James Ruse Agricultural High School

2004 TRIAL HIGHER SCHOOL CERTIFICATE EXAMINATION

BIOLOGY MARKING GUIDELINES

PART A

| Mult | tiple (| Choic | e: Qu | estio | ns 1-1 | 15 (1 | mark | each |) Tota | al mar | ks: 1 | 5 mar | KS | | | |
|------|---------|-------|-------|-------|--------|-------|------|------|--------|--------|-------|-------|----|----|----|--|
| | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9* | 10 | 11 | 12 | 13 | 14 | 15 | |
| (A) | Х | | | | | | Х | Х | Х | | | | Х | | | |
| (B) | | Х | Х | | | | | | Х | | | | | | | |
| (C) | | | | Х | Х | Х | | | | | | | | | | |
| (D) | | | | | | | | | | Χ | Χ | Х | | X | X | |

* pulmonary and systemic circulations not stipulated hence either option correct.

PART B

| PART B | |
|---|-------|
| Total marks: 60 marks | |
| 16. | Marks |
| Discuss the use of antibiotics in treating disease. <i>Discuss: identify issues and provide points for and/or against</i>. Negative: Variation exists in disease-causing bacteria; some survive a certain antibiotic while the antibiotic destroys some. Thus antibiotics are not 100% effective. The bacteria that survive the antibiotic will thrive due to lack of competition. The excessive use of antibiotics allows the proliferation of antibiotic-resistant bacteria. Not completing course of antibiotics allows bacteria to flourish. | 1 |
| • Positive: Antibiotics are effective in treating specific bacterial infections e.g., throat infection. Non-invasive, quick acting, specificity. | 1 |
| • Elaborations (two) of either the positive or negative point. E.g., "The <i>excessive</i> use of antibiotics allows the proliferation of antibiotic-resistant bacteria." | 1 |
| Or, description of how antibiotics function: "chemicals that destroy/inhibit bacterial cells" | I |

| 17. (a) | Marks |
|---|-------|
| State one early hypothesis for the cause of malaria. | 1 |
| • Romans and ancient Greeks suspected that stagnant water (marsh water), or the | |
| gases associated with this water, caused malaria. Another hypothesis was that insects | |
| caused malaria. Pathogen in water. | |
| | |

| 17. (b) | Marks |
|--|-------|
| Describe one piece of evidence that led to the discarding of the hypothesis you stated | |
| in (a). Describe: provide characteristics and features. | 1 |
| • An early experiment to test the hypothesis that malaria is caused by drinking | |
| marsh water involved some Italian volunteers to consume the marsh water, or have it | |
| sprayed into their noses. These volunteers did not develop malaria | |

| 17. (c) | Marks |
|--|-------|
| Describe the evidence that proved the cause of malaria. | |
| • In 1880, Laveran, using a microscope slides preparations, observed certain forms | 1 |
| of the protozoan, <i>Plasmodium</i> , in blood samples of malaria patients. Further work | |
| proved that malaria was caused by <i>Plasmodium</i> . Or, in the 1890s, Ronald Ross | |
| analysed the Anopheles species of mosquito via dissection and discovered that | |
| Plasmodium is taken into the mosquito when it sucks the blood of people containing | |
| Plasmodium, that is, malaria sufferers. | |
| | |
| 17. (d) | Marks |
| | |

| 17. (u) | | Wiai KS |
|---|------|---------|
| State one way of preventing malaria. | | |
| • Eradicate mosquito larvae in water bodies via the introduction of fish that fee | d on | |
| mosquito larvae; Use of insecticides; Use of drugs to destroy certain forms of | | |
| Plasmodium; Use of protective nets and clothing. | | |

| 17. (e) | Marks |
|---|-------|
| Explain how the method you describe in (e) works. | |
| • The strategy of eradicating mosquito larvae via the introduction of fish that feed on | 1 |
| mosquito larvae is aimed at destroying the vector, the Anopheles species of | |
| mosquito. | |

| 18. | Marks |
|---|-------|
| Explain why the immune response is suppressed in organ transplant patients. | |
| • The immune system is suppressed to minimise rejection of the organ. | 1 |
| • The organ being transplanted is likely to have non-identical MHCs (major | |
| histocompatibility complexes), which makes the MHCs antigenic and thus triggering | 1 |
| the immune response. The immune response will destroy the organ. | |
| | |
| 19. | Marks |
| State the name of a disease that results from an imbalance of microflora in humans. | |
| • Candidiasis (thrush). | 1 |

| 19. (b) | Marks |
|--|--------|
| Explain how the disease you stated in (a) occurs. Explain: relate cause and effect | t. |
| • Candidiasis (thrush) is caused by a fungus, <i>Candida albicans</i> . | 1 |
| • C. albicans and other microbes exist in humans but when an imbalance occurs, | |
| such as when using an antibiotic to treat some other disease, C. albicans can incr | ease 1 |
| in number. (Other reasons for imbalance: diabetes, immune suppression, steroids | , |
| oral contraceptives, and pregnancy.) | |
| | |

| 20 (a) | Marks |
|--|-------|
| Describe one other way in which water can be treated so that it is suitable for | |
| drinking. | |
| • Water purification – large scale: Step 1: filter water to remove undissolved solids; | |
| Step 2: add a flocculating agent to trap small particles and bacteria, filter this away; | 1 |
| Step 3: add chlorine to water in order to destroy bacteria and any other organism. | |
| | |

| 20. (b) | Marks |
|---|-------|
| Explain how the method you described in (a) reduces the risk of infection by | |
| organisms such as bacteria, Giardia and Cryptosporidium. | 1 |
| • The flocculating agent in Step 2 forms a thick gelatinous mass that traps bacteria, | |
| protozoans and small particles. This mass is filtered from the water thus removing | |
| the bacteria and protozoans. | |
| • Chlorine is an oxidising agent, killing bacteria and protozoans. | 1 |
| • "Filters", "Traps" | |

| 21. (a) | Marks |
|--|-------|
| Outline a process used to produce a transgenic species. Outline: sketch in general | |
| terms; indicate the main features of. | |
| Animals: | |
| • This method involves the direct microinjection of a chosen gene construct (a single | 1 |
| gene or a combination of genes) from a different species, into the pronucleus of a | |
| fertilized ovum. | |
| • The DNA construct (usually about 100 to 200 copies in 2 pl of buffer) is | |
| introduced by microinjection through a fine glass needle into the male pronucleus - | |
| the nucleus provided by the sperm before fusion with the nucleus of the egg. The | 1 |
| diameter of the egg is 70 μ m and that of the glass needle is 0.75 μ m; the | |
| experimenter performs the manipulations with a binocular microscope at a | |
| magnification of 200x. | |
| • The insertion of DNA is, however, a random process, and there is a high | |
| probability that the introduced gene will not insert itself into a site on the host DNA | 1 |
| that will permit its expression. The manipulated fertilized ovum is transferred into | |
| the oviduct of a recipient female, or foster mother that has been induced to act as a | |
| recipient by mating with a vasectomized male. | |
| | Or, |
| Plants: | |
| • The transgene is spliced into an Agrobacterium's plasmid. | 1 |
| • The Agrobacterium then inserts the plasmid into a plant cell's chromosomal DNA. | 1 |
| • The targeted plant cell is then treated so that it will divide and in so doing replicate | 1 |
| the transgenic DNA into its daughter cells. | |
| | |
| | 361 |

| 21. (b) | Marks |
|---|-------|
| Give one reason for producing the transgenic species you described in (a). | |
| • Usually, the new piece of DNA contains a gene that codes for a specific protein. | |
| As a result, transgenic organisms usually gain a new function or trait. For example, | |
| crops have been generated that have a new gene that provides a resistance to insects, | 1 |
| animals have been generated that produce proteins in their milk that can be | |
| harvested and used to treat life-threatening ailments, and some bacteria are able to | |
| safely break down toxic waste. | |

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|--------------|--------------|
| \sim | h |
| \mathbf{X} | |
| | |

| 22. | Marks |
|---|-------|
| "The evolution of organisms can be affected by changes to the environment and the | |
| use of modern reproductive technologies." Discuss this statement. Discuss: identify | |
| issues and provide points for and/or against. | |
| • The theory of organic <i>evolution</i> (biological evolution) contends that all living | |
| organisms arose in the course of history from earlier forms. Usually, many groups of | |
| organisms have a common ancestor. As the earth's environments altered over a long | |
| period of time, organisms gradually change, or evolved, into other types of | |
| organisms. | |
| Changes to the environment: | |
| • Organisms that have suitable characteristics to survive a particular environment | |
| may survive, successfully reproduce and thus their offspring will inherit the | |
| appropriate characteristics. In this way the species will evolve as the environments it | |
| encounters change or vary. Since these processes are random evolution will | |
| continue. | |
| • E.g., Changes in the fossil record of horses are correlated with changes in the | |
| environment. During the existence of the little <i>Hyracotherium</i> , the land was marshy | |
| and the main vegetation was leaves; the teeth of <i>Hyracotherium</i> were adapted for | |
| browsing. In the Miocene, the grasslands began to spread; horses whose teeth had | |
| become adapted to grinding grasses (which had much coarser blades than dicots) | |
| survived, whereas those who remained browsers did not. The placement of the eye | |
| higher in the head may have facilitated watching for predators while grazing. | |
| • E.g., Biston betularia, the peppered moth. Against the light-coloured background | |
| of lichen-covered trees and rocks, the light colouring of this moth made it practically | |
| invisible, concealing it from predatory birds. Virtually no black-coloured <i>B</i> . | |
| <i>betularia</i> existed. With the increasing industralisation of England, smoke particles | |
| began to pollute the forests, killing lichens and leaving tree trunks bare. During this | |
| period, more and more black <i>B. betularia</i> were found. E.g., kangaroos have changed | |
| due to Australia drying out. E.g., convergent or divergent evolution processes. | |
| Modern reproductive technologies: | |
| • These include processes such as artificial insemination, artificial pollination and | |
| cloning. All these are different to environmental influences since man determines | |
| what organisms will breed. These selective breeding processes may reduce the | |
| genetic diversity of a species and thus reduce the potential of the species to evolve. | |
| • E.g., artificial insemination is a technique involving the artificial injection of | |
| sperm-containing semen from a male into a female to cause pregnancy. Artificial | |
| insemination is often used in animals to multiply the possible offspring of a <i>prized</i> | |
| <i>animal</i> and for the breeding of endangered species. Advantages: genetically superior | |
| offspring; reduced disease; reduced expenses (feed, facilities, damage, vet, etc.); | |
| reduced risk of injury. | |
| Disadvantages: acquired skill; management intensive; cost of technician or | |
| equipment; less genetic variation due to the artificial selection of traits for breeding. | |
| (1): statement about the role of the environment in affecting evolution of species | |
| (1): environment elaboration mark ("because") or example | |
| (1): statement about the role of one modern reproductive technology in affecting the | |
| evolution of species ("reduces the genetic diversity") | |
| (1): modern reproductive technology elaboration mark ("because") or example | |
| (1): adequate demonstration of the concept of evolution: gradual change. | |
| (1): any additional example or elaboration given | |
| (1): stance statement provided "I support" or "I do not agree with the statement" | |
| | |

| 23. | Marks |
|---|-------|
| Construct a flow chart that shows the changes in DNA sequences can result in | |
| changes in cell activity. | |
| • Changing adenine to thymine in the gene for one of the chains of haemoglobin | |
| results in a haemoglobin chain containing the amino acid valine instead glutamic | |
| acid (normal). This results in the haemoglobin molecules forming end-to-end | |
| associations and the red blood cells become a sickle-shape. | |
| | |
| (1): flowchart provided (arrows and sequencing of information). | |
| (1): correct DNA structure shown (bases) or "different mRNA made". | |
| (1): demonstration of how a change in DNA sequence leads to a change in the amino | |
| acid (probable change). | |
| (1): demonstration of how a change in amino acid sequence can alter a protein and | |
| hence affect the cell's activity. | |
| | |

| 24. | Marks |
|---|-------|
| Explain one way by which the environment may affect the phenotype (expression of | |
| a gene) of an organism. | |
| • One of the main reasons for the effects of temperature is that many enzymes are | |
| thermolabile, that is they do not function at high temperatures, even within the | |
| normal physiological range. | |
| • Siamese cats have their coat colour determined by an enzyme that functions | |
| adequately in cooler, peripheral areas of the body, such as the ears, nose, paws, and | |
| the tip of the tail, but becomes inactive in the warmer areas of the body. In the | |
| northern seal, the newborns are white, as a result of their developing at a warm | |
| (internal) temperature. The newborns cannot swim and so are restricted to the ice | |
| where their white coats provide them with colour camouflage. By the time they can | |
| swim, their coats have turned brown (new fur is actually grown) and so blend with | |
| the dark artic waters. Many lizards change their colour from light colours in the hot | |
| summer to darker colours in the cooler winter months. | |
| | |
| (1): reason as to how the environment affects the phenotype: "enzymes affected by | |
| temperature". | |
| (1): relevant example given. | |
| | |

| 25. (a) | Marks |
|--|-------|
| Give an example of hybridisation within a species. | |
| • Various strains of corn. | 1 |
| | |

| 25. (b) | Marks |
|---|-------|
| Explain the purpose of the hybridisation in the example you gave in (a). | |
| • Improves certain qualities of the plant such as increased resistance to diseases; | 1 |
| ability to grow in poor soils; desired colours, better growth, etc. | |
| • The corn strains crossed are heterozygous for many of the genes responsible for | |
| growth etc. This leads to an increase in the desirable combination of alleles for | 1 |
| various traits. Hybrid vigour occurs. | |

| npare the type of nitrogenous wastes in terrestrial mammals and Australian cts. Compare: show how things are different or similar. errestrial mammals excrete nitrogenous waste products largely as urea, which ammonia or uric acid comes principally from the breakdown of amino acids. a, unlike uric acid, must be dissolved in water for excretion. sects eliminate nitrogenous wastes in the form of uric acid or uric acid salts, ch can be excreted as crystals. (b) lain the differences in the type of nitrogenous wastes produced by terrestrial nmals and Australian insects. e excretory malpighian tubules of insects are highly efficient at removing the nitrogenous waste ucts from blood and to function in osmoregulation. Insects have two to several hundred rightan tubules projecting from the junction of the midgut and hindgut. They lie bathed in clear (haemolymph) of the insect's body cavity. K ⁺ and Na ⁺ are actively pumped into the tubule, e water uric acid salts, and several other substances follow by passive transport. Water and some are reabsorbed in the hindgut, while uric acid precipitates out as a paste and is passed out of the along with the faecal material. The ability to conserve water by excreting solid uric acid has led insects to colonise arid environments. e initial product in the deamination of amino acids in the mammal liver is ammonia, but this is is mely toxic in mammals and is converted to urea, the main nitrogenous waste product. In |
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| mery loxic in mammals and is converted to mea, the main nitrogenous waste broduct. In |
| mals and other vertebrates, the organs of nitrogenous excretion are the kidneys. These contain |
| rons, the structure that filters the blood of urea, excess salts and water, amongst other excretory |
| tances. |
| Insects have malpighian tubules as the organs of excretion. |
| Insects excrete uric acid since this substance does not need to be dissolved in |
| er in order to be excreted from the insect. |
| Mammals use nephrons in kidneys to excrete nitrogenous waste. |
| Mammals excrete nitrogenous waste in the form of urea since they have water |
| lable to dissolve and transport urea. |
| |
| |

| 27. (a) | Marks |
|---|-------|
| Draw a labelled diagram of a mammalian nephron. | |
| (1): correct anatomy. | |
| (1): two parts of the nephron labelled correctly. | |
| | |

| 27. (b) | Marks |
|--|-------|
| Explain how the process of passive transport is involved in the nephron's ability to | |
| regulate body fluid composition. | |
| • Filtration occurs at Bowman's capsule. | 1 |
| • This process allows the diffusion of water, salts, urea from the blood. | 1 |
| (1): correct location | |
| (1): correct description of processes occurring for the location. | |

| 27. (c) | Marks |
|---|-------|
| Explain how the process of active transport is involved in the nephron's ability to | |
| regulate body fluid composition. | |
| • The walls of the proximal tubule and the descending branch of the loop of Henle | |
| are freely permeable to water and contain carrier proteins that pump Na ⁺ and K ⁺ out | 1 |
| of the tubule by active transport, with the negative ions such as Cl ⁻ and HCO ₃ ⁻ | |
| following passively. | |
| • As the ions move into the plasma –by either an active or a passive process –an | 1 |
| osmotic gradient is generated across the tubule wall with the consequence that water | |
| flows passively into the capillaries. | |
| | |
| | |
| 28. (a) | Marks |
| State the name of the plant tissue shown in diagram 1 and describe one theory about | |
| the movement of materials in it. | |
| • Phloem. | |
| • Translocation may occur via the pressure-flow hypothesis (accounting for the | |
| source-to-sink pattern). This means that the sugar of the leaves (the source) attracts | |
| water, the pressure of the water consequently causing it to flow to other tissues | |
| lacking in sugar (the sink). This flow of water from source to sink drags sugar | |
| molecules. | |
| (1): sugar attracts water. | |
| (1): water pressure results in flow of sugar solution to other parts of the plant. | |
| If only "source-to-sink" is stated then a maximum of 1 was awarded | |

If only "source-to-sink" is stated then a maximum of 1 was awarded.

| 28. (b) | Marks |
|---|-------|
| State the name of the plant tissue shown in diagram 2 and describe one theory about | |
| the movement of materials in it. | |
| • Xylem. | |
| • Evaporation (transpiration)-cohesion-tension theory accounts for the movement of | |
| water, and thus ions, in the xylem. Water molecules attract each other and are also | |
| attracted to the walls of the xylem. As water evaporates from the leaf, each molecule | |
| of water drags another upwards. This stream carries ions as well. | |
| (1): water attracts water. | |
| (1): water movement occurs a result of the evaporation of water from the leaf or | |
| other thus tugging the solution to other parts of the plant. | |

| 29. (a) | Marks |
|--|-------|
| Using a specific example, describe how the theory of evolution is supported by | |
| biochemical studies. | |
| • Many types of organisms have the same type of molecules in their cells that | |
| perform the same function. This similarity suggests that all living things have a | |
| common ancestry and that these molecules perform essential functions in cells. Such | |
| molecules include proteins such as the enzymes, those involved in the formation of cell membranes, DNA, and RNA. | |
| • Evolutionary relationships between species can be characterised by comparing the | |
| amino acid sequences of their respective plasma proteins and enzymes. Cytochrome | |
| C, a respiratory chain enzyme, has been studied extensively in this regard, and many | |
| species have been compared. The fact that certain crucial amino acids are | |
| consistently present in the same sites in all species suggests that the different | |
| cytochrome sequences arose from through mutations from an ancestral molecule. | |
| Also from the amino acid data, it is possible to calculate the minimum number of | |
| nucleotide differences that would be necessary to account for the amino acid | |
| differences. Presumably, the greater the number of amino acid (or nucleotide) | |
| differences between any two organisms, the more distant their evolutionary | |
| relationship; conversely, the smaller the number of differences, the closer their | |
| relationship. The results confirm well, but not perfectly, with phylogenies | |
| constructed by traditional methods. Chimps and humans have a very small number | |
| of amino acid differences in their cytochrome C enzyme compared to other primates | |
| suggesting that humans and chimps diverged from a common ancestral type after | |
| gorillas. | |
| (1): relevant example of a biochemical feature stated. | |
| (1): description of how the biochemical feature supports the theory of evolution. | |

| 29. (b). | Marks |
|---|-------|
| Using a specific example, describe how the theory of evolution is supported by | |
| palaeontological studies. | |
| • Palaeontological evidence refers to the fossil record and this provides support for | |
| the theory of evolution. For example, the modern horse, Equus, has had several | |
| ancestors, as found in the fossil record. | |
| • These horse fossils show gradual changes over the course of some 65 million | |
| years. | |
| • The earliest horse fossil for instance, Hyracotherium, had a three-toe foot structure | |
| whilst the younger horse fossil, Miohippus, had evolved to have mainly a single-toe | |
| foot with two side bones; the Equus has a single toe foot. These changes are thought | |
| to be the result of the horse evolving in an environment that was changing from | |
| marsh-like to one that was dry and of hard ground. | |
| (1): relevant example of a fossil stated. | |
| (1): description of how the fossil record supports the theory of evolution. | |
| (1): elaboration of the description with respect to specific features of the fossils | |
| involved. | |

Question 33: Genetics The code is broken?

| 33. (a) | Marks |
|---|-------|
| Explain how DNA fingerprinting can be used for paternity testing. | |
| • Paternity testing: determining if a man is the father of a child. DNA fingerprinting | |
| involves the analysis of minisatellites or Variable Number Tandem Repeats | 1 |
| (VNTRs) (1) of DNA. These regions of DNA are short, highly repeated sequences | |
| of base pairs that are found throughout a person's chromosomes. Because they are | |
| derived from the chromosomes donated from both parents (1), minisatellites or | 1 |
| VNTRs can be used to identify if two pieces of DNA are linked via heredity or are | |
| from the same person. A child, for instance, will inherit minisatellites or VNTRs | |
| from the mother or father or a combination of their sequences. By analysing the | |
| DNA of the child and the suspected male parent, specifically the minisatellites or | |
| VNTRs, via the use of gel electrophoresis and <u>Southern Blotting techniques</u> , (1) a | 1 |
| comparison can be made in terms of the number of similar minisatellites or VNTRs | |
| and the pattern formed on the gel. The more similar (1) the pattern between a male's | 1 |
| and the child's DNA the more likely that male is the father of the child. | |

| 33. (b) | Marks |
|---|-------|
| Discuss the benefits and limitations of the Human Genome Project. | |
| • The Human Genome Project aims to construct detailed physical and genetic maps | 1 |
| of the human genome. | |
| Negative: However, knowing the location and DNA sequence of genes has | |
| limitations. For example, polygenic traits are influenced by more than one gene. It is | |
| difficult to measure the interactive effects of various genes on a trait (1) so if a | |
| person has a defective gene that is involved for a trait, it does not necessarily mean | |
| that the person will suffer from a disease associated with that trait; other genes may | |
| have greater influence than the defective gene on the trait. Other examples of | |
| limitations: having a defective gene(s) for a trait and not having the appropriate | |
| treatment for the condition that will develop; will the data be accessible to insurance | |
| companies or potential employers, who in turn will use it to deny coverage or | |
| employment? | |
| Positive: The Human Genome Project will develop tools to identify the genes | |
| involved in both rare and common diseases. Such discoveries, in turn, are likely to | |
| bring improvements in the early detection and treatment of disease and new | |
| approaches to prevention. Once the molecular basis of a disease is revealed, | |
| scientists have a far better chance of defeating it. One approach is to design highly | |
| targeted drugs that act on the cause, not merely the symptoms, of disease. Another is | |
| to correct or replace the altered gene through gene therapy. Even before that, | |
| however, gene discovery can lead to predictive tests that can tell a person's | |
| likelihood of getting a disease long before symptoms appear. In some cases, | |
| preventive actions can then be undertaken that may avert the disease entirely or else | |
| detect it at its earliest stages, when treatment is more likely to be successful. | |
| (1): definition of the human genome project. | |
| (1): negative (limitation) aspect statement of the human genome project. | |
| (1): negative statement elaboration. | |
| (1): positive (benefit) aspect statement of the human genome project. | |
| (1): positive statement elaboration. | |

| 33. (c) (i) | Marks |
|--|-------|
| Give one example of a mutation that is a result of chromosomal rearrangements. | |
| • Down's syndrome. Or, deletion, duplication, inversion, translocation, trisomy. | 1 |
| | |

| 33. (c) (ii) | Marks |
|---|------------------|
| Explain how this mutation may occur. | |
| • Non-disjunction in meiosis results in a trisomy at position 21. | |
| | |
| 33. (d) | Marks |
| UV radiation (from the sun) is mutagenic, however DNA has the ab | oility to repair |
| itself when affected by UV radiation. | |
| Describe how this repair is accomplished in eucaryotes. | |

• Photoactivation is the simplest mode of DNA repair containing thymine dimers and has been demonstrated in eucaryotes and procaryotes.

• The photoactivating enzyme binds to the region of the DNA containing the dimer to form an enzyme-DNA chromophore that absorbs visible light (300 to 600nm) and then catalyses cleavage of the cyclobutyl pyrimidine dimer. As a result, the thymine residues are restored and the enzyme dissociates from the DNA.

(1): use of undamaged DNA strand as template.

(1): enzyme synthesises DNA (e.g., DNA polymerase)

| 33. (e) | Marks |
|---|-------|
| Explain the effects of the following type of genetic mutation on human health: | |
| Sickle cell anaemia. | |
| • Sickle cell anaemia is due to a base substitution in which a thymine is substituted | 1 |
| for an adenine. | |
| • This mutation causes a different amino acid to be used to form haemoglobin. | 1 |
| • This alteration causes haemoglobin to fold differently and the altered proteins | |
| connect end-to-end thus causing the red blood cell to become a sickle-shape. This | 1 |
| leads to anaemia-less oxygen carriage by the red blood cell. | |

| 33. (f) | Marks |
|---|-------|
| Discuss the research about the evolution of human haemoglobin genes and their | |
| actions. | 1 |
| • Transposable genetic elements are segments of DNA, including gene(s), which | |
| move to other chromosomes or to another place on the same chromosome. Human | |
| haemoglobin is made of four polypeptides – two alpha chains and two beta chains. | |
| The ancestral form of haemoglobin is thought to have had only one type of gene, | |
| which was found on homologous chromosomes. One of these genes underwent | |
| mutation to form into either the alpha or beta polypeptide gene. Thus two genes | |
| were formed. (1) (Similarities in the sequences of the alpha chain and beta chain | |
| genes indicate they had a common ancestral gene.) One of these genes then | |
| underwent transposition (conservative transposition), which caused it to be | |
| transferred to another chromosome. (1) After this, these two genes underwent | |
| duplication and mutations to form into the various haemoglobin chain genes that | |
| exist today: foetus, adult and embryo. Each of these genes is expressed at some time | |
| in the development of humans. | |
| 1 | |
| • It can be <u>argued that the mutation that caused the formation of the different form</u> | |
| of gene responsible for the different polypeptide of haemoglobin (either the alpha or | |
| beta chains) has had more impact on the evolution of haemoglobin than | |
| transposition. Transposition results in the same gene being moved to another place | |
| in the cell (another chromosome). This gene will still be expressed. Consequently, | |
| the mutation event will have the biggest impact on haemoglobin. However, since the | |
| gene for one type of haemoglobin polypeptide is on another chromosome, it may be | |
| influenced by events that occur on that chromosome. | |
| | |
| (1): ancestral globin gene. | |
| (1): mutation occurred resulting in alpha chain and beta chain genes. | |
| (1): transposition of the alpha chain and beta chain genes. | |
| Or, (1): duplications and mutations of the alpha chain and beta chain genes. | |
| (1): different versions of these genes are expressed at different times in | |
| development. | |
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| 33. (g) | Marks |
|--|-------|
| Describe how recombinant DNA technology can be used to identify the position of a | |
| gene on a chromosome. | |
| • The chromosome that contains the gene of interest is obtained and cut into large | |
| fragments using a restriction enzyme. These fragments are cloned using recombinant | |
| DNA technology. These fragments are cut with two restriction enzymes and the | |
| fragments formed are cloned using recombinant DNA technology. The fragments | |
| from each restriction enzyme form two libraries (library 1 and library 2). | |
| | |
| Cloning of fragments using DNA technology: The fragments are cut from a human | |
| cell with a restriction enzyme and mixed with bacterial plasmids that have been cut | |
| by the same restriction enzyme. The plasmids have been removed from bacterial | |
| cells. Next, DNA ligase is added to the mixture so that the fragment combines with | |
| the plasmids to form the recombinant DNA. The plasmids, with the fragment, are | |
| reinserted into the bacterial cells, which are allowed to reproduce. The fragments are | |
| then extracted from the bacterial plasmids using a restriction enzyme. | > |
| | |
| • Radioactive DNA probe is prepared based on a certain part of the protein | |
| expressed from the gene (first 30 amino acid of protein are analysed and sequenced | |
| to work out the complimentary DNA probe sequence). | |
| • The DNA probe is mixed with the single-stranded DNA of the chromosome | |
| fragments being used. Both libraries are used. | |
| | |
| • The end of the fragment that had the DNA probe bound to it is then sequenced and | |
| a second radioactive DNA probe is made (using a 'gene machine') complimentary to | |
| this. DNA probe two is added to the fragments from the other library. | |
| • The above step is repeated until the desired physical map of the chromosome for | |
| the genes or DNA sequences of interest is made. | |
| | |
| (1): chromosome cut using two restriction enzymes. | |
| (1): cloning of fragments via recombinant DNA technology via use of same | |
| restriction enzyme used to splice plasmid and fragment. | |
| (1): DNA probe used based on sequence of protein from gene. | |
| (1): Fragment probe binds to is analysed and a second probe made for its end. | |
| (1): DNA probe two is added to the other library and the step above is repeated. | |
| Or, (1): gene machine makes DNA sequence of gene (assuming the protein is known | |
| and isolated). | |
| (1): complimentary DNA probe (radioactive) made. | |
| (1): radioactive probe added to single-stranded chromosome. | |
| (1): recombinant DNA technology is used to make multiple copies of chromosomal | |
| fragments. | |
| (1): use microscope and radiography to identify the chromosomal fragment that the | |
| radioactive probe binds to. | |
| | · |
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| \checkmark | |