| Surname | Centre Number | Candidate Number |
|---|---|---|
| First name(s) | | 2 |

# GCE A LEVEL

1400U40-1

S24-1400U40-1

**FRIDAY, 14 JUNE 2024 – MORNING**

## BIOLOGY – A2 unit 4
### Variation, Inheritance and Options

2 hours

| | For Examiner's use only | | |
|---|---|---|---|
| | **Question** | **Maximum Mark** | **Mark Awarded** |
| **Section A** | **1.** | **11** | |
| | **2.** | **15** | |
| | **3.** | **13** | |
| | **4.** | **10** | |
| | **5.** | **12** | |
| | **6.** | **9** | |
| **Section B** | **Option** | **20** | |
| | **Total** | **90** | |

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01

**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.

You may use a pencil for graphs and diagrams only.

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 additional page(s) 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**: 20 marks; Options. Answer **one option only**. You are advised to spend 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 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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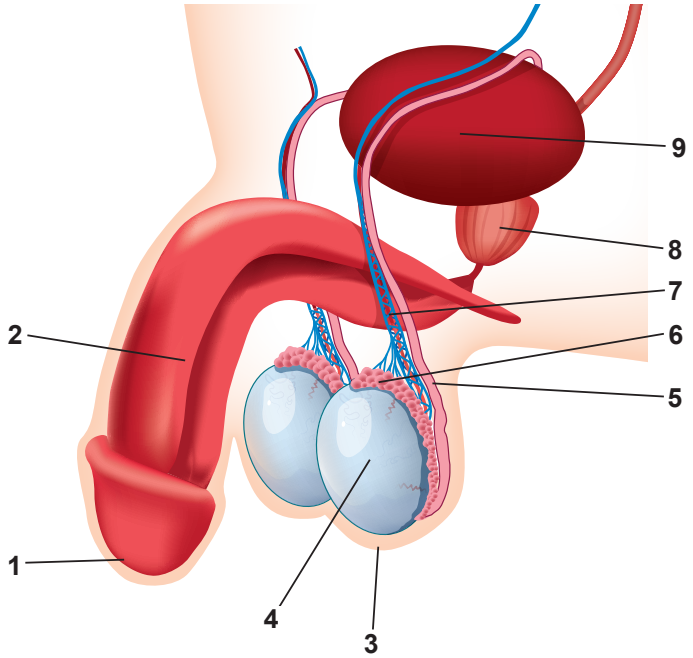
## SECTION A

Answer **all** questions.

**1.** The male reproductive system consists of a number of sex organs that play a role in the process of human reproduction. These organs are located on the outside of the body and within the pelvis as shown in **Image 1.1**.

**Image 1.1**




(a) Using the numbers in **Image 1.1**, identify the following structures and briefly describe **one** function for each structure. [5]

| Structure | Number from **Image 1.1** | Function |
|---|---|---|
| vas deferens | ..... | ..... |
| scrotum | ..... | ..... |
| testes | ..... | ..... |
| epididymis | ..... | ..... |
| prostate gland | ..... | ..... |



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(b) Compare the function of the urethra in males and females. [2]

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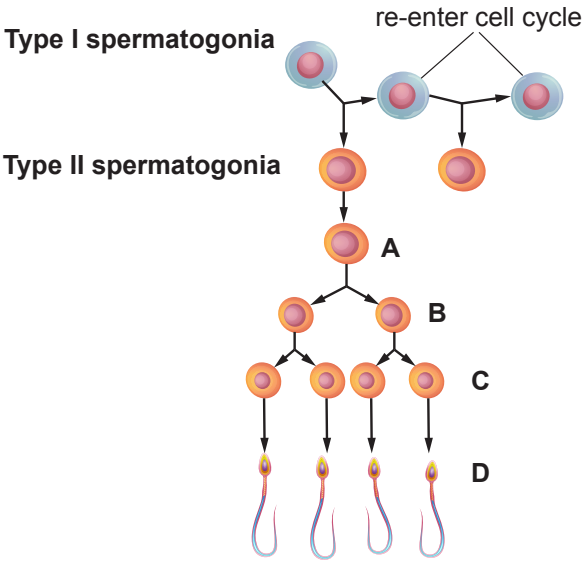
**Image 1.2** shows the process of spermatogenesis.

**Image 1.2**




(c) (i) Name the cells **A**, **B**, **C** and **D** in **Image 1.2**. [2]

| Cells | Name |
|---|---|
| **A** | ..... |
| **B** | ..... |
| **C** | ..... |
| **D** | ..... |

(ii) High numbers of spermatozoa must be constantly produced to ensure fertility. Use your knowledge of **cell division** to explain why it is important that all type I spermatogonia do not become type II spermatogonia. [2]

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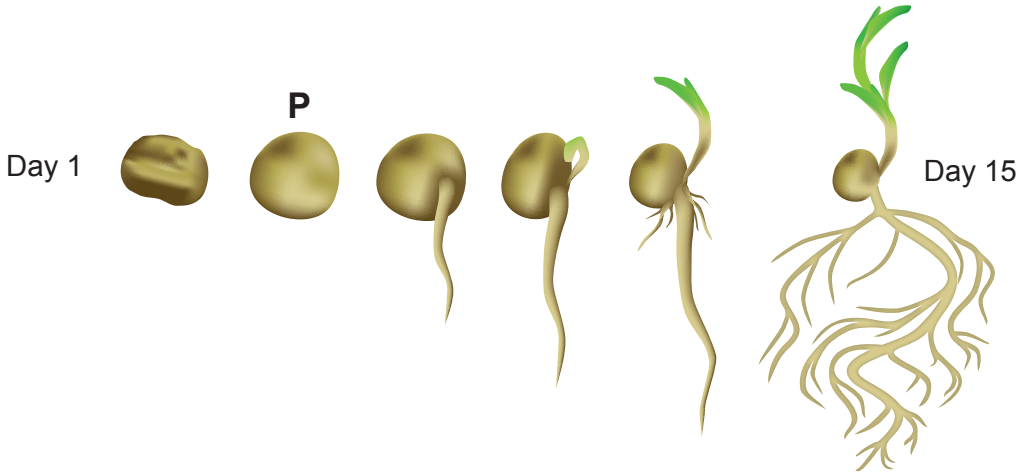
**2.** Germination involves the rapid onset of biochemical activity and growth of a seedling until the plant can carry out photosynthesis and become independent of the food stores contained in the cotyledons. **Image 2.1** shows some stages of a germinating pea plant over a 15-day period.

**Image 2.1**




(a) (i) The germinating pea seed labelled **P** in **Image 2.1** was cut open. This is shown in **Image 2.2.**

**Image 2.2**

**Add three labelled lines** to identify the **cotyledon, plumule and radicle** on **Image 2.2**. [2]

(ii) State **two** reasons why it is important that the radicle emerges prior to the plumule. [2]

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(b) Briefly describe the stages of germination in a **non-endospermic** seed such as the pea. [3]

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**Question continued overleaf**



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Gibberellin is required for maize kernel (seed) germination. It is blocked by another plant hormone, abscisic acid (ABA) whilst the kernel is maturing on the cob. Mutant kernels which cannot produce ABA germinate whilst still on the cob. This is shown in **Image 2.3**.

**Image 2.3**

(c) With reference to the function of gibberellin, explain why the mutant kernels germinate causing the appearance of the corn cob in **Image 2.3**. [3]

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(d) Field trials were carried out to investigate the effect of increasing soil temperature on the average number of wheat plants that grow. The results are shown in **Table 2.4**. In each field, the wheat grains were planted at a depth of 4 cm at a rate of 100 kg per hectare and the temperature was measured in each field at a depth of 5 cm.

**Table 2.4**

| Mean maximum soil temperature /°C | Number of mature wheat plants per $m^2$ |
|---|---|
| 20.2 | 315.3 |
| 33.2 | 256.7 |
| 42.2 | 89.8 |

(i) One simple conclusion from this data is that higher soil temperatures reduce the number of mature wheat plants per square metre.
State **four** other factors that could affect the growth and so your confidence in this conclusion. [2]

I. .....

II. .....

III. .....

IV. .....

(ii) Assuming that this was a valid conclusion, state why the results of this research are of serious significance to worldwide wheat production. [3]

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**3.** An artificial gene coding for human growth hormone (HGH), was synthesised in a laboratory. The synthesised gene was used to genetically modify *E. coli* bacteria, which then produced HGH. The synthesised HGH was identical to natural HGH in size and biological activity.

(a) The human genome project determined the base pairs that make up human DNA. Explain how the results from the human genome project have allowed the synthesis of the artificial gene to become possible. [1]

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(b) State **three** advantages of using an artificially synthesised gene rather than extracting the gene from the human genome. [3]

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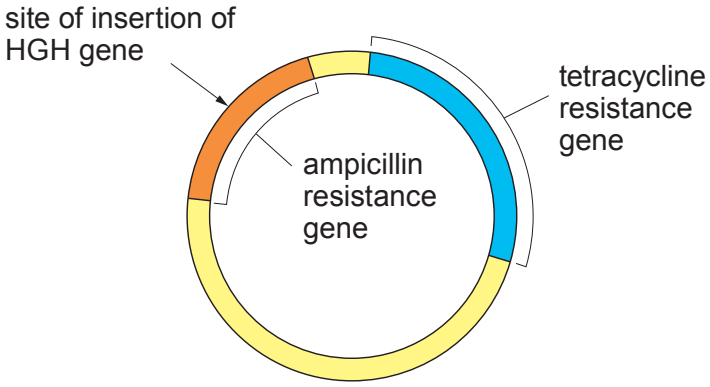
The plasmid used to genetically modify the *E. coli* is shown in **Image 3.1**. It also shows the positions of two antibiotic resistance genes and the site of insertion of the HGH gene.

**Image 3.1**




(c) Name the **two** types of enzymes needed for the successful insertion of the human growth hormone gene and state their function in the process. [2]

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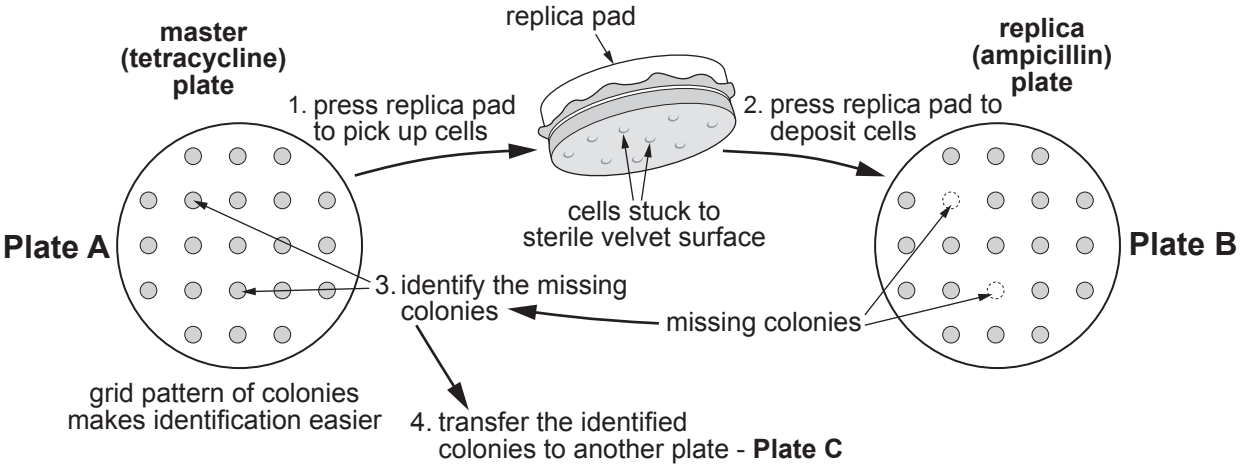
To test that the plasmid has transferred successfully, the bacteria were grown on separate agar plates containing the two antibiotics. The bacteria were initially grown on agar containing tetracycline and the colonies were then transferred, using a replica pad, to agar containing ampicillin as shown in **Image 3.2**.

**Image 3.2**




(d) (i) Explain the need for **Plate A** in the process. [1]

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(ii) Explain the results shown on **Plate B**. [3]

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(iii) Suggest **one** vital property of the cells on **Plate C** which would need to be confirmed prior to their use in the production of HGH. [1]

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(e) Each *E. coli* cell transformed with this recombinant plasmid produced $2.9 \times 10^6$ molecules of HGH. If there are $1 \times 10^8$ bacteria in the culture vessel, calculate the number of HGH molecules being produced by the cells. **Give your answer in standard form**. [2]

Number of HGH molecules = .....

13



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**4.** Bones can be preserved as fossils so research into evolutionary patterns and processes can include extinct vertebrates. **Image 4.1** shows how the skeleton of the modern horse has evolved from a horse-like mammal over the last 38 million years.

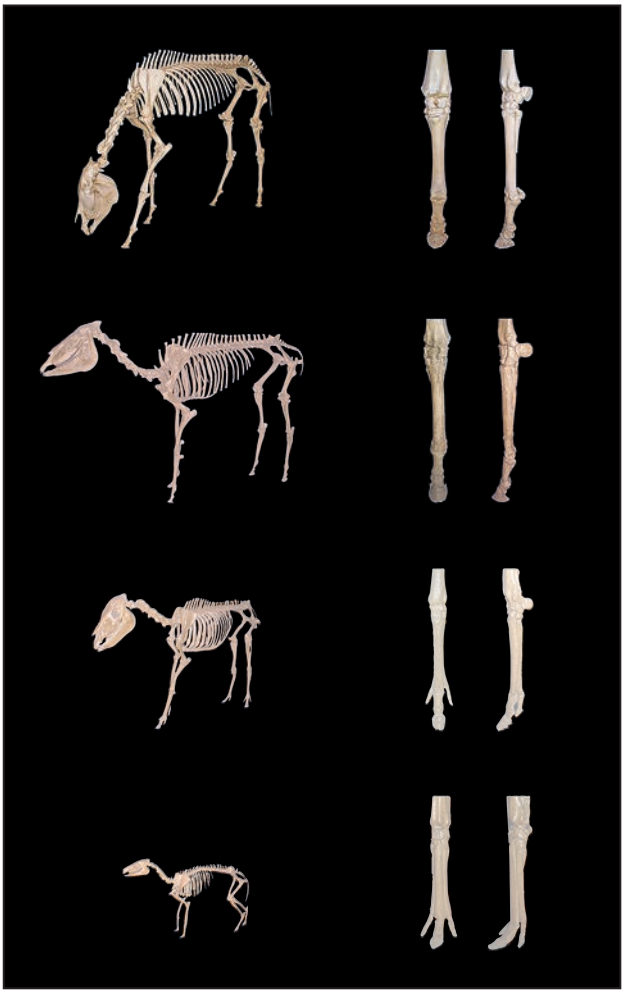
**Image 4.1**

***Equus***
Recent

***Pliohippus***
Late Miocene
7 MYA

***Merychippus***
Middle Miocene
11 MYA

***Mesohippus***
Late Eocene
38 MYA



MYA = Million Years ago

(a) Explain how the information in **Image 4.1** supports Darwin's theory of evolution. [3]

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Ancient horses had many coat colours. One type of gene controlling depth of colour in horses is the dun allele (**D**). For example the bay horse in **Image 4.2** is homozygous for the **d** allele but a horse is a dun if it has at least one **D** allele **(Image 4.3)**.

**Image 4.2** Bay (**dd**)

**Image 4.3** Dun (**DD** or **Dd**)

(b) (i) In a sample of 900 wild horses, 891 were dun and 9 were bay.
Calculate the frequencies of the three genotypes within this population, assuming that the population is in Hardy-Weinberg equilibrium. [3]

$p + q = 1$

$p^2 + 2pq + q^2 = 1$

where
p = frequency of the dominant allele (**D**)
q = frequency of the recessive allele (**d**)

Frequency of **dd** = .....

Frequency of **DD** = .....

Frequency of **Dd** = .....



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(ii) The population in (b)(i) consisted of randomly mating horses in semi-desert conditions.
Use all the information given to give **two** reasons why this population may not be in Hardy-Weinberg equilibrium. [2]

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**Question continued overleaf**

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**Images 4.4A** and **4.4B** show photographs of a mule and a hinny.

**Image 4.4A – Mule**

**Image 4.4B – Hinny**

(c) Mules are considered more patient, hardy and long-lived than horses and are described as less obstinate and more intelligent than donkeys. They can carry heavy loads for long distances so are an ideal pack-animal.

A mule is a hybrid resulting from crossing a male donkey (*E. asinus*) with a female horse (*E. caballus*).

(i) Explain why a breeder would need to use a donkey and a horse **each time** they wished to produce a mule. [1]

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(ii) The opposite cross (a hinny) can also be carried out, but it is done very rarely. The mother is the much smaller donkey compared with the larger horse mother in a mule cross. Explain why the mule cross is more suitable for a load-bearing pack animal. [1]

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**5.** There are many sex-linked genetic diseases in humans which cause changes in phenotype.

X-linked inheritance means that the gene is located on the X chromosome.

X-linked severe combined immunodeficiency (X-SCID) is a recessive immunodeficiency disorder in which the body produces very few lymphocytes.

(a) (i) I. Use the symbols $\mathbf{X^D}$ for unaffected by X-SCID and $\mathbf{X^d}$ for affected by X-SCID to draw a genetic diagram to show how parents who are both unaffected can produce a son with X-SCID. [2]

| | | | |
|---|---|---|---|
| Parental phenotype | unaffected male | × | unaffected female |
| Parental genotypes | ..... | | ..... |
| Gametes | ..... | | ..... |

Space for genetic diagram

II. **Complete the table** below linking the **four** different genotypes from your genetic diagram with their correct phenotype. [2]

| Offspring genotype | Offspring phenotype |
|---|---|
| ..... | ..... |
| ..... | ..... |
| ..... | ..... |
| ..... | ..... |

(ii) The estimated UK incidence of X-SCID is 1 in 48 933. The population of Wales in 2018 was 3 139 000.
Calculate the number of cases of X-SCID that would have been expected in Wales in 2018.
**Give your answer to an appropriate number of significant figures.** [2]

Number of cases = .....

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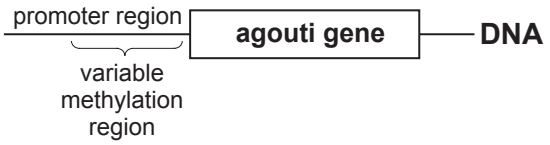
There are other mechanisms, such as epigenetic changes, which can alter the phenotype of organisms. One example is DNA methylation, where a methylated promoter region decreases the expression of a gene and demethylation increases the expression of a gene. This is shown in **Image 5.1**.

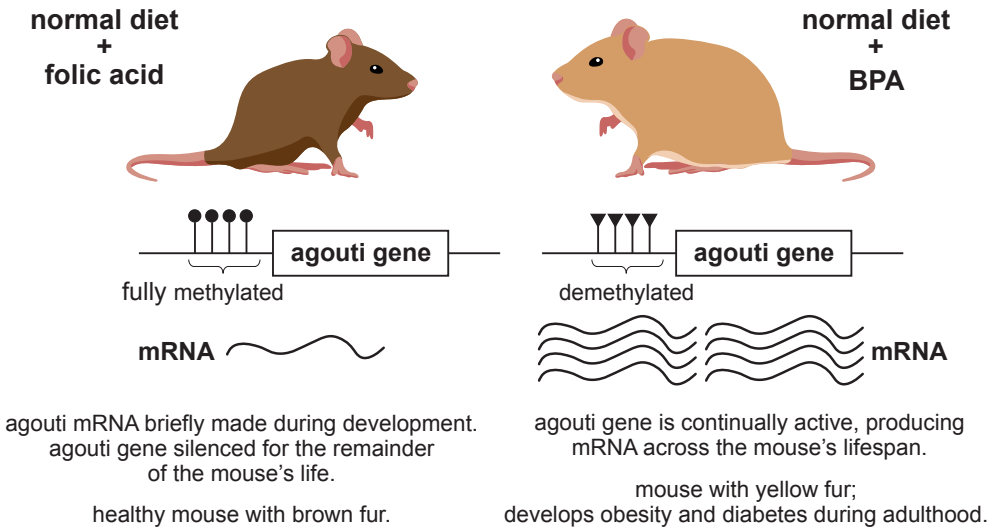
**Image 5.1**




Epigenetic control mechanisms can be seen in coat colour in mice which is influenced by the nutrition of the pregnant female.

When the pregnant females are fed compounds such as BPA (used in plastic production) with their normal diet, this can cause more yellow offspring to be born. When the pregnant females' normal diets are supplemented with high folic acid, more brown offspring are born. This is summarised in **Image 5.2**.

**Image 5.2**




When mice were fed the two different diets, the following results were obtained, with ranges in coat colour as shown in **Image 5.3** and number of each type shown in **Graph 5.4**.

**Image 5.3**

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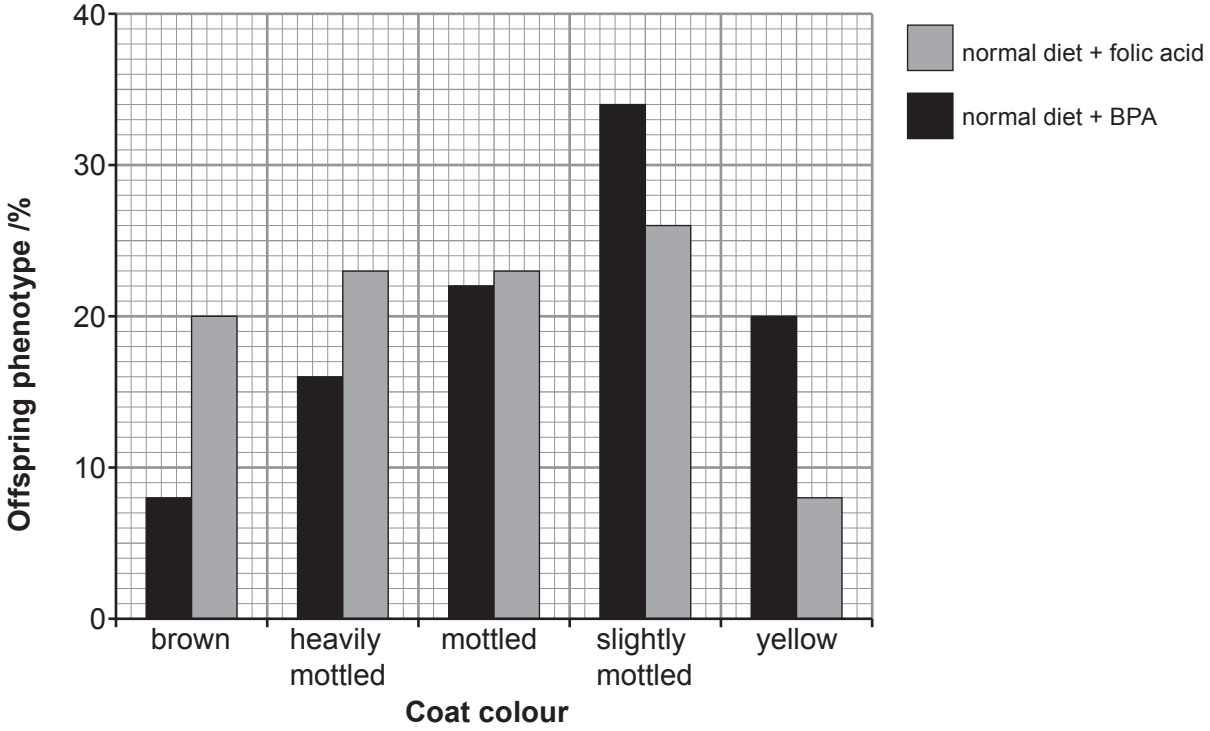
**Graph 5.4**




(b) (i) Using **Images 5.1, 5.2, 5.3 and Graph 5.4**, explain the effect of the **two** different supplements on gene expression and coat colour in mice. [4]

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(ii) Use all of the information to suggest how the results of this study might influence the use of BPA in the production of food containers and packaging. [2]

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**6.** Mutations in the DNA of an organism may have a profound effect on the phenotype. There are two main types of mutations; gene and chromosome.

Explain what is meant by the term mutation and describe some factors which increase the rate of mutations.

Describe the different types of gene and chromosome mutations and, using suitable examples, explain the different effects that gene and chromosome mutations may have on the phenotype. [9 QER]



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## 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.**





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## Option A: Immunology and Disease

**7.** Tuberculosis (TB) is the leading cause of death by an infectious disease worldwide.

(a) (i) State why tuberculosis is called an infectious disease. [1]

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(ii) To prevent the spread of TB, the BCG vaccination was introduced in 1953 for children aged 10–14 years old. Define the term vaccine. [1]

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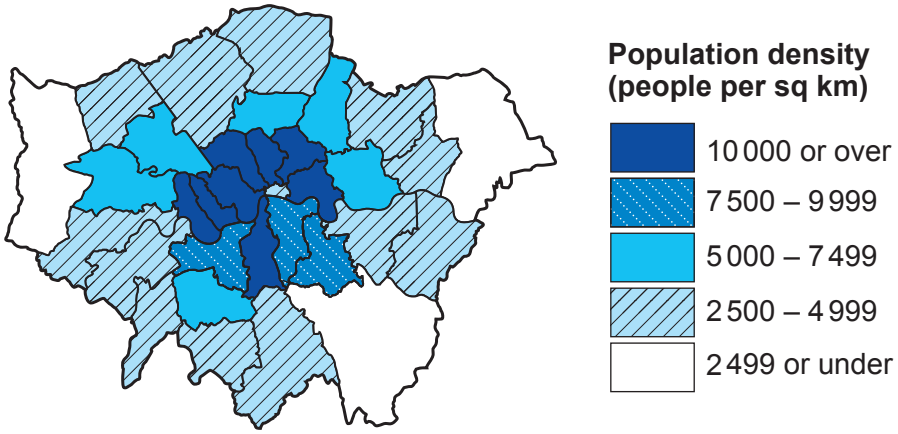
The BCG vaccination is no longer part of childhood immunisations. It is only offered to those who are at higher risk of TB; for example, children under 5 who live in an area of the UK with a high rate of TB, as in some areas of London. **Image 7.1** shows the population density of different areas of London.

**Image 7.1**




(iii) Using the map in **Image 7.1**, suggest why the TB incidence is so high in some areas of London. [2]

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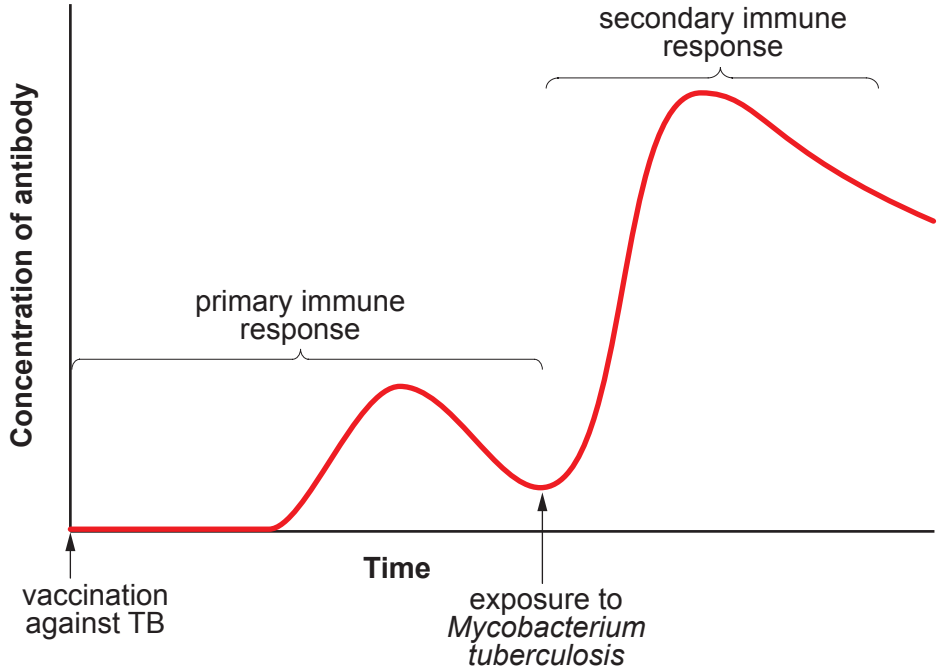
**Graph 7.2** shows the concentration of antibodies released by the body following a BCG vaccination and then after exposure to the live infectious micro-organism, *Mycobacterium tuberculosis*.

**Graph 7.2**




(b) The BCG vaccine initiates an active immune response which leads to the production of antibodies by the patient.

(i) Describe and explain the shape of the part of **Graph 7.2** which shows the primary immune response. [4]

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(ii) With reference to **Graph 7.2**, explain how vaccination prevents development of TB when exposed to *M. tuberculosis*. [4]

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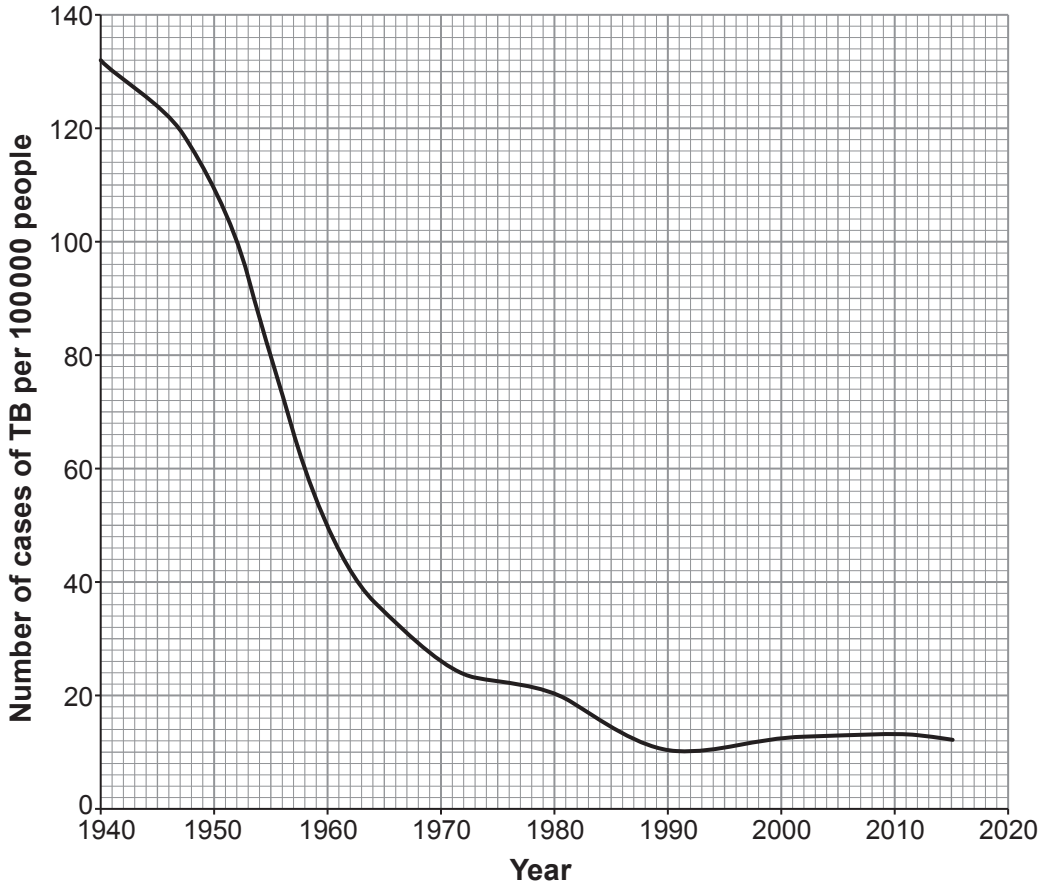
(c) Since the 1940s, antibiotics have been used to treat TB. **Graph 7.3** shows the change in the numbers of diagnosed TB cases in England and Wales from 1940 to 2015.

**Graph 7.3**




(i) Calculate the percentage decrease in **TB incidence** between 1940 and 2015. [2]

Percentage decrease in incidence of TB = ..... %



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*Mycobacterium tuberculosis* can develop into a dormant state, referred to as latent TB. Whilst dormant, *M. tuberculosis* is resistant to antibiotics. Patients with TB receive a combination of antibiotics to kill active bacteria rapidly and to try to prevent latent TB.

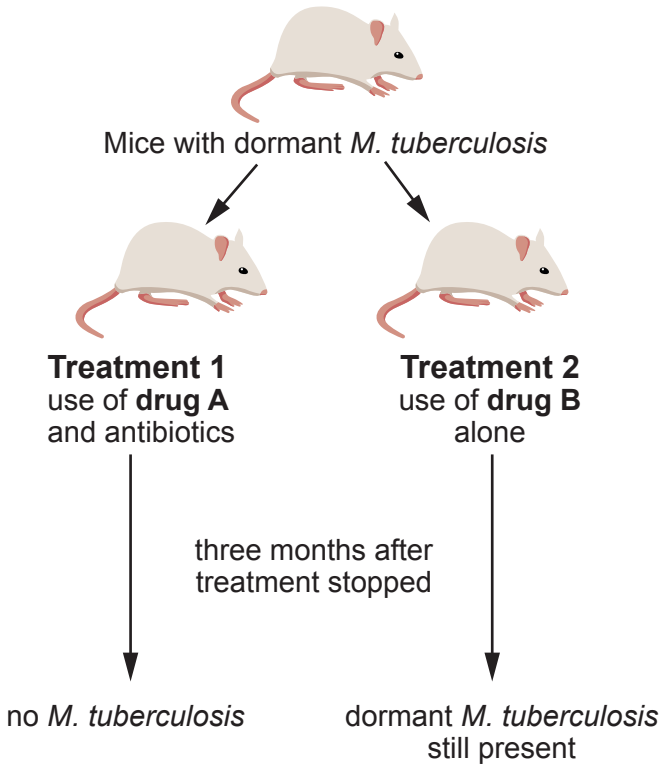
Two new treatments have been proposed to target dormant *M. tuberculosis* to prevent TB recurring in a patient.

**Treatment 1**
Use of **drug A** to activate the dormant bacteria as well as treating with high doses of antibiotics to kill all activated bacteria.

**Treatment 2**
Use of **drug B** that will inhibit the activation of any dormant bacteria while the drug is present.

These treatments have been trialled in mice as shown in **Image 7.4**.

**Image 7.4**




(ii) Complete the table below by stating **one** advantage and **one** disadvantage for each of the treatments. [4]

| Treatment | Advantage | Disadvantage |
|---|---|---|
| **Drug A** and antibiotics | ..... ..... | ..... ..... |
| **Drug B** alone | ..... ..... | ..... ..... |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(d) The methods of treating TB can also be used to treat malarial infections.
Explain why these methods are less effective when they are used in the treatment of malarial infections. [2]

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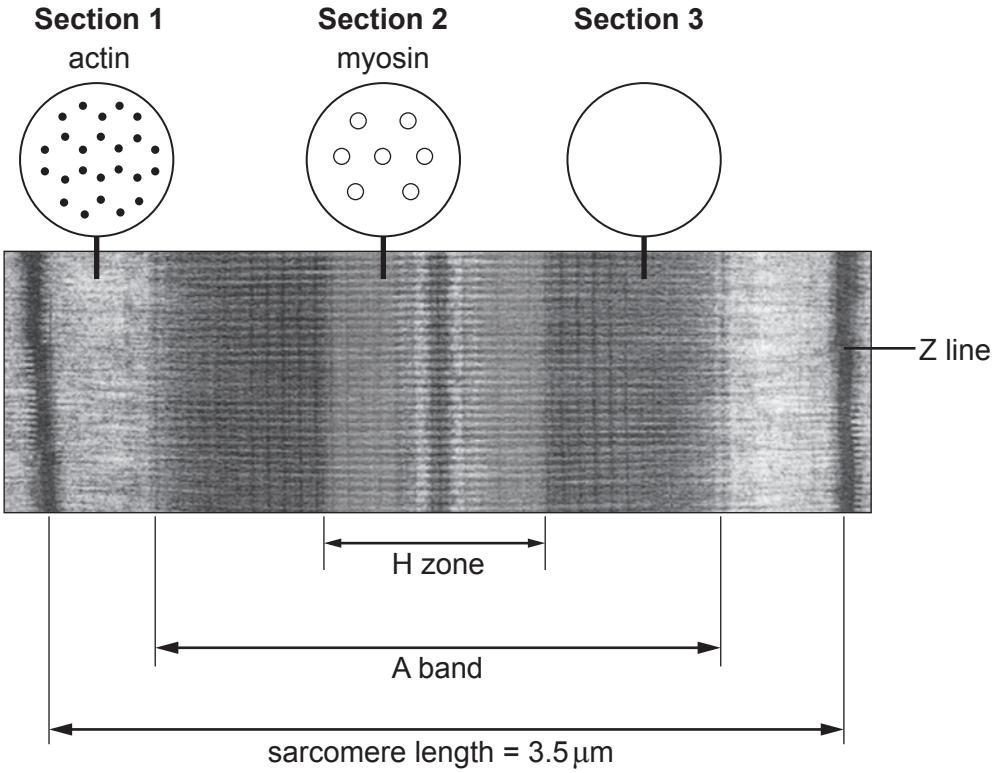
## Option B: Human Musculoskeletal Anatomy

**8.** The human musculoskeletal system is comprised of a number of different tissues, including muscle, cartilage and bone.

**Image 8.1** shows the structure and appearance of a sarcomere in relaxed, striated muscle. It also shows transverse sections of the actin and myosin filaments in the sarcomere.

**Image 8.1**

**Transverse sections across sarcomere**




(a) (i) **Complete the appearance** of transverse **Section 3** shown on **Image 8.1**. [1]

(ii) During contraction of the muscle, the sarcomere shortens. Describe and explain what happens to the H zone and A band. [4]

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(iii) When the sarcomere is contracted, its length is 1.5 μm long.
Use **Image 8.1** to calculate the percentage decrease in length when the sarcomere contracts. [2]

Percentage decrease in length of sarcomere = .....

(iv) Describe how the structure of actin allows cross bridges to form during contraction of the muscle and explain why cross bridges are unable to form in a resting muscle. [2]

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(b) Cartilage is connective tissue found in the skeleton. There are several forms of cartilage.

(i) Explain how the structure of hyaline cartilage relates to its function in the skeleton. [2]

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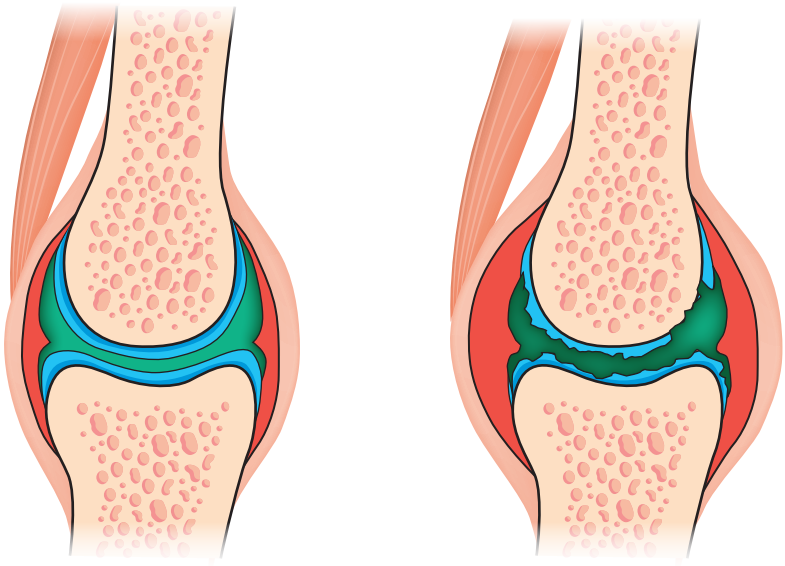
**Image 8.2** shows a normal knee joint and a joint affected by rheumatoid arthritis.

**Image 8.2**



normal knee joint

knee joint with rheumatoid arthritis

(ii) Describe what is meant by rheumatoid arthritis and name **two** types of tissues affected by rheumatoid arthritis as shown in **Image 8.2**. [2]

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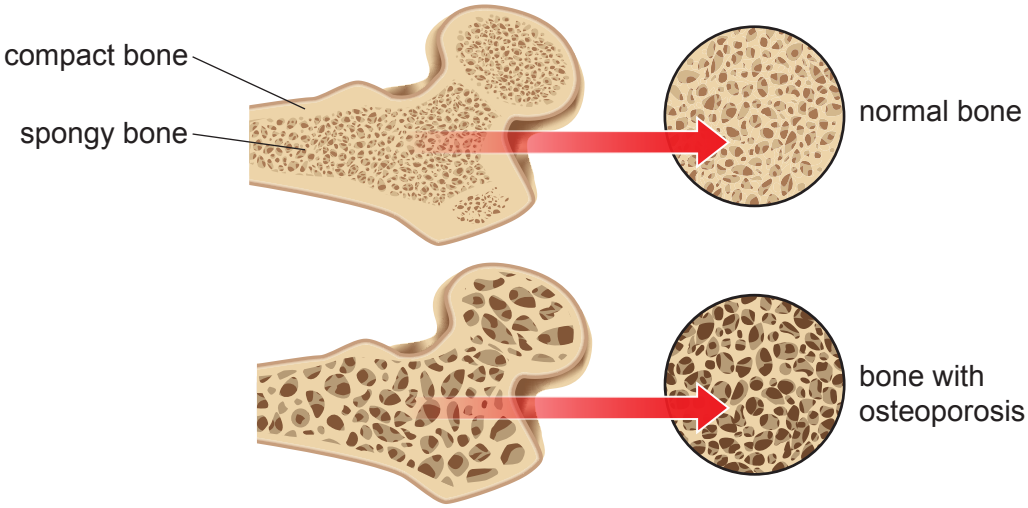
(c) Osteoporosis is a form of brittle bone disease in humans. It is caused by the loss of calcium from the bones leading to decreased bone density. **Image 8.3** shows a cross section of bone from a healthy patient, and one from a patient suffering from osteoporosis.

**Image 8.3**




(i) Using **Image 8.3** and your own knowledge, describe the effect of osteoporosis on the composition of the bones. [2]

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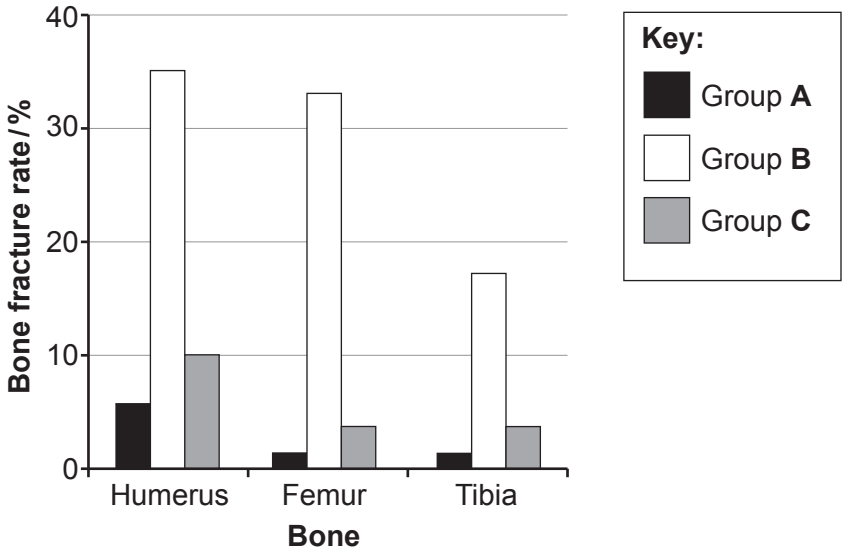
Amniotic fluid contains a form of stem cell that can differentiate into osteoblasts. Research has been carried out to assess the effectiveness of stem cells in treating osteogenesis imperfecta (OI) in mice.
Three groups of mice were tested. The treatment received by each group is shown in **Table 8.4**.

**Table 8.4**

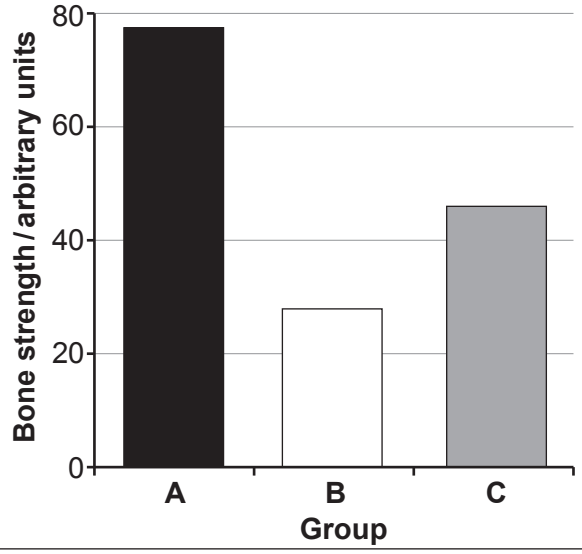
| Group of mice | Osteogenesis imperfecta (OI) present | Received amniotic stem cells at birth |
|---|---|---|
| **A** – healthy mice | No | No |
| **B** – control group | Yes | No |
| **C** – experimental group | Yes | Yes |

The results of the research are shown in **Graphs 8.5** and **8.6**.

**Graph 8.5**




**Graph 8.6**





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(ii) Summarise the effect of stem cell treatment in the experimental group **C** in comparison to the healthy mice (**A**) and the control group (**B**) shown in **Graphs 8.5 and 8.6**.

[3]

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(iii) The use of human stem cells is being considered for clinical trials in humans to treat osteoporosis. Using all of the information provided, suggest how stem cell treatment could improve the condition of bones in osteoporosis patients. [2]

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## Option C: Neurobiology and Behaviour

**9.** Prairie dogs (*Cynomys ludovicianus*) are rodents, native to the grasslands of North America, which live in large colonies. Members of a family group interact through a variety of social behaviours such as kissing and grooming one another. **Image 9.1** shows prairie dogs sharing a greeting kiss.

**Image 9.1**

(a) Prairie dogs are said to demonstrate social behaviour.

(i) Define the term social behaviour. [1]

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Prairie dogs communicate with other members of the colony by making different sounding barks. When an intruder approaches, the first prairie dog gives a sharp bark, bobs up and down, barks again, and then plunges into a burrow. Other lookouts further away repeat this behaviour alerting other members of the colony along the route taken by the intruder.

(ii) The social behaviour of 'barking and bobbing' is an example of a fixed action pattern. Explain how such a fixed action pattern is generated and why it may be advantageous to the animals within the social group. [2]

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(iii) State **two** differences between a fixed action pattern and a reflex action. [2]

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(b) Social behaviour involves a large number of learnt responses.

Researchers investigated the behaviour of prairie dogs in colonies in Colorado. They were trying to find out if prairie dogs became habituated to the arrival of a human in their territory.

(i) Define the term habituation and explain why it is an advantage to the prairie dogs. [2]

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The researchers selected two colonies of prairie dogs:

Colony **A** From a rural area, out of town, and not used to human presence.

Colony **B** From an urban area, close to city parks, where the prairie dogs would occasionally encounter humans.

Humans slowly approached each colony 100 times over 200 days. They recorded the concealment distance – this is the distance from the lookout animal to the human when the prairie dogs went down into the burrows. The researchers expected that the prairie dogs would become habituated to the presence of humans.



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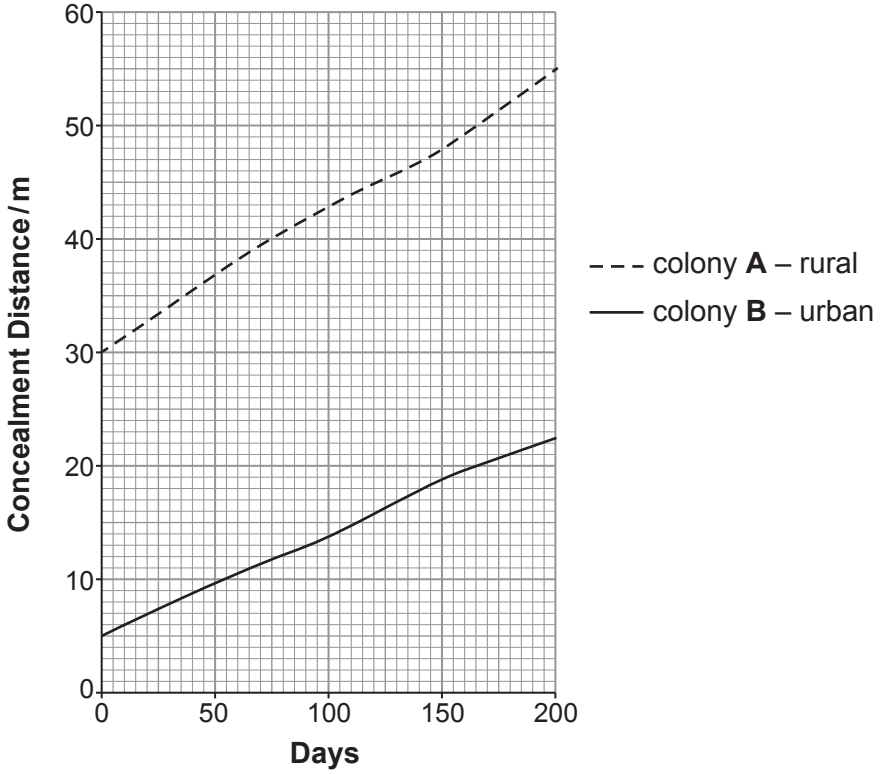
The results are shown in **Graph 9.2**.

**Graph 9.2**




(ii) Using **Graph 9.2**, calculate the percentage increase in concealment distance for the rural prairie dogs over the 200 days. [2]

Percentage increase in concealment distance = .....

(iii) Use the evidence from **Graph 9.2** to conclude whether the researchers' expectations were correct. [3]

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(c) **Images 9.3** and **9.4** show fMRI scans of two patients taken whilst carrying out language skill tasks.

**Image 9.3**

fMRI taken from a healthy adult patient

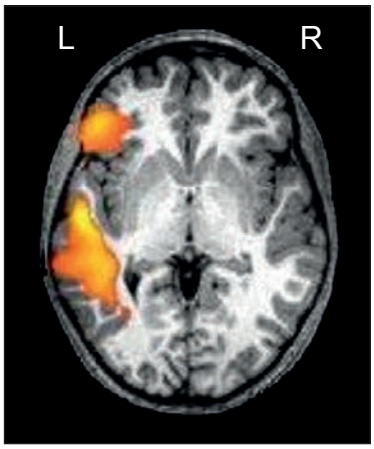
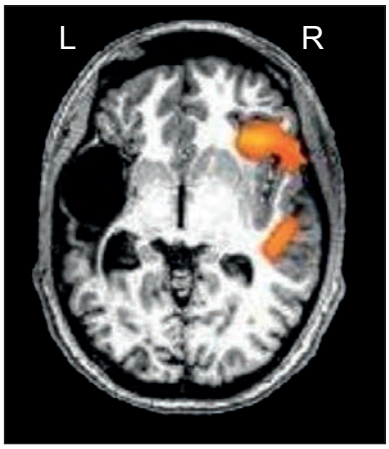



**Image 9.4**

fMRI taken from an adult patient who had a stroke shortly after birth




(i) State why an fMRI scan was the most suitable form of brain scan to show where the patients were processing language. [1]

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(ii) Suggest the area of the brain where the stroke took place and explain what has happened in the brain of the stroke patient in **Image 9.4** to account for the fMRI scan result. [4]

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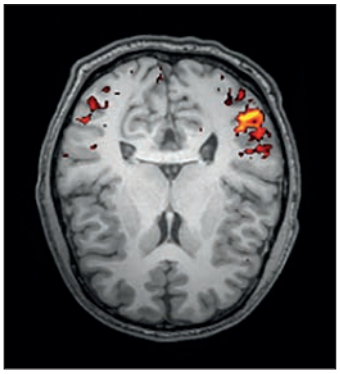
The patient in **Image 9.4** regained all aspects of language. **Image 9.5** shows an fMRI scan from a patient who had a stroke as a mature adult.

**Image 9.5**



(iii) Using **Image 9.5** and your own knowledge, explain why it would be more difficult for this patient to fully regain all aspects of language. [3]

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