InkBiology

ANSWER BOOK

Mutation and cancer: what goes wrong, and where it goes wrong

Gene regulation, genomics and biotechnology · Unit B17

AQA 3.8.1, 3.8.2.1, 3.8.2.3 · OCR A 6.1.1, 6.1.2 · Edexcel A Topic 3 · CAIE 6.2, 16.2

5 questions · 17 marks

NAVY EDITION

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SECTION A · Short, 1 to 2 marks

A1 **B1** a nonsense substitution, which ends translation early. **B1** the code is degenerate, so more than one triplet codes for the same amino acid. [2]

SECTION B · Standard, 3 to 5 marks

B1 **B1** the code is non-overlapping and read in triplets from a fixed starting point. [4]
B1 losing one base moves every base after it one place forward, so the reading frame is shifted and every triplet downstream is read from a new position. **B1** all the amino acids after the deletion are therefore potentially wrong, and a stop codon usually appears early by chance, so the polypeptide is shortened as well as scrambled and will not fold into anything. **B1** three is a multiple of three, so the frame is not shifted: one amino acid is removed and every triplet after it is read exactly as before.

B2 **B1** a proto-oncogene codes for a protein that stimulates cell division when a growth signal arrives, whereas a tumour suppressor gene codes for a protein that halts the cell cycle at a checkpoint or triggers apoptosis. **B1** a proto-oncogene does harm when a mutation makes it permanently active or present in too many copies, whereas a tumour suppressor gene does harm when its product is lost. **B1** one faulty allele is enough for an oncogene, because a permanently active protein acts whatever the other allele makes, whereas both alleles of a tumour suppressor gene must go, because one working copy still makes the protein. **B1** in both cases the outcome is the same: cells pass checkpoints they should not and divide repeatedly, so a mass of cells accumulates. [4]

B3 **B1** for: embryonic stem cells are pluripotent and can give rise to any cell type of the body, so they offer a range of treatment that multipotent adult stem cells cannot. **B1** for: the embryos are surplus from IVF and would otherwise be discarded, and many people hold that there is a moral difference between using such an embryo and creating one in order to destroy it. **B1** against: those who hold that a human embryo has the full moral status of a person from fertilisation onwards regard its destruction as the deliberate ending of a human life, which the good done with the cells does not license. **B1** judgement: British law takes a middle position, licensing such research through the Human Fertilisation and Embryology Authority up to fourteen days; induced pluripotent cells narrow the disagreement, since they are pluripotent and need no embryo, though they can carry mutations acquired in the adult cell and a tendency to form tumours. [4]

B4 **B1** with fewer methyl groups on the promoter, transcription factors and RNA polymerase can bind to it again, so the gene is transcribed and mRNA is made. **B1** the tumour suppressor protein is then synthesised and the checkpoint it enforces is applied again, so cells with damaged DNA are held for repair or sent into apoptosis instead of dividing; no base had changed, so nothing needed repairing. **B1** the drug reduces methylation across the whole genome and in every cell it reaches, so genes that were properly silenced in healthy cells are switched on as well. [3]