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]

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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.

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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]

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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]

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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]

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]

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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]

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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]

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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]

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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]

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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]

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]

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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]

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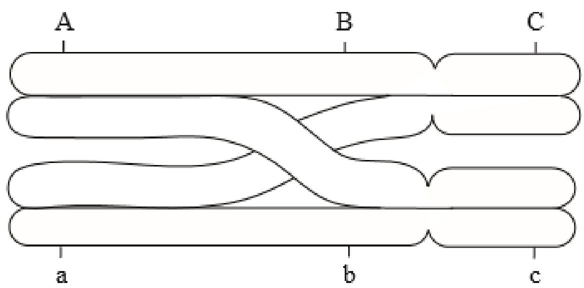
a. Predict the genotypic and phenotypic ratios of the possible offspring of a male hemophiliac and a female carrier using suitable symbols for the alleles in a Punnett grid. [3]

Genotypic ratio:

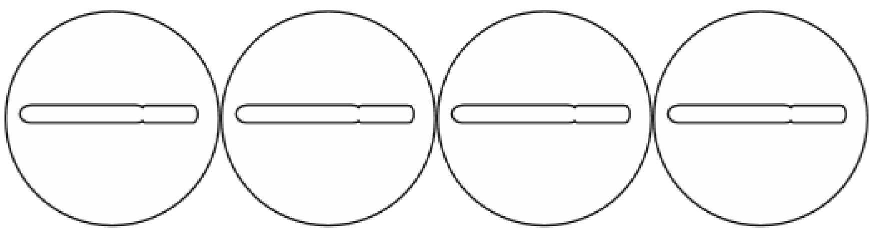
Phenotypic ratio:

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

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]

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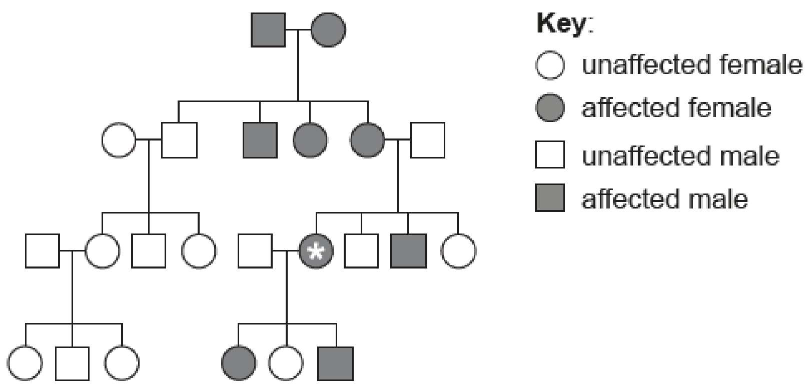
a. Explain chemiosmosis as it occurs in photophosphorylation. [8]

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]

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.

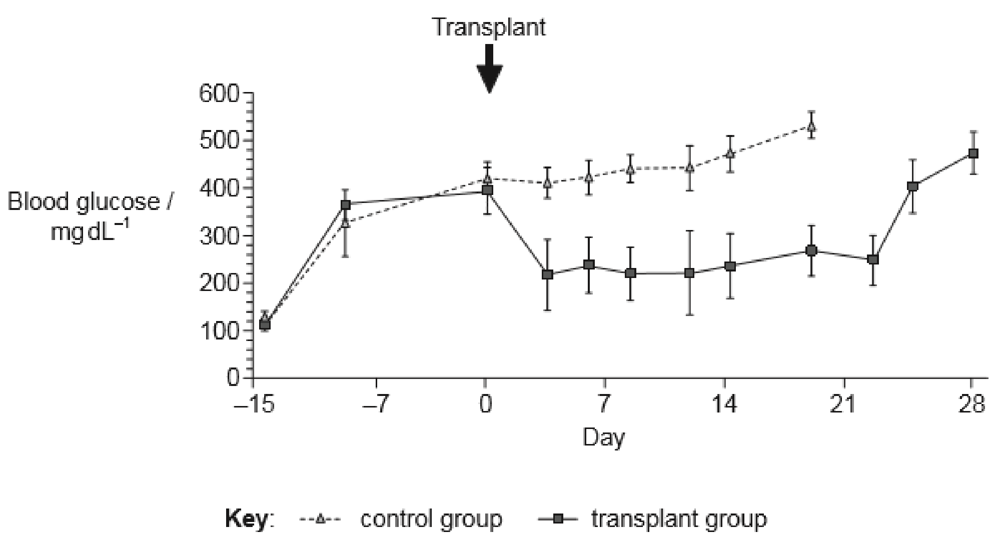


a. Using the pedigree chart, deduce the type of allele that causes hypophosphatemia. [2]

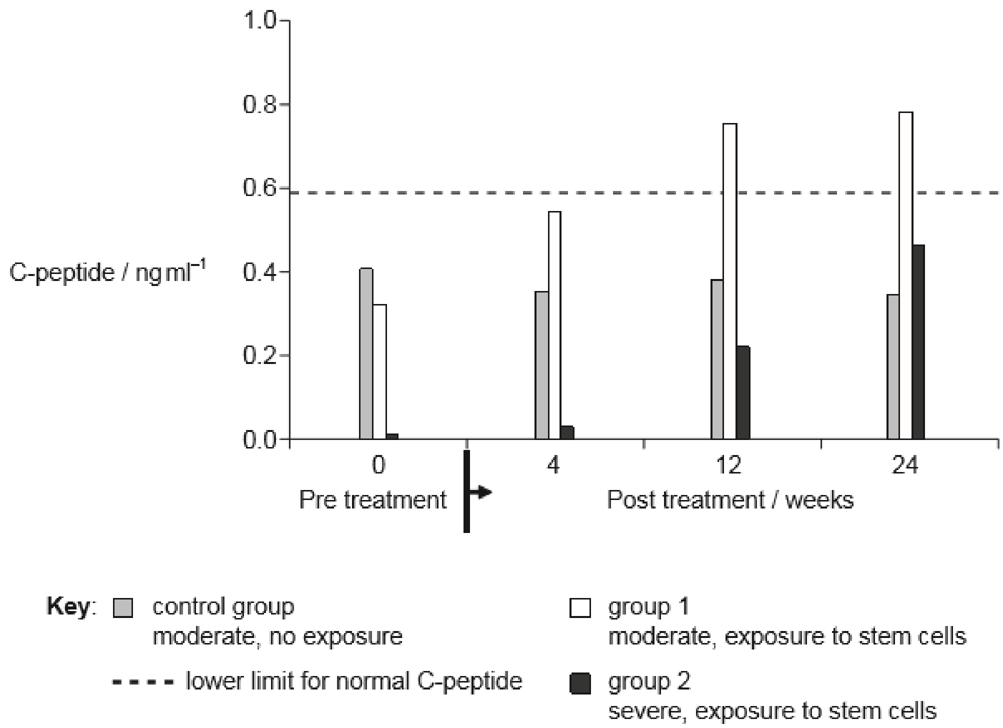
b. Identify the genotype of the individual marked with a star in the pedigree chart, using appropriate symbols for your answer. [1]

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.



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]

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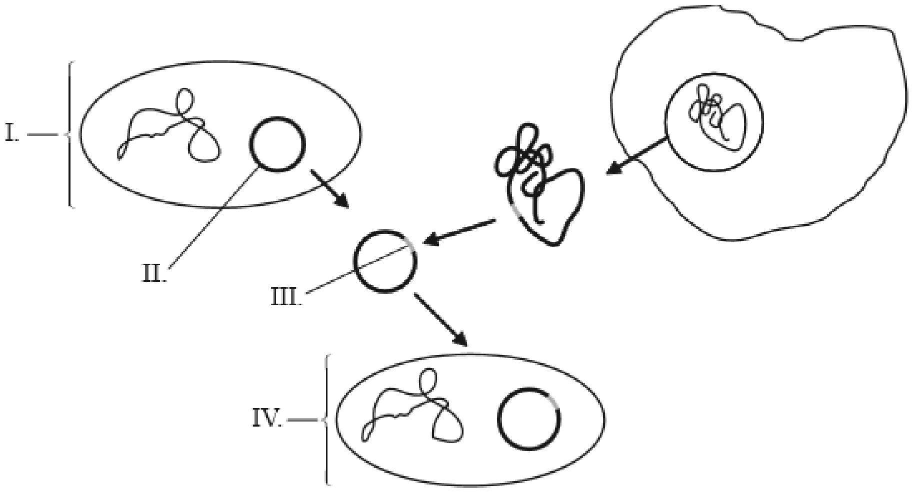
a. Explain why carriers of sex-linked (X-linked) genes must be heterozygous. [2]

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]

The diagram shows a human karyotype.

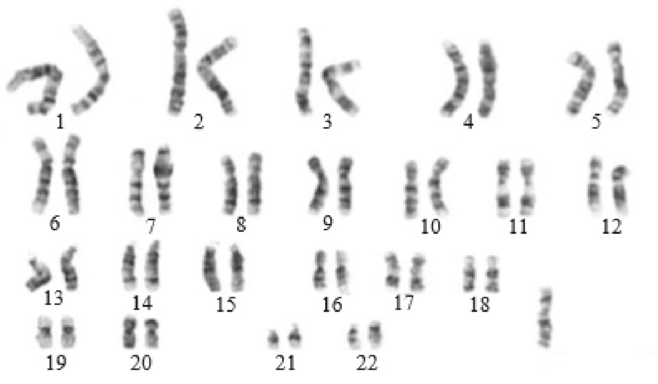
[Source: http://en.wikipedia.org/wiki/File:NHGRI_human_male_karyotype.png, courtesy of the National Human Genome Research Institute.]

a. Analyse this karyotype. [2]

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

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

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]

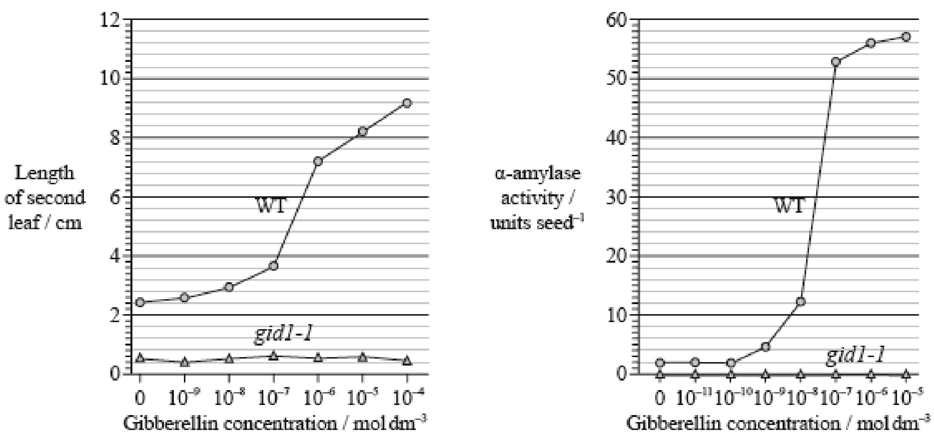
Identify the phenotypes of each part of the phenotypic ratio.

Ratio	Phenotypes
9	
3	
3	
1	

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

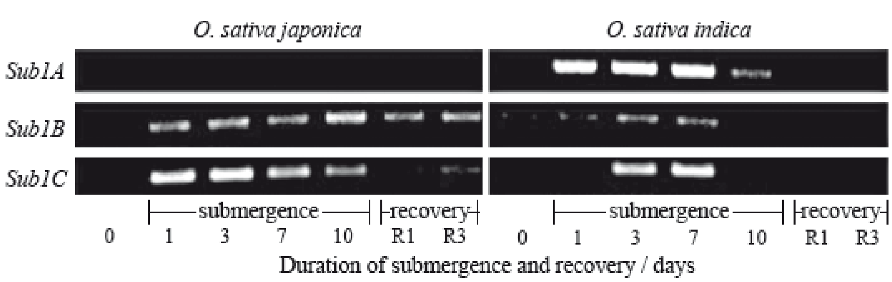
b (ii) Determine, with a reason, whether this karyotype shows that non-disjunction has occurred. [1]

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.

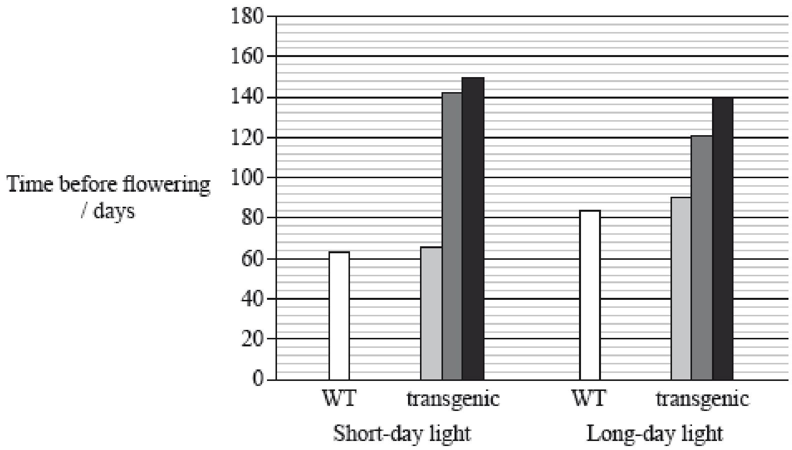


[Source: Adapted from "Sub1A is an ethylene-response-factor-like gene that confers submergence tolerance to rice" (2006) Kenong Xu, Xia Xu, Takeshi Fukao, Patrick Canlas, Reycel Maghirang-Rodriguez et al. Nature, 442, pp. 705—708. Adapted by permission from Macmillan Publishers Ltd (c) 2006.]

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]

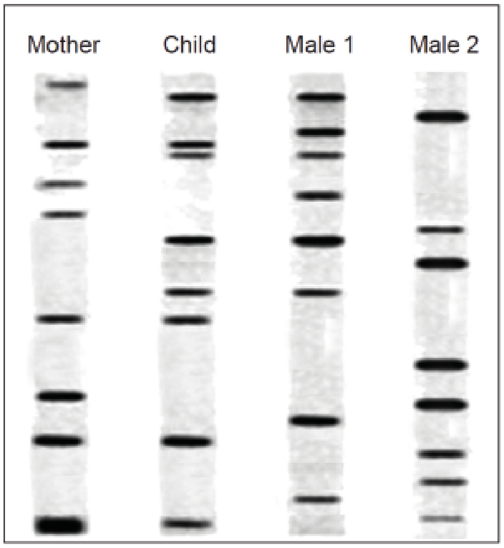
e(i). State the overall effect of overexpression of the *OsGI* gene in plants treated with short-day light. [1]

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]

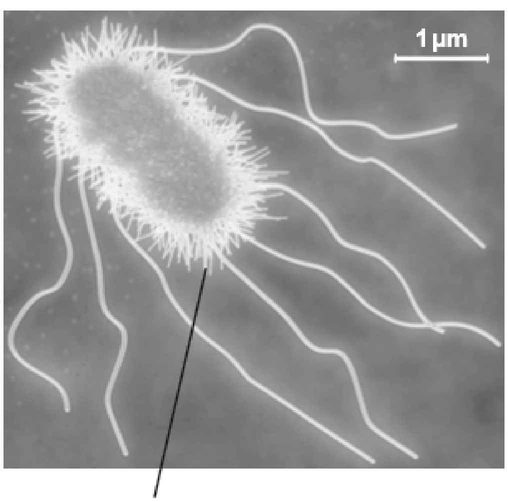
The image shows data collected in order to determine the paternity of a child.



[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]



. . . .

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

DNA ligase

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

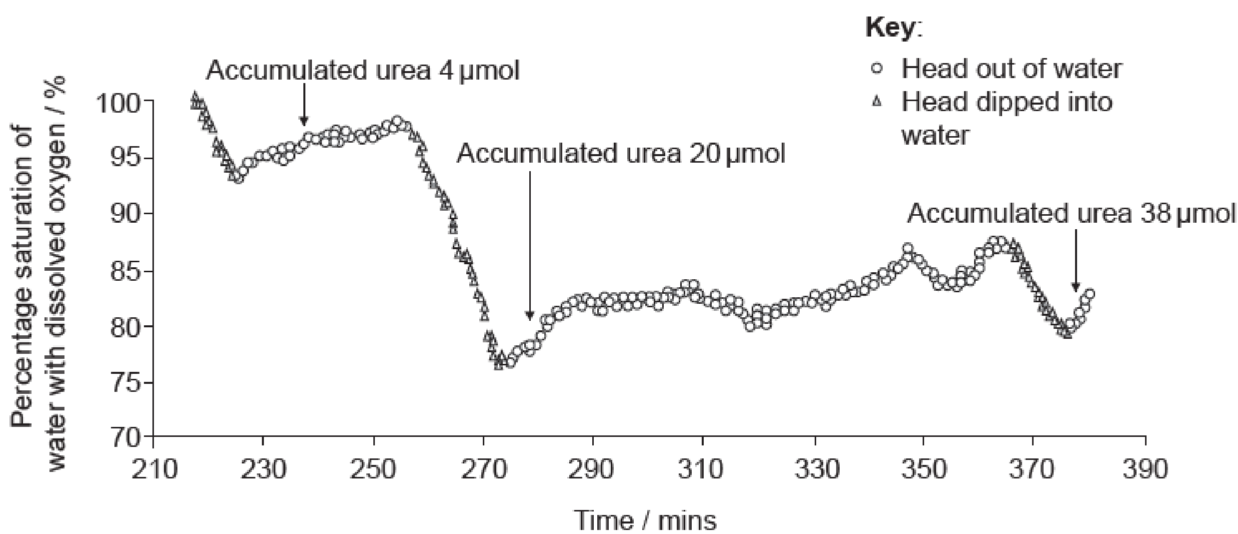
These turtles have fully developed lungs and kidneys, however, many microvilli have been discovered in the mouth of *P. sinensis*. A study was undertaken to test the hypothesis that oxygen uptake and urea excretion can simultaneously occur in the mouth.

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

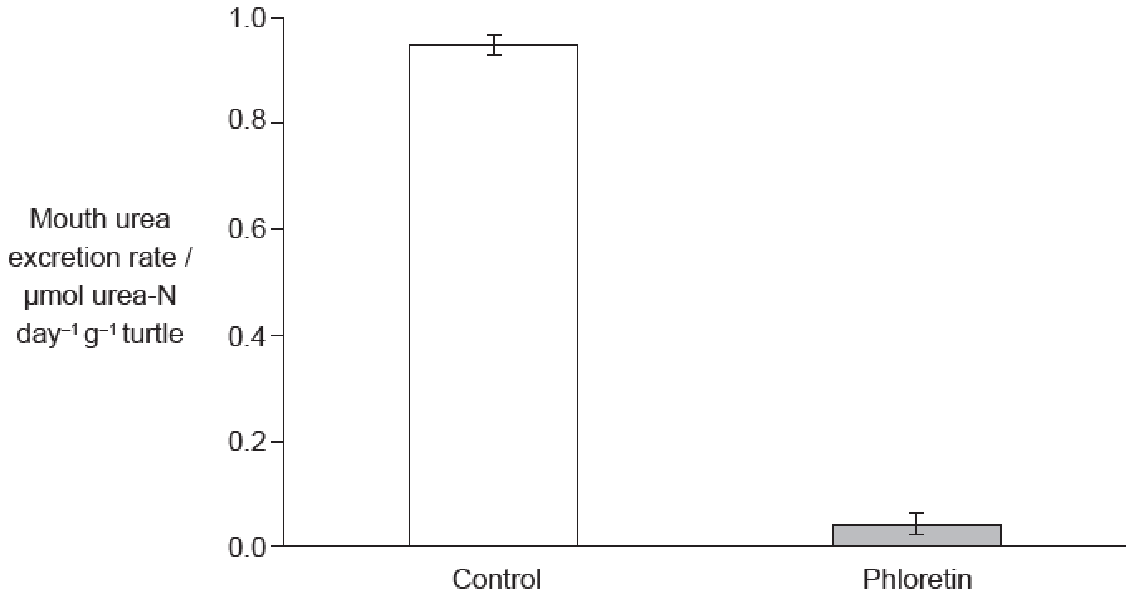
[Source: Reproduced with permission, Y. Ip *et al*. (2012) *The Journal of Experimental Biology*, 215, pages 3723—3733. jeb.biologists.org. doi: 10.1242/jeb.068916]

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.

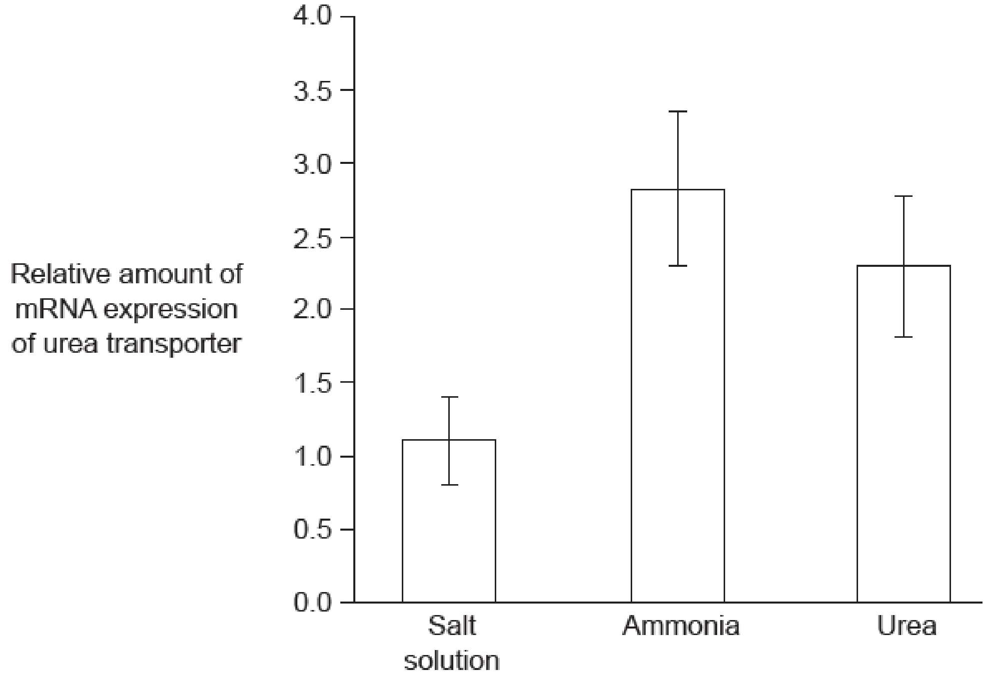


[Source: Reproduced with permission from Y. Ip *et al*. (2012) *The Journal of Experimental Biology*, 215, pages 3723–3733. jeb.biologists.org.]

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]

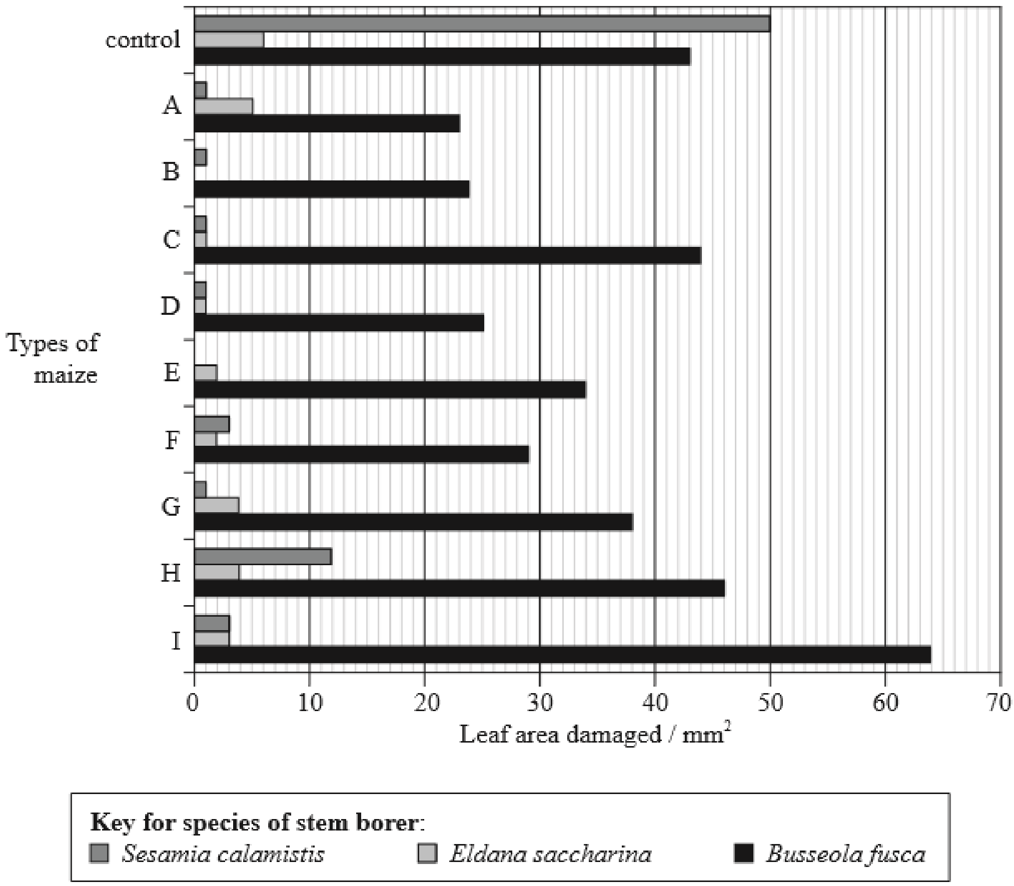
f.i. Identify which of these turtle groups represent the control, giving a reason for your answer. [1]

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]

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.

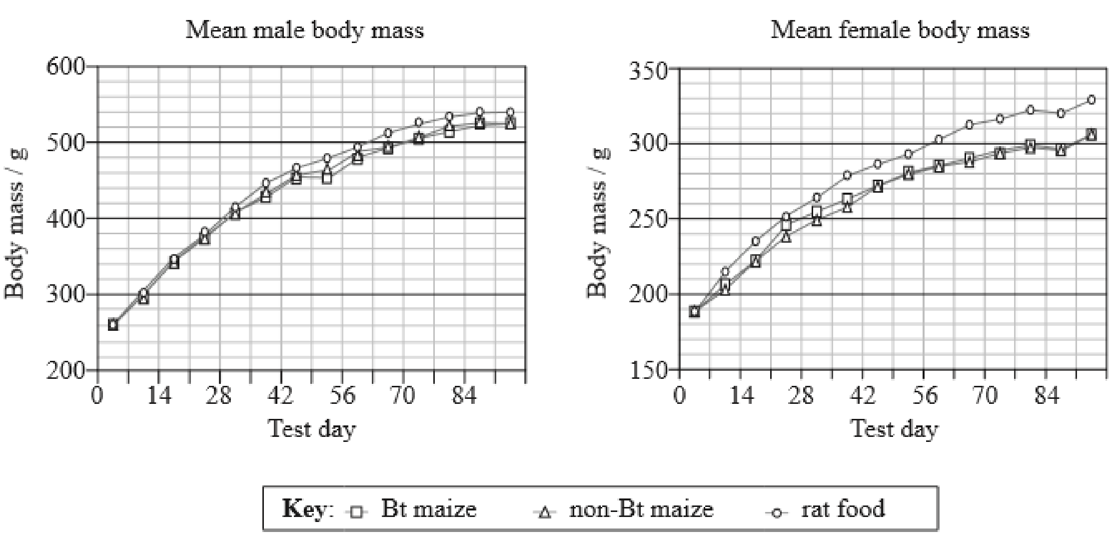


[Source: adapted from S Mugo, *et al.*, (2005), *African Journal of Biotechnology*, **4** (13), pages 1490–1504]

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]

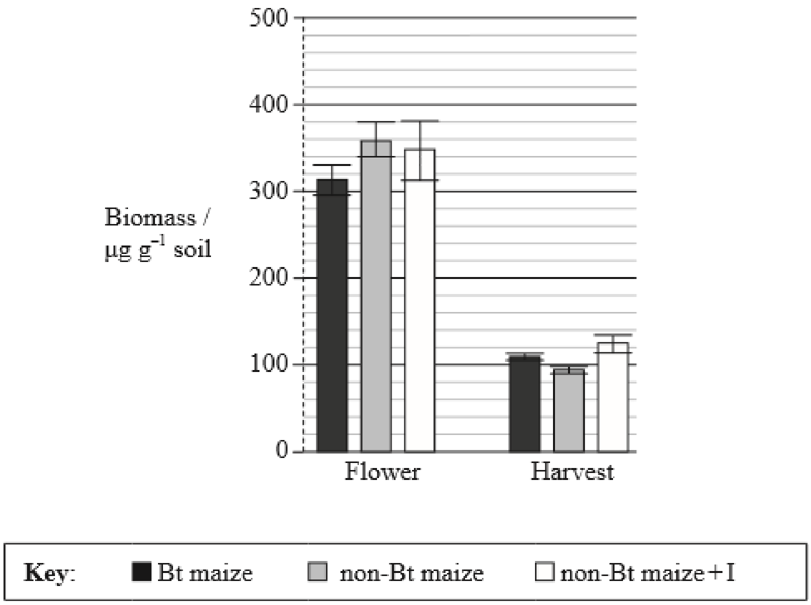
Studies have shown that Bt proteins are released by plant roots and remain in the soil. One study looked at the biomass of microorganisms in soil surrounding the roots of:

• Bt maize

• non-Bt maize

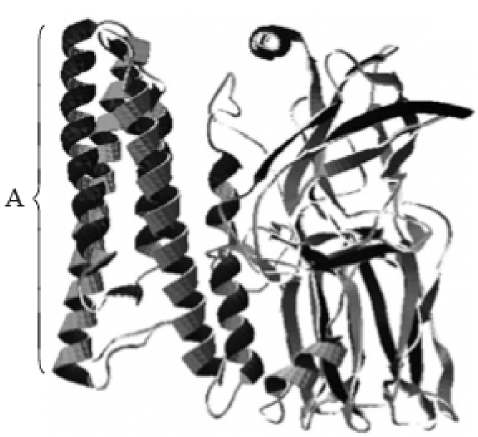
• non-Bt maize with an insecticide (I).

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]