19.2 Genetic Technology Applied to Medicine

Question Paper

Course	CIE A Level Biology
Section	19. Genetic Technology
Topic	19.2 Genetic Technology Applied to Medicine
Difficulty	Medium

Time allowed: 50

Score: /35

Percentage: /100

Question 1a

BRCA1 and BRCA2 are genes that code for proteins involved with the repair of DNA mutations

(i)
State how mutations occur in DNA.

[1]

(ii)
Suggest how mutations in BRCA1 and BRCA2 genes could increase the likelihood of tumour development.

[3]

[4 marks]

Question 1b

Individuals with a family history of breast cancer will often have genetic screening to identify mutations in the BRCA1 and BRCA2 genes.

Outline how genetic screening can reduce the risk of breast cancer in high risk individuals.

[2 marks]

Question 1c

Fig. 1 shows a genetic screening technique.

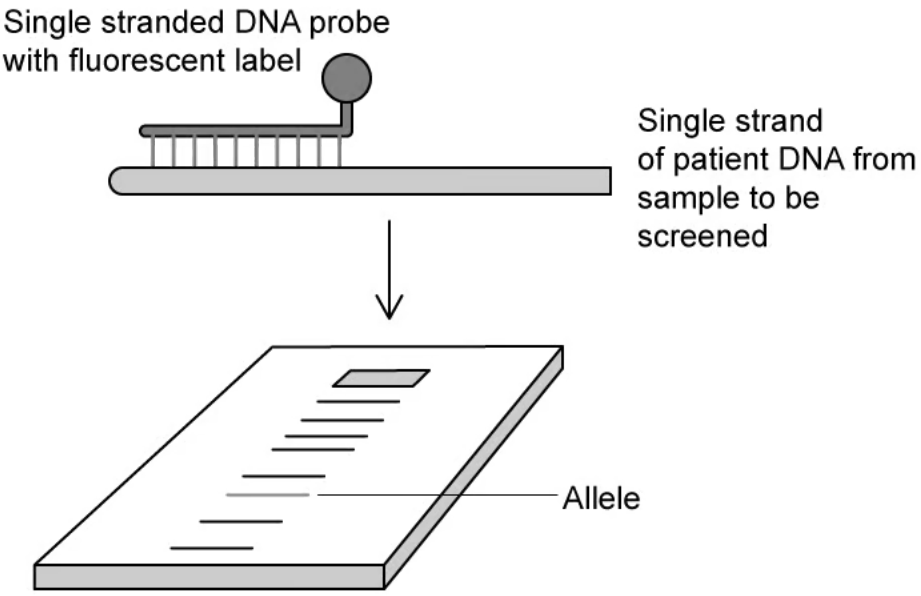

Fig. 1

Use Fig. 1 to suggest how a DNA probe can be used to show the presence of a particular allele.

[2 marks]

Question 1d

Screening for BRCA1 and BRCA2 is carried out in adults, but some other genetic traits are tested for in foetuses prior to birth. In either case the results can bring up difficult decisions and individuals considering or undergoing such screening will often receive help from a genetic counsellor to help them think through ethical and social issues.

Outline the social and ethical issues around genetic screening.

[3 marks]

Question 2a

Adult haemoglobin is a protein made up of four polypeptide chains; two alpha chains and two beta chains.

Beta thalassemia is a genetic condition in which a mutation in the gene for the beta chain of adult haemoglobin either reduces or prevents its production.

Suggest **three** symptoms of beta thalassemia.

[3 marks]

Question 2b

A form of gene therapy is being trialled for use in the treatment of beta thalassemia. The treatment involves harvesting stem cells from the bone marrow of patients before altering them in the lab and infusing the cells back into the patient's body.

(i)
Describe the stem cells found in bone marrow.

[2]

(ii)
Outline the process by which the stem cells can be altered.

[3]

[5 marks]

Question 2c

One difficulty with carrying out safe and effective gene therapy is of ensuring that any new DNA is inserted into the right part of a patient's DNA.

Suggest why the insertion of DNA in the wrong location could be a problem.

[2 marks]

Question 2d

Give **three other** concerns related to gene therapy.

[3 marks]

Question 3a

Adenosine deaminase (ADA) is an enzyme involved with multiple chemical reactions inside cells. One effect of ADA deficiency is a lack of lymphocyte maturation.

Outline how ADA deficiency can lead to severe combined immune deficiency (SCID).

[4 marks]

Question 3b

Treatment options for SCID caused by an ADA deficiency include:

- Enzyme replacement therapy (ERT), in which patients receive injections of recombinant ADA enzymes.
- Haemoatopoietic stem cell transplant (HSCT), in which patients receive a bone marrow transplant from a donor.

Complete the table below by suggesting an advantage and a disadvantage of each of these treatment types.

	Advantage	**Disadvantage**
ERT		
HSCT		

[4 marks]

Question 3c

More recent treatment for ADA SCID involves the use of gene therapy.

Describe the process of gene therapy for ADA SCID.

[3 marks]