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VCE Biology ¾
AOS 1 Revision [1.0]
SAC 7

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)

Scientists will often use restriction enzymes to help them with their analysis or 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)

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

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)

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

Name of stage	Temperature used	Brief description of what happens

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.

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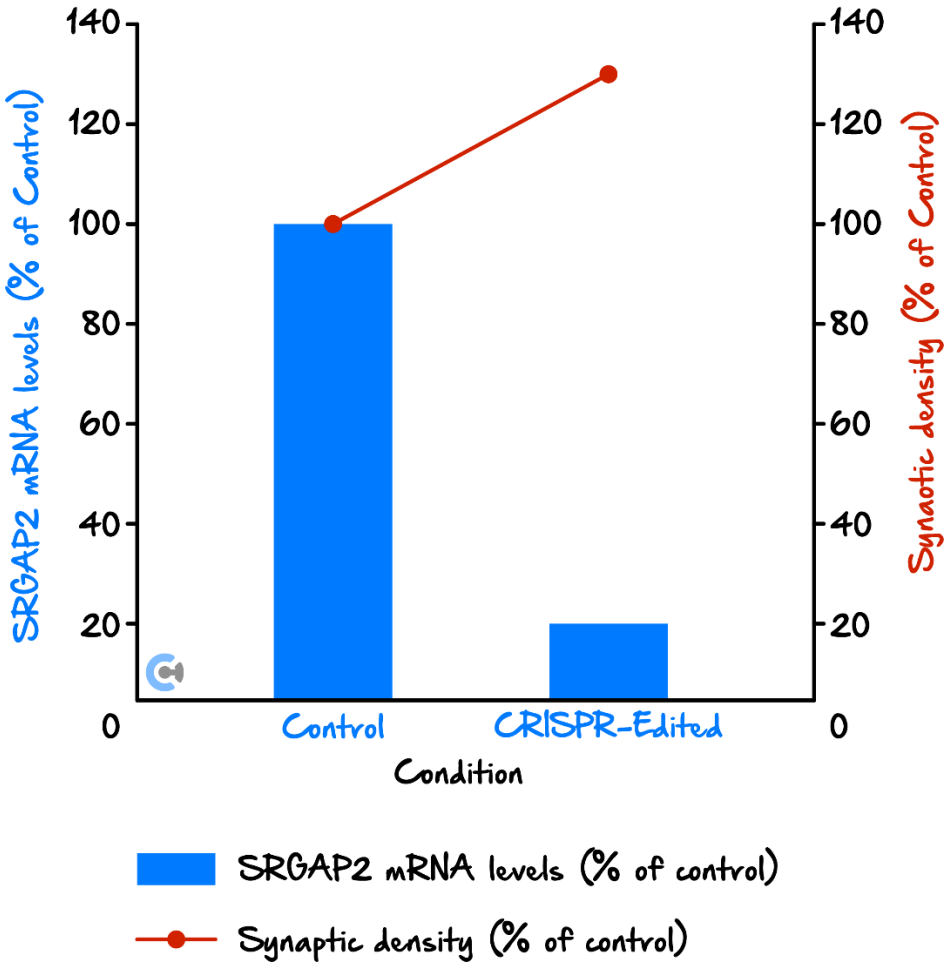
Question 3 (4 marks)

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)

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)

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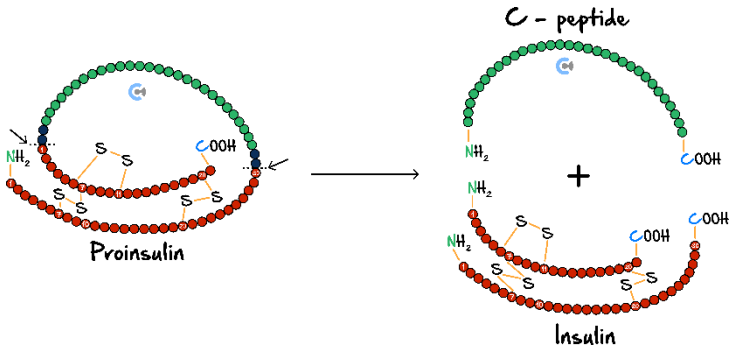
Question 4 (14 marks)

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)

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

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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)	
Golgi Apparatus	
Secretory Vesicle	

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)

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)

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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 β-galactosidase protein, which allows for the detection of successful gene insertion. β-galactosidase is coded for by a gene called lacZ. If β-galactosidase is present, the transformed bacteria will form blue colonies. If β-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)

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