# HL Paper 2

a. Outline a possible cause of Down syndrome. [4]

b. Outline the processes involved in oogenesis within the human ovary. [8]

c. Discuss the ethical issues surrounding IVF. [6]

# Markscheme

a. non-disjunction (can cause Down syndrome);

occurs when pair of homologous chromosomes fails to separate during meiosis;

one gamete/daughter cell receives two chromosomes / diagram showing this;

occurs in anaphase I/II of meiosis;

fertilization involving this gamete leads to change in chromosome number/47 chromosomes;

most common form of Down is trisomy 21/extra chromosome 21;

increase risk of Down syndrome with increased age of mother;

b. oogenesis is process by which female gametes/eggs are produced;

begins during fetal development; oogonia/large number of cells formed by mitosis;

oogonia/cells enlarge/undergo cell growth/become primary oocytes;

begin first meiotic division but stop in Prophase I;

until puberty;

(at puberty) some follicles develop each month in response to FSH;

(primary oocyte) completes first meiotic division;

forms two cells of different/unequal sizes / unequal distribution of cytoplasm;

(creating a) polar body;

polar body eventually degenerates;

larger cell/secondary oocyte proceeds to meiosis II;

stops at prophase II;

meiosis II completed if cell is fertilized;

ovum and second polar body formed;

c. *To award full marks, discussion must contain both pro and con considerations.*

*pros/positive considerations:* ***[3 max]***
chance for infertile couples to have children;
decision to have children is clearly a conscious one due to difficulty of becoming pregnant;
genetic screening of embryos could decrease suffering from genetic diseases;
spare embryos can safely be stored for future pregnancies/used for stem cell research;

*cons/negative considerations:* ***[3 max]***
IVF is expensive and might not be equally accessible;
success rate is low therefore it is stressful for the couple;
it is not natural/cultural/religious objections;
could lead to eugenics/gender choice;
could lead to (unwanted) multiple pregnancies with associated risks;
production and storage of unused embryos / associated legal issues / extra embryos may be used for (stem cell) research;
inherited forms of infertility might be passed on to children;
*Accept any other reasonable answers.*

# Examiners report

a. Most of the candidates gave trisomy 21 and non-disjunction, but fewer were able to accurately describe how it comes about. Confusion between genetic mutation and chromosome mutation was common especially when discussing causes.

b. Quite a few candidates described the process of ovulation in detail, gaining no marks. Most candidates did refer to the formation of the polar body in oogenesis, but missed out on many of the changes given in the mark scheme. Detailed discussions of the menstrual cycle were common. Some obviously understood it but could not write clearly and logically/a general lack of detail.

c. Most candidates gained a fair number of marks, but often limited their score by focusing on the negatives of IVF rather than the positives. But then again, the mark scheme only listed 4 pros vs 7 cons.

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a. Define *linked genes*. [1]

b. In cats, the allele for curled ears (C) is dominant over the allele for normal ears (c). The allele for black colour (B) is dominant over the allele for grey colour (b). A cross occurs between two cats that are both heterozygous for these unlinked traits. [3]

Using a Punnett grid, predict the ratio of phenotypes of offspring in the next generation.

# Markscheme

a. Genes located on the <u>same</u> chromosomes.

b. a. Punnett grid correctly drawn / correct gametes;

b. phenotype ratio 9:3:3:1;

c. correct identification of expected phenotypes (9) black cats with curled ears, (3) grey cats with curled ears, (3) black cats with normal ears, (1) gray cat with normal ears;

# Examiners report

a. A surprising number failed to define this term correctly.

b. Most candidates earned at least some marks on this question. A number of candidates could not correctly identify gametes. Some made errors in counting the numbers of different phenotypes in the grid leading to incorrect ratios. Only a few candidates failed to identify the phenotypes.

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a. Outline the role of condensation and hydrolysis in the relationship between amino acids and polypeptides. [4]

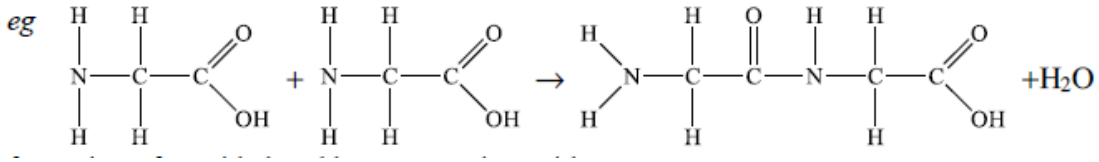
b. The protein hemoglobin transports oxygen to cells. Describe the processes that occur in the mitochondria of cells when oxygen is present. [8]

c. Sickle-cell anemia affects the ability of red blood cells to transport oxygen.Explain the consequence of the mutation causing sickle-cell anemia in relation to the processes of transcription and translation. [6]

# Markscheme

a. *Award [3 max] for condensation reactions*

*Condensation reactions:*
condensation is two molecules joining (by a covalent bond) with the loss of a water molecule;
example of condensation reaction;




formation of peptide bond between amino acids;
(covalent) bond between carboxyl end of one amino acid molecule and amino end of other;
many amino acids joined by condensation to form polypeptide;

*Hydrolysis reactions:*
hydrolysis is the addition of water to break a large molecule into smaller ones;
polypeptide broken down into amino acids/dipeptides by hydrolysis;
*Award any of the above points for a clearly drawn correctly annotated diagram.*

b. pyruvate decarboxylated/$CO_2$ is removed and reduced NAD/NADH + $H^+$ is formed (when entering mitochondrion);

*(both needed)*

2-C molecule/acetyl group reacts with (reduced) coenzyme A to form acetyl CoA;

acetyl CoA enters Krebs cycle;

2 $CO_2$ molecules removed (as waste);

energy/electron rich NADH + $H^+$/$FADH_2$ formed;

for each turn of cycle/each pyruvate, 3 NADH + $H^+$ and 1 $FADH_2$ formed;

1 ATP formed per pyruvate each turn (by substrate-level phosphorylation);

reduced NAD/NADH + $H^+$ and $FADH_2$ enter electron transport chain/ETC;

oxidative phosphorylation uses energy released by ETC to synthesise ATP;

as electrons move along ETC, protons/$H^+$ move into intermembrane space;

creates $H^+$ gradient across the membrane;

ATP synthesized by flow of $H^+$ back across membrane through ATP synthase;

ATP synthesized by chemiosmosis;

ETC reduces oxygen/oxygen is final hydrogen (and electron) acceptor forming water;
*Award any of the above points for a clearly drawn correctly annotated diagram.*
*Accept reduced NAD and $NAD^+ + H^+$ as alternatives to each other.*

c. caused by single base substitution (mutation);

mutation in gene coding for (one of) polypeptide chain in hemoglobin/HbA;

GAG (on sense strand of DNA) mutated to GTG;

when transcribed, RNA sequence/codon becomes GUG rather than GAG;

during translation, have one amino acid substituted for another;

causes glutamic acid/glutamate to be replaced by valine;

change alters folding of Hb protein/makes RBCs sickle-shaped (in low oxygen);

sickle shaped cells block capillaries/cause tissue damage and pain;

*Award any of the above points for a clearly drawn correctly annotated diagram.*

*(Plus up to [2] for quality)*

# Examiners report

a. Most were able to gain some marks on hydrolysis and condensation. Very few diagrams/ structures were completely correct.

b. The processes inside the mitochondria were well known by the better prepared candidates, who were able to explain in detail. Several candidates just tried to draw a diagram of the Krebs cycle without any annotation, hoping that the examiners would find some marks. A well annotated diagram can achieve full marks, but it must be clear. Many risked losing the quality marks by describing glycolysis in great detail, thus giving the impression that they were simply writing down everything they knew instead of answering the question.

c. Most knew that Sickle Cell Anaemia is due to a mutation, but only the better ones were able to correctly state that it was a single base substitution. Few correctly described that the mutation was in (one of) the polypeptide chain of Haemoglobin(A), with many vague statements attributing it to erythrocyte instead. Nearly every candidate remembered that glutamic acid was replaced by valine. Unfortunately this was the only mark gained by many.

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a. Describe the causes of Down syndrome. [5]

b. Describe how human skin colour is determined genetically. [5]

c. Explain the causes of sickle-cell anemia. [8]

# Markscheme

a. Down syndrome is caused by non-disjunction;

occurs during meiosis;

chromosome pairs fail to separate in meiosis I / chromatids in meiosis II / anaphase II;

some gametes have an extra chromosome;

can lead to zygotes/individuals with an extra chromosome / individual has 47 chromosomes;

in Down syndrome this would be trisomy 21/extra chromosome 21;

increased probability with increased age of mother/ages of parents;

b. skin colour is an example of polygenic inheritance;

many/more than two genes contribute to a person's skin colour;

due to the amount of melanin in the skin;

combination of alleles determines the phenotype;

allows for range of skin colours / continuous variation of skin colour;

phenotypes do not follow simple Mendelian ratios of dominance and recessiveness;

the environment also affects gene expression of skin colour / sunlight/UV light stimulate melanin production;

the more recessive alleles there are, the lighter the skin colour; (*vice versa*)

c. caused by gene mutation;

(sickle-cell anemia) due to a base substitution (mutation);

changes the code on the DNA;

which leads to a change in transcription / change in mRNA;

DNA changes from CTC to CAC/GAG to GTG / mRNA changes from GAG to GUG; (*accept DNA changes from CTT to CAT/GAA to GTA / mRNA changes from GAA to GUA*)

which (in turn) leads to a change in translation / change in polypeptide chain/ protein;
(the tRNA) adds the wrong amino acid to the polypeptide chain;
glutamic acid replaced by valine;
produces abnormal hemoglobin;
causing abnormal red blood cell/erythrocyte shape / sickle shape;
which lowers the ability to transport oxygen;
sickle-cell allele is codominant;
homozygote/$Hb^S$ $Hb^S$ have sickle cell anemia/is lethal / heterozygote/$Hb^S$ $Hb^A$ has the sickle trait/is carrier (and is more resistant to malaria);

# Examiners report

a. In part (a) most candidates got several marks but few the full five for describing the causes of Down syndrome. There was confusion as to when

this occurs and how,. The most common statements were mp e, f and g.

b. Many candidates lost marks in part (b) by not knowing skin colour is an example of polygenic inheritance and describing a dihybrid inheritance.

Others simply gave an incomplete account for 5 marks and many were confused as to the difference between alleles and genes.

c. Answers to part (c) were good in many cases. In these scripts, there was complete detail on the cause, the DNA and amino acid changes and the effect on haemoglobin. This is obviously a topic that is well taught in many centres although some students are confused about the effect of the mutation on haemoglobin and its subsequent effect on the shape of the red blood cell.

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a. Draw a labelled diagram of a mature human egg. [5]

b. Outline a technique used for gene transfer. [5]

c. Explain how evolution may happen in response to environmental change with evidence from examples. [8]

# Markscheme

a. *Award **[1]** for each structure accurately drawn and correctly labelled.*

a. haploid nucleus;

b. cytoplasm – with nucleus-to-membrane distance >4 times nucleus diameter;

c. centrioles – two must be shown but only one needs to be labelled;

d. cortical granules – needs to be drawn in vicinity of plasma membrane;

e. plasma membrane – shown as a single line and approximately circular overall;

f. polar cell / (first) polar body – needs to be drawn outside the egg cell;

g. zona pellucida / layer of gel (outside the cell membrane);

h. follicle cells / corona radiata (outside the cell membrane);

i. size shown as 100 μm/0.1mm; *(accept 90 μm to 120 μm)*

b. a. plasmid used for gene transfer/removed from bacteria;

b. plasmid is a small/extra circle of DNA;

c. restriction enzymes/endonucleases cut/cleave DNA (of plasmid);

d. each restriction enzyme cuts at specific base sequence/creates sticky ends;

e. same (restriction) enzyme used to cut DNA with (desired) gene;

f. DNA/gene can be added to the open plasmid/sticky ends join gene and plasmid;

g. (DNA) ligase used to splice/join together/seal nicks;

h. recombinant DNA/plasmids inserted into host cell/bacterium/yeast;

c. a. variation in population;

b. (variation is) due to mutation/sexual reproduction;

c. valid example of variation in a specific population;

d. more offspring are produced than can survive / populations over-populate;

e. competition / struggle for resources/survival;

f. example of competition/struggle for resources;

g. survival of fittest/best adapted (to the changed environment)/those with beneficial adaptations / converse;

h. example of changed environment and adaptation to it;

i. favourable genes/alleles passed on / best adapted reproduce (more) /converse;

j. example of reproduction of individuals better adapted to changed environment;

k. alleles for adaptations to the changed environment increase in the population;

l. example of genes/alleles for adaptations increasing in a population;

m. evolution by natural selection;

n. evolution is (cumulative) change in population/species over time / change in allele frequency;

*Suitable examples are antibiotic resistance and the peppered moth but any genuine evidence-based example of adaptation to environmental change can be credited.*

# Examiners report

a. Of the four drawings on this exam, the egg drawings were in general the weakest. The nucleus was in almost all cases far too large and cortical granules were often distributed throughout the cytoplasm rather than being located close to the plasma membrane. Structures outside the plasma membrane were often muddled, perhaps because it was necessary to use three or more concentric circles to represent them.

b. Strong candidates had no difficulty in scoring full marks here by describing gene transfer using plasmids, restriction enzymes and DNA ligase. The weakest candidates wrote on a wide range of other topics.

c. Answers ranged from impressive, with a secure understanding of evolution by natural selection and effective use of examples, to very confused. In contrast to some previous exams most candidates chose appropriate examples such as the evolution of antibiotic resistance in bacteria or the development of melanism in peppered moths. Descriptions of the development of the giraffe's neck or speciation in Galapagos finches were not accepted because they do not correspond with any specific environmental change for which we have good evidence. It is particularly important to base accounts of evolution on strong evidence rather than speculation, because of the objections to the theory that are still being raised.

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b. Explain, using a **named** example, how polygenic inheritance gives rise to continuous variation. [2]

c. Describe the inheritance of colour blindness in humans. [3]

# Markscheme

b. human skin colour can vary from pale to very dark / amount of melanin varies;

skin colour/melanin controlled by (alleles from) at least three/several genes;

no alleles are dominant / alleles are co-dominant / incomplete dominance;

many different possible combinations of alleles;

skin colour controlled by cumulative effect/combination of genes/alleles;

*Award the above marking points for any other valid example.*

c. sex linked condition;

carried on an X chromosome / absent from Y chromosome;

if present in male causes colour blindness;

(allele is) recessive so heterozygous females are not colour blind;

homozygous females are colour blind;

*Do not allow carried on sex chromosome.*

# Examiners report

b. Answers were varied in quality. Some candidates were not clear about the nature of continuous variation and therefore either described how a small number of skin colours could arise, or described another example of variation with only a small number of phenotypic variants. The best answers explained how continuous variation results from the alleles of different genes acting in combination, with no single allele being dominant over the others. As there is considerable uncertainty about the number of genes influencing the quantity of melanin in human skin, the mark scheme accepted a wide range of answers.

c. This was well known by the stronger candidates, who had no difficulty in scoring three marks. There were some long answers describing particular mating and the offspring that they could produce, which sometimes scored few marks, as they did not make general points about the inheritance of colour blindness. Where crosses are used in an answer to a general question about the inheritance of a trait, they should be used to exemplify the pattern of inheritance, with annotation to make general points, rather than focusing too much on specific ratios.

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a. Describe the process of blood clotting. [4]

b. Factor IX is a blood clotting protein which some hemophiliacs lack. In the future hemophilia could be treated using clotting factors synthesized by genetically modifiedbacteria.Outlinethebasictechniqueusedforthisgenetransfer. [6]

c. Explain how males inherit hemophilia and how females can become carriers for the condition. [8]

# Markscheme

a. *Remember, up to TWO "quality of construction" marks per essay.*

a. clotting factor released by platelets/damaged tissue/cells;
b. cascade/series of reactions;
c. prothrombin (activated) to thrombin;
d. soluble fibrinogen to insoluble fibrin / thrombin converts fibrinogen to fibrin;
e. mesh of fibrin/fibres seals wound/traps platelets/red blood cells;

b. *Remember, up to TWO "quality of construction" marks per essay.*

a. mRNA/gene coding for factor IX extracted from human cell/tissue;
b. mRNA copied to DNA/cDNA (using reverse transcriptase);
c. plasmids used (for gene transfer);

d. restriction enzyme/endonuclease used to open plasmid/cut DNA;
e. complementary bases/sticky ends on gene and plasmid/link gene to plasmid;
f. sealed using ligase;
g. recombinant plasmid/plasmid containing desired gene taken up by bacteria;
h. isolate/clone the recombinant/transformed bacteria;
i. bacteria cultured/grown in fermenter to produce factor IX;

c. *Remember, up to TWO "quality of construction" marks per essay.*

a. hemophilia is due to a recessive allele/is a recessive trait / $X^H$ is normal allele and $X^h$ is hemophilia allele;
b. hemophilia is sex linked;
c. allele/gene is on the X chromosome;
*Reject disease/hemophilia carried on X chromosome.*
d. (sex chromosomes in) females are XX while males are XY;

e. Y chromosomes do not have the allele/hemophiliac males are $X^hY$;
f. males inherit their X chromosome from their mother/do not pass the allele to sons;
g. males have only one copy so recessive trait/allele is not masked;
h. males have a 50% chance of hemophilia/receiving the allele if mother is a carrier;

i. carrier is heterozygous for the gene/is $X^HX^h$;
j. dominant/normal allele masks the recessive allele (so clotting is normal);
k. females inherit one X chromosome from father and one from mother;
l. affected/hemophiliac males have carrier daughters;
m. hemophilia allele could have been inherited from either parent;

*Accept the points above explained either in text or clearly using a Punnett grid or genetic diagram, but not for simply reproducing an unlabeled Punnett grid or diagram without explanation.*

# Examiners report

a. There were many general accounts of the sealing up of cuts with clotted blood but what was needed here was the process that leads to clotting. The programme specifies which stages in the cascade of reactions are expected and better answers included these and scored full marks without difficulty.

b. Marks for this part of the question covered the whole range. Among weaker candidate there were various misunderstandings about gene transfer and many confused gene transfer with gene therapy, describing the transfer of the Factor IX gene to haemophiliacs rather than to bacteria. In almost every case the method of gene transfer described in successful answers was that using plasmids. There were some detailed and accurate accounts of this process.

c. Almost all candidates knew something about the inheritance of hemophilia. The mark scheme rewarded a wide range of relevant points as long as they were clearly made. Punnett grids could be used to illustrate particular points but they did not score marks in themselves. One area of confusion among weaker candidates was the difference between genes and chromosomes, with answers referring to dominant or recessive X chromosomes or implying that X was the gene for hemophilia.

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a. Outline the action of enzymes. [4]

b. Explain the roles of specific enzymes in prokaryote DNA replication. [7]

c. Many genetic diseases are due to recessive alleles of autosomal genes that code for an enzyme. Using a Punnett grid, explain how parents who do not show signs of such a disease can produce a child with the disease. [4]

# Markscheme

a. Catalyse/speed up reactions

Substrate-specific

Lower the activation energy «of a chemical reaction»

Substrate collides with/binds to <u>active site</u>

Enzyme–substrate complex formed
***OR***
transition state formed
***OR***
bonds in substrate weakened

b. «DNA» gyrase/topoisomerase «II» prepares for uncoiling/relieves strains «in the double helix»

Helicase uncoils/unwinds the DNA/double helix

Helicase separates/unzips/breaks hydrogen bonds between the two strands of DNA

«DNA» primase adds an RNA primer/short length of RNA *Accept RNA primase.*

DNA polymerase III adds «DNA» nucleotides/replicates DNA/synthesizes complementary strand in a <u>5' to 3' direction</u>

DNA polymerase III starts replication/adding nucleotides <u>at the primer</u>

DNA polymerase I removes the primer
***OR***
replaces RNA with DNA

«DNA» ligase seals the nicks
***OR***
links sections of replicated DNA
***OR***
links Okazaki fragments

DNA polymerase I/DNA polymerase III proofreads for mistakes

c. Key or text giving alleles with upper case for dominant allele and lower case for recessive allele/allele causing disease

Reject key showing a sex linked gene such as hemophilia.
*Reject if X or Y chromosomes are shown with the alleles.*
*Accept Aa or any other upper and lower case letters.*

Punnett grid showing that both parents can pass on either a dominant or a recessive allele in their gamete

*For example row and column headings with A and a.*
*This mark can be awarded if X or Y chromosomes are shown but each parent has one recessive and one dominant allele as if for autosomal inheritance.*

Four possible genotypes for child correctly shown on grid

*AA, Aa, aA and aa for example.*
*This mark can be awarded if X or Y chromosomes are shown but the genotypes are correct for autosomal inheritance.*

Double/homozygous recessive shown having the disease

*Cannot be awarded with sex linkage.*

25 % or 0.25 or 1/4 chance of inheriting the disease

*This mark can be awarded if X or Y chromosomes are shown but the ratio is correct for autosomal inheritance.*

# Examiners report

a. This was generally well answered with most candidates able to give enough of the important features of enzyme action to score well. One mistake seen in a number of responses was to state that the active site is on the substrate rather than on the enzyme.

b. Knowledgeable candidates had no difficulty in scoring full marks by giving an accurate description of the role of enzymes in DNA replication. It was not necessary to focus on the leading and lagging strands as the action of the various enzymes is largely the same, though of course primers are repeatedly added to the lagging strand and then replaced. Some candidates were obviously concerned that they were being asked about prokaryote DNA replication. This is of course the type of DNA replication that is specified by the programme and has been for many years. It is worth making sure that candidates know that they have learned about this rather than eukaryote replication.

c. This part was very well answered with many candidates scoring full marks. There were a few errors in notation with different letters of the alphabet used for alleles of the same gene or X and Y chromosomes indicating confusion between autosomal and sex-linked genes.

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a. Describe the characteristics of stem cells that make them potentially useful in medicine. [5]

b. Outline a technique of gene transfer resulting in genetically modified organisms. [5]

c. Explain the use of karyotyping in human genetics. [8]

# Markscheme

a. (stem cells) have/retain the capacity to divide;

can be used to produce cell cultures/large number of identical cells;

can be used to repair/replace damaged/lost cells/tissue;

(stem cells) are undifferentiated / have not yet differentiated/specialized;

can differentiate/specialize in different ways / are pluripotent/totipotent;

can be used to form a variety of different tissues / form organs;

used in medical research;

used in treatment of (named) disease;

b. gene transfer takes a gene from one species/organism and inserts it into another;

using plasmid/viral vector/ballistic impregnation/electroporation;

use of reverse transcriptase to obtain gene from mRNA;

restriction enzyme/endonuclease used to cut out/excise gene;

(same) restriction enzyme used to cut open plasmid;

sticky ends used to link DNA/link gene to plasmid;

DNA ligase used to seal nicks/splice;

bacterium takes in plasmid / plasmid transferred to bacterium/plant/host cell;

valid documented example (*e.g.* human insulin from bacterium/yeast / salt-tolerant tomato plant / carotene/vitamin A in rice / herbicide/Roundup/glyphosate resistance in crop plants / factor IX/clotting factor in sheep milk / low phosphate feces in pigs;

c. *Definition and construction of karyotypes:*

karyotype is the number and type / image of chromosomes in a cell;

cells collected from chorionic villus / by amniocentesis;

requires cells in metaphase / stimulate cells to divide and reach metaphase;

burst cells and spread chromosomes / photo taken of chromosomes;

chromosomes are arranged in pairs;

according to size/structure/position of centromere/banding pattern;

*Uses for karyotypes:*
karyotypes used to identify sex/gender;
male is XY and female XX;
used to identify chromosome mutations/abnormal numbers/non-disjunction;
Down syndrome due to extra chromosome 21 / other trisomy/aneuploidy example;
used for pre-natal diagnosis of chromosome abnormalities;
may lead to a decision to abort the fetus;
prepare for consequences of abnormality in offspring;

# Examiners report

a. The characteristics of stem cells and their uses were generally well known. Almost all candidates mentioned that they are undifferentiated cells and that they can differentiate in different ways. Some distinguished pluripotent from totipotent stem cells which was impressive. Fewer candidates than expected mentioned the ability of stem cells to divide repeatedly. Some candidates who were struggling to find much to include in their answer wrote extensively about the ethics of stem cell research, which was not required.

b. This was answered more poorly than expected. The examining team was anticipating thorough accounts of gene transfer using reverse transcriptase, restriction enzymes, plasmids, sticky ends and plasmids but few of these were seen. In many cases the techniques were not well understood, with errors and omissions in candidates‟ answers. The word splice was often used to mean slice or cut, when it actually means linking together. One use of this word before the days of molecular biology was the act of joining the ends of ropes by weaving together their strands –this image might help candidates see the word splice is used for joining together fragments of DNA using sticky ends and DNA ligase.

c. Despite an apparently narrow focus, this was one of the better answered questions in Section B. Most candidates at least knew that Down syndrome is due to trisomy of chromosome 21. The best answers included details both of how karyotypes are prepared and also what can be learned from them. The weakest candidates mostly wrote in vague terms about testing for genetic defects and did not appreciate the difference between gene and chromosome mutations.

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a. Define the terms *chromosome*, *gene*, *allele* and *genome*. [4]

b. Compare the genetic material of prokaryotes and eukaryotes. [6]

c. Explain the process of DNA replication. [8]

# Markscheme

a. *chromosome:* structure formed by DNA and proteins;

*gene:* a heritable factor that controls a specific characteristic;

*allele*: one specific form of a gene occupying the same gene locus as other alleles of the gene;

*genome*: the whole of the genetic information of an organism;

b.

| ***prokaryotic DNA*** | ***eukaryotic DNA*** |
|---|---|
| circular | linear; |
| one chromosome | many chromosomes; |
| not associated with proteins / naked DNA / no nucleosomes | associated with proteins / histones / nucleosomes; |
| plasmids present | no plasmids present; |
| no introns | introns and exons; |
| found in nucleoid region | contained in nucleus; |
| one replication/initiation point | many replication/initiation points; |
| mitochondrial and chloroplast DNA similar to prokaryotic DNA; | |
| both use DNA as their genetic material; | |

*Responses do not need to be shown in a table format.*

c. occurs during (S phase of) interphase/in preparation for mitosis/cell division;

DNA replication is semi-conservative;

unwinding of double helix/separation of strands by helicase;

hydrogen bonds between two strands are broken;

each strand of parent DNA used as template;

deoxynucleoside triphosphate provides energy;

synthesis continuous on leading strand but not continuous on lagging strand;

resulting in formation of Okazaki fragments (on lagging strand);

synthesis occurs in 5'→3' direction;

RNA primer synthesized on parent DNA using RNA primase;

DNA polymerase III adds the nucleotides (to the 3' end);

complementary base pairing;

adenine pairs with thymine and cytosine pairs with guanine; (*both pairings required*) (*do not accept letters alone*)

DNA polymerase I removes the RNA primers and replaces them with DNA;

DNA ligase joins Okazaki fragments/seals nicks (in sugar-phosphate backbone);

*Accept any of the above points shown in a clearly annotated diagram.*

# Examiners report

a. This was the most popular question by far. It also tended to be the best answered. Most candidates were attempting to describe chromosome, gene, allele and genome, rather than defining as asked. The definitions in the syllabus were expected or very close alternatives.

b. Better-prepared candidates scored well on part b, being able to competently compare the genetic material in prokaryotes and eukaryotes. Weak answers were caused by missing the word "genetic material" and just compared the two, scoring very few marks. A large number inappropriately defined naked DNA as being DNA that is not enclosed within a nucleus rather than DNA that is not associated with histones.

c. The explanation of DNA replication was well known by all but the least well prepared candidates. Many gave answers of textbook quality. It should be mentioned that if diagrams are included they should be clear and well labelled.

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a. Outline the processes that occur during the first division of meiosis. [6]

b. Prior to cell division, chromosomes replicate. Explain the process of DNA replication in prokaryotes. [8]

c. Outline outcomes of the human genome project. [4]

# Markscheme

a. **Remember, up to TWO "quality of construction" marks per essay.**

a. (consists of) prophase, metaphase, anaphase and telophase;
b. chromosome number halved/reduced/(diploid) to haploid;
c. homologous chromosomes pair up/form a bivalent/synapsis in prophase;
d. crossing over between non-sister chromatids/chromatids of different homologues;
e. nuclear envelope breaks down (at end of prophase/start of metaphase);
f. tetrads/bivalents/homologous pairs move to/align on equator/cell centre/on metaphase plate in metaphase; (accept homologous chromosomes without pairs if pairing has already been described)
g. attachment of spindle fibres/microtubules to centromeres/kinetochores;
h. (homologous) chromosomes separate/pulled to opposite poles in anaphase;
i. nuclear envelopes reform/do not reform (because of meiosis II) in telophase;
*Accept the above points in a series of annotated diagrams. Reject answers with single chromatids forming pairs in metaphase or separating or moving to opposite poles in anaphase.*

b. **Remember, up to TWO "quality of construction" marks per essay.**

a. DNA replication is semi-conservative;
b. each (molecule formed) has one new strand and one from parent molecule;
c. helicase uncoils DNA;
d. helicase separates the two strands by breaking hydrogen bonds between bases; (reject unzips as an alternative to uncoils but accept as alternative to separates if breakage of hydrogen bonds is included)
e. RNA primase adds primer / primase adds (short) length of RNA;
f. DNA polymerase III binds to/starts at (RNA) primer;
g. DNA polymerase (III) adds nucleotides/bases in a 5' → 3' direction;
h. bases according to complementary base pairing / A–T and C–G;
i. (leading strand) built up continuously (towards the replication fork);
j. (lagging strand) built up in pieces/short lengths/Okazaki fragments;
k. DNA polymerase I removes RNA/primers and replaces them with DNA;
l. ligase seals gaps between nucleotides/fragments/makes sugar-phosphate bonds;
m. nucleoside triphosphates provide the energy to add nucleotides;
*Accept the above points in annotated diagrams.*

c. **Remember, up to TWO "quality of construction" marks per essay.**

a. complete human DNA/chromosomes sequenced;
b. identification of all human genes / find position/map (all) human genes;
c. find/discover protein structures/functions;
d. find evidence for evolutionary relationships/human origins/ancestors;
e. find mutations/base substitutions/single nucleotide polymorphisms;
f. find genes causing/increasing chance of/develop test for/screen for diseases;
g. develop new drugs (based on base sequences) / new gene therapies;
h. tailor medication to individual genetic variation / pharmacogenomics;
i. promote international co-operation/global endeavours;

# Examiners report

a. First division of meiosis

Most candidates knew the names of the four phases and many knew some of the events in them, but there were few really convincing accounts and some confusion between mitosis and meiosis. Few candidates made it clear in their answer than the two nuclei produced in the first division are haploid. The chromosome/chromatid terminology in mitosis and meiosis is rather awkward, but was expected to be used correctly in answers

to this question. In past mark schemes there has often an easy mark for simply mentioning crossing over, whether in context or not. In this case candidates had to say that it occurs between non-sister chromatids.

b. DNA replication in prokaryotes

Some candidates were confused by the specification that replication should be described in prokaryotes. This is of course the only type of replication included in the IB Biology program. There were some very good answers and stronger candidates did not have difficulty in reaching full marks. Abler candidates seemed to have chosen question 5, perhaps because they knew they could cope with the complexities of DNA replication and knew that they had enough to say for 8 marks.

c. Outcomes of the human genome project

There were some good answers to this question also. Candidates often referred to the complete sequencing of the genome, evidence on human ancestry and the discovery of genes causing diseases or of genes that increase the incidence of a disease.

---

The biological insights of Mendel and Darwin in the 19th century remain important to this day.

a. Discuss the role of genes and chromosomes in determining individual and shared character features of the members of a species. [7]

b. Outline the process of speciation. [4]

c. Describe, using **one** example, how homologous structures provide evidence for evolution. [4]

# Markscheme

a. *Genes*

a. mutation changes genes/causes genetic differences

b. genes can have more than one allele/multiple alleles
***OR***
alleles are different forms/versions of a gene

c. different alleles «of a gene» give different characters
***OR***
variation in alleles between individuals

d. eye colour/other example of «alleles of» a gene affecting a character

e. alleles may be dominant or recessive
***OR***
dominant alleles determine trait even if recessive allele is present

f. both alleles influence the characteristic with codominance
***OR***
reference to polygenic inheritance

g. all members of a species are genetically similar/have shared genes
***OR***
certain genes expressed in all members of a species

h. reference to epigenetics/methylation/acetylation / not all genes are expressed «in an individual»

i. genes are inherited from parents/passed on to offspring/passed from generation to generation

*Chromosomes*

j. same locus/same position of genes
***OR***
same sequence of genes/same genes on each chromosome «in a species»

k. same number of chromosomes «in a species»/all humans have 46 chromosomes/differences in chromosome number between species

l. some individuals have an extra chromosome/Down syndrome/other example of aneuploidy
***OR***
polyploidy divides a species/creates a new species

m. X and Y/sex chromosomes determine the sex/gender of an individual

n. meiosis/independent assortment/fertilization/sexual reproduction give new combinations «of chromosomes/genes»

b. a. speciation is the splitting of a species «into two species»

b. reproductive isolation/lack of interbreeding

c. isolation due to geography/«reproductive» behavior/«reproductive» timing

d. polyploidy can cause isolation

e. gene pools separated

f. differences in/disruptive selection cause traits/gene pools to change/diverge

g. gradualism / speciation/changes accumulating over long periods

h. punctuated equilibrium / speciation/changes over a short time period

c. a. similar structure but different function «in homologous structures»

b. pentadactyl limbs/limb with five digits/toes / other example

c. similar bone structure/example of similarity of bones «in pentadactyl limbs» but different uses/functions

d. two examples of use of pentadactyl limb by a vertebrate group

e. suggests a common ancestor «and evolutionary divergence»

f. process called adaptive radiation

# Examiners report

a. [N/A]
b. [N/A]
c. [N/A]

---

a. Distinguish between autosomes and sex chromosomes in humans. [4]

b. Describe the inheritance of hemophilia including an example using a Punnett grid. [6]

c. Explain how meiosis results in an effectively infinite genetic variety of gametes. [8]

# Markscheme

a. X and Y chromosomes determine sex;

females XX and males XY;

X chromosome is larger than / carries more genes than the Y chromosome;

22 types/pairs of autosomes;

males and females have same types of autosomes;

b. sex-linked / due to gene on the X chromosome;

more common in males who only receive one X chromosome;

female is hemophilic if homozygous recessive / homozygous recessive normally fatal;

$X^H$ for dominant/normal allele and $X^h$ for recessive/ hemophilia allele; *(accept in Punnett grid/square)*

example in Punnett grid/square with correct parental genotype and gametes;

correct genotypes of offspring;

correct phenotype ratio or percentage;

*eg*

| | $X^H$ | $X^h$ |
|---|---|---|
| $X^H$ | $X^H X^H$ | $X^H X^h$ |
| Y | $X^H Y$ | $X^h Y$ |

half the males are hemophilic and half of the females are carriers / *OWTTE*;

*Allow marks for correct genotypes if the alleles are not shown superscript on an X, as long as the Y chromosome is indicated.*
*Do allow marking point d. if the letters for the dominant and recessive allele are not upper and lower case versions of the same letter.*

c. one (homologous) chromosome is from the mother and one from the father;

homologous chromosomes pair (in prophase I);

crossing over/chiasma formation in prophase I;

recombination of linked genes / alleles/genes swapped;

many possible points of crossing over;

crossing over occurs at random positions;

due to crossing over the two chromatids of metaphase I chromosomes are not identical;

random orientation (of bivalents) in metaphase I;

in anaphase/at end of metaphase I chromosomes move to opposite poles;

independent assortment of chromosomes/genes;

$2^n/2^{23}$ combinations (without considering crossing over);

four genetically different nuclei/gametes from each meiosis;

*Accept any of the above points in a clearly annotated diagram.*

# Examiners report

a. A usual guideline for examiners is to have 50% more points on the mark scheme than raw marks in Section B questions. There were fewer points than that for part (a) of this question and only the strongest candidates found enough to say to reach a total of four. A point that was almost always missed was that males and females do not differ in the autosomes that they possess. This is a significant distinction between the sex chromosomes and autosomes.

b. For part (b), a small proportion of candidates forgot or did not know that hemophilia is a sex linked condition and so scored few marks here. Most candidates who did know that sex-linkage is involved used the expected notation of an upper case X to represent the X chromosome with superscript upper case and lower case letters to show the alleles. If an upper case Y is also shown, even though it does not carry a copy of the gene, it makes mistakes much less likely when working out possible outcomes from a cross between two parents. The most significant cross is one between an unaffected male and a carrier female as this is how almost all cases of hemophilia are derived. Most candidates showed this. Parental genotypes were often missing and gametes on the Punnett grid were usually shown but not labelled as gametes. The best answers showed the phenotypes of each possible type of offspring, together with the genotype on the Punnett grid. It was also useful to add a ratio or percentages below the grid. Candidates who showed a series of different crosses rarely scored any more marks after the first cross.

c. Part (c) is a standard question but even so, answers were very variable, probably because meiosis is complicated and there are multiple causes of genetic variety, which some candidates struggle to understand. Terminology was sometimes used rather loosely. The best candidates distinguished between random orientation of bivalents in metaphase I and independent assortment of genes due to random orientation or crossing over, depending on whether pairs of genes are on different or the same type of chromosome.

---

a. Predict the genotypic and phenotypic ratios of the possible offspring of a male hemophiliac and a female carrier using suitable symbols for the alleles in a Punnett grid. [3]

Genotypic ratio:

Phenotypic ratio:

b. Hemophilia is a disorder where the ability to control blood clotting or coagulation is impaired. Describe the process of blood clotting. [2]

# Markscheme

a. correctly constructed Punnett square with correct gamete genotypes;

*e.g.*

| (male gametes) \ (female gametes) | $X^H$ | $X^h$ |
|---|---|---|
| $X^h$ | $X^HX^h$ | $X^hX^h$ |
| Y | $X^HY$ | $X^hY$ |

*genotypic ratio*: 1 $X^HX^h$ : 1$X^hX^h$ : 1 $X^HY$ : 1 $X^hY$; (can be inferred from cells of Punnett square)

*phenotypic ratio*: 1 female hemophiliac : 1 female carrier/non-hemophiliac :
1 male hemophiliac : 1 male normal/non-hemophiliac / 50 % hemophiliac :
50 % non-hemophiliac;

*Allow ECF. Award **[2 max]** if notation used does not indicate sex linkage, i.e. if cross is Hh×hh.*

b. release of clotting factors from platelets/damaged cells;

conversion of prothrombin to thrombin;

thrombin catalyses the conversion of fibrinogen into fibrin;

(insoluble) fibrin (net) captures blood cells;

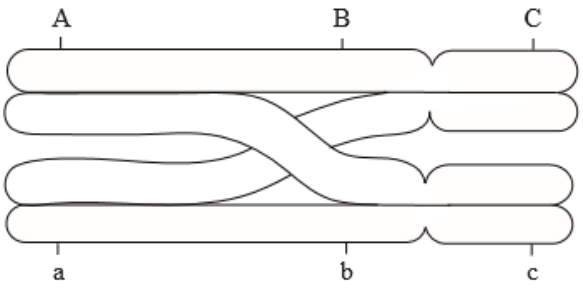
# Examiners report

a. The words "hemophiliac" and "female carrier" should have been enough to remind the students of sex linkage. Many did not know that hemophilia was sex-linked The candidates were allowed an "error carried forward" mark if they had completed the Punnett square correctly but with the incorrect parents.

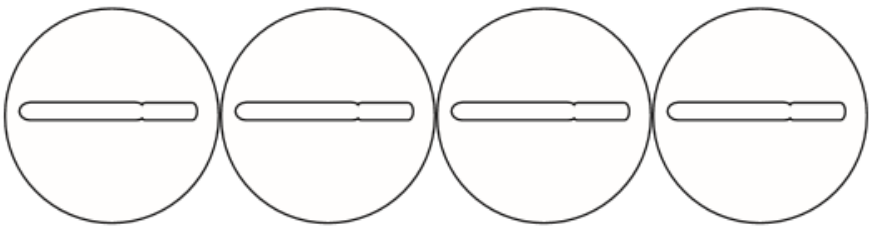
b. The blood clotting process was not at all well known. Many interchanged fibrin and fibrinogen in terms of function and properties.

---

The diagram below shows a pair of chromosomes during meiosis in a cell in the human testis. The position of the alleles of some genes is indicated.




At the end of meiosis, each of the chromatids shown in the diagram will be in a different haploid cell. The diagrams below represent the chromatids inside the haploid cells.



a (i) Deduce with reasons for your answer, whether the chromosomes are autosomes or sex chromosomes. [1]

a (ii) Deduce with reasons for your answer, whether the chromosomes are homologous or non-homologous. [1]

b. State the stage of meiosis of a cell if it contains pairs of chromosomes as shown in the diagram. [1]

c. Determine the combinations of alleles that would be present on each chromatid. Use the diagrams to indicate your answer. [2]

d. State the pattern of inheritance shown by the three genes. [1]

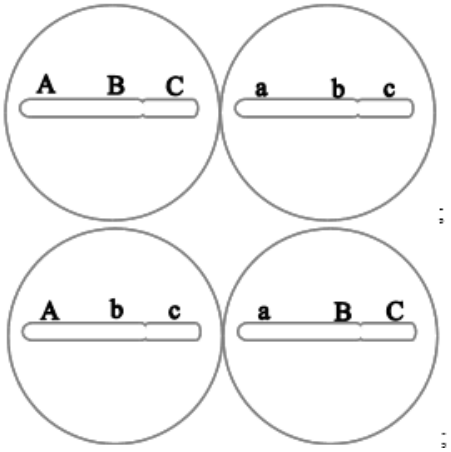
# Markscheme

a (i) autosomes because the sex chromosomes/X and Y chromosomes would be different lengths/sizes / would have different genes

a (ii) homologous because they have paired/formed a bivalent / tetrad / there is crossing over between the chromosomes / they have the same genes (in the same sequence) / they are the same size and shape

b. first prophase/first metaphase/prophase I/metaphase I

c.




*Allow **[1]** only if the C allele is not on the short arm or the A and B alleles are not on the long arm. Use a maximum of two ticks in your marking.*

d. (gene) linkage / autosomal linkage

# Examiners report

a (i) For question 3 a (i) and (ii) Too many were just guessing and, even if correct, did not give a reason. Many argued that crossing over does not occur in sex chromosomes. A failure to use the introductory information “in the human testis” caused a problem for many.

a (ii) For question 3 a (i) and (ii) Too many were just guessing and, even if correct, did not give a reason. Many argued that crossing over does not occur in sex chromosomes. A failure to use the introductory information “in the human testis” caused a problem for many.

b. Although this was most commonly answered correctly, there were a surprising number that did not know what should have been a very straightforward bit of factual knowledge.

c. This was quite commonly answered correctly though many candidates should be advised to take greater care in the construction of lower case and upper case letters in genetics problems.

d. A surprising number indicated polygenic inheritance showing conceptual misunderstanding.

---

a. Explain chemiosmosis as it occurs in photophosphorylation. [8]

b. Draw an annotated graph of the effects of light intensity on the rate of photosynthesis. [4]

c. Using a **named** example of a genetically modified crop, discuss the specific ethical issues of its use. [6]

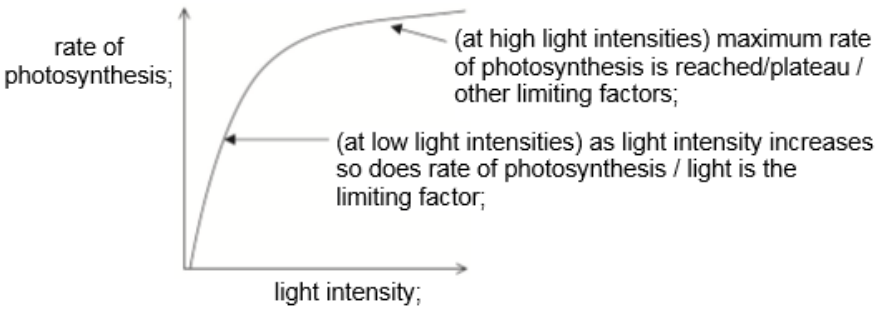
# Markscheme

a. *Remember, up to TWO "quality of construction" marks per essay.*

a. photophosphorylation is the production of ATP;
b. (some of the) light absorbed by chlorophyll / photosystem II;
c. photolysis/splitting of water separation of hydrogen ion from its electron;
d. the electron transport system moves the electrons through a series of carriers;
e. (electron transport system occurs) in the thylakoid membrane;
f. electron transport linked to movement of protons into thylakoid space;
g. a proton gradient builds up (in the thylakoid space);
h. small thylakoid space enhances the gradient;
i. hydrogen ions move by diffusion through the ATP synthase;
j. ADP + inorganic phosphate (Pi) forms ATP;
k. (the kinetic energy from) movement of hydrogen ions (through ATP synthase) generates ATP;
l. ATP synthase is a protein complex in the thylakoid membrane;
m. formation of proton gradient / ATP synthesis linked to electron transport is chemiosmosis;

*Award marks for a clearly drawn correctly annotated diagram.*

b.




a. vertical axis labelled as "rate of photosynthesis" and horizontal axis labelled as "light intensity";
b. drawn showing that at low light intensities, increased intensity leads to increased rate of photosynthesis (sharply);
c. drawn with plateau formed at high light intensities;
d. plateau annotated as maximum rate of photosynthesis;
e. curve intersecting horizontal axis at a value above zero;
f. arrows added to axes or student annotates axis with "rate of photosynthesis increases" and "light intensity increases"

c. *Remember, up to TWO "quality of construction" marks per essay.*

a. named example of verified genetically modified crop; *eg*, Bt corn / golden rice;
*Example must be verifiable.*
b. specific gene added / new protein synthesized by the crop plant / specific modification; *eg* gene from *Bacillus thuringiensis* / cry protein;
c. biological effect of the modification; *eg*, makes the plant toxic to (herbivorous) insects / insect pests / corn borers;
***[2 max]*** *for benefits and* ***[2 max****] for harmful effects / costs;*

d. a benefit of specific genetic modification; *eg*, increased crop yields / less land needed;
e. a second benefit of this specific modification; *eg*, reduced need for use of chemical pesticides;
f. a harmful effect of specific genetic modification; ingestion of toxin by nontarget species;
g. another specific harmful effect; *eg*, concerns about contamination of neighbouring non-GMO crops affecting trade;

*To award* ***[6]*** *responses need to address the name, description and the effect of the modification. Effects have to be linked to the specific example discussed. Marks have to be all linked to one example. Assistant examiners are required to research examples.*

# Examiners report

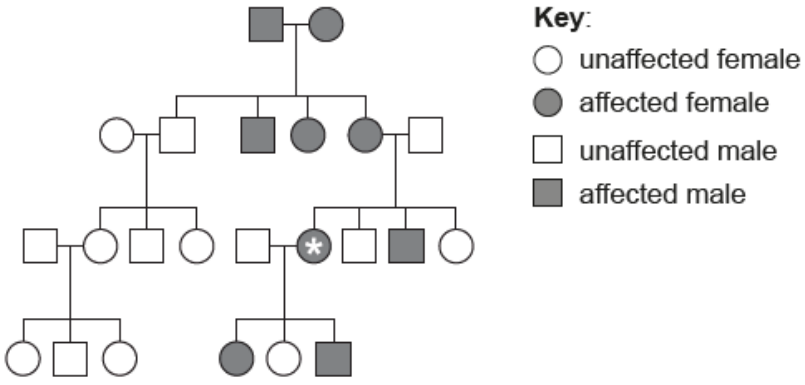
a. Students appear to have a general understanding of mechanisms but make a number of errors in terms of the location of events such as where the proton gradient builds up.

b. This was well answered by most students. Many did not draw the curve intersecting the horizontal axis at a value above zero. Many constructed a diagram of the curve but provided text below the curve in a paragraph rather than annotating the curve itself with explanations of what was occurring at various levels of light intensity.

c. The best answers outlined the biology of the example well though a very large number dealt in hypothetical or speculative costs and benefits of genetic modification.

---

This is a pedigree chart of a family with hypophosphatemia, an X-linked condition, in which bone deformities occur because of poor absorption of phosphates into the blood.




a. Using the pedigree chart, deduce the type of allele that causes hypophosphatemia. [2]

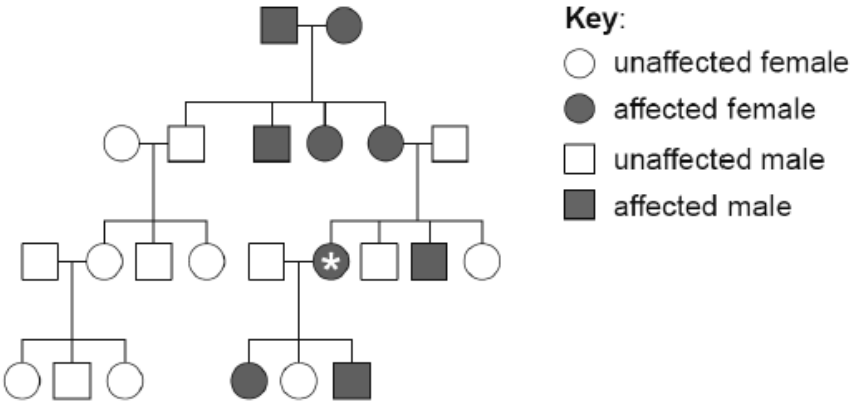
b. Identify the genotype of the individual marked with a star in the pedigree chart, using appropriate symbols for your answer. [1]

# Markscheme

a. a. dominant (allele)

*Reject dominant disease/homozygous dominant.*

b. all the offspring of the first generation would be affected if the allele was recessive (and one son is unaffected) / affected mothers could only have affected sons if the allele was recessive (and the pedigree shows that they can have both affected and unaffected sons) / affected mothers who have an unaffected son must be carriers of allele for being unaffected so the allele for being affected must be dominant / unaffected fathers could not have affected sons/daughters/children if the unaffected allele was dominant (and the pedigree shows that they can)




*There must be a coherent argument here and not just observations about individuals on the pedigree chart, but the argument can be expressed in various ways and can be shown using a Punnett square or other genetic cross diagram. Do not accept arguments that involve ratios between the phenotypes.*

b. $X^HX^h$ «where H = hypophosphatemia and h = normal «absorption of phosphate»»

*For the mark, allow any upper and lower case versions of the same letter, as long as they are shown superscript to an X to indicate sex-linkage.*

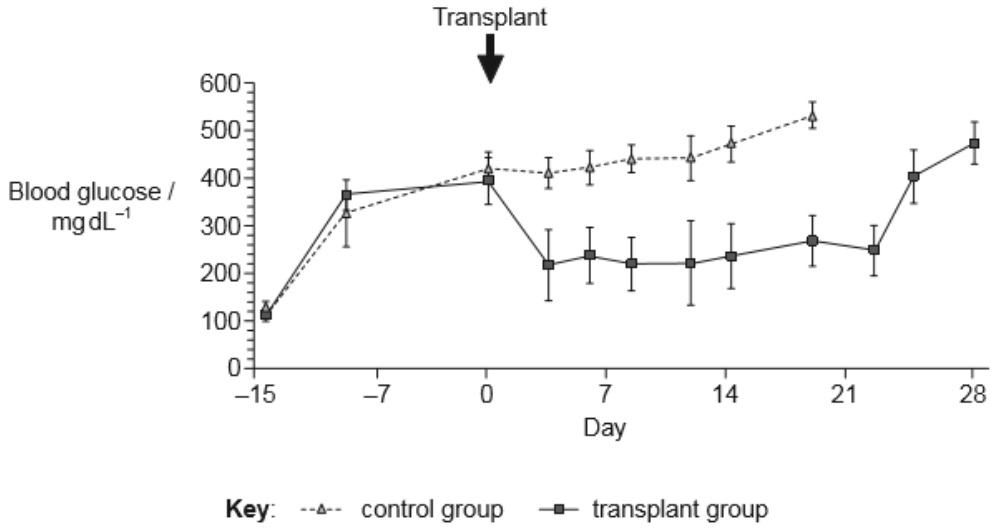
# Examiners report

a. [N/A]
b. [N/A]

---

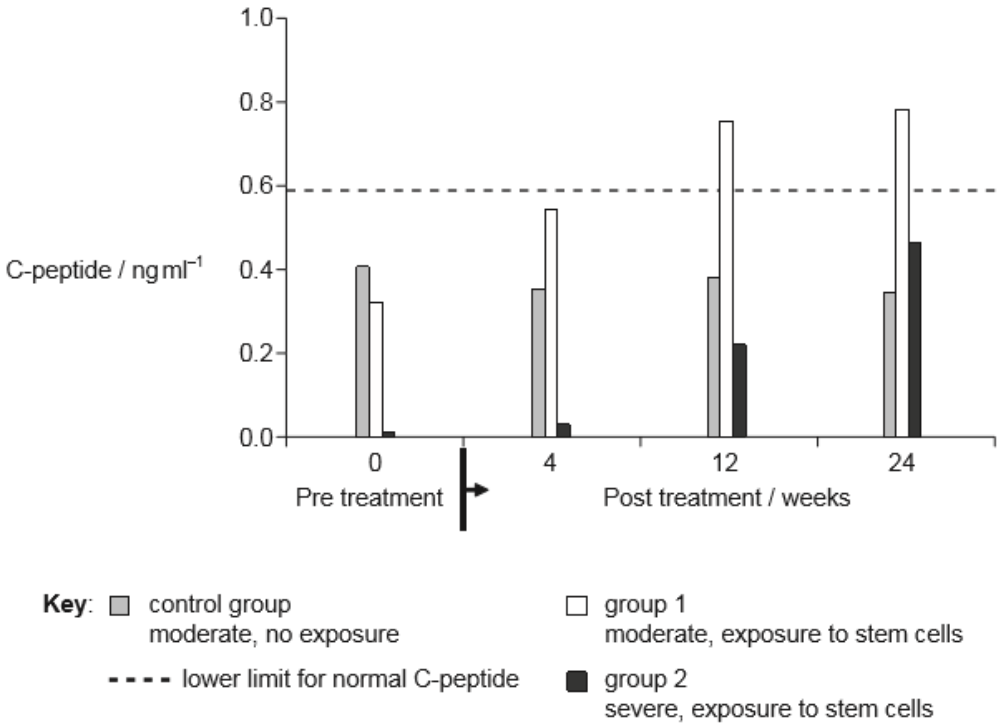
Type I diabetes is a leading cause of death in advanced countries and is associated with various severe or fatal complications, including blindness, kidney failure, heart disease, stroke, neuropathy, and amputations. Embryonic stem cells are considered to be a powerful tool in the treatment of diabetes.

In a study, embryonic stem cells were grown in culture and tested for insulin mRNA. A drug was injected into two groups of healthy mice in order to simulate type I diabetes 15 days prior to the transplant of embryonic stem cells. The mice in the transplant group received embryonic stem cells that produce insulin mRNA. The control group did not receive the transplant. The graph shows the blood glucose concentration in both groups.




[Source: Reprinted from *The American Journal of Pathology*, Vol 106, no. 6, Takahisa Fujikawa *et al.*, "Teratoma Formation Leads to Failure of Treatment for Type I Diabetes Using Embryonic Stem Cell-Derived Insulin-Producing Cells", pp. 1781–1791, Copyright © 2005 American Society for Investigative Pathology. Published by Elsevier Inc. All rights reserved.]

A few years later, a third study used a treatment with umbilical cord stem cells on patients who had suffered from moderate or severe type I diabetes for an average of 8 years. They were divided into two groups: group 1 had moderate diabetes and group 2 had severe diabetes. The patients' blood was circulated outside the body and exposed to umbilical cord stem cells before returning to the patients' circulation. The control group had moderate diabetes and received the same treatment but without umbilical cord stem cells.




[Source: doi:10.1186/1741-7015-10-3
Zhao *et al.*: Reversal of type 1 diabetes via islet ß cell regeneration following immune modulation by cord blood-derived multipotent stem cells. *BMC Medicine* 2012 10:3.]

a. State the highest mean concentration of blood glucose in the mice with transplants. [1]

.....mg dL$^{-1}$

b. Outline the cause of type I diabetes in humans. [1]

c. Describe the reason for testing for insulin mRNA in the embryonic stem cell cultures. [1]

d. Compare and contrast the concentration of blood glucose resulting from the embryonic stem cell transplant with the control. [2]

e. Evaluate the effectiveness of the embryonic stem cell treatment in controlling blood glucose. [2]

h. Compare and contrast the results of the treatment on group 1 with the results of the treatment on group 2. [3]

i. Suggest an ethical advantage of using this type of therapy over embryonic stem cell therapy. [1]

j. Using the data from all three studies, evaluate the use of embryonic stem cells as a treatment for type I diabetes. [4]

# Markscheme

a. 470. *Accept answers in the range of 460 to 480 «mg $dL^{-1}$»*.

b. «Autoimmune» destruction of beta/β cells. (*Accept B cells instead of β cells).*

Reduced/insufficient/no production of insulin

c. Indicates «stem» cells can produce insulin

***OR***

is needed for insulin production

***OR***

shows insulin gene is working/being translated.

Insulin is needed to treat type I diabetes
***OR***
insulin is needed to bring blood glucose level down

*Answers must relate to insulin mRNA.*

d. Decrease in transplant group «after treatment» in contrast to control group which does not decrease/decreases only very slightly

«initially»/increases/is higher than treatment group

Glucose «remains» lower in transplant group «than control group» for 2 max 2 weeks/3 weeks/for a time

«in the 4th week» transplant group rises back to level before transplant/to higher level than before transplant/to «near» level of control group

*The answer must include some indication of time or non-permanency.*

e. Glucose level still higher than normal/higher than 100 «mg»/higher than it was before the drug injection

Effective/lowers blood glucose for 3 weeks/temporarily/for a short time. *This can either be positive (the treatment is effective for a while) or negative (it isn't effective permanently).*

***OR***
glucose level rises back in 4th week/by day 28
***OR***
rises back to level of control group
***OR***
rises again but not above control group

*There must be a correct indication of the timing of the effects.*

h. C-peptide increases after treatment in both groups. *There must be an explicit comparison.*

***OR***

treatment effective in both groups

***OR***

both groups rose higher than the control

Similar/same overall/total increase «in both groups»
***OR***
quoted figures to show this

Smaller percentage/% increase «pre to post treatment» in group 1 «than group 2» *Reject answers relating to rates of increase*.
***OR***
quoted figures to show this

Initial increase is greater in group 1
***OR***
increases slowing/finished/rate of increase reduced by end of study/by week 24 in group 1 but continuing in group 2

Group 1 rose above lower limit «by week 12» and group 2 remained below it «even at week 24»

i. Umbilical cord «stem» cells are discarded/die if not harvested

***OR***

harvesting umbilical cord cells does not harm the baby

***OR***

taking «stem» cells from an embryo may harm/kill it

*Do not accept answers relating to consent.*

j. Study 1/study with mice/embryonic stem cell study shows treatment can cause increased insulin production/ reduce blood glucose levels

«Insulin production/reduction in blood glucose in study 1 was» only temporary/did not reduce glucose to normal levels

Study 2 shows increases in C-peptide/insulin
***OR***
some type I diabetes patients required no insulin after treatment

Study 2 shows treatment effective for a long time/2 years

«Stem cell treatment in study 2» was more successful in some patients than others
***OR***
more successful for moderate «than more severe» diabetes

Study 3 shows that stem cells can cause C peptide/insulin levels to double/rise significantly/rise above lower limit «for normal C-peptide»/rise and stay raised

«Study 3» does not give evidence for embryonic stem cells
***OR***
used umbilical cord rather than embryonic stem cells

# Examiners report

a. Most candidates successfully identified the highest mean blood glucose concentration.

b. Answers here were varied with many candidates correctly stating either that insulin is not produced or that beta cells are destroyed. A common

incorrect answer was to state that diabetes is a purely genetic disease.

c. There were also varied answers here with some candidates not appreciating the significance of the presence of insulin mRNA or not stating it clearly enough.

d. Many candidates failed to pick out more than one significant trend here. The data before the transplant was not relevant so the three phases that could have been described were the initial drop in blood glucose in the transplant group, the period when both groups remain relatively constant but the transplant group stayed lower and finally the rise in the transplant group to the original level before the transplant. For the latter two points the timing was expected.

e. Very few candidates scored both marks here. The point commonly made was that the drop in the transplant group is temporary. Very few candidate also made the significant point that the transplant did not cause the blood glucose concentration to drop down to the level before diabetes had been induced, so even in the early stages the treatment was not fully effective.

h. This was another place where many candidates failed pick out enough significant similarities and differences. One similarity and four significant differences were included in the mark scheme but most candidates scored only one or two marks (out of three).

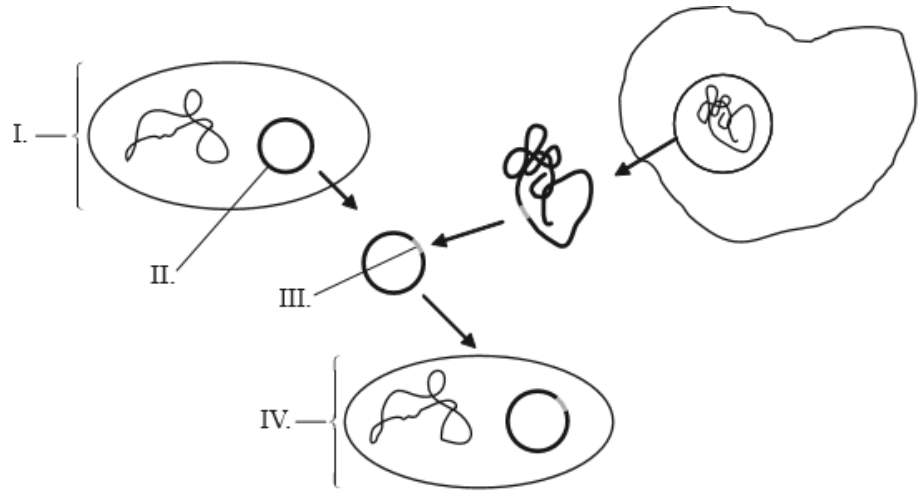
i. Most realised that there are ethical concerns if an embryo is damaged or killed and not if stem cells are taken from the placenta and umbilical cord before they are discarded, but the phrasing of answers was often too imprecise for a mark to be awarded. Terminology was frequently vague or incorrect. The terms embryo and fetus are not interchangeable, for example.

j. Many teachers correctly commented on G2 forms that there was an ambiguity in this question. It instructed candidates to use data from all three studies and also to evaluate the use of embryonic stem cells. Only study 1 had specifically been carried out using embryonic stem cells and study 3 was certainly done with umbilical cord rather than embryonic stem cells. It was therefore very important for candidates to quote which study they were using for a particular point and not all did this. Another weakness of some answers was to mention trends in the data without making them clearly a strength or weakness as is expected in an evaluation. The best answers coped well with the ambiguity in the question and scored full marks.

---

a. Explain why carriers of sex-linked (X-linked) genes must be heterozygous. [2]

b (i) Label the diagram below which shows a basic gene transfer. [2]




I. .....
II. .....
III. .....
IV. .....

b (ii) State **two** general types of enzymes used in gene transfer. [1]

# Markscheme

a. carrier has (one copy of) a recessive allele;

must also have a dominant allele to prevent having the condition/disease;

***or***

cannot be homozygous dominant or they would not carry the recessive allele;

cannot be homozygous recessive or they would have the condition/disease;

b (i) *Award **[1]** for every two correct answers.*

I. bacterial cell/bacterium/prokaryote;

II. plasmid;

III. inserted/engineered/cloned/desired DNA/gene / DNA/gene from donor cell;

IV. genetically modified/transformed/GM/recombinant organism/cell/ bacterium/host cell containing recombinant plasmid;

b (ii) restriction enzymes / endonucleases;

ligases;

reverse transcriptase;

*Award **[1]** for two correct answers.*

# Examiners report

a. There was a significant challenge here in explaining why carriers of sex-linked genes must be heterozygous. The inclusion of sex-linkage in this question was something of a “red herring” to use an English idiom, as the answer to the question would have been the same for a gene that was not sex linked. It was not possible to explain the answer effectively without referring to recessive and dominant alleles.

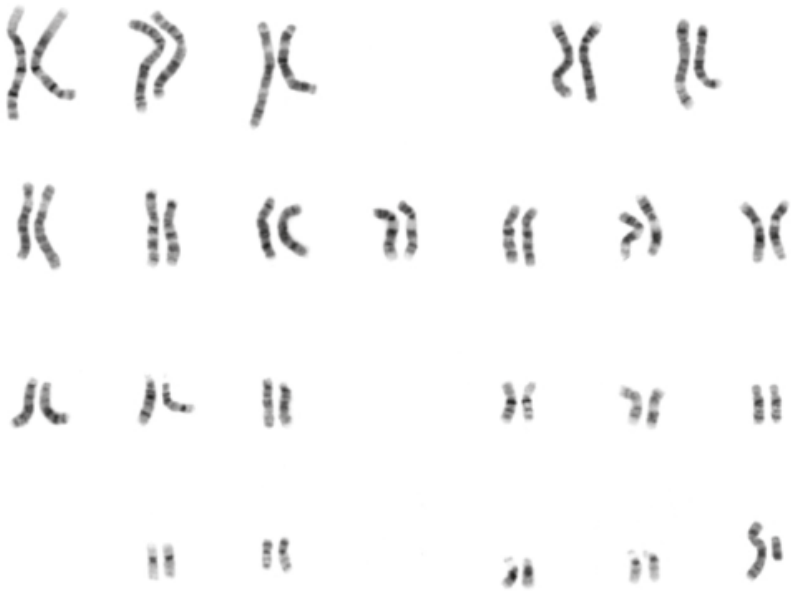
Some candidates referred to genes rather than alleles, but as long as the meaning of the answer was clear, marks were awarded. Understandably some candidates took more notice of sex-linkage than was necessary and in some cases did little more than explain gender determination.

b (i) Approximately equal numbers of candidates scored two, one and no marks here. Many could recognise the bacterial cell (I) and the plasmid within it (II) but fewer were able to give acceptable names for the gene transferred from the eukaryotic cell or the genetically modified bacterium.

b (ii) This generally well answered with most candidates naming two enzymes involved in gene transfer. Restriction enzymes and DNA ligase were the most obvious ones but reverse transcriptase was also accepted as it is used to produce DNA copies of genes from mRNA. DNA polymerase and RNA polymerase were the commonest answers that were not accepted.

---

The diagram shows a human karyotype.



[Source: http://en.wikipedia.org/wiki/File:NHGRI_human_male_karyotype.png, courtesy of the National Human Genome Research Institute.]

a. Analyse this karyotype. [2]

b. Outline the inheritance of hemophilia in humans. [2]

c. Using an example, describe polygenic inheritance. [3]

# Markscheme

a. Male has (one X and) one Y chromosome / X chromosome is bigger than Y chromosome;

non-disjunction leads to three copies of chromosome 13/trisomy 13.

b. sex-linked/on X chromosome;

recessive allele / Xh;

more common in males than females;

heterozygous females are carriers / only females can be carriers;

c. more than one gene contribute to/control same characteristic;

as number of genes increase so does possible number of phenotypes;

leads to continuous variation;

specific example; *(eg human skin color (due to differing amounts of melanin))*

*Award **[2 max]** for general points with no example.*

# Examiners report

a. The fact that there was a trisomy 13 as a result of non-disjunction eluded the majority, who seemed to register that pair 21 was OK, therefore nothing else could be wrong. Many lost a mark for not explaining why it was a male. Better prepared candidates were able to explain haemophilia and polygenic inheritance. For some candidates it seemed to be the first time that they had encountered them.

b. The fact that there was a trisomy 13 as a result of non-disjunction eluded the majority, who seemed to register that pair 21 was OK, therefore nothing else could be wrong. Many lost a mark for not explaining why it was a male. Better prepared candidates were able to explain haemophilia and polygenic inheritance. For some candidates it seemed to be the first time that they had encountered them.

c. The fact that there was a trisomy 13 as a result of non-disjunction eluded the majority, who seemed to register that pair 21 was OK, therefore nothing else could be wrong. Many lost a mark for not explaining why it was a male. Better prepared candidates were able to explain haemophilia and polygenic inheritance. For some candidates it seemed to be the first time that they had encountered them.

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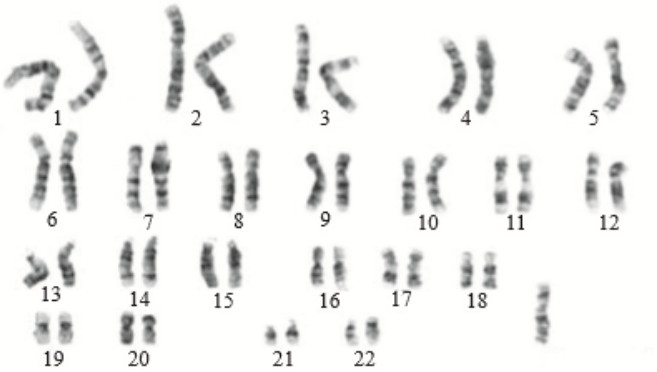
The image shows the karyotype of a person who developed as a female.




[Source: http://en.wikipedia.org/wiki/File:45,X.jpg]

a (i) In a strain of soybeans, high oil content (H) in seeds is dominant to low oil content (h) and four seeds in a pod (F) is dominant to two seeds in a pod (f). A farmer crosses two soybean plants, both with high oil content and four seeds in a pod. The offspring have a phenotypic ratio of 9 : 3 : 3 : 1. [1]

Identify the genotypes of the soybean plants with high oil content and four seeds in a pod that were used in the cross.

a (ii) In a strain of soybeans, high oil content (H) in seeds is dominant to low oil content (h) and four seeds in a pod (F) is dominant to two seeds in a pod (f). A farmer crosses two soybean plants, both with high oil content and four seeds in a pod. The offspring have a phenotypic ratio of 9 : 3 : 3 : 1. [2]

Determine the genotypes of the gametes and offspring using a Punnett grid.

a (iii) In a strain of soybeans, high oil content (H) in seeds is dominant to low oil content (h) and four seeds in a pod (F) is dominant to two seeds in a pod (f). A farmer crosses two soybean plants, both with high oil content and four seeds in a pod. The offspring have a phenotypic ratio of 9 : 3 : 3 : 1. [2]

Identify the phenotypes of each part of the phenotypic ratio.

| Ratio | Phenotypes |
|---|---|
| 9 | |
| 3 | |
| 3 | |
| 1 | |

b (i) Deduce the reason for the person developing as a female. [1]

b (ii) Determine, with a reason, whether this karyotype shows that non-disjunction has occurred. [1]

# Markscheme

a (i). HhFf HhFf □ / (both) HhFf;

a (ii).

| gametes | HF | Hf | hF | hf |
|---|---|---|---|---|
| HF | HHFF | HHFf | HhFF | HhFf |
| Hf | HHFf | HHff | HhFf | Hhff |
| hF | HhFF | HhFf | hhFF | hhFf |
| hf | HhFf | Hhff | hhFf | hhff |

all gametes shown correctly on Punnett grid;
all offspring genotypes correct;

a (iii).

| | *ratio* | *phenotypes* |
|---|---|---|
| a. | *9* | high oil four seeds; |
| b. | *3* | high oil two seeds; |
| c. | *3* | low oil four seeds; |
| d. | *1* | low oil two seeds; |

*Award **[1]** for any **two** correct phenotypes.*

b (i). no Y chromosome.

b (ii). yes as there is only one X chromosome/chromosome missing/only 45 chromosomes

# Examiners report

a (i). Most wrote the correct genotype, though there were a surprising number that did not follow notation conventions such as writing HFhf or using linkage notation.

a (ii). The majority of students could answer this question. Where students were not answering correctly, it was due to a lack of conceptual understanding of segregation; i.e. writing gametes as HH or ff for example.

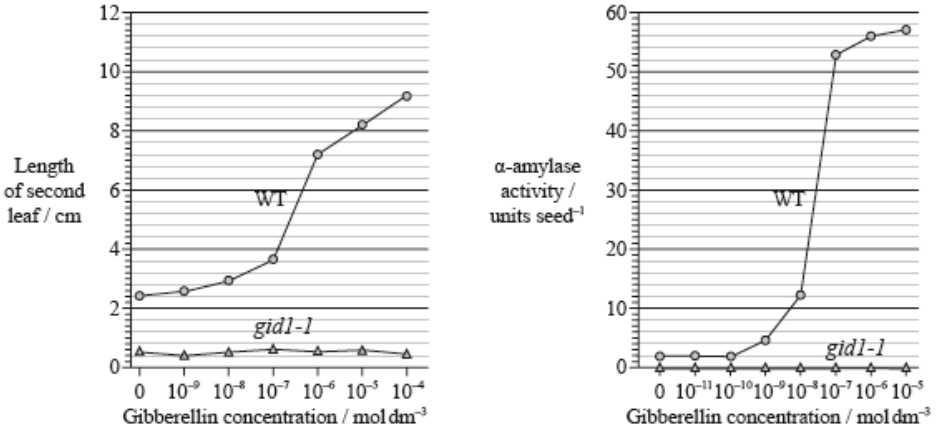
a (iii). This question was most commonly answered correctly.

b (i). Approximately half of students answered this correctly. A number did not recognize the condition for determining a female was the absence of the Y chromosome rather than the presence of the X chromosome.

b (ii). This was more commonly answered correctly than i). Here a common misunderstanding was that nondisjunction could only be present if additional chromosomes were present rather than if one were missing.

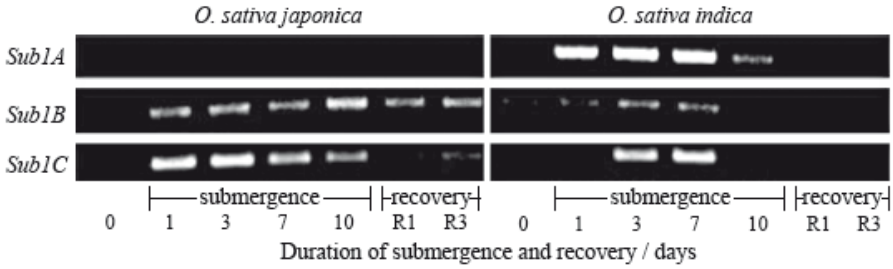
---

Gibberellin promotes both seed germination and plant growth. Researchers hypothesize that the gene *GID1* in rice (*Oryza sativa*) codes for the production of a cell receptor for gibberellin. The mutant variety *gid1-1* for that gene leads to rice plants with a severe dwarf phenotype and infertile flowers when homozygous recessive. It is suspected that homozygous recessive *gid1-1* plants fail to degrade the protein SLR1 which, when present, inhibits the action of gibberellin. The graphs show the action of gibberellin on the leaves and α-amylase activity of wild-type rice plants (WT) and their *gid1-1* mutants.




[Source: adapted from M. Ueguchi-Tanaka et al. (2005) 'Gibberellin-insensitive dwarf1 encodes a soluble receptor for gibberellin'. Nature, 437, pp. 693—698. Adapted by permission from Macmillan Publishers Ltd (c) 2005.]

Most rice varieties are intolerant to sustained submergence under water and will usually die within a week. Researchers have hypothesized that the capacity to survive when submerged is related to the presence of three genes very close to each other on rice chromosome number 9; these genes were named *Sub1A*, *Sub1B* and *Sub1C*. The photograph below of part of a gel shows relative amounts of messenger RNA produced from these three genes by the submergence-intolerant variety, *O. sativa japonica*, and by the submergence-tolerant variety, *O. sativa indica*, at different times of a submergence period, followed by a recovery period out of water.

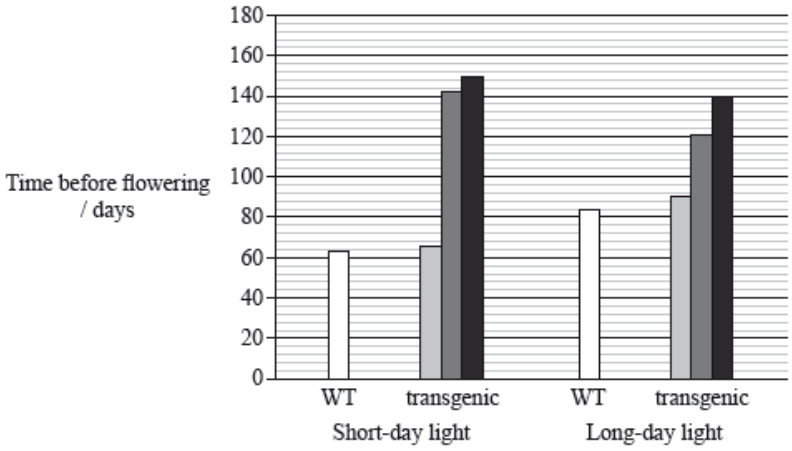



[Source: Adapted from "Sub1A is an ethylene-response-factor-like gene that confers submergence tolerance to rice" (2006) Kenong Xu, Xia Xu, Takeshi Fukao, Patrick Canlas, Reycel Maghirang-Rodriguez et al. Nature, 442, pp. 705—708. Adapted by permission from Macmillan Publishers Ltd (c) 2006.]

The *OsGI* gene causes long-day flowering and the effect of its overexpression has been observed in a transgenic variety of rice. Some wild-type rice (WT) and transgenic plants were exposed to long days (14 hours of light per day) and others to short days (9 hours of light per day).

The shades of grey represent the genotypes of the transgenic plants, where:

- –/– do not have the overexpressed *OsGI* gene
- +/– are heterozygous for the overexpressed *OsGI* gene
- +/+ are homozygous for the overexpressed *OsGI* gene.




[Source: adapted from R. Hayama, S. Yokoi, S. Tamaki, M. Yano and K. Shimamoto (2003) 'Adaptation of photoperiodic control pathways produces short-day flowering in rice.' Nature, 422, pp. 719—722. Adapted by permission from Macmillan Publishers Ltd (c) 2003.]

a(i). State which variety of rice fails to respond to gibberellin treatment. [1]

a(ii). The activity of α-amylase was tested at successive concentrations of gibberellin. Determine the increment in gibberellin concentration that produces the greatest change in α-amylase activity in wild-type rice plants (WT). [1]

b. Discuss the consequence of crossing *gid1-1* heterozygous rice plants amongst themselves for food production. [3]

c(i). Determine which gene produced the most mRNA on the first day of the submergence period for variety *O. sativa japonica*. [1]

c(ii). Outline the difference in mRNA production for the three genes during the submergence period for variety *O. sativa indica*. [2]

d. Using only this data, deduce which gene confers submersion resistance to rice plants. [2]

e(i). State the overall effect of overexpression of the *OsGI* gene in plants treated with short-day light. [1]

e(ii). Compare the results between the plants treated with short-day light and the plants treated with long-day light. [2]

e(iii). State, giving **one** reason taken from the data opposite, if unmodified rice is a short-day plant **or** a long-day plant. [1]

g. Evaluate, using all the data, how modified varieties of rice could be used to overcome food shortages in some countries. [2]

# Markscheme

a(i). *gid1-1*

a(ii). between $10^{-8}$ and $10^{-7}$ mol $dm^{-3}$ *(units required)*

b. a. 25% / 1 in 4 / 1:3 seeds produced would be homozygous recessive;

b. no response to/inhibits gibberellin in homozygous recessives results in less germination;

c. less growth / dwarf plants produced; *(must be in context)*;

d. would produce plants with infertile flowers that cannot produce rice grains;

e. would lower rice production/less yield because infertile plants cannot produce seeds (that humans can eat);

c(i) *Sub1C*

c(ii) a. *Sub1A* is expressed strongly/the most / *Sub1A* produces the most RNA;

b. *Sub1B* (always) has the lowest expression/produces least mRNA;

c. *Sub1A* expressed/produces mRNA for the longest time/days 1 to 10;

d. *Sub1C* expressed/produces mRNA for the shortest time/days 3 to 7;

d. a. *Sub1A*;

b. is only expressed in *indica* / *Sub1B* and *SubC* are expressed in both rice varieties;

c. *indica* is the variety showing submersion tolerance / vice versa for *japonica*;

e(i) it increases the length of time <u>before</u> flowering

e(ii) a. long-day light exposure increases time before flowering only if (*OsGI*) gene is not overexpressed/in WT and –/–;

b. long-day light exposure decreases time before flowering for +/– and/or +/+;

c. length of day does not make much difference/makes least difference for +/+;

d. overexpression for +/– reduces time before flowering;

e. –/– acts as a control / has nearly the same length of time before flowering as WT;

*Accept numerical answers if they are making a clear comparison*.

e(iii) is a short-day plant <u>because</u> WT has shortest time/shorter time before flowering in shorter days than longer days / as it takes less time to flower under short day conditions;

g. a. the mutant *gid1-1* would not be useful because it produces sterile plants;

b. genetically modified rice/rice with *Sub1A* is more tolerant to submersion/can withstand seasonal flooding/torrential rain;

c. *OsGI*+ varieties adapted to different latitudes / day length could be produced (to overcome food shortages);

d. short flowering time possibly means more crops per year;

# Examiners report

a(i) The word “increment” seemed to confuse the weaker candidates who stated a value rather than a range. In addition there were a large number who omitted or misquoted the units. In spite of being clearly stated in topic 9.3.5, very few candidates correctly gained the mark in part (iii) for saying that the amylase catalysed the breakdown of starch to maltose. Many answered glucose instead of maltose, but a surprising number did not even realise that amylase is an enzyme.

a(ii) The word “increment” seemed to confuse the weaker candidates who stated a value rather than a range. In addition there were a large number who omitted or misquoted the units. In spite of being clearly stated in topic 9.3.5, very few candidates correctly gained the mark in part (iii) for saying that the amylase catalysed the breakdown of starch to maltose. Many answered glucose instead of maltose, but a surprising number did not even realise that amylase is an enzyme.

b. Most of the better candidates realised that it was a simple monohybrid cross (although several thought it was dihybrid) and realised that 25% would produce dwarf plants, but did not explain the consequences on potential yield in sufficient detail for the third mark.

c(i). In spite of doubts from the G2 forms, candidates had little difficulty in interpreting the photograph.

In part (i) most correctly answered *Sub1C*.

c(ii). The answers to (ii) tended to be descriptive, not making clear <u>differences</u>, as asked.

d. Most candidates correctly identified *Sub1A* with a correct reason.

e(i). Most answered correctly that it increased the time <u>before</u> flowering.

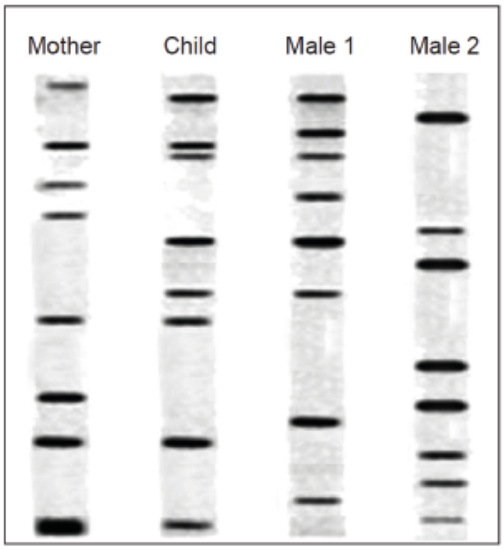
e(ii). In (ii) almost every correct answer was from the first two mark points.

e(iii). In (iii) most candidates identified it as a short-day plant with reasons.

g. In spite of the stem saying “using all the data”, most of the answers were very vague and did not use the data. The ideas that the mutant *gid1-1* should be avoided as it produces sterile plants and those modified with *Sub1A* would withstand seasonal flooding were missed by most candidates.

---

The image shows data collected in order to determine the paternity of a child.




[Source: © International Baccalaureate Organization 2015]

a. State the name of the process used to produce the pattern of bands seen in the image. [1]

b. Determine, with a reason, which male is the father of the child. [1]

# Markscheme

a. gel electrophoresis/DNA profiling

b. male 1 because all child's bands / alleles match either mother or male 1 / (approximately) half of bands match male 1 [1]

*Do not accept reference to genes.*

# Examiners report

a. Most candidates answered this question correctly.

b. If candidates had difficulty with this question it was due to communication issues. Some referred to the bands as genes and others found it difficult to clearly express their rationale for identifying Male 1 as the father.

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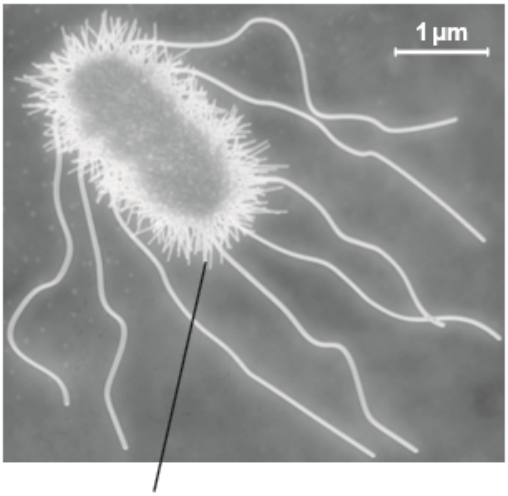



a. Outline the cell theory. [2]

b (i). Annotate the electron micrograph of the *Escherichia coli* cell with the function of the indicated structure. [1]

b (ii). Calculate the magnification of the electron micrograph. [1]

c (i). Explain the role of the following enzymes in DNA replication. [1]

Helicase

c (ii). Explain the role of the following enzymes in DNA replication. [1]

DNA ligase

# Markscheme

a. a. living things are composed of cells;

b. cells are the basic/smallest unit of life;

c. cells come from pre-existing cells;

*Do not accept cells are the "smallest organisms".*

*Do not accept "cells are the building blocks" of life on its own.*

b (i) attachment to surfaces / holds bacteria together / conjugation

*Do not accept "exchange material" on its own.*

*If more than one function is given, mark the first answer only.*

b (ii) × 15 000 *(accept answers in the range of × 14 000 to × 16 000)*

c (i) helicase: unwinds /unzips the DNA (into two strands) / breaks H bonds;

c (iv) DNA ligase: joins/seals the nick between the (Okazaki) fragments;

# Examiners report

a. Most students earned these marks. A small number demonstrated knowledge of the properties of cells but seemed to be unfamiliar with the cell theory itself.

b (i) A number failed to state a correct function. The pilus plays a role in adhering to surfaces and in bacterial conjugation. A number annotated the picture with the name of the structure without stating a function.

b (ii) About half of candidates correctly answered this question. A number were making order of magnitude errors such as writing 150 000x and 1500x. Some were unfamiliar with the interpretation of the metric prefix.

c (i) Most were able to explain the function of helicase.

c (iv) Similar to primase, the mechanism of action of ligase was very rarely accurately described, most limiting it to bond formation between Okazaki fragments, not acknowledging that ligase has a role on the leading strand as well.

---

The Chinese soft-shelled turtle, *Pelodiscus sinensis*, lives in salt water marshes. The turtle can live under water and out of water.

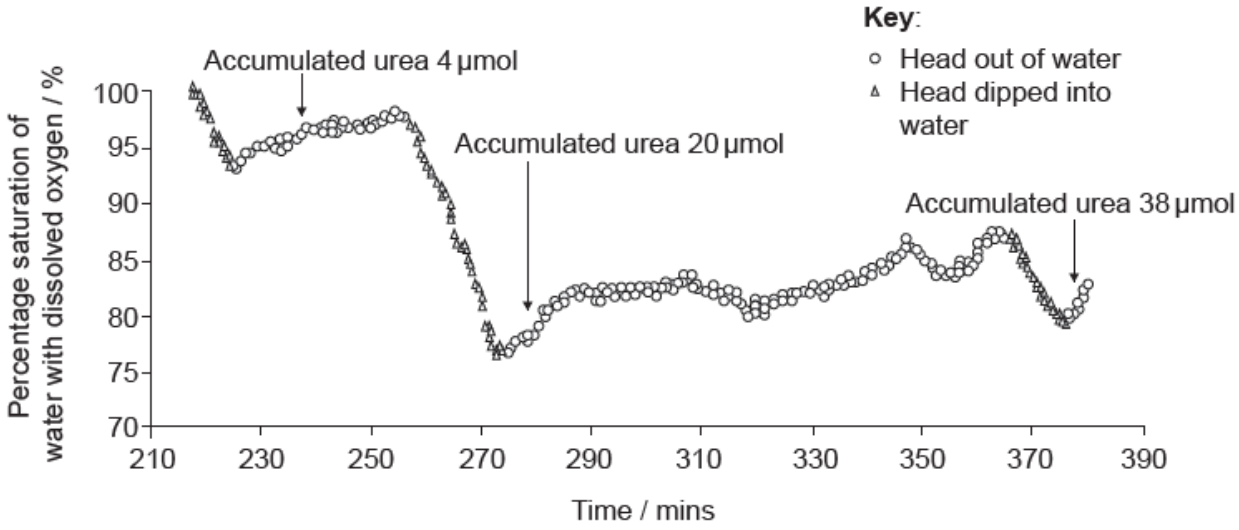
These turtles have fully developed lungs and kidneys, however, many microvilli have been discovered in the mouth of *P. sinensis*. A study was undertaken to test the hypothesis that oxygen uptake and urea excretion can simultaneously occur in the mouth.

Initial experiments involved collecting nitrogen excretion data from *P. sinensis*. The turtle urinates both in water and out of water. When in water it allows waste products to be washed out of its mouth. When out of water it regularly dips its head into shallow water to wash its mouth. The table shows the mean rates of ammonia and urea excretion from the mouth and kidney over six days.

| | Excretion of nitrogen by the mouth / μmol day$^{-1}$ g$^{-1}$ turtle | | Excretion of nitrogen by the kidney / μmol day$^{-1}$ g$^{-1}$ turtle | |
|---|---|---|---|---|
| | Turtle submerged in water | Turtle out of water | Turtle submerged in water | Turtle out of water |
| **Ammonia** | 0.29 | 0.30 | 0.63 | 0.54 |
| **Urea** | 0.90 | 1.56 | 0.07 | 0.73 |

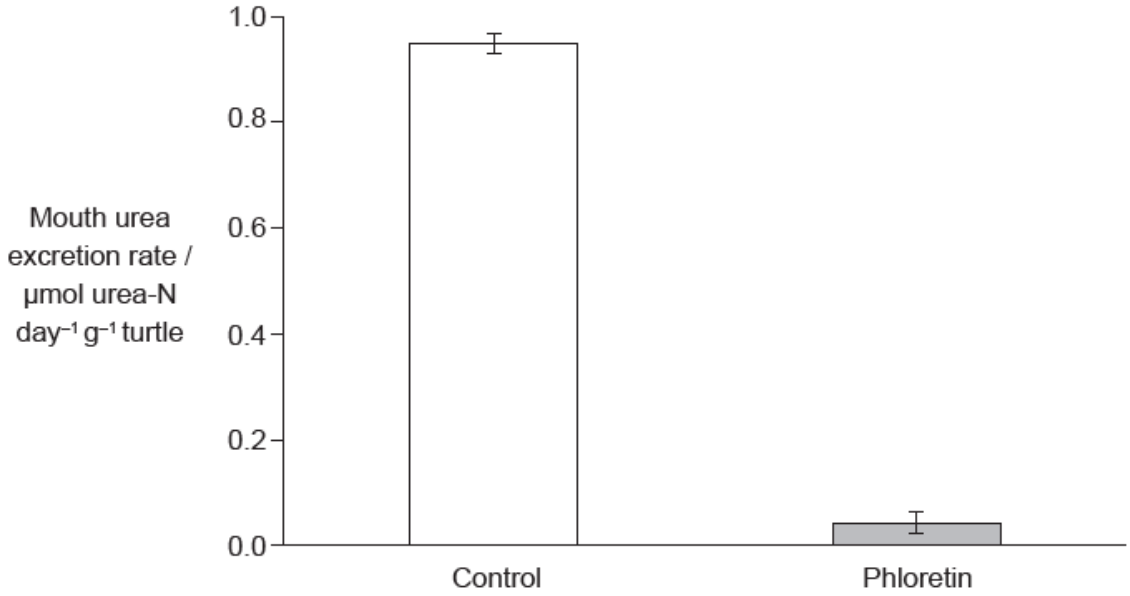
[Source: Reproduced with permission, Y. Ip *et al*. (2012) *The Journal of Experimental Biology*, 215, pages 3723—3733. jeb.biologists.org. doi: 10.1242/jeb.068916]

It was noted that during long periods out of water, turtles rhythmically moved their mouths to take in water from a shallow source and then discharge it. Changes in the dissolved oxygen and the quantity of accumulated urea in the rinse water discharged by the turtles were monitored over time as shown in this graph.




[Source: adapted with permission from Y. Ip *et al*. (2012) *The Journal of Experimental Biology*, 215, pages 3723–3733.]

In order to test whether a urea transporter was present in the mouth tissues of the turtles, phloretin (a known inhibitor of membrane proteins that transport urea) was added to the water in which a further set of turtles submerged their heads. The results of that treatment are shown.

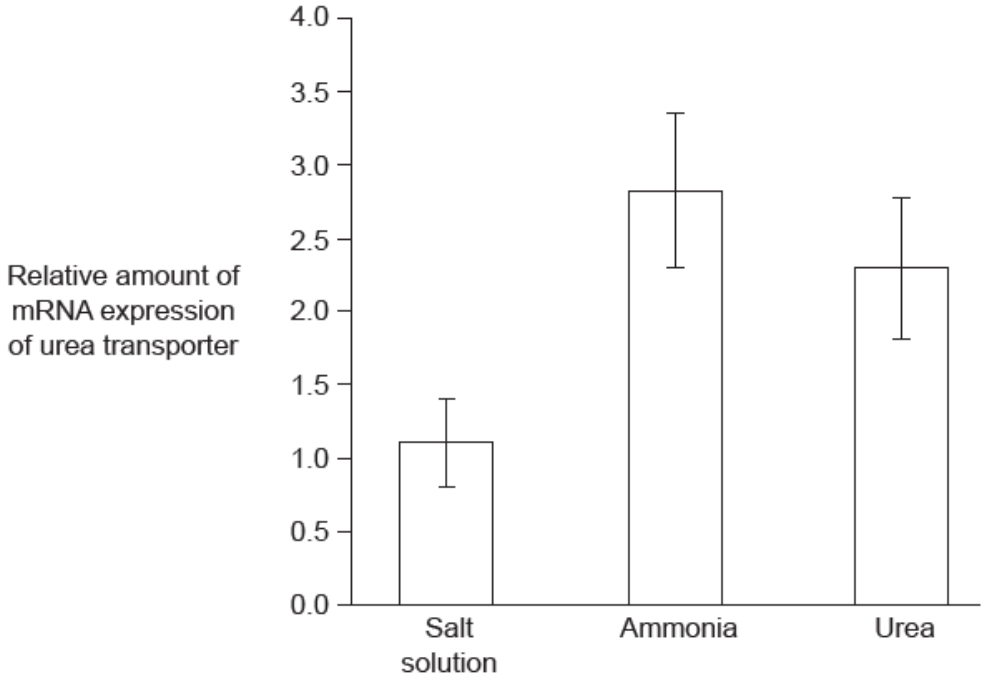



[Source: Reproduced with permission from Y. Ip *et al*. (2012) *The Journal of Experimental Biology*, 215, pages 3723–3733. jeb.biologists.org.]

Further research was conducted to determine where mRNA expression of a urea transporter gene might be occurring in *P. sinensis*. Gel electrophoresis was used to analyse different tissue samples for mRNA activity.


[Source: Reproduced with permission from Y. Ip *et al.* (2012) *The Journal of Experimental Biology*, 215, pages 3723–3733. jeb.biologists.org.]

Expression of the urea transporter gene by cells in the turtle's mouth was assessed by measuring mRNA activity. Turtles were kept out of water for 24 hours and then injected with either a salt solution that matched the salt concentration of the turtle, dissolved ammonia or urea, followed by another 24 hours out of water.




[Source: © International Baccalaureate Organization 2017]

a. Deduce whether the excretion of ammonia or urea changes more when a turtle emerges from water. [2]

b. Compare and contrast the changes in urea excretion in the mouth with the changes in urea excretion in the kidney when a turtle emerges from the water. [3]

c.i. Describe the trends shown by the graph for dissolved oxygen in water discharged from the mouth. [1]

c.ii. Suggest reasons for these trends in dissolved oxygen. [2]

d. Deduce with a reason whether a urea transporter is present in the mouth of *P. sinensis*. [2]

e. Outline the additional evidence provided by the gel electrophoresis results shown above. [2]

f.i. Identify which of these turtle groups represent the control, giving a reason for your answer. [1]

f.ii. Suggest a reason for the greater expression of the gene for the urea transporter after an injection with dissolved ammonia than an injection of urea. [2]

g. The salt marshes where these turtles live periodically dry up to small pools. Discuss the problems that this will cause for nitrogen excretion in the turtles and how their behaviour might overcome the problems. [3]

# Markscheme

a. a. urea

b. for both mouth and kidney

c. percentage change/change in μmol $day^{-1}$ $g^{-1}$ greater with urea/other acceptable numerical comparison

b. a. both higher/increased on emergence from/with turtle out of water

b. both increased by 0.66 «$\mu mol^{-1}$ $g^{-1}$ when turtle emerges from water»

c. % increase is higher in kidney / kidney 940% versus mouth 73/75% / increase is higher proportionately higher in kidney / kidney x10 versus mouth nearly double/x1.73

d. urea excretion by mouth greater than kidney out of water «despite larger % increase in kidney excretion»

c.i. decrease «when head is submerged» and increase when head is out of water

c.ii. a. oxygen absorbed from water/exchanged for urea when head dipped in water«so oxygen concentration decreases»

b. lungs cannot be used with head in water / can «only» be used with head out of water

c. oxygen from water «in mouth» used in «aerobic cell» respiration

d. oxygen from air dissolves in water when head out of water «so oxygen concentration increases»

d. a. urea transporter is present

b. less urea «excreted»/ lower rate «of urea excretion» / excretion almost zero when phloretin/inhibitor was present

e. a. mRNA only in mouth and tongue/in mouth and tongue but not esophagus intestine kidney or bladder

b. bands / lines indicate mRNA for/expression of urea transporter gene

c. urea transporter gene expressed / urea transporters in mouth/tongue / not expressed/made in esophagus/intestine/kidneys/bladder

d. mRNA/transcription/gene expression/urea transporters higher in tongue/more in tongue «than mouth»

f.i. salt solution is control because it does not contain a nitrogenous/excretory waste product / it matches the salt concentration of the turtle / the turtle's body already contains salt / because the turtle lives in salt water/salt marshes / because nothing has been altered

f.ii. a. ammonia is «highly» toxic/harmful

b. ammonia is more toxic than urea/converse

c. ammonia converted to urea

d. urea concentration raised «by injecting ammonia»

e. difference between ammonia and urea «possibly» not «statistically» significant

g. *Problems:*

a. urea becomes more concentrated «in small pools» / lower concentration gradient «between tongue/mouth and water»

b. less water available for urine production/excretion by kidney

***OR***

less water in ponds for mouth rinsing/more competition for pools (to use for mouth rinsing)

*Behaviour to overcome problems:*

c. «still able to» dip mouth into/mouth rinse in water/pools

d. «still able to» excrete urea «though the mouth» in the small pools

e. more conversion of ammonia to urea/urea excretion rather than ammonia

f. more urea transporters/expression of urea transporter gene

g. urea excreted «in mouth/via microvilli» by active transport/using ATP

h. excretion with little/no loss of water

# Examiners report

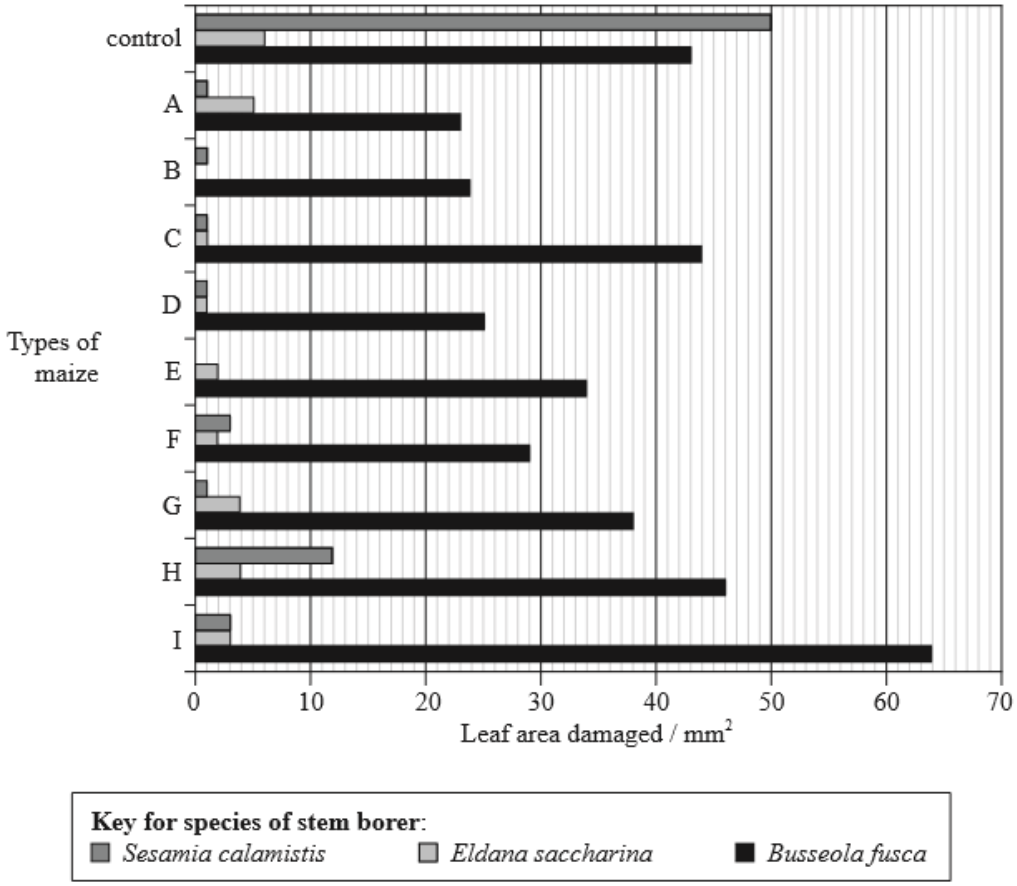
a. [N/A]
b. [N/A]
c.i. [N/A]
c.ii. [N/A]
d. [N/A]
e. [N/A]
f.i. [N/A]
f.ii. [N/A]
g. [N/A]

---

Genetic engineering allows genes for resistance to pest organisms to be inserted into various crop plants. Bacteria such as *Bacillus thuringiensis* (Bt)

produce proteins that are highly toxic to specific pests.

Stem borers are insects that cause damage to maize crops. In Kenya, a study was carried out to see which types of Bt genes and their protein products would be most efficient against three species of stem borer. The stem borers were allowed to feed on nine types of maize (A–I), modified with Bt genes. The graph below shows the leaf areas damaged by the stem borers after feeding on maize leaves for five days.

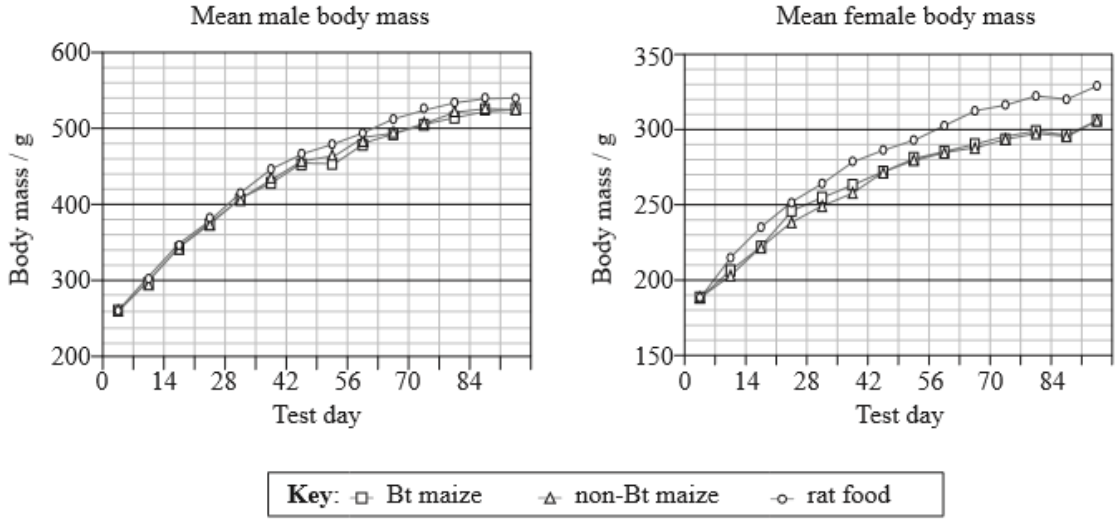



[Source: adapted from S Mugo, *et al.*, (2005), *African Journal of Biotechnology*, **4** (13), pages 1490–1504]

Before the use of genetically modified maize as a food source, risk assessment must be carried out. A 90-day study was carried out in which adult male and female rats were fed either:

- seeds from a Bt maize variety
- seeds from the original non-Bt maize variety
- commercially prepared rat food.

All the diets had similar nutritional qualities.




[Source: adapted from L A Malley, *et al.*, (2007), *Food and Chemical Toxicology*, **45**, pages 1277–1292]

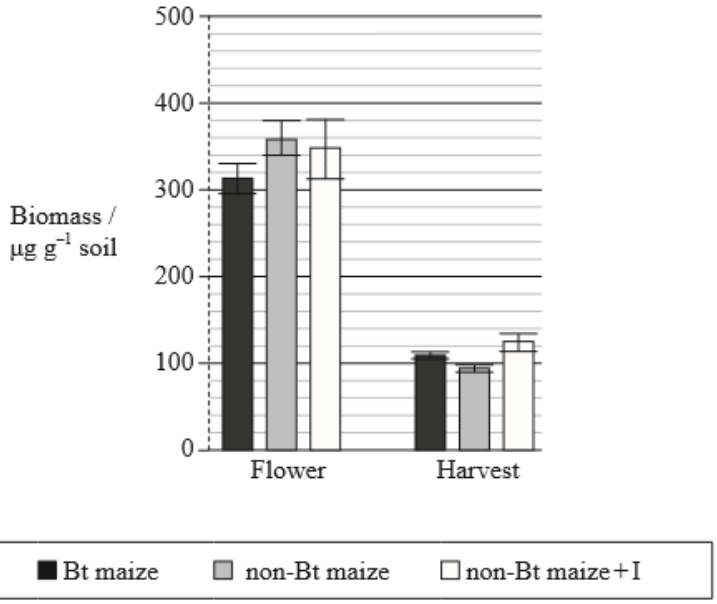
Studies have shown that Bt proteins are released by plant roots and remain in the soil. One study looked at the biomass of microorganisms in soil surrounding the roots of:

• Bt maize

• non-Bt maize

• non-Bt maize with an insecticide (I).

The graph below shows the biomass of microorganisms at two different times in the growth cycle of the plants (Flower and Harvest). Error bars represent standard error of the mean.




[Source: adapted from M Devare, *et al.*, (2007), *Soil Biology and Biochemistry*, **39**, pages 2038–2047]

Bt proteins act as toxins to insects, primarily by destroying epithelial cells in the insect's digestive system. Below is the three-dimensional structure of one such protein.


[Source: M Soberon, *et al.*, (2007), *Toxicon*, **49**, pages 597–600]

a. Calculate the percentage difference in leaf area damaged by *Sesamia calamistis* between the control and maize type H. Show your working. [2]

b. Discuss which species of stem borer was most successfully controlled by the genetic engineering of the maize plants. [3]

c. Calculate the change in mean mass of male and of female rats fed on Bt maize from day 14 to 42. [2]

d. Evaluate the use of Bt maize as a food source on the growth of the rats. [2]

e. Comment on the use of Bt maize as a food source compared to the other diets tested. [1]

g. Compare the biomass of microbes in the soils surrounding the roots of Bt maize and non-Bt maize. [2]

h. The researchers' original hypothesis stated that microorganisms would be negatively affected by the Bt protein released by the plant roots. [2]

Discuss whether the data supports the hypothesis.

i (i). State the type of structure shown in the region marked A in the diagram above. [1]

i (ii). Outline how this structure is held together. [2]

i (iii). Region A inserts into the membrane. Deduce, with a reason, the nature of the amino acids that would be expected to be found in this region. [2]

# Markscheme

a. 50 12 38 (mm ); *Accept 12 50 = 38*

(38 50) 100 ( )76(%); (ECF)

b. *Sesamia* (was most successfully controlled);

in control plants *Sesamia* caused most damage;

all types of Bt/genetically modified maize/A–I show (significant) decrease in damage by *Sesamia*;

mark for correct numerical comparison;

*Sesamia* caused no damage to type E/ in one instance;

*Busseola* not controlled/affected by Bt/genetically modified maize/caused largest amount of damage in types A–I/increased damage in some varieties;

*Eldana* controlled by some types of maize / B/C/D but not others / *Eldana* caused least damage in control and not much difference in many maize types;

c. *males*: (440 – 325 =)115g ; (*Accept answers in range 105–125 g*)

*females*: (268 – 215 =)53g ; (*Accept answers in range 51–57 g*)

*Units required, no workings required*.

d. (promotes) highest rate of growth at start of study / tapering off later in the study;

Bt maize appears to cause less growth/mass gain than rat food / vice versa;

more pronounced difference in females;

no difference in growth/mass gain between Bt and non-Bt maize;

e. (Bt) maize may not be as good as the (commercially prepared) rat food;

Bt maize appears to be as good a food source as non-Bt maize;

Bt maize an acceptable/safe food source;

*Answers require a judgement about Bt maize as a food source rather than a description.*

g. (for both groups) overall biomasses were higher during flowering than harvest / vice versa

the microbial biomass for the Bt crop was (slightly) lower than for the non-Bt crops at flower time;

the microbial biomass for the Bt crop was (slightly) higher than for the non-Bt crops at harvest time;

h. data does not support the hypothesis as there is little difference between biomass found in the soil (surrounding) roots (of the Bt and non-Bt) at either time;

data does not support the hypothesis as there is a slightly positive effect at harvest;

data supports hypothesis as there is a slightly negative effect at flowering;

i (i). helix / alpha helix

i (ii). hydrogen bonds;

between the turns of the helix (rather than between R-groups);

bonds between carboxyl and NH groups/C-O---H-N;

i (iii). non-polar amino acids/R-groups;

(inner part of phospholipid) bilayer is hydrophobic/non-polar;

# Examiners report

a. In comparison to similar questions in previous years, candidates were relatively successful in answering this question. Where candidates did not answer correctly, it was due to a lack of ability to calculate percent difference rather than a problem with interpreting the data.

b. Most candidates scored at least one mark. A common error was to interpret the results without comparison to the control.

c. Most candidates calculated the mean masses correctly and included the correct units.

d. Most candidates scored at least one mark. A common error was to focus on the difference between male and female rats rather than the food source and to not make reference to growth.

e. Most candidates gained the mark, but some simply repeated their answer to (d). The command term "comment" requires candidates to give a judgment. Commonly, candidates mistakenly described the data in response to this command term.

g. Most candidates gained both the marks by recognizing the difference between harvest and flowering. Like answer (f), word choice affected performance with candidates referring to the biomass of flowers for example rather than biomass of soil microbes.

h. Many candidates scored both marks. A common error was to answer without reference to the hypothesis.

i (i). Many candidates identified the alpha helix, though a surprising number referred to the double helix.

i (ii) Most candidates identified hydrogen bonds as stabilizing the structure but very few could identify the parts of the molecule that were connected by H-bonds.

i (iii) Only a minority of candidates recognized the importance of the hydrophobic nature of membrane proteins.