Mutations and Stem Cells: AQA A Level Biology Exam Questions by Topi

16 questions · 43 marks · about 56 minutes. Answer in the spaces, then check the mark schemes at the end.

Question 1 · A level Paper 3 · June 2019 · Q04.1 · 1 mark

0 4 Mitochondrial DNA (mtDNA) is a small circular DNA molecule located in mitochondria. It is 16 569 nucleotides long and contains 37 genes and a control region.

Sports scientists investigated whether a mutation in the control region of mtDNA in human males was related to an ability to exercise for longer.

- The males in Group **T** had thymine at nucleotide position 16 519
- The males in Group **C** had a mutation resulting in cytosine at nucleotide position 16 519

0 4 . 1 The control regions of Group **T** and Group **C** were the same length.

Name the type of gene mutation that is most likely to have occurred at nucleotide position 16 519

[1 mark]

Question 2 · A level Paper 3 · November 2020 · Q03.1 · 2 marks

0 3 In Europe, viruses have infected a large number of frogs of different species. The viruses are closely related and all belong to the Ranavirus group.

Previously, the viruses infected only one species of frog.

0 3 . 1 Suggest and explain how the viruses became able to infect other species of frog.

[2 marks]

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Question 3 · A level Paper 2 · June 2025 · Q08.1 · 2 marks

0 8 . 1 A study of cells from a human embryo found that 2.66×10^{-7} % of the genome of one cell had changed due to mutation.

You can assume that:

- the genome contains 3×10^{9} nucleotides
- the mean rate of mutation is 1.14 mutations per cell cycle
- the cell cycle lasts 24 hours.

Use this information to calculate the age of the embryo in days.

Show your working.

[2 marks]

Answer _____ days

Question 4 · A level Paper 2 · June 2019 · Q02.1 · 3 marks

0 2 Sickle cell disease (SCD) is a group of inherited disorders. People with SCD have sickle-shaped red blood cells. A single base substitution mutation can cause one type of SCD. This mutation causes a change in the structure of the beta polypeptide chains in haemoglobin.

0 2 . 1 Explain how a single base substitution causes a change in the structure of this polypeptide.

Do **not** include details of transcription and translation in your answer.

[3 marks]

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Question 5 · A level Paper 2 · June 2025 · Q04.1 · 4 marks

0 4 . 1 People with the inherited blood disorder haemophilia A (HA) have a reduced ability to clot blood. One variant of HA is due to an inversion mutation in the *F8* gene. This mutation causes the production of a non-functional form of the protein, factor VIII. Factor VIII is **not** an enzyme.

Explain how an inversion mutation can cause the production of non-functional factor VIII.

[4 marks]

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Question 6 · A level Paper 3 · June 2024 · Q05.1 · 2 marks

0 5 Leigh syndrome (LS) is a rare, recessive, inherited condition.

LS is caused by a mutation in any one of more than 75 different genes coding for proteins involved in oxidative phosphorylation.

In 80% of people with LS, these mutations occur in nuclear DNA. In 20% of people with LS, these mutations occur in mitochondrial DNA (mtDNA).

15% of the nuclear DNA mutations that cause LS occur in the *SURF1* gene. A mutated *SURF1* gene codes for a shorter polypeptide than a non-mutated *SURF1* gene.

0 5 . 1 Name **one** type of *SURF1* gene mutation and explain how this mutation could lead to production of a shorter polypeptide.

[2 marks]

Type of mutation _____

Explanation _____

_____

_____

_____

_____

Question 7 · information from earlier in the question

0 1 In one species of squirrel, *Sciurus carolinensis*, fur colour is controlled by one gene, with two codominant alleles. C^G represents the allele for grey fur colour, and C^B represents the allele for black fur colour.

Table 1 shows the three possible phenotypes.

Table 1

Genotype	Phenotype
C^GC^G	Grey fur
C^GC^B	Brown-black fur
C^BC^B	Black fur

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Question 7 · A level Paper 3 · November 2021 · Q01.4 · 2 marks

The mutation that caused the C^B allele was due to a 24 base-pair deletion from the C^G allele.

0 1 . 4 The protein coded for by the C^B allele is 306 amino acids long.

Calculate the percentage reduction in size of the protein coded for by the C^B allele compared with the protein coded for by the C^G allele.

Give your answer to 3 significant figures and show your working.

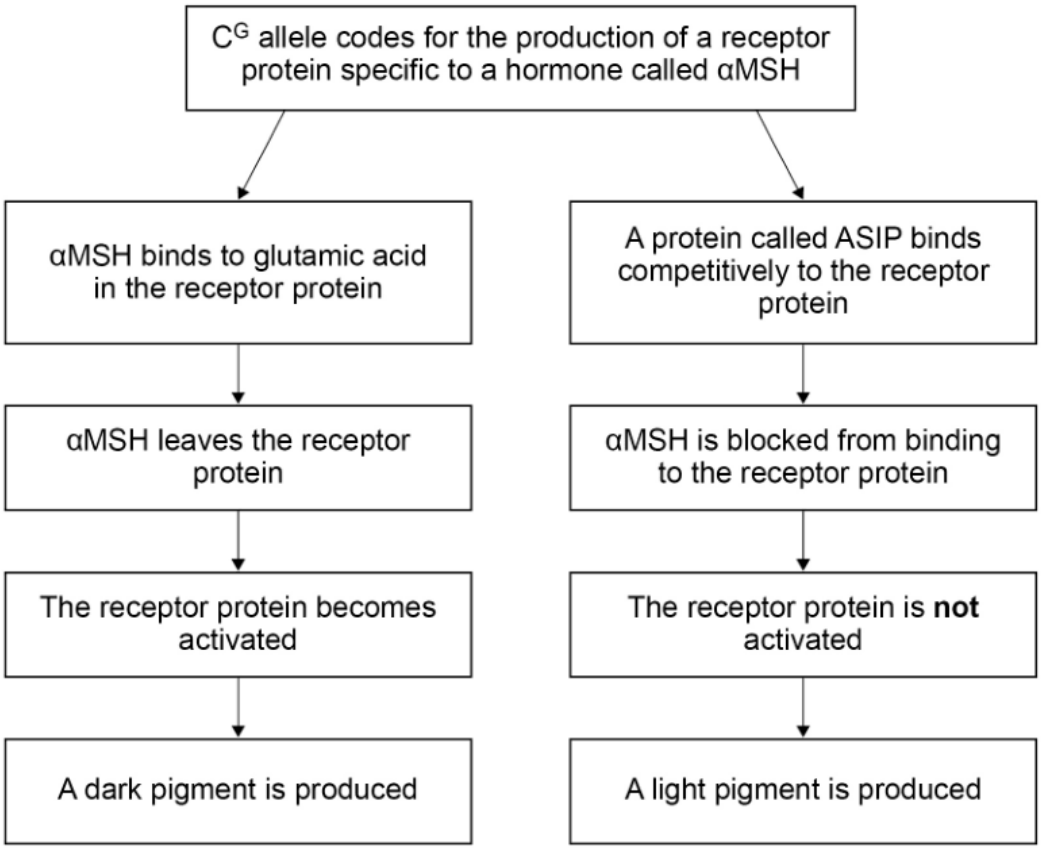
[2 marks]

Answer _____

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In *S. carolinensis*, fur colour depends on the distribution and relative amounts of light pigments and dark pigments in the hairs of the fur. **Figure 1** shows how the protein produced from the $\mathbf{C^G}$ allele can result in the production of a light pigment or a dark pigment.

Figure 1



The deletion mutation in the $\mathbf{C^B}$ allele results in the production of a receptor protein that does not have glutamic acid. The lack of glutamic acid in the receptor protein has the same effect as αMSH leaving the receptor protein.

0 1 . 5 Use **Figure 1** and this information to suggest why *S. carolinensis* with the genotype $\mathbf{C^BC^B}$ have black fur rather than grey fur.

[3 marks]

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Question 9 · information from earlier in the question

0 2 Sickle cell disease (SCD) is a group of inherited disorders. People with SCD have sickle-shaped red blood cells. A single base substitution mutation can cause one type of SCD. This mutation causes a change in the structure of the beta polypeptide chains in haemoglobin.

Question 9 · A level Paper 2 · June 2019 · Q02.2 · 3 marks

Haematopoietic stem cell transplantation (HSCT) is a long-term treatment for SCD. In HSCT, the patient receives stem cells from the bone marrow of a person who does not have SCD. The donor is often the patient's brother or sister. Before the treatment starts, the patient's faulty bone marrow cells have to be destroyed.

0 2 . 2 Use this information to explain how HSCT is an effective long-term treatment for SCD.

[3 marks]

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Question 10 · information from earlier in the question

1 0 Read the following passage.

Complete achromatopsia is a form of complete colour blindness. It is caused by having only rods and no functional cone cells. People with complete achromatopsia have difficulty in seeing detail. Complete achromatopsia is caused by an autosomal recessive allele and is usually very rare in populations with only one in 40 000 being affected. However on the Pacific island of Pingelap ten percent of the population are affected.

One form of red-green colour blindness is caused by a sex-linked recessive allele which affects more men than women. People with this red-green colour blindness are unable to distinguish between red and green, and also between other colours. They have green-sensitive cones but the photoreceptive pigment they contain does not function.

Scientists investigated the use of gene therapy to correct red-green colour blindness in monkeys. They injected viruses containing the gene for the green-sensitive pigment directly into the eyes of the monkeys. Although the monkeys maintained two years of colour vision, there is debate on whether this form of gene therapy is worthwhile. No clinical trials of this procedure have been carried out on humans. Current research into the treatment of red-green colour blindness involves the use of induced pluripotent stem cells (iPS cells). The use of iPS cells could have advantages over the use of gene therapy.

Use the information in the passage and your own knowledge to answer the following questions.

Question 10 · A level Paper 2 · June 2018 · Q10.5 · 2 marks

1 0 . 5 Current research into the treatment of red-green colour blindness involves the use of induced pluripotent stem cells (iPS cells) (lines 17–19).

Suggest how iPS cells could correct red-green colour blindness.

[2 marks]

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Question 11 · A level Paper 2 · June 2018 · Q10.6 · 3 marks

1 0 . 6 The use of iPS cells could have advantages over the use of gene therapy to correct red-green colour blindness (lines 19–20).

Using the information from the passage, suggest and explain reasons why.

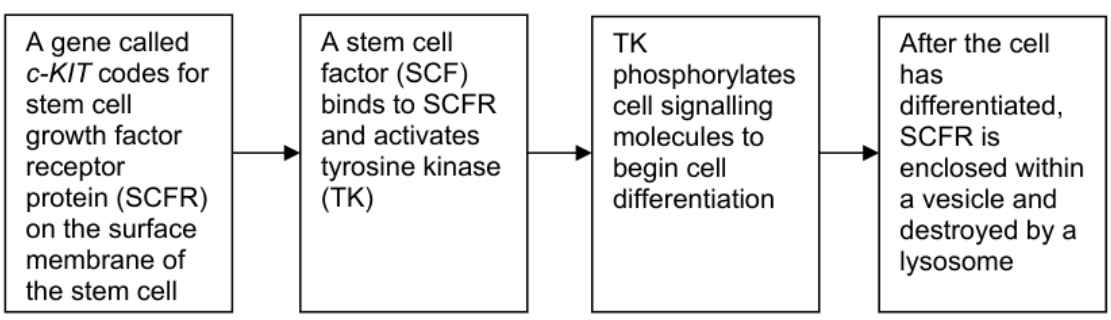
[3 marks]

Question 12 · information from earlier in the question

0 4 Following a body injury, bone marrow stem cells move to the site of damage and undergo cell differentiation.

Figure 5 shows how this differentiation occurs.

Figure 5



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After a heart attack, cardiomyocytes (cardiac muscle cells) die, and become infarcted tissue. Infarcted tissue cannot contract.

Stem cells in bone marrow **cannot** move to the infarcted tissue and differentiate into cardiomyocytes.
Scientists used laboratory rats to investigate if bone marrow stem cell transplants could be used to repair infarcted tissue resulting from a heart attack.

They split the rats into three groups.

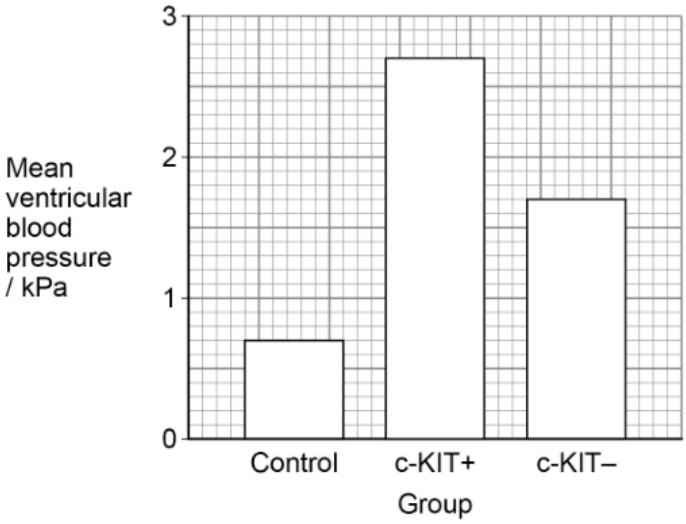
- **Control** group did not get a transplant of bone marrow stem cells.
- **c-KIT+** group got a transplant of bone marrow stem cells with a functioning *c-KIT* gene.
- **c-KIT–** group got a transplant of bone marrow stem cells with **no** functioning *c-KIT* gene.

After 9 days, the scientists measured the mean ventricular blood pressure of each of the three groups.

Figure 6 shows their results.

The differences between the groups were all statistically significant.

Figure 6



0 4 . 2 Using all of the information, suggest explanations for the results for the **Control** group and the **c-KIT–** group shown in **Figure 6**.

[4 marks]

Control _____

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Question 13 · A level Paper 3 · June 2023 · Q04.3 · 2 marks

0 4 . 3 Nine days after transplantation, the **c-KIT+** group showed that 68% of infarcted tissue was made up of new cardiomyocytes. The control group had **no** new cardiomyocytes.

Assuming that mean ventricular blood pressure is directly proportional to the number of cardiomyocytes, calculate the percentage of infarcted tissue that was made up of new cardiomyocytes in the **c-KIT–** group.

[2 marks]

Answer _____ %

All new cardiomyocytes produced:

- Connexin-43, a channel protein that allows electrical impulses to pass between cardiomyocytes
- GATA-4, a transcriptional factor that stimulates the expression of genes for actin and myosin.

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0 6 . 3 Scientists investigated the use of induced pluripotent stem cells (iPS cells) to treat type I diabetes in mice. The scientists used four transcription factors to reprogramme skin cells to form iPS cells. The scientists then stimulated the *in vitro* differentiation of iPS cells into pancreatic cells.

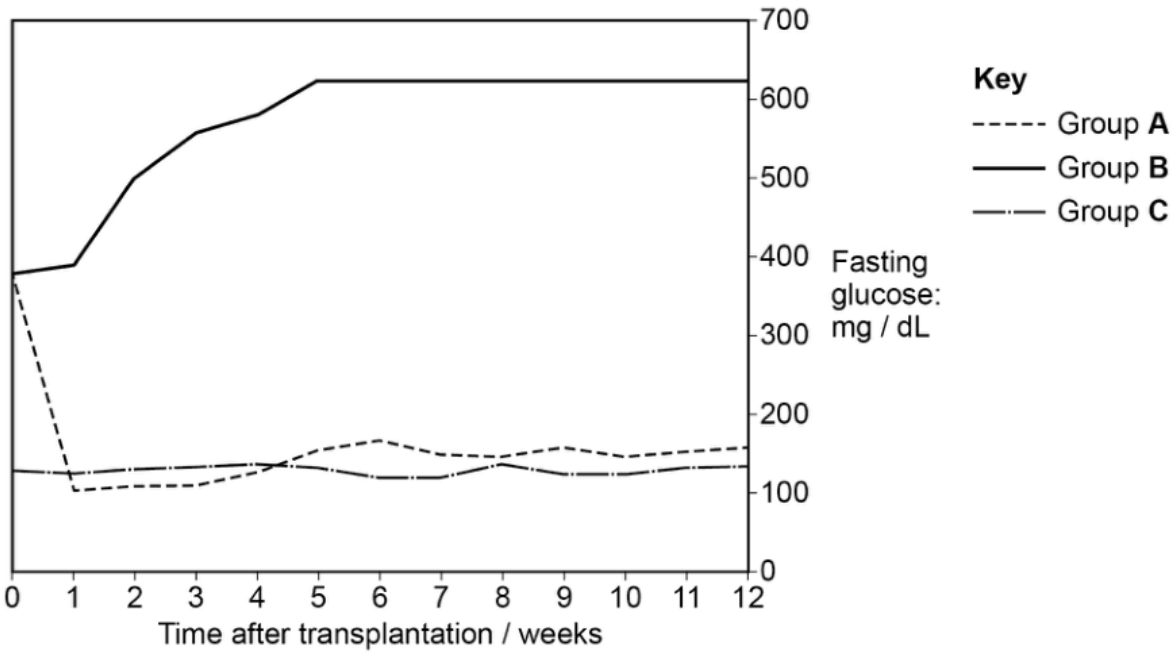
The scientists set up three experimental groups:

- Group **A** – 30 mice with type I diabetes received pancreatic cell transplants derived from iPS cells.
- Group **B** – 30 mice with type I diabetes were left untreated.
- Group **C** – 30 mice without diabetes were left untreated.

The scientists measured the blood glucose concentration of all the mice on a weekly basis for 12 weeks.

The results the scientists obtained are shown in **Figure 6**.

Figure 6



Suggest how transcription factors can **reprogramme** cells to form iPS cells.

[2 marks]

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Question 14 · A level Paper 2 · June 2017 · Q06.4 · 4 marks

0 6 . 4 Using all the information provided, evaluate the use of iPS cells to treat type I diabetes in humans.

[4 marks]

Question 15 · information from earlier in the question

0 8 Alport syndrome (AS) is an inherited disorder that affects kidney glomeruli of both men and women. Affected individuals have proteinuria (high quantities of protein in their urine).

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Scientists investigated the use of transplanted stem cells to treat AS in mice.

The scientists set up four experimental groups.

Group **A** – 40 wild type* mice
Group **B** – 40 AS mice
Group **C** – 40 AS mice that received stem cells from AS mice
Group **D** – 40 AS mice that received stem cells from wild type mice

*Wild type mice are mice **not** affected by AS.

After 20 weeks, the scientists measured the quantity of protein in the urine using a scale from 0 (lowest quantity) to +++++ (highest quantity).

The results the scientists obtained are shown in **Table 2**.

Table 2

Group	Maximum quantity of protein in urine at 20 weeks	Percentage of mice with this quantity of protein
A	0	100
B	+++++	97.5
C	+++++	100
D	++	68

0 8 . 3 Using all the information, evaluate the use of stem cells to treat AS in humans.

[4 marks]

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Question 16 · A level Paper 2 · November 2020 · Q08.4 · 2 marks

0 8 . 4 The scientists carried out further work to investigate how the transplanted stem cells developed after transplantation.

- The scientists transplanted stem cells from wild type male mice into AS female mice.
- After 20 weeks, they found that the quantity of protein in the urine of these female mice had significantly decreased.
- They examined cells from glomeruli in the female mice. Some of these cells contained a Y chromosome.

Suggest how the transplanted stem cells reduce proteinuria.

[2 marks]

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Mark schemes

Question 1 · A level Paper 3 · June 2019 · Q04.1 · 1 mark

04.1	Substitution;	1	Accept inversion or translocation Ignore 'point mutation'

Question 2 · A level Paper 3 · November 2020 · Q03.1 · 2 marks

03.1	1. Mutation in the viral DNA/RNA/genome/genetic material; 2. Altered (tertiary structure of the) viral attachment protein; 3. Allows it/attachment protein/virus to bind (to receptors of other species);

2 max	1. Accept named examples mutations 2. Accept 'antigen' for 'attachment protein' 2. Accept causes antigenic variability 3. Accept descriptions of binding eg is complementary

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08.1	Correct answer of 7 (days) = **2 marks**;; Incorrect answer but shows 7 to incorrect magnitude e.g. 700 (ignore preceding/subsequent zeros and position of decimal point i.e. did not divide by 100 or incorrect use of order of magnitude) = **1 mark** **OR** Incorrect answer of 168 (incorrectly multiplied by 24) = **1 mark** **OR** Incorrect answer shows 9 ignoring subsequent numbers (multiplied rather than divided by 1.14) = **1 mark** **OR** Incorrect but shows 6.93 (incorrect rounding of 7.98) = **1 mark** **OR** Incorrect but shows 7.98 or 8 (did not divide by 1.14) = **1 mark;**	2 (2 x AO2)

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02.1	1. Change in (sequence of) amino acid(s)/primary structure; 2. Change in hydrogen/ionic/disulfide bonds; 3. Alters <u>tertiary/3^0</u> structure;	3

1. Reject amino acids are formed.

1. Reject amino acids code.

3. Reject active site.

3. Ignore quaternary.

3. Ignore 3D.

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Question 5 · A level Paper 2 · June 2025 · Q04.1 · 4 marks

04.1	1. Change in sequence of DNA bases **OR** DNA base sequence inverted/reversed; 2. Change in (sequence of) amino acid(s) **OR** Change in primary structure; 3. Change in hydrogen/ionic/disulfide bonds; 4. Alters tertiary/3° structure (of protein);	4 (4 x AO2)	1. Accept if answer refers to DNA and shows base sequence inverted e.g. TATGC to CGTAT 1. Accept nucleotides for bases 1. Accept changes DNA triplet(s) 2. Reject amino acids are 'formed' or 'produced' 2. Accept shorter amino acid sequence or terminated 4. Reject active site/enzyme 4. Ignore quaternary 4. Ignore 3D

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Question 6 · A level Paper 3 · June 2024 · Q05.1 · 2 marks

05.1	**Mark as pairs: 1 and 2 OR 3 and 4** 1. Deletion/translocation; 2. Could mean triplet(s)/codon(s) missing **OR** Could mean amino acid(s) missing (from the polypeptide/SURF1); 3. Substitution/inversion/addition/duplication/ deletion/translocation; 4. Could result in a (premature) stop triplet/codon;	2 max (2 x AO2)

2. Reject could mean an amino acid is not produced

Question 7 · A level Paper 3 · November 2021 · Q01.4 · 2 marks

01.4	1. 2.55% **= 2 marks**;; 2. 2.61% **= 1 mark** (question misread ie 8/306x100) **OR** Evidence of dividing by 314 or 942 **= 1 mark** **OR** Answers **not** given to three significant figures **= 1 mark;**	2

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Question 8 · A level Paper 3 · November 2021 · Q01.5 · 3 marks

01.5	1. Mutation/lack of glutamic acid leads to (permanent) activation of the receptor/protein; 2. (Because) the receptor/protein does not require the binding/leaving of αMSH (to become activated); 3. ASIP (might) not (be) able to bind to the receptor/protein; 4. (Only) the dark <u>pigment</u> is produced	3 max

2. Answer must convey the idea that binding/leaving is not required

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02.2	1. Produce healthy (red blood) cells **OR** Produce (normal) polypeptide/haemoglobin; 2. No sickle/faulty/SCD (red blood) cells (produced) **OR** No defective polypeptide/haemoglobin; 3. Stem/marrow cells (continuously) divide/replicate **OR** Less chance of rejection (from brother/sister);	3

1. Produce only healthy (red blood) cells is only equivalent to mark point 1.

1. Accept produce 'normal'/non-SCD cells.

1. and 3. Ignore type of stem cell e.g. pluripotent.

3. Differentiate is not equivalent to divide/replicate.

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10.5	1. (iPS cells) divide; 2.(iPS cells) develop/differentiate into (green sensitive) cones;	2	2. Accept 'produce'/'specialise' 'turn in to' / 'genes switched on' / 'turned on' for 'develop' but ignore 'grow' 2. Reject develop into 'green cones'/blue' cones'/'red cones' Ignore develop/differentiate into (blue/red sensitive) cones; Reject reference to develop in to 'green pigment'/ 'blue pigment/ 'red pigment

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10.6	1 (Use of iPS cells) long-term; 2. (Use of iPS cells) less chance of rejection/immune response; 3. (Use of iPS cells) single treatment; 4. Harm/side effects from using viruses (in gene therapy);	3 max

1. Accept ‘gene therapy short-term’ or ‘only two years’ 1. Accept ‘permanent’ 3. Accept ‘gene therapy ‘regular/frequent treatment’’

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Control

1. Cardiomyocytes/(cardiac muscle) cells have **not** been replaced

   OR

   Infarcted tissue is **not** repaired/replaced

   OR

   (The contraction of the ventricle is weak as) there is a small number of cardiomyocytes/ (cardiac muscle) cells still alive

   OR

   (The ventricular pressure is low as) cardiomyocytes/(cardiac muscle) cells are damaged/dead;

2. (Pressure is not zero as) not **all** cardiomyocytes/(cardiac muscle) cells died

   OR

   (Pressure is not zero as) not **all** cardiomyocytes/(cardiac muscle) cells became infarcted tissue;

04.2 — 4 (4 x AO3)

c-KIT–

3. Higher than control, **so** (some) stem cells (must) have been able to differentiate

   OR

   Higher than control, **so** (some) cardiomyocytes/(cardiac muscle) cells have been replaced/infarcted tissues have been repaired;

4. (So) *c-KIT* is not the only gene responsible for differentiation

   OR

   (So) SCF must be able to bind to something other than SCFR

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Question 13 · A level Paper 3 · June 2023 · Q04.3 · 2 marks

04.3	A correct answer 34% **= 2 marks**;; Evidence of 2 and 1 **= 1 mark** An answer of 42.8/43% **= 1 mark** (answer that did not subtract the 'control' from read values)	2 (2 x AO2)

Question 14 · A level Paper 2 · June 2017 · Q06.4 · 4 marks

06.4	1. (Effective as) group A/with iPS/treated lower than group B/with diabetes; 2. (Effective as) group A similar to group C/without diabetes; 3. (Investigation) done on mice not humans; 4. Only shows results for 12 weeks/short-time period / long-term effects not known;	4	Ignore: Only one study / not repeated / sample size. 2. Accept: 'healthy' or 'normal' or control for group C.

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Question 15 · A level Paper 2 · November 2020 · Q08.3 · 4 marks

08.3	1. Effective as D has lower protein (than B/C); 2. Not fully effective as D has higher protein than A; 3. Do not know all results for other mice in D **OR** Only shows results for 68% of mice; 4. Some of D mice may have been cured **OR** Some of D may have died; 5. Do not know actual/numerical quantity of protein; 6. (Investigation) only on mice **OR** (Investigation) not on humans; 7. Rejection may occur; 8. Only shows results for 20 weeks/short-time period **OR** Long-term effects not known;

	Accept descriptions of each group e.g. A = wild type mice. B = AS mice. C = AS mice that received AS stem cells. D = mice that received wild type stem cells. Accept 'healthy' or

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Question 16 · A level Paper 2 · November 2020 · Q08.4 · 2 marks

08.4	1. (Transplanted stem cells) differentiate/specialise; 2. Reduce loss of protein at the glomerulus **OR** Prevents protein moving into filtrate;	2	2. Accept Bowman's/renal capsule.

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