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VCE Biology  $\frac{3}{4}$   
AOS 1 Revision [1.0]  
**SAC 7 Solutions**

40 Marks. 5 Minutes Reading. 60 Minutes Writing.

## Section A: SAC Questions (40 Marks)



### Case Study: The Controversial Use of PGD

- Over the past two decades, advances in genetic sequencing, embryo screening, and gene-editing techniques have transformed reproductive medicine. One of the most widely used technologies is Preimplantation Genetic Diagnosis (PGD), which is performed alongside in vitro fertilisation (IVF) to screen embryos for genetic abnormalities before implantation. PGD involves extracting a single cell from an early-stage embryo and amplifying its DNA using polymerase chain reaction (PCR). This allows scientists to analyse specific genes for inherited diseases or other genetic traits.
- Initially, PGD was used to prevent serious genetic disorders such as cystic fibrosis, Huntington's disease, and Tay-Sachs disease. However, recent advancements have expanded the capabilities of genetic screening, allowing for the detection of genes associated with intelligence, physical attributes, and disease predisposition. This has led to widespread ethical debates over whether genetic selection should be restricted to preventing severe medical conditions or whether parents should have the right to select embryos based on non-medical traits.
- In 2023, a couple undergoing IVF and PGD to prevent their child from inheriting a genetic disorder made headlines when they disclosed that they had selected their embryo not only for health but also for predicted intelligence and specific physical traits. Their decision sparked an intense public debate.
- Supporters of expanded genetic selection argue that parents should be able to maximise their child's potential using available technologies. They believe that selecting for intelligence, height, or athletic ability could improve future generations and reduce genetic diseases linked to lower cognitive function or physical disabilities.
- However, critics warn that such practices could lead to a rise in genetic inequality, where only wealthy families can afford enhancements, leading to a genetically stratified society. Others raise concerns about unintended genetic consequences and the loss of genetic diversity, questioning whether scientists truly understand the long-term effects of selecting for complex traits.
- Ethicists and policymakers are now struggling to answer the question:
- Where should we draw the line between medical necessity and genetic enhancement?

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**Question 1** (18 marks)

Preimplantation genetic screening involves DNA analysis, often using short tandem repeats (STRs).

- a.** Short tandem repeats (STRs) are widely used in genetic screening for diseases such as those mentioned in the article.

Explain what STRs are and why they are useful in analysing genetic differences between individuals. (3 marks)

STRs (Short Tandem Repeats) are short sequences of DNA (typically 2–6 base pairs long) that are repeated multiple times in a row at specific locations in the genome. (1 mark)

They are useful in analysing genetic differences because the number of repeats varies between individuals, making them highly polymorphic. (1 mark)

This variation allows STRs to be used in DNA profiling for forensic analysis, paternity testing, and identifying individuals with high accuracy. (1 mark)

Scientists will often use restriction enzymes to help them with their analysis of selected genetic material.

- b.** Describe what a restriction enzyme does and the two types of ends that could be produced when using one. (2 marks)

A restriction enzyme cuts DNA at specific recognition sequences, breaking the sugar-phosphate backbone. (1 mark)

It can produce either blunt ends, where the DNA is cut straight across both strands, or sticky ends, where the cuts are staggered, leaving single-stranded overhangs. (1 mark)

- c.** Explain how restriction enzymes could be used to identify a disease-causing abnormal gene being present in the DNA. (2 marks)

Restriction enzymes cut DNA at specific recognition sites, and a mutation in a disease-causing gene may create or remove these sites. (1 mark)

By comparing the fragment patterns using gel electrophoresis, scientists can determine if the abnormal gene is present. (1 mark)

The polymerase chain reaction (PCR) is used to amplify a DNA sample.

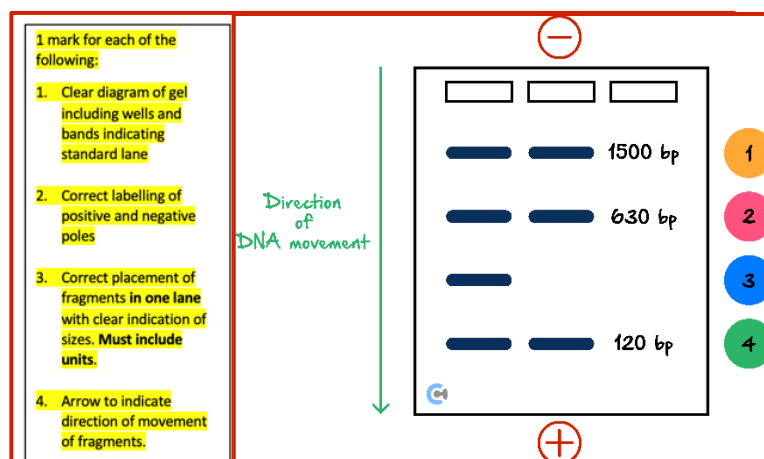
d. PCR involves the use of primers. Describe what a primer is. (2 marks)

A primer is a short, single-stranded sequence of DNA that is complementary to a specific target region of DNA. (1 mark)  
It provides a starting point for DNA polymerase to begin DNA replication during PCR. (1 mark)

e. PCR involves three stages. Complete the following table. (5 marks)

Name of Stage	Temperature	Description of Process
Denaturation	94°C	H bonds between double strands of DNA are broken; this separates DNA into single strands.
Annealing	55°C	Primers attach to 3' ends of single-stranded DNA.
Extension	72°C	Tag Polymerase adds complementary nucleotides to (5'-3'), using primers as a starting point, joining phosphate backbone via phosphodiester bonds forming double stranded DNA copy.

f. In this case, the scientists used a restriction enzyme to cut the DNA sample, and three fragments obtained were 1500 bp, 630 bp and 120 bp in length. These fragments were separated using a technique known as gel electrophoresis. In the space below, draw a labelled diagram of the outcome of the gel run. (4 marks)



**Question 2** (4 marks)

A couple undergoing **IVF and Preimplantation Genetic Diagnosis (PGD)** to prevent their child from inheriting a genetic disorder, ultimately decided to **select an embryo not only for health but also for predicted intelligence and specific physical traits**. The couple defended their choice, stating:

*"If we have the ability to give our child the best possible future by selecting the best possible genetic combination, why shouldn't we?"*

**Describe one bioethical approach and one concept** that is relevant to this situation.

Beneficence is the ethical principle of acting in the best interest of others. In this case, the couple may argue that selecting an embryo with traits they believe will provide the best future for their child aligns with beneficence, as it aims to improve their well-being. However, critics may argue that prioritizing non-medical traits could lead to unintended social and ethical consequences.

Genetic determinism is the belief that an individual's traits, such as intelligence or appearance, are primarily determined by their genes. This concept is relevant because the couple assumes that selecting certain genetic traits will guarantee a better future for their child, overlooking the influence of environmental and social factors on development.

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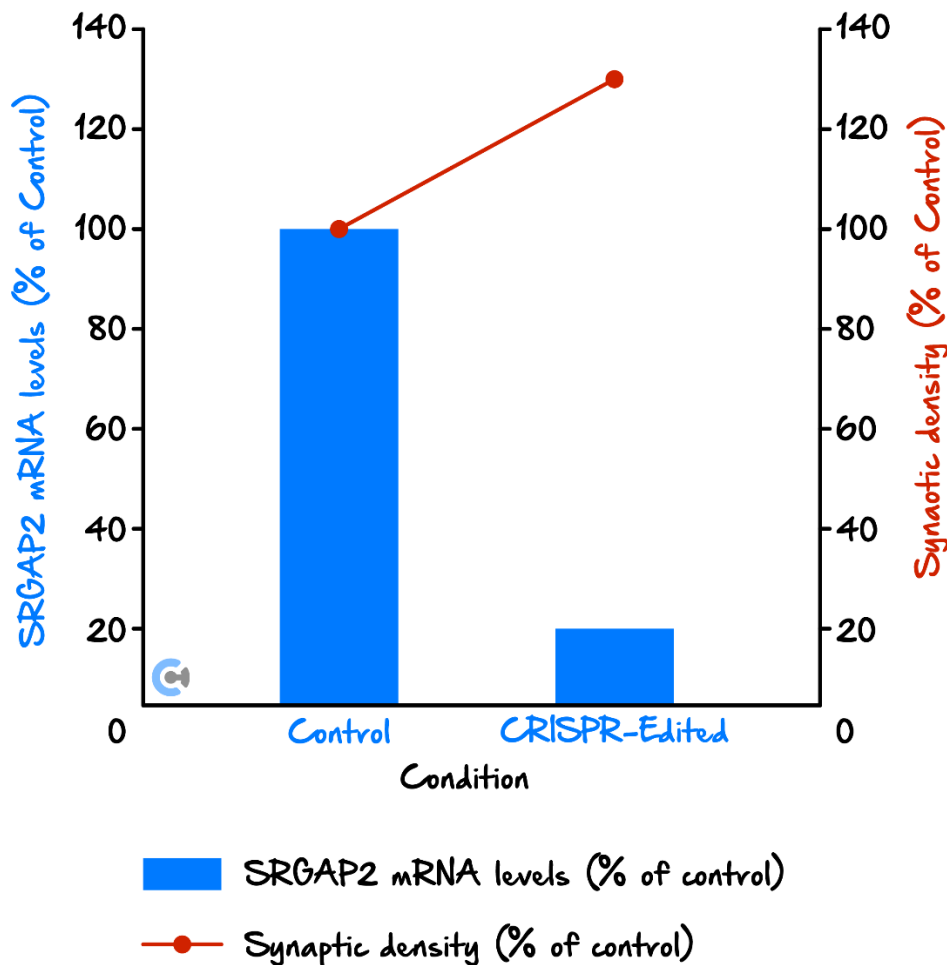
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**Question 3** (4 marks)

Following their decision to **genetically enhance their child**, the couple in the previous scenario participated in an **experimental CRISPR-Cas9 trial to silence a gene** linked to cognitive impairment. The scientists targeted the **SRGAP2 gene**, which is involved in **neurodevelopment** and has been suggested to affect **synaptic formation and brain plasticity**.

The researchers used **CRISPR-Cas9 with a guide RNA (gRNA) targeting the coding region of SRGAP2**. The goal was to prevent the gene from being expressed. After editing 30 **embryos**, they monitored the effectiveness of gene silencing by measuring **SRGAP2 mRNA levels and synaptic density in neural precursor cells derived from these embryos**.



- a. Explain how CRISPR-Cas9 can be used to silence the SRGAP2 gene, including the role of Cas9, guide RNA, and the cell's repair mechanisms. (2 marks)

CRISPR-Cas9 silences the SRGAP2 gene by using a guide RNA (gRNA) to direct the Cas9 enzyme to a specific DNA sequence, where it creates a double-strand break. (1 mark)  
 The cell repairs the break through non-homologous end joining (NHEJ), which introduces mutations that disrupt the gene's function, effectively silencing it. (1 mark)

- b. Using the graph provided, analyse the relationship between SRGAP2 mRNA levels and synaptic density in CRISPR-edited embryos. What conclusions can be drawn from the data regarding the effectiveness and potential consequences of silencing this gene? (2 marks)

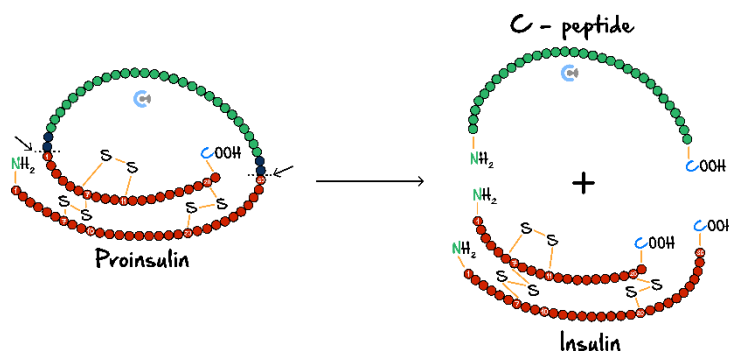
The graph shows an inverse relationship between SRGAP2 mRNA levels and synaptic density—CRISPR editing reduced SRGAP2 mRNA to 20%, while synaptic density increased to 130% compared to the control.

The data suggest that CRISPR successfully silenced SRGAP2, leading to increased synaptic density. This indicates that SRGAP2 normally restricts synapse formation, and its silencing may alter brain function.

#### Question 4 (14 marks)

Insulin is a peptide hormone that regulates blood glucose levels in the body. Research suggests a potential link between insulin levels in the brain and the development of neurodegenerative diseases such as Alzheimer's.

In pancreatic beta cells, insulin is initially synthesised as proinsulin. To become active, a segment known as the C-peptide is removed, forming mature insulin. This process is illustrated in the diagram below:



- a. Disulfide bonds play a crucial role in stabilising the structure of active insulin. What type of interaction occurs between the amino acids forming these bonds, and why is this important for protein structure? (1 mark)

Disulfide bonds form through covalent bonding between sulfur atoms of cysteine residues, stabilizing insulin's tertiary and quaternary structure to maintain its functional shape. (1 mark)

- b. Proinsulin undergoes post-translational modifications before becoming functional insulin. Describe one key modification that occurs and explain its significance. (1 mark)

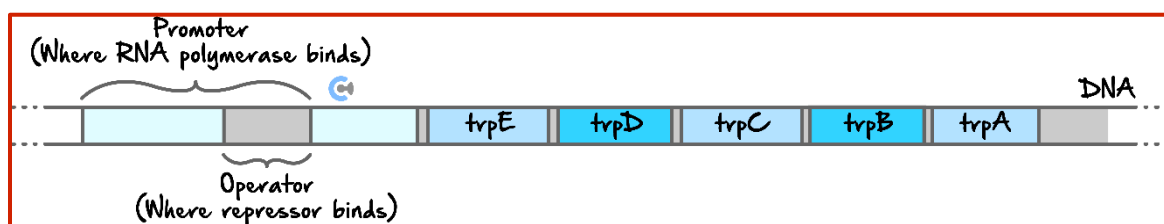
The C-peptide is cleaved from proinsulin, allowing insulin to adopt its active structure and regulate blood glucose levels. (1 mark)

- c. Several specialised cell organelles contribute to the production, processing, and secretion of insulin from beta cells. Complete the table below by describing the function of each organelle in insulin synthesis and secretion. (3 marks)

Organelle	Role in Insulin Synthesis & Secretion
Rough Endoplasmic Reticulum (RER)	Synthesizes <b>proinsulin</b> as ribosomes on the RER translate the insulin mRNA. The protein undergoes initial folding and formation of disulfide bonds.
Golgi Apparatus	Processes and <b>packages proinsulin</b> , cleaving the <b>C-peptide</b> to produce active insulin, which is then sorted into secretory vesicles.
Secretory Vesicle	Stores and <b>transports insulin</b> to the plasma membrane, where it is released into the bloodstream via <b>exocytosis</b> in response to glucose levels.

- d. The expression of the insulin gene in humans is regulated by proteins that are coded for by regulatory genes. In bacteria, gene regulation involves operons.

In the space below, draw a labelled diagram of the trp operon. (3 marks)



Required Components include:

- Promoter
- Operator
- Structural genes



- e. Part of the trp operon includes a leader sequence. When tryptophan levels are high in E. coli cells, attenuation of the operon will occur. Briefly describe why this happens and what it would look like. (3 marks)

When tryptophan levels are high, ribosomes quickly translate the leader sequence (trpL) because tryptophan-loaded tRNA is abundant.

This rapid translation allows the formation of a terminator hairpin (a stem-loop structure) in the mRNA, followed by a U-rich sequence, causing RNA polymerase to detach.

Transcription of the trp operon structural genes stops, preventing unnecessary tryptophan synthesis when it is already abundant.

50 years ago, diabetics had to inject insulin extracted from cows or pigs. In 1978, pharmaceutical companies commenced the production of human insulin using genetic engineering techniques.

The insulin genes are inserted into the plasmids next to a gene for  $\beta$ -galactosidase protein, which allows for the detection of successful gene insertion.  $\beta$ -galactosidase is coded for by a gene called lacZ. If  $\beta$ -galactosidase is present, the transformed bacteria will form blue colonies. If  $\beta$ -galactosidase is not present, white colonies are produced.

Two separate cell lines are created. One containing a plasmid with the DNA coding for the insulin A subunit and one containing a plasmid with the DNA coding for the insulin B subunit.

- f. Describe how functional insulin is obtained once these two cell lines have been established. (3 marks)

The bacteria of the two cell lines will produce the insulin A subunit and the insulin B subunit respectively. (1 mark)

These subunits are extracted from the bacteria and purified. (1 mark)

The two subunits are then combined to make functional insulin. (1 mark)

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