technique outlined in the following diagram.



Using all of the information provided explain why a child produced by this method has been referred to as a **'three parent baby'**. [3]

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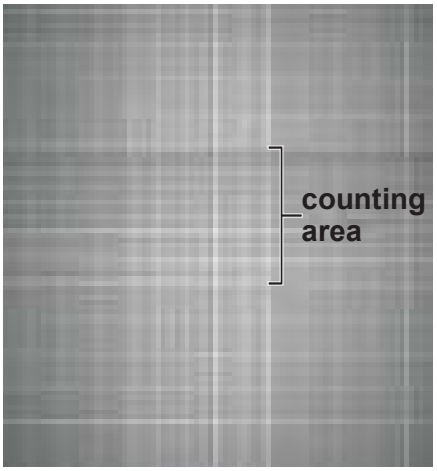
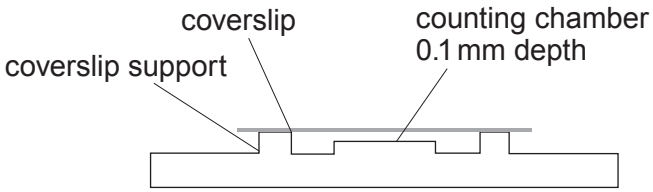
Examiner only

(b) A haemocytometer can be used to calculate the number of sperm in a sample of semen.

A haemocytometer is a microscope slide with a rectangular chamber at the centre which is engraved with a grid as shown below. A coverslip is supported over the chamber and the depth of the chamber is known. The number of sperm cells in a specific volume of semen can be counted.





The counting area is observed using a microscope and the number of sperm cells is counted. The sperm are killed before counting.

(i) Explain why this technique is not suitable for counting live sperm. [1]

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Examiner only

(ii) The semen sample was diluted by adding 0.1 cm^3 of semen to 9.9 cm^3 of solution. The number of sperm cells in the diluted sample in the counting area was 40. The counting area is 1 mm^2 with a depth of 0.1 mm.

I. Calculate the number of mm^3 in 1 cm^3. [1]

number = ...

II. Given that in each 0.1 mm^3 there are 40 sperm cells, calculate the number of sperm cells in 1 cm^3. [1]

number = ...

III. The semen sample was diluted by adding 0.1 cm^3 of semen to 9.9 cm^3 of solution. Calculate the number of sperm cells in the original 1 cm^3 of semen. [1]

number = ...

12

Turn over.

Examiner only

4. In addition to the ABO blood group system in humans there is another system called MN. It is caused by codominant alleles, M and N which code for proteins in the membrane of red blood cells.

(a) (i) The Hardy-Weinberg principle states that allele frequencies in a population remain constant from generation to generation providing certain conditions exist. State **three** of these conditions. [3]

1. .....

2. .....

3. .....

(ii) The frequency of allele N in Germany is 45%.
Calculate the following given that:

p = frequency of allele M,
q = frequency of allele N,
$p + q = 1$
$p^2 + 2pq + q^2 = 1$

I. The % frequency of allele M; [1]

.....

II. The number of individuals in a population of 10 000 with blood group M; [1]

.....

III. The number of individuals in a population of 10 000 with blood group N; [1]

.....

IV. The number of individuals in a population of 10 000 with blood group MN. [1]

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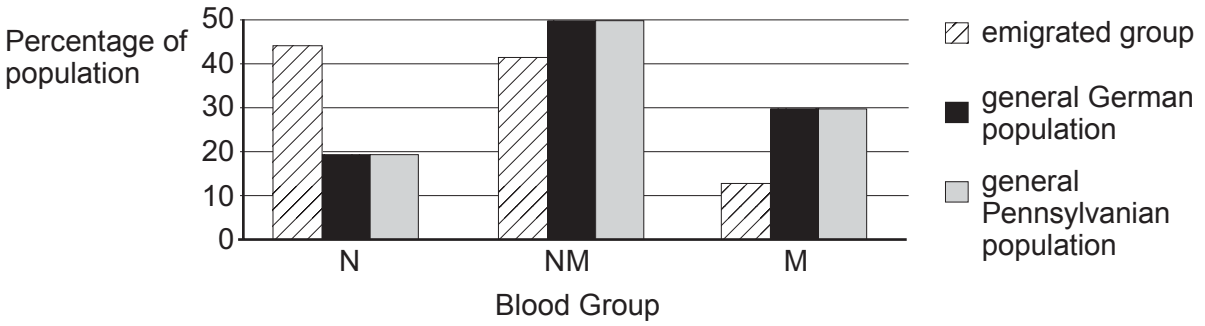
Examiner only

(b) During the eighteenth century, some groups of people emigrated from Germany to America. Since then, these people have married almost exclusively within their own communities.
In 1950, a blood group analysis of 200 members from one of these groups in Pennsylvania (a state in America) was undertaken. Their blood groups were compared with current German and American populations. Blood group analyses produced the following results:



Explain why the emigrated population shows allele frequencies which are uncharacteristic of either the American or German populations. [3]

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Examiner only

5. As maize seeds germinate they produce the enzyme amylase which hydrolyses starch in the endosperm into maltose. This can be demonstrated in the laboratory using the following method.

- Maize seeds are soaked in water.
- Seeds are cut in half and placed onto starch agar.
- After 24 hours the seeds are removed and iodine solution added to the starch agar.
- Clear zones around the position of the seeds indicate amylase activity.
- Boiled then cooled seeds are set up as a control.

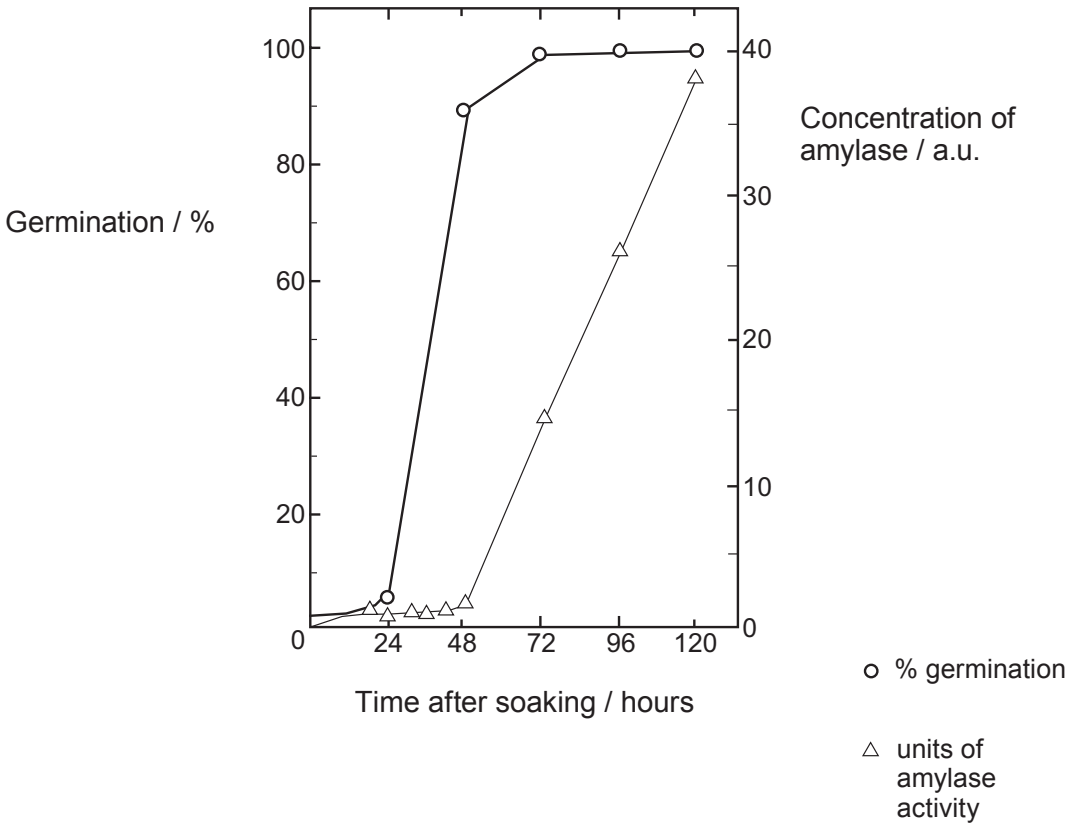
(a) Describe how you would develop and refine this practical procedure to determine whether treatment of seeds with a 0.1 mmol dm^{-3} solution of gibberellic acid increased production of amylase. [4]

Examiner only

(b) The following experiment was carried out to test the hypothesis that gibberellic acid stimulates amylase synthesis which then causes germination.

Oat seeds were soaked in 0.1 mmol dm^{-3} gibberellic acid and placed on moist filter paper. They were kept at 23°C and the number of germinated seeds were counted at various intervals after soaking. As soon as germination occurred the seeds were homogenised (blended) and the concentration of amylase in them was determined.

The results are shown in the graph below.



Using the evidence from the graph, state whether you would accept or reject the hypothesis. Explain your answer. [2]

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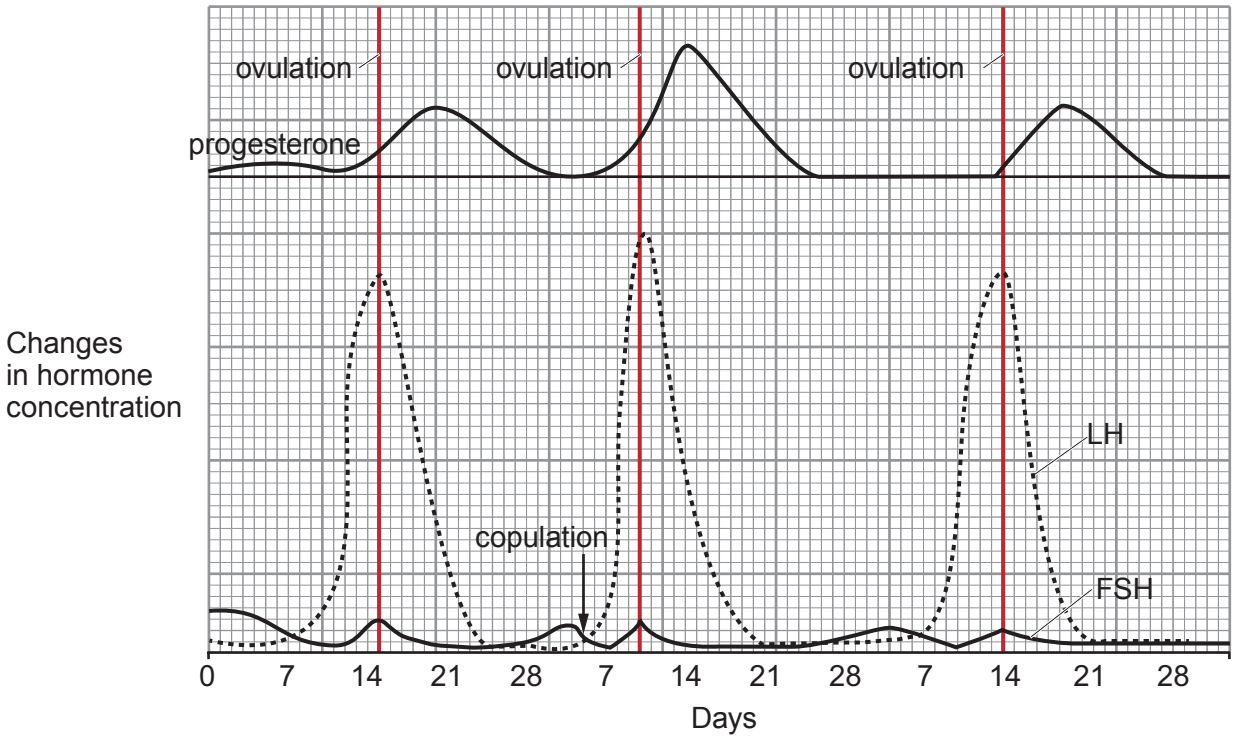
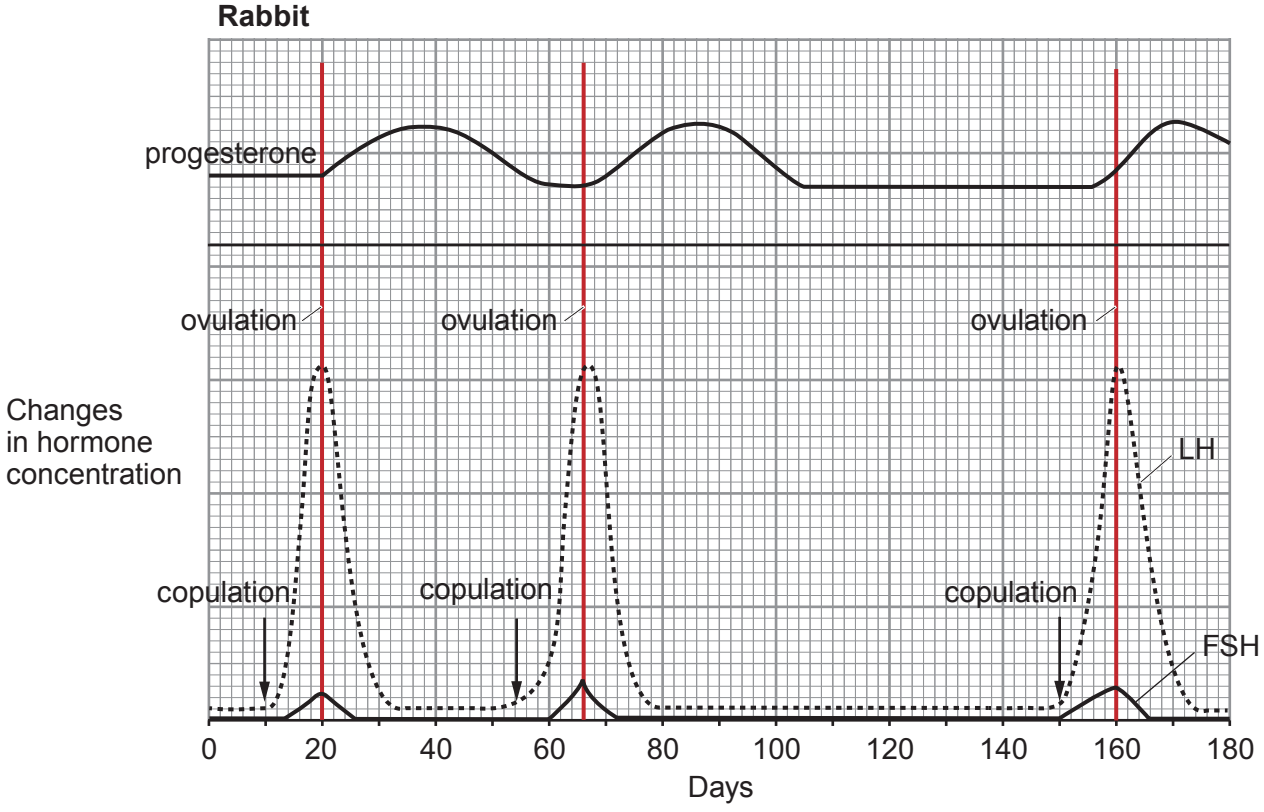
6. Mammals have evolved different patterns of ovulation which are controlled by changes in hormone concentrations and affected by copulation (sexual intercourse). The graphs below show the changes in concentrations of LH, FSH and progesterone during the menstrual cycles of a human and a rabbit.

Human



Rabbit



Examiner only

Using the graphs and your knowledge of hormonal control of the human reproductive cycle, explain the effects of the hormonal changes which are shown. Discuss the similarities and differences in the control of ovulation in rabbits and humans. [9 QER]

Turn over.

Examiner only

Examiner only

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9

SECTION B: OPTIONAL TOPICS

Option A: **Immunology and Disease** ☐

Option B: **Human Musculoskeletal Anatomy** ☐

Option C: **Neurobiology and Behaviour** ☐

Answer the question on **one topic only**.

Place a tick (✓) in **one** of the boxes above, to show which topic you are answering.

You are advised to spend about 25 minutes on this section.

Examiner only

Option A: Immunology and Disease

7. Lyme disease, or Lyme borreliosis, is caused by the bacteria *Borrelia burgdorferi* and is endemic in some areas. There are a number of strains of this bacterium. The bacteria are transmitted to humans through the bite of an infected tick. Reported cases in England and Wales rose from 268 in 2001 to 959 in 2011, with an increase in the number of cases in Snowdonia National Park.

Ticks are tiny, spider-like creatures found in woodland and heath areas. They feed on the blood of birds and mammals, including humans. Ticks that carry the bacteria responsible for Lyme disease are found throughout the UK and in other parts of Europe and North America.

(a) State what is meant by the term *endemic*. [1]

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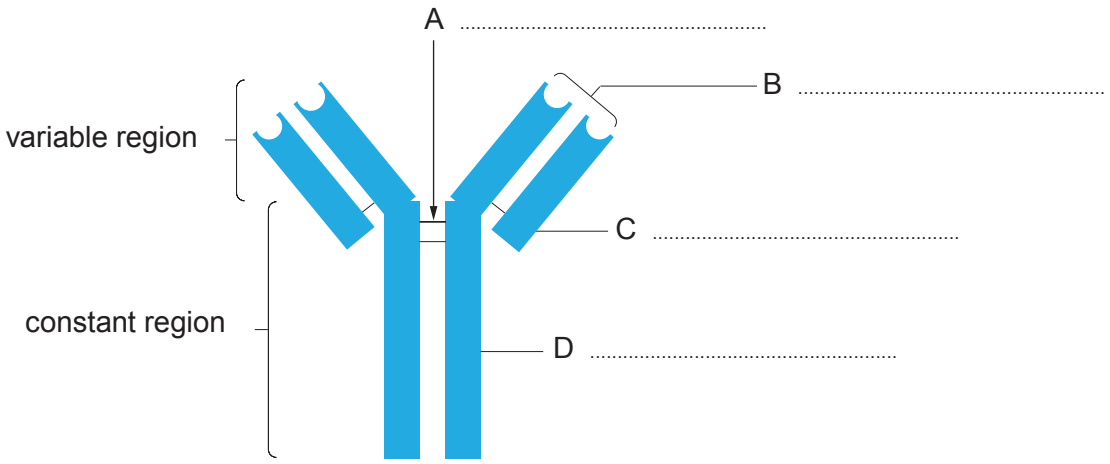
The immune system of an infected person will react by producing specific antibodies to the bacterial antigens.

(b) (i) Label the diagram below to identify the regions of the antibody. [3]



(ii) Define the term *antigen*, and explain the meaning of an antigen-antibody complex. [2]

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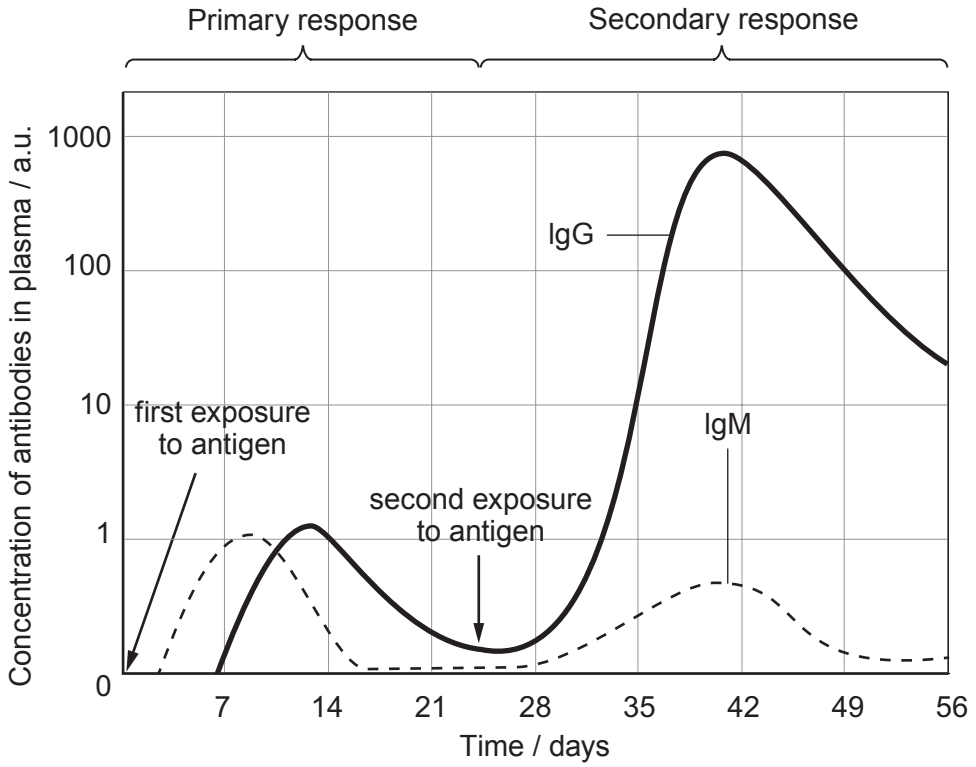
Examiner only

(c) The body's response to infection shows both a primary response and a secondary response to the foreign antigens. Both responses can be identified by measuring the levels of two antibodies found in the blood, IgM and IgG.
IgM is produced mainly by B-cells on initial exposure; IgG antibodies are produced in higher levels during the secondary response.



(i) Explain why the level of IgG is higher after the second exposure to the antigen than the first exposure. [1]

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(ii) Lyme disease can persist in patients for many years and the IgM levels remain high. Suggest possible reasons why. [2]

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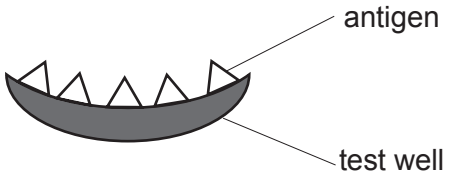
Examiner only

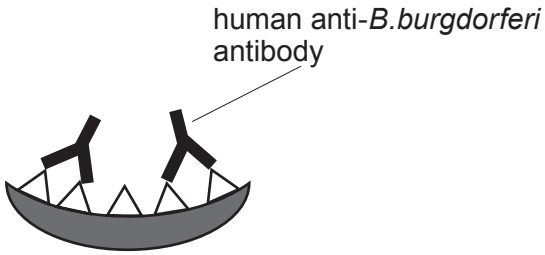
(d) The symptoms of Lyme disease include fever, chills, fatigue, headaches and muscle aches. As a consequence, patients with Lyme disease are often misdiagnosed.

An ELISA (enzyme-linked immunosorbent assay) can be carried out to diagnose Lyme disease using a blood sample from the patient around **two weeks** after infection. It is based on detecting the antibodies made in response to being exposed to *B.burgdorferi*.

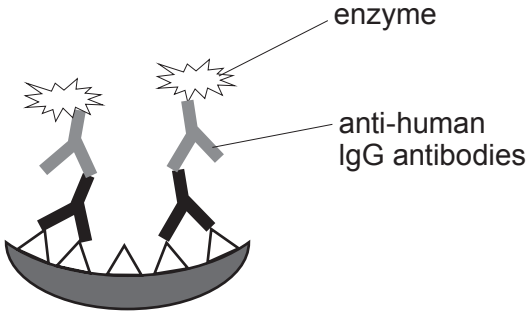
The results of the assay show the concentration of IgG antibodies in the patient's blood. The steps involved in the ELISA are shown below.



Specific antigen from *B.burgdorferi* attached to the surface of the test well.



Patient's blood plasma added to the well. Human antibodies bind to the *B.burgdorferi* antigen.



Enzymes attached to anti-human IgG antibodies cause a colour change if an antigen-antibody complex forms. Greater colour intensity indicates a higher IgG concentration.

(i) Using the graph, explain why carrying out an ELISA would not detect the IgG antibodies in the days immediately after the tick bite. [2]

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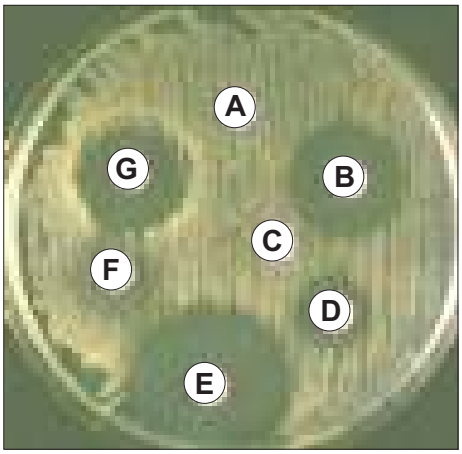
Examiner only

(ii) The ELISA can produce false negative results for Lyme disease. In a false negative result there is no colour change, even if the patient is infected with the bacteria.

Suggest why, even if there are anti-*B.burgdorferi* antibodies in the patient's plasma, the enzyme may not cause a colour change. [5]

Examiner only

(e) *B.burgdorferi* is a Gram-negative bacterium and can be treated using a bacteriostatic antibiotic which stops protein synthesis.
The image below is of an agar plate showing the results of testing various antibiotics on *B.burgdorferi*.



(i) State the temperature at which the culture should be incubated.
Explain your answer. [1]

.....

.....

(ii) The most effective antibiotic was found to be E. The diameter of the zone of inhibition was 22 mm. Calculate the area of the zone of inhibition caused by E to a suitable level of precision. [2]

Formula for the area of a circle: πr^2

$\pi = 3.14$

answer = ...

(iii) With reference to the image, suggest what assumption is being made when making this calculation. [1]

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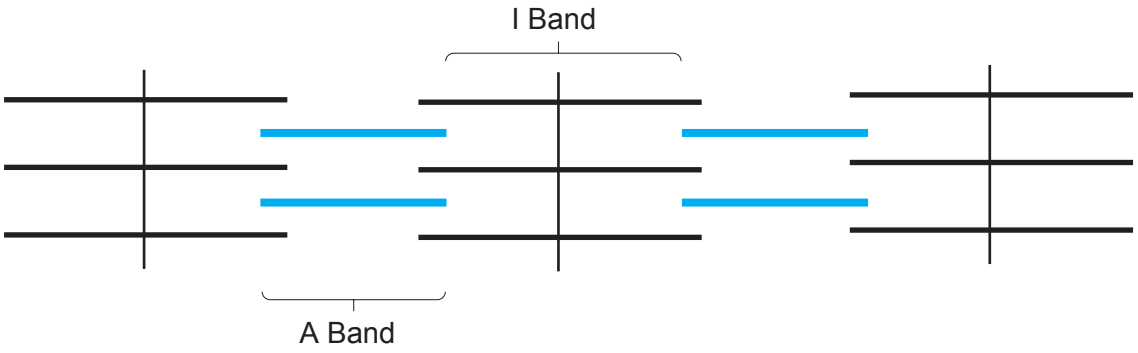
Option B: Human Musculoskeletal Anatomy

8. In 1953, Huxley introduced the sliding filament theory to explain muscle contraction. This theory was based on the idea that muscle proteins slide past each other to generate tension. Below is a diagram of two sarcomeres.



(a) (i) Identify the main proteins found within each of the regions using ticks (✓) to complete the following table. [2]

	Actin	Myosin	Troponin	Tropomyosin
A Band				
I Band				

(ii) Explain how the interaction between actin and myosin results in the contraction of muscle. Reference to the roles of troponin and tropomyosin are **not** required. [2]

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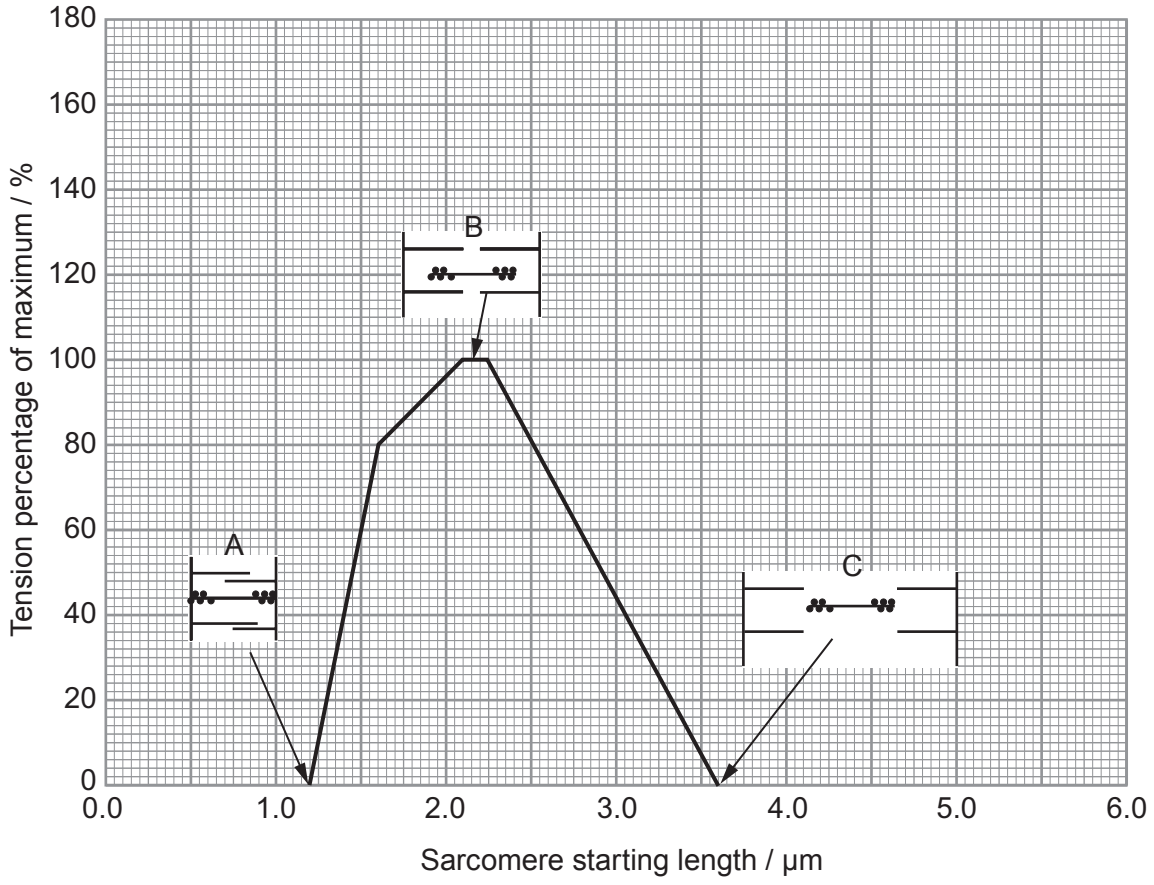
Examiner only

Muscle samples can be analysed in order to produce a length-tension curve using the following procedure:

- muscle fibres are suspended in a solution
- muscles are positioned so that sarcomeres are at different lengths as shown at A, B and C on the graph below
- muscle fibres are stimulated to contract
- the tension (force) generated as a percentage of the maximum is measured at each sarcomere length (A, B and C)

The results are shown on the graph below **(diagrams of sarcomeres are not drawn to scale)**.



(b) (i) To determine the length of sarcomeres at different resting positions, myofibrils were viewed using a high-powered microscope. The muscle proteins were stained using fluorescent chemicals that bonded to the actin and myosin. Explain why at least 20 sarcomeres were measured. [1]

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Examiner only

(ii) Use the **graph** to calculate the length of the actin fibre. [3]

actin length = ...

(iii) Physiologists conducting these experiments used tissue from the same organism. Suggest **two** other factors that would need to be controlled when carrying out the investigation. [2]

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(iv) Suggest an explanation as to why no tension is generated at **C** on the graph. [2]

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Examiner only

(c) Movement of the legs in humans involves the use of muscles attached by tendons to the bones. The quadriceps and hamstring muscles work antagonistically to move the knee joint and raise the leg. The image below shows the arrangement of muscles and tendons in the knee joint.

(i) Explain why skeletal muscles are arranged in antagonistic muscle pairs. [1]

.....

Scientists studied patients with osteoarthritis and a control group without the condition. Some of the measurements made are shown in the table below.

	Osteoarthritis Group	Control Group
Mean age	65	65
Maximum voluntary contraction force / N	444.50	486.10
Stair climbing time / s	13.08	8.82

(ii) With reference to the role of the quadriceps in movement of the lower leg, explain the differences between the osteoarthritis and control groups. [2]

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Examiner only

Body mass index (BMI) is a measure that relates body mass to height. The mean BMI of the two groups was calculated:

Mean BMI of osteoarthritis group = 30.6

Mean BMI of control group = 24.1

A healthy BMI range is between 21 and 25. Values in excess of 30 are classified as obese.

(iii) Explain the effect of a higher BMI on the results in the table and suggest why increased physical activity would reduce the impact and progression of osteoarthritis. [5]

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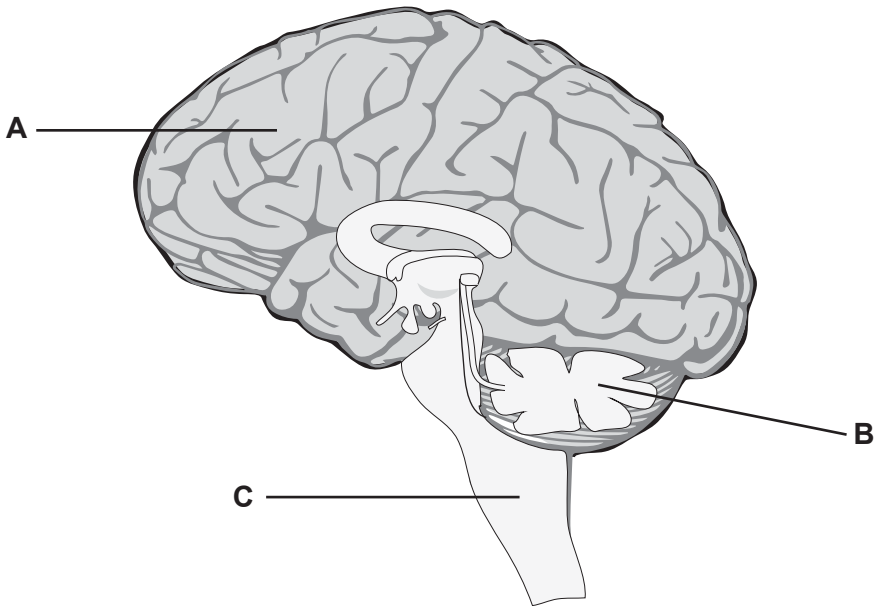
Examiner only

Option C: Neurobiology and Behaviour

9. The diagram below shows a section through the human brain.



(a) Identify **A**, **B** and **C** on the diagram above. [1]

A .....

B .....

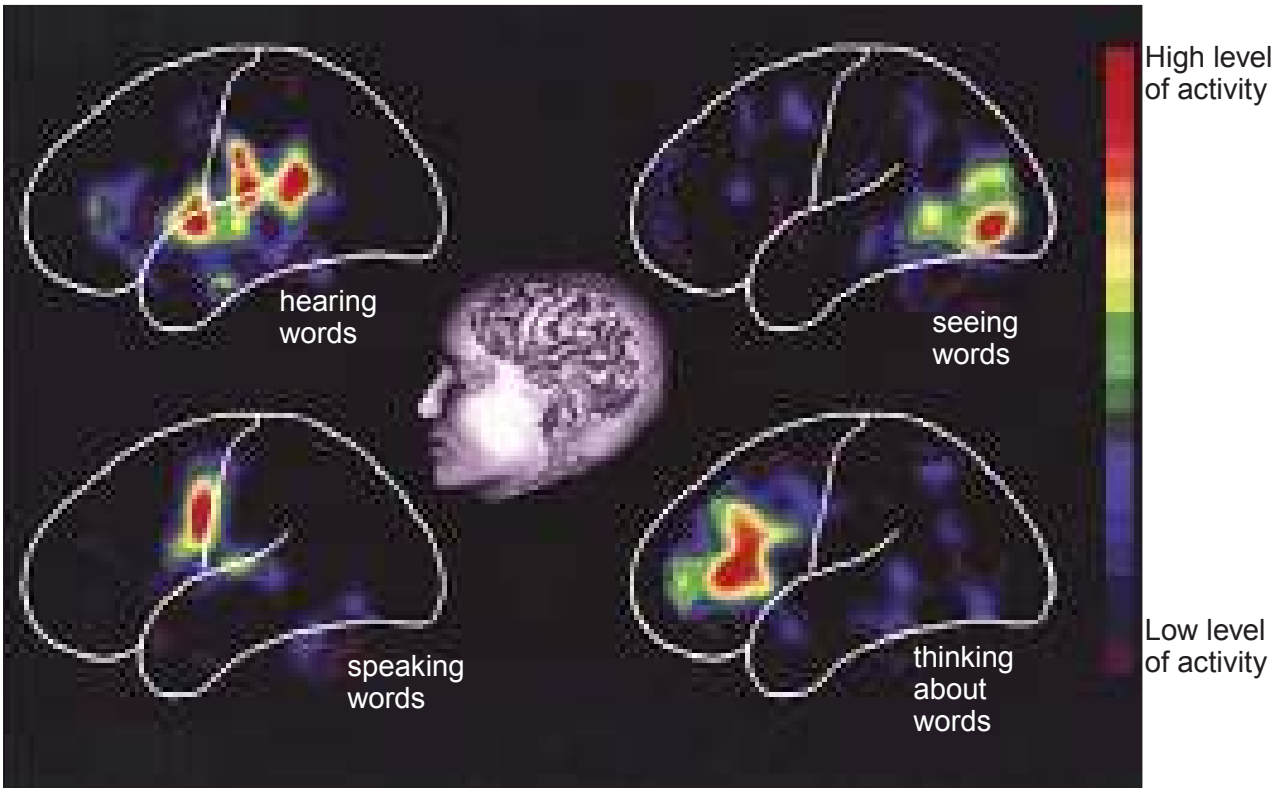
C .....

Examiner only

(b) It is estimated that there are 125 000 deaf adults in the UK who use British Sign Language (BSL). The language involves movement of the hands, body, face and head. Some people are born deaf and others become deaf during their lives. There are many causes of deafness, including damage to the auditory nerve between the ear and the brain.

Positron emission tomography (PET) can be used to generate images of the brain using radioactive tracers which have a short half-life. There is a PET scanner at Cardiff University.

The image below shows the results of a PET scan when a hearing patient was given tasks associated with language.



(i) Explain why a PET scan would be suitable for investigating activity levels within the brain whereas an MRI scan would not. [2]

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Examiner only

(ii) With reference to the image of the PET scan and your knowledge of brain regions involved in language, suggest and explain how the PET scan of a deaf person, actively using BSL, would compare to that of a hearing person. [5]

(iii) Suggest **two** medical details that would be needed when interpreting PET scans from patients with hearing problems. [2]

(iv) Explain why PET scans of adults with total hearing loss have revealed activation of regions of the cortex related to hearing. [3]

Examiner only

(c) The three-spined stickleback, *Gasterosteus aculeatus*, is common to lakes and rivers in Wales. The male stickleback has a red belly during the breeding season and behaves aggressively when defending its territory.

The behaviour of 12 male sticklebacks was investigated during the breeding season. Each male stickleback was exposed to model sticklebacks with red or silver bellies and the number of times each model was bitten was recorded.

Male and female sticklebacks during the breeding season, in Llyn Frongoch, Ceredigion

(i) State **one** advantage to male sticklebacks of defending their territory. [1]

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The table below shows the results of the investigation.

Model colour	Mean number of bites
red belly	119.8
silver belly	60.4

(ii) State what is meant by a **sign stimulus** and based on this information, identify what it would be for the stickleback. Explain your answer. [3]

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Examiner only

Model colour	Mean number of bites	$\sum(x-\bar{x})^2$	Standard Deviation
red belly	119.8	524.7	6.9
silver belly	60.4	343.3	.....

(iii) **Complete** the table above by calculating the standard deviation for the silver belly stickleback using the formula below: [2]

$$\textit{Standard Deviation} = \sqrt{\frac{\sum(x-\bar{x})^2}{N-1}}$$

where:

$\bar{x}$ = mean
$\sum$ = sum of
N = number of samples

(iv) The standard deviation for both models was high. Explain how this could affect your confidence in the conclusion. [1]

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