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VCE Biology $\frac{3}{4}$

AOS 1 Revision [1.0]

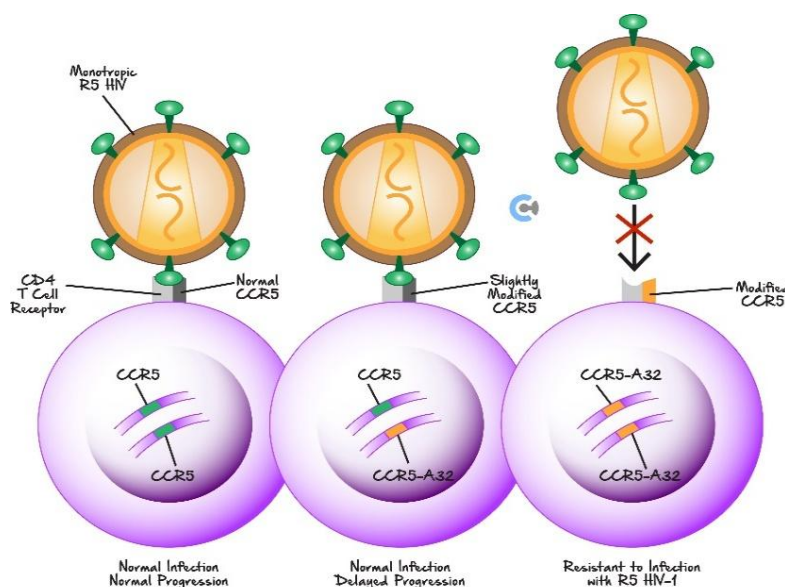
SAC 6 (JMSS) Solutions

33 Marks. 10 Minutes Reading. 40 Minutes Writing.

Section A: Bioethical Issue - Scientific Concepts (20 Marks)



- HIV (Human Immunodeficiency Virus) is the virus that causes AIDS (Acquired Immune Deficiency Syndrome) and is one of the most pressing health concerns facing the modern world. Since the first reported case of HIV/AIDS in 1981, over 42 million people have died. HIV infects human cells by binding to the CC chemokine receptor 5 (CCR5) which is widely expressed in several immune cells, including T cells (a type of white blood cell), involved in the inflammatory responses.
- Out of the millions of people infected each year with the HIV virus, a few have shown HIV/AIDS resistance. A genetic mutation known as the CCR5Δ32 (Delta 32) allele, an alternative form of the CCR5 gene, delays the progression of AIDS and in some cases even brings about immunity. The CCR5Δ32 mutation is characterised by a 32 base-pair (bp) deletion of the CCR5 gene coding region. The mutation inhibits HIV's ability to infect immune cells and delays the progress of AIDS. Several studies have shown that the number of the CCR5Δ32 (Delta 32) allele that each individual carries affects his/her susceptibility to HIV infection (Figure shown below).



Susceptibility to HIV-1 infection of different CCR5 genotypes.

GENOTYPE	NUMBER OF GENE COPIES IN GENOME		SUSCEPTIBILITY TO HIV INFECTION
	Normal CCR5 allele	CCR5Δ32 (Delta 32) allele	
Homozygous wildtype	2	0	Normal
Heterozygous	1	1	Normal
Homozygous CCR5Δ32	0	2	Resistant

The table above shows the summary of the effect of CCR5Δ32 gene copy number on the susceptibility to HIV infection.

Question 1 (1 mark)

Viruses are able to infect all living organisms, ranging from simple prokaryotes (e.g., bacteria) to complex eukaryotes (e.g., humans). Viruses hijack the host cell machinery to make proteins that they need. Name the feature of the genetic code that viruses are exploiting.

Genetic code is **universal**.

Question 2 (14 marks)

Scientists used the polymerase chain reaction (PCR) in a study to determine the percentage of people in a population who carry the CCR5Δ32 (Delta 32) mutation.

a. Complete the following table by listing the component (reagent) used in a PCR or its function. (4 marks)

COMPONENT (reagent)	FUNCTION
	Synthesises the new DNA molecule.
DNA Primers	
	Basic building blocks used to make a polymer.
DNA template	

Component and function (what does it do?).

Component: **Taq /thermostable DNA polymerase** is a special type of DNA polymerase used in PCR.

Function: Primers allow (DNA polymerase/enzyme) to **add nucleotides/starting point** for DNA synthesis.

Component: (Free) **DNA nucleotides** are the building blocks for making new DNA.

Function: DNA template provides a copy of the **gene/region of interest** to be copied/to which **primers** can bind.

- b. Complete the table below to explain the temperature changes in a thermocycler (machine in which PCR is completed). (3 marks)

Stage	Temperature	Explain the purpose for using this temperature range
1	90°C - 95°C	
<p><i>90-95°C: Denatures/breaks H-bonds → separates double-stranded DNA template/makes DNA single-stranded</i> <i>55-60°C : Complementary primer/s → bind to the DNA template/strand</i> <i>72°C: Optimal temperature for Taq DNA polymerase → extend/build the new DNA molecule.</i></p>		
3	72°C	

- c. The PCR in the study was designed to produce a DNA fragment that spans across the Delta 32 mutation site within the CCR5 gene. If the wildtype CCR5 gene produces a 420 bp (base pairs) PCR product, what would be the size (in bp) of the PCR product arising from the Delta 32 allele? (1 mark)

388 bp

- d. The PCR products were analysed using gel electrophoresis which separates DNA fragments of different lengths. In terms of your understanding of the DNA molecule, explain why this process requires an electrical current to be applied to the gel electrophoresis chamber. (2 marks)

DNA is **negatively charged** → move **towards the positive**/away from negative electrode (cause & effect)
Smaller DNA fragments will move further than larger fragments

e. During gel electrophoresis, scientists used a 1000- bp DNA ladder as a reference to estimate the size of the PCR products. The ladder (L) consists of ten DNA fragments of known sizes that differ from each other by 100 bp. On the figure below: (4 marks)

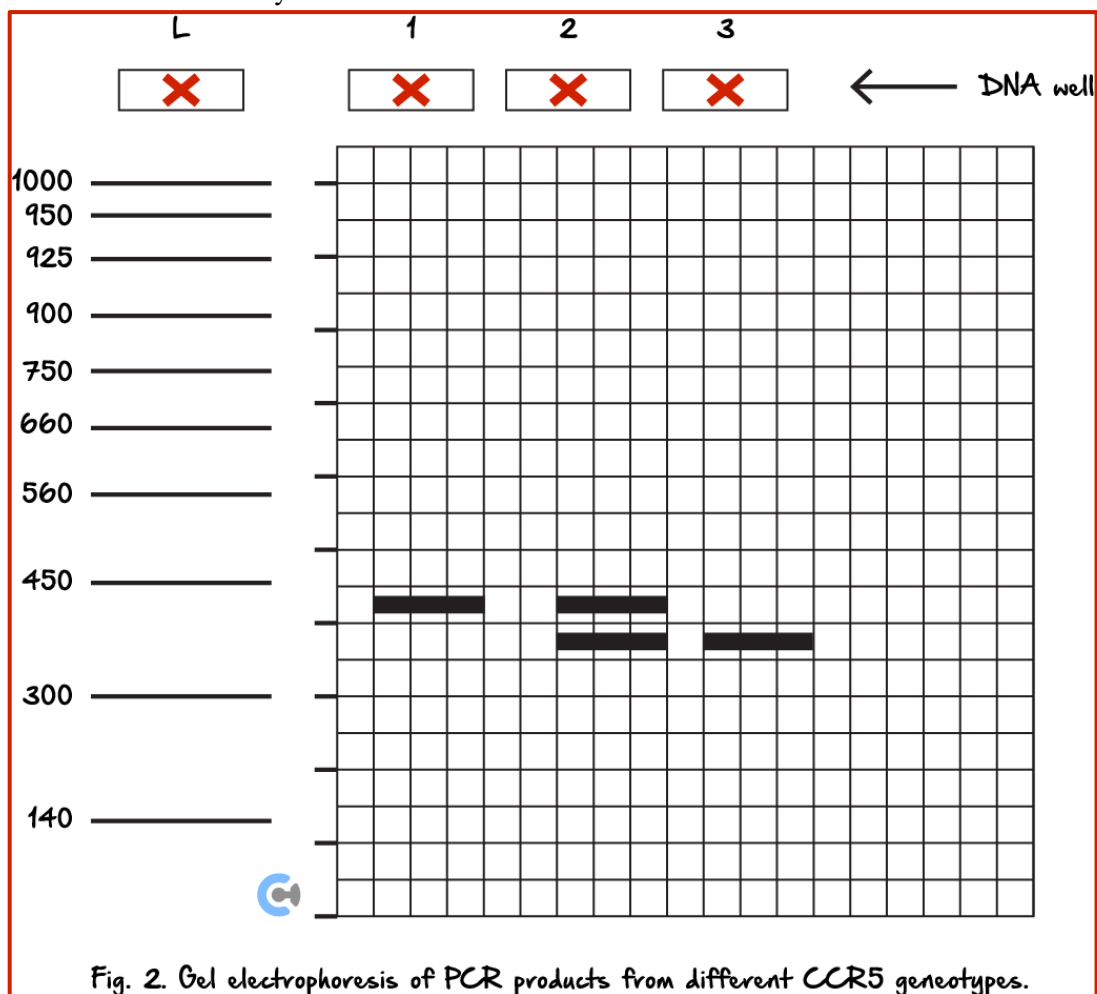
i. Write the size of each DNA fragment in the ladder on the left-hand side.

ii. Using the information in **part 2.c.**, draw the expected PCR products in lanes 1-3 for three different individuals in the study:

➤ L: DNA ladder Individual 1: Has only wildtype CCR5 alleles.

➤ Individual 2: Has one wildtype CCR5 allele and one Delta 32 allele.

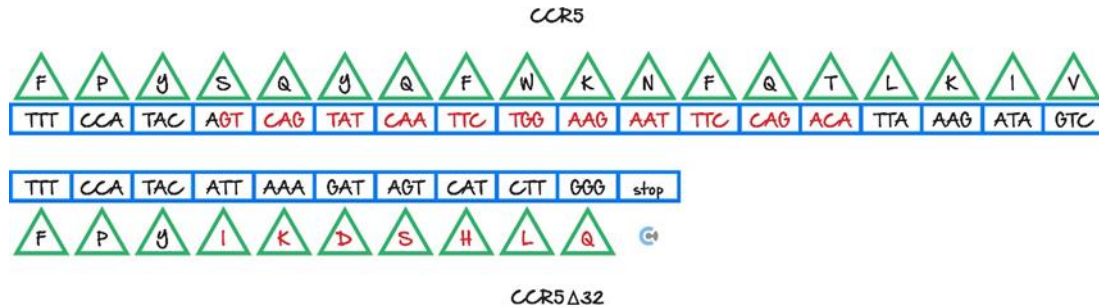
➤ Individual 3: Has only Delta 32 alleles.



Correctly label 100 & 1000 bp markers in lane L
 Correctly show Individual 1 has a single band at 420 bp
 Correctly show Individual 2 has two bands at 388 bp & 420 bp
 Correctly show Individual 3 has a single band at 388 bp
 Penalised 1/2 mark if remaining marker fragments not correctly shown (10 in total)
 Penalised 1/2 mark if bands are not all aligned to match MW ladder/standard
 Penalised 1/2 mark if both homozygous genotypes are not shown as a single band
 Penalised 1/2 mark if not all bands are aligned with similar sized bands (Individuals 1-2)

Question 3 (3 marks)

CCR5 is a transmembrane receptor protein consisting of a single polypeptide chain. The CCR5 Δ 32 (Delta 32) deletion (shown in red in the figure below) introduces a premature stop codon into the coding sequence, resulting in a nonfunctional receptor.



In the hierarchy of protein structure, how many levels of the CCR5 protein structure are affected by the CCR5 Δ 32 (Delta 32) deletion? Provide evidence in your answer.

☐ 3 levels of protein structure.

Any 2 of the following:

☐ **Primary:** amino acid sequence changed due to a premature stop codon/refer to a.acids changed (cause & effect).

☒ **Secondary:** alpha-helices and beta-pleated sheets affected (\rightarrow overall 3D structure is changed as a result).

☐ **Tertiary:** single polypeptide chain i.e. not quaternary which has multiple chains)

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Question 4 (2 marks)

The CCR5 gene is predominantly expressed in T cells (a type of white blood cell) but not in other cell types. Describe two advantages for this example of gene regulation.

Saves **energy**

Ensures that gene is expressed at the right **time/amount/location**

Penalised ½ mark if context not shown e.g. function of CCR protein (refer to white blood cells/immune response/infection)

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Section B: Bioethical Issue - Ethical Concepts (13 Marks)



- Since the discovery of its role in adaptive immunity, CRISPR – Cas9 (clustered regularly interspaced short palindromic repeats-CRISPR associated protein) has drawn great interest due to its potential use as a genome editing tool. The CRISPR – Cas9 system consists of a series of palindromic repeats separated by non-repetitive sequences called spacers, to which cas genes (CRISPR-associated genes) are associated. These genes encode endonuclease enzymes capable of cutting DNA strands in a site-specific manner.
- Gene editing strategies using the CRISPR – Cas9 system is currently being trialled to treat many diseases including HIV/AIDS. One such strategy is to edit the CCR5 gene by creating a mutation which would result in a non-functional CCR5 protein. The rationale behind this approach is that people with both mutated alleles of CCR5, such as the CCR5 Δ 32/ Δ 32 genotype, are protected against viruses such as HIV, smallpox, and flavivirus. They also have faster recovery from brain injury following trauma or stroke, and reduced inflammatory responses during infection. The health costs of carrying the double CCR5 Δ 32/ Δ 32 mutant alleles are having increased susceptibility to viruses that cause Influenza and West Nile fever and abnormal bone growth due to the increased activity of osteoclasts (cells that degrade bone).
- The CRISPR – Cas9 system offers precise and affordable technology to edit the genomes of individuals affected by numerous genetic disorders. However, multiple studies have reported unexpected off-target mutations generated by CRISPR – Cas9. These are unintended mutations on other genes that are not the original targets of the gene edits, resulting in unexpected effects. Additional ethical concerns with the use of CRISPR – Cas9 include the editing of germline (reproductive) cells, as any genetic modification can be inherited and passed onto future generations and the issue of eugenics to improve the genetic 'quality' (whatever that may be) of certain individuals or populations.

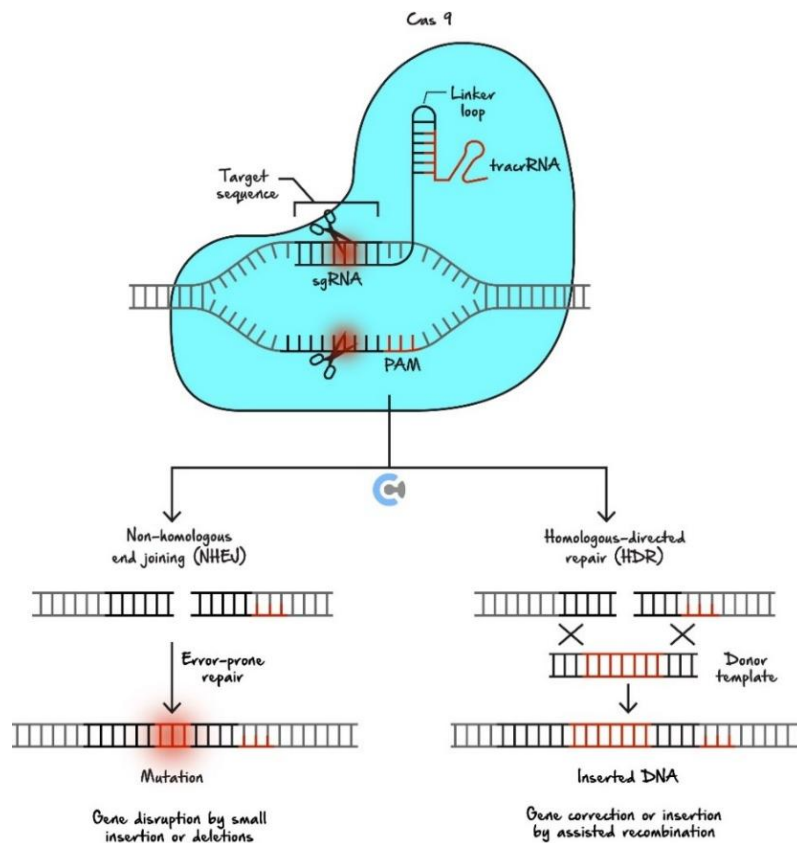
Question 5 (2 marks)

Restriction enzymes (e.g. EcoRI) and Cas9 are different types of endonucleases. Describe one similarity and one difference between them.

Similarity: both cut DNA at a **specific sequence/site** OR both can make double-stranded DNA breaks
 Differences: Cas9 uses a **guide RNA** (not RE) OR Cas9 cutting site can be **customised/modified** (not RE) OR RE can make **sticky ends**/Cas9 only makes blunt-ended cuts.

Question 6 (4 marks)

In November 2018, Chinese scientist Jiankui He announced that he had used CRISPR – Cas9 to edit two human embryos in an attempt to disable the gene for CCR5, which codes for a receptor that HIV uses to enter cells. He said that two girls had been born a few weeks earlier. The father of the twins was HIV-positive. Describe the procedure of how Jiankui He may have carried out the experiment by referring to only **ONE** of the pathways (NHEJ or HDR) shown below.



- ☒ (Single) guide/sgRNA/gRNA made **complementary** to the target (CCR5) gene.
- ☐ sgRNA/Cas9 complex recognises/attaches to the **PAM** sequence (in embryos).
- ☐ Cas9 makes a (double-stranded) **DNA break/cut** within the target (CCR5) gene.
- ☒ Accurately describe **ONE** of the repair processes (ideally name pathway):
 - ☒ **NHEJ:** (random) **mutation** is introduced during **DNA repair** → silence/switched off (CCR5) gene
 - ☐ **HDR:** mutant/Delta 32 version/388 bp segment of gene use as **template for DNA repair** → mutation created/(CCR5) gene function silenced.
- ☒ Penalised 1 mark for not providing context at least once i.e. gene targeted by sgRNA/Cas9 is CCR5 gene
- ☐ Penalised 1/2 mark for indicating whether the results of this technology switches the gene off OR on/repairs it.

Question 7 (4 marks)

For each comment raised below, indicate ONE ethical concept that best describes the type of implication/issue. Each concept may only be used ONCE.

Comments and questions raised by individuals	Choose from: Integrity/Justice/Beneficence/ Non-maleficence/Respect
Infertile couples often undergo in-vitro fertilisation (IVF) treatment to increase their chance of having a baby. An IVF company offers these couples the option of using CRISPR – Cas9 for gene therapy to edit any disease gene (e.g. cystic fibrosis). For this service, the company charges a fee of three times more than the standard IVF treatment, a cost which very few can afford.	
An ethical committee is debating the use of gene editing tools to mutate the CCR5 gene. One argument put forward is that people with the CCR5 mutation have increased resistance to viruses such as HIV, smallpox, and flavivirus.	<div> Justice - affordability of treatment costs Beneficence - increased disease resistance Non-maleficence - side/unintended effects Integrity - honest reporting of research outcomes </div>
Another argument in the debate of using CRISPR – Cas9 in gene editing is that people carrying mutated copies of the CCR5 gene are more susceptible to infection from viruses such as Influenza and West Nile and are more likely to suffer from abnormal bone growth.	
Billionaire Donald Trump is suffering from an unspecified incurable disease. He offers to donate US\$100 million dollars to any research institution who can find a cure and in his words, make him 'great again'. To receive the money, the institution must publish their research data as supporting evidence in a scientifically accepted journal.	

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Question 8 (3 marks)

Gene editing experiments on human embryos are currently banned in all countries internationally. Using a consequences-based approach to bioethics, suggest two possible reasons for why international governments have agreed on the ban of gene editing in human embryos.

explicitly articulated. Students should avoid repeating/paraphrasing information from the Q7 table.

- ☒ Consequences-based approach aims to **maximise positive outcomes while minimising negative outcomes / weigh positives vs negatives.**
- ☐ Two possible reasons that match the context of the case study (embryonic gene editing) and government ban which would focus on **negative outcomes**. Suggested answers include:
 - ☐ Alternatives to gene editing (context e.g. diseases like HIV can be easily treated or spread prevented).
 - ☒ Unintended off-target mutations (context e.g. passed on/can lead to other diseases or health complications).
 - ☐ Social and economic costs (context e.g. money spent on gene editing could be spent on other programs like vaccine development which is cheaper/helps more people).
 - ☐ Another suitable argument against gene editing, which is in context.
- ☐ Penalised 1 mark (max) for not providing context e.g. reflect stakeholder perspective (government) OR gene editing in embryos

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