

## LIFE SCIENCES: PAPER I

Time: 21/2 hours

150 marks

## PLEASE READ THE FOLLOWING INSTRUCTIONS CAREFULLY

- 1. This question paper consists of 12 pages and a yellow Answer Booklet of 7 pages (i vii). Please check that your question paper is complete. Detach the yellow Answer Booklet from the middle of the question paper.
- 2. This question paper consists of five questions.
- 3. Question 1 must be answered in the yellow Answer Booklet provided. Questions 2, 3, 4 and 5 must be answered in your Answer Book.
- 4. Read the questions carefully.
- 5. Number the answers exactly as the questions are numbered.
- 6. Use the total marks that can be awarded for each of Questions 1, 2, 3 and 4 as an indication of the detail required.
- 7. It is in your own interest to write legibly and to present your work neatly.
- 8. Please hand in this question paper.

2.1 Study the diagram below of a cell undergoing the reduction division phase of meiosis and answer the questions that follow.



## Diagram showing the first stages of meiosis

[Adapted from: <www.course1.winona.edu>]

2.1.1	(a)	Explain clearly the function of meiosis in the human body.	(2)
	(b)	How many cells would be formed from cell A by the end of the complete process of meiosis?	(1)
	(c)	What term (based on chromosome number) describes the original cell at A?	(1)
2.1.2	(a)	Name the biological process by which a chromosome duplicates before meiosis takes place.	(1)
	(b)	Write a brief description of this process you named in Question 2.1.2 (a) to explain how the chromosomes make duplicate chromatids as seen in diagram B.	(6)
2.1.3	Provide the correct biological terms for each of the following:		
	(a) (b)	pair of chromosomes at $\mathbf{X}$	
	(0) $(c)$	structure at <b>Z</b>	(3)

2.1.4 The diagram below is from a micrograph of part of a cell in the testis of a human male. It illustrates a process known as 'crossing over' as seen in cell B in the diagram on the previous page.

#### Diagram showing crossing over of chromosomes during meiosis



[Adapted from: <bioinfo.org.cn>]

- (a) Give a brief description of what happens during crossing over of chromatids.
- (b) What is the biological importance of the 'crossing over' of chromatids in the cells of the testis?
- 2.2 Scientists investigated the sperm production of the 'mink', a small mammal living in the Northern Hemisphere. They discovered that sperm production in male minks decreases during the cold winter months and increases in the warm spring and summer months during their breeding season.



The graph below illustrates some of the scientific findings.

## Graph showing the relationship between sperm and testosterone production in minks



[Adapted from: <www.sciencedirect.com>]

(4)

(3)

2.2.1	What evidence is provided by the graph to suggest that meiosis is affected by the changes in seasons?	(2)
2.2.2	Suggest and explain a possible reason for the differences in both sperm and testosterone production as shown by the graph.	(4)
2.2.3	Write a suitable hypothesis for this investigation.	(3) [ <b>30</b> ]

3.1 There are many factors which contribute to the process of evolution of organisms over time.

Over the last 30 years scientists have studied many organisms on the Galapagos Islands. One of these, the Medium Ground Finch, *Geospiza fortis (G. fortis)*, is a bird on one of the smaller islands, Daphne Major. This species of ground finch is a seedeater. During a study over an 18 month period, from June 1976 to the end of 1977, a severe drought occurred on this island. The drought resulted in a shortage of seeds, especially small seeds. To survive, the finches had to turn to large, hard seeds for food. Only birds with large beaks were able to crack and eat the large seeds. These large-beaked birds were able to survive the drought, while the small-beaked birds died. When the drought eased in 1978, the Medium Ground Finch population recovered and the average beak size was found to be greater than in 1976.

[Adapted from: Oxford University Press; 2006]

3.1.1	Explain clearly, using appropriate scientific terminology, how Darwin's theory of evolution would account for the average increase in beak size of <i>G. fortis</i> .			
3.1.2	(a)	State a suitable aim for the investigation which occurred from June 1976 to the end of 1977.	(2)	
	(b)	What is the independent variable in this investigation?	(1)	
	(c)	Could this Medium Ground Finch population that had a greater average beak size after the drought be termed a new species of finch? Give a scientific reason for your answer.	(2)	
	(d)	Do you think that the scientists could publish their findings on the effects of the drought on <i>G. fortis</i> on Daphne Major in a scientific journal? Explain your answer.	(3)	

3.2 The graph below illustrates the variation in beak length in a population of *G. fortis*. Use the graph and your own knowledge to answer the following questions:



[Adapted from: Oxford University Press; 2006]

3.2.1 Would beak length be considered an example of a trait displaying continuous or discontinuous variation? Use evidence from the graph to explain your answer.

(2)

(2)

(1)

(4)

- 3.2.2 Is beak length likely to be controlled by one or many genes? Give a reason for your answer.
- 3.2.3 (a) What was the longest beak length recorded in this sample of *G. fortis?* 
  - (b) How many birds had the most common beak length? (1)
- 3.2.4 The investigation referred to in Question 3.1 could contribute to scientists' understanding of evolutionary theory. Describe TWO ideas that could have developed from the investigation.
- 3.3 If a volcanic eruption had occurred on an island inhabited by a population of tortoises and the population of tortoises became separated into two groups, what type of speciation could occur? Explain how this could lead to the formation of a new species of tortoise.

(6) [**30**]

4.1 Sickle-cell anaemia is an autosomal disorder that affects the production of haemoglobin, the oxygen-carrying chemical found in red blood cells. Faulty haemoglobin causes red blood cells to have a sickled (distorted) shape instead of their normal disc-shape. The cause and various effects of sickle-cell anaemia are summarised below.



4.1.1 Explain what is meant by an 'abnormal gene' in the diagram above. (2)

- 4.1.2 According to the diagram, why does sickling of red blood cells cause anaemia?
- 4.1.3 From the information provided in the diagram, explain clearly how abnormal haemoglobin in red blood cells leads to a life-threatening disorder.

4.1.4

The gene for normal haemoglobin is dominant and may be indicated as **H**. The gene that causes abnormal haemoglobin may be indicated as **h**. An individual having TWO copies of the abnormal gene will develop sickle cell disease. An individual with ONE copy of the abnormal gene in the genotype shows some sickling without all the effects shown in the diagram above.

(a) Give the genotype for an adult that produces normal haemoglobin and has no distorted red blood cells.

(1)

(4)

- (b) Use a diagram (such as a Punnet diagram or genetic cross) to show the result of a cross between an **Hh** individual and the normal condition in Question 4.1.4 (a).
- (c) (i) Use your Punnett diagram/genetic cross to determine what percentage of the offspring are likely to suffer from some sickling?
  - (ii) What percentage of the offspring are likely to show no sickling?
- 4.2 The following diagram is an example of a karyotype from a normal human individual.



[Adapted from Toole, G and S]

- 4.2.1 Is the karyotype showing the haploid or diploid state of the chromosomes? (1)
- 4.2.2 Explain why a karyotype must be made from cells which are in the process of cell division.
- 4.2.3 What evidence is there that this is a karyotype of a 'human male'? (2)
- 4.2.4 Describe TWO ways in which an abnormal karyotype could appear different to the example shown above. (2)

(2)

(4)

(1)

(1)

4.3 Read the information below on Jesse Gelsinger, one of the first recipients of gene therapy and then answer the following questions.

A medical study at the University of Pennsylvania took a turn for the worse when Jesse Gelsinger, age 18, died from complications related to a gene therapy he had received. At the time, this was the first known death directly attributable to gene therapy. Gelsinger was a voluntary participant in an experimental gene therapy trial with the aim of treating a fatal liver disorder known as OTC.

The therapy consisted of the healthy OTC gene being packaged in a virus vector. The vector was then injected into an artery that leads directly into the liver. Preliminary studies on mice, baboons and monkeys showed success with this approach, with mild (but temporary) side effects. Before this trial no one had injected viruses directly into the human bloodstream. Jesse was in the study's group which received the highest dosage. Within 24 hours, Jesse's liver began to show serious signs of distress and he slipped into a coma. Four days after the injection there was no sign of brain activity and after the withdrawal of life support, Jesse died almost instantly.

The researchers involved in the study determined that there was no evidence of human error and that Jesse's death was the result of an unusual immune response triggered by the virus. Upon investigation, the two governing agencies responsible for overseeing gene therapy trials found that four monkeys in the preliminary studies had a similar reaction and consequently died. Researchers modified the virus and lowered the dosage in the human trials as a result. Jesse's death opened a number of questions about gene therapy and halted all gene therapy trials in the US for a while. The agencies found that deaths in other, different trials had not been reported. Furthermore, reports of adverse reactions or deaths that were reported were kept confidential. Many in the scientific and medical community believe that sharing information about gene therapy trials might prevent something like this from ever happening again.

[Adapted from http://gslc.genetics.utah.edu]

4.3.1 Explain what is meant by gene therapy in the above article.

- (3)
- 4.3.2 'The researchers involved in the study determined that there was no evidence of human error ...'. Using information from the article explain why some scientists might not agree with this statement.

(6) [**30**]

# Do you think that South Africa should establish a national DNA database that is freely available to any interested party, for example, crime investigators, insurance companies and employment agencies?

Read the source material carefully and present a debated argument to illustrate your point of view.

To answer this question you are expected to:

- select relevant information from sources A to G below. Do not attempt to use all the detail provided.
- integrate your own relevant biological knowledge. However, do not write an essay based solely on your own knowledge.
- take a definite stand on the question and arrange the information to best develop your argument.
- write in a way that is scientifically appropriate and communicates your point of view clearly.

[20]

Write an essay of not more than  $1\frac{1}{2}$  to 2 pages to answer the question.

# SOURCE A PROCESS OF ELECTROPHORESIS USED IN CONSTRUCTING A DNA PROFILE



[<http://palmer-dna-technology.com>]

# SOURCE B THE INVENTOR OF DNA FINGERPRINTING, PROFESSOR SIR ALEC JEFFREYS, OF THE UNIVERSITY OF LEICESTER, HAS VOICED HIS CONCERNS OVER THE ETHICS OF A DNA DATABASE

- He said: "The national DNA database is a very powerful tool in the fight against crime, but recent developments such as keeping innocent people's DNA on file raises significant ethical and social issues."
- "Now hundreds of thousands of entirely innocent people are populating that database." "This was not the initial purpose of the database."
- "There are also issues concerning familial searching, where the database is used to identify possible relatives of an unknown suspect in a criminal investigation. This is a violation of human rights and a right to privacy."

[Adapted from: Science Daily, 11 January 2008]

## SOURCE C HISTORY OF DNA DATABASES

A national DNA database is a government database of DNA profiles which can be used by law enforcement agencies to identify suspects of crimes.

- The first government database was set up by the United Kingdom in April 1995.
- The second one was set up in New Zealand.
- France set up theirs in 1998.
- In the USA, the FBI has organised the CODIS database. Originally intended for sex offenders, it has since been extended to include almost any criminal offender.
- In England and Wales, anyone arrested on suspicion of a recordable offence must submit a DNA sample, the profile of which is then stored on the DNA database as a permanent record.

Many are now calling for national DNA databases that would include the DNA of all citizens regardless of whether they have been arrested and convicted of a crime.

- Portugal, for example, has plans to introduce a DNA database of its entire population.
- In Denmark, the Danish Newborn Screening Biobank keeps a blood sample from people born after 1981. The purpose is to test for **inheritable** disease and other disorders. But it is also used for DNA tests to identify criminals with diseases that need treatment in prison and to identify suspected criminals.



The potential use to crime-fighting and health studies has caused many individuals, politicians, and organisations to call for national DNA databases. Yet, many oppose the idea, considering it a violation of privacy rights. Other concerns include the price-tag of collecting DNA data and maintaining a DNA database.

## A technician working with DNA samples in a laboratory

[<http://debatepedia.idebate.org>]

## SOURCE D SOUTH AFRICA'S NATIONAL DNA DATABASE

- Currently holds the DNA profiles of certain suspects arrested and convicted and DNA profiles collected from some crime scenes.
- South Africa is in the early stages of recognising the importance of maximising the size of its National DNA Database because the greater the number of DNA Profiles on the Database, the greater the chance of solving crimes and catching criminals.
- The expansion of our National DNA Database requires certain changes in our law, and currently Parliament is reviewing an important new Bill called the **Criminal Law (Forensic Procedures) Amendment Bill** which, when passed, will ensure that every person arrested for an alleged offence as well as all convicted offenders, will have their DNA profiles loaded onto the Database.
- Given the number of repeat offenders in South Africa, there is a strong possibility that eventually, the individual who committed the crime already has his or her DNA profile on the National DNA Database.
- **THE DNA PROJECT** was started by Vanessa Lynch in conjunction with the Matthews Family following the tragic murders of each of their family members, John Lynch and Leigh Matthews, in 2004. They determined that successful criminal justice systems worldwide all had implemented and developed a National DNA Criminal Intelligence Database.

[Adapted from SA National Database – Proposed Legislation]

## SOURCE E LETTING THE DEAD SPEAK

Across the world there are areas that suffer under ruthless governments. Opponents of these regimes are often abducted, murdered and buried in unmarked graves. DNA analysis plays an important role in identifying the victims and is a key technology in the process of mapping human genocide.

[Adapted from QUEST; 2005]

## SOURCE F

## The loss of life in the Haiti earthquake

On 12 January 2010 Haiti experienced a major earthquake. The earthquake killed around 300 000 people and left one million people homeless and without basic needs. Spain helped Haiti with a DNA fingerprinting system to help reunite lost children with their families, and combat the kidnapping and sale of lost children (trafficking) that was occurring.

The DNA testing system, called DNA-Prokids, has already helped identify more than 230 victims of child trafficking in 12 countries.



[Adapted from: <http://www.news24.com>]

## SOURCE G GENE DISCRIMINATION

Once all the disease causing genes are identified, researchers say it will be possible for people at high risk of cancer or heart disease to be identified by their genetic code. The genes, in effect, could forecast years in advance who will get sick and who will not.

Employers and health insurance companies could save millions of dollars by not hiring or enrolling people whose genes show them to be highly susceptible to diseases, experts say.

Gene discrimination could, therefore, join the list of other forms of discrimination – racial, ethnic, and sexual.

'While we might have the scientific basis for a whole new revolution in medicine, we might find that nobody wants to participate because of their fears of misuse,' said Dr Francis Collins, Head of the National Human Genome Research Institute. 'That would be a terrible tragedy.'

Already people are expressing their concerns. A Time/CNN poll appearing in Time magazine found that 75% of Americans did not want insurance companies to know their genetic code and 84% didn't want the government to know.

[Adapted from: <www. BBC.UK>]

#### Total: 150 marks