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ANSWER BOOK

# Homeobox genes, Hox genes and apoptosis

Cell cycles, reproduction and development · Unit B06

OCR A 6.1.1

15 questions · 45 marks

NAVY EDITION

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## SECTION A · Short, 1 to 2 marks

**A1** HG-Q7

**B1** the homeobox is 180 base pairs long. **B1** the homeodomain is 60 amino acids long, because three bases code for one amino acid. [2]

**A2** HG-Q14

**B1** colinearity is the match between the order of the Hox genes along the chromosome and the order of the body regions they specify, from head to tail. **B1** a human has four Hox gene clusters, on four different chromosomes. [2]

**A3** HG-Q15

**B1** the gene is **p53**. **B1** it is a tumour suppressor gene. [2]

## SECTION B · Standard, 3 to 5 marks

**B1** HG-Q1

**B1** enzymes break down the cytoskeleton, so the cell shrinks and the cytoplasm becomes dense with closely packed organelles. **B1** the chromatin condenses, the nuclear envelope breaks down and the DNA is cut into fragments. **B1** the cell surface membrane blebs, bulging outwards without breaking, and the cell separates into apoptotic bodies each wrapped in membrane. **B1** the apoptotic bodies are engulfed by phagocytosis and their contents are digested and reused. [4]

**B2** HG-Q2

**M1** three bases code for one amino acid, so divide 180 by 3. **A1** 60 amino acids. **M1** 180 divided by 1200, multiplied by 100. **A1** 15 per cent. [4]

**B3** HG-Q8

**B1** the legs are complete and normally formed, so the genes that build a leg are intact and functioning correctly. **B1** what has changed is where those leg-building genes are expressed: they are switched on in the head region instead of the region that normally specifies legs. **B1** this shows that a Hox gene's role is to specify the identity of a region, activating the developmental programme appropriate to it, rather than to build any structure directly. **A1** it is called a homeotic mutation because one region develops with the identity of another region, rather than because any structure has been lost or deleted. [4]

**B4** HG-Q3

**B1** the homeodomain binds to DNA, so the protein coded for by a homeobox gene is a transcription factor. **B1** it controls the transcription of many other genes, and those genes are the ones that build the structures of the region. **A1** a change to the transcription factor therefore changes which set of genes is switched on in every cell of that region, so the region can develop with the identity of a different region. [3]

**B5** HG-Q4 [3]

**B1** a cell undergoing apoptosis shrinks, whereas a cell undergoing necrosis swells. **B1** in apoptosis the cell surface membrane stays intact and wraps every fragment, whereas in necrosis the membrane breaks and the cell contents escape. **B1** apoptosis is controlled by signals and by the cell's own enzymes and causes no inflammation, whereas necrosis follows injury or a shortage of oxygen or nutrients and damages neighbouring cells, causing inflammation.

**B6** HG-Q5 [3]

**B1** mitosis produces the cells of the whole hand plate, including the tissue lying between the developing digits. **B1** the cells of the tissue between the digits receive a signal and undergo apoptosis, and the apoptotic bodies are removed by phagocytosis. **A1** the five digits are left free of one another; the number of digits is unchanged, because it is the webbing between them that is removed.

**B7** HG-Q6 [3]

**B1** a cell with DNA damage that cannot be repaired is normally removed by apoptosis, so that the damage is not passed on. **B1** if apoptosis does not happen the damaged cell survives and continues through the cell cycle, and its daughter cells inherit the damage. **B1** further mutations accumulate in those descendants, the rate of division exceeds the rate of cell death, and a mass of cells builds up.

**B8** HG-Q9 [3]

**B1** it shows that the two species inherited the homeobox sequence from a shared common ancestor that already possessed it. **B1** it shows the sequence and its function have been conserved across an enormous span of evolutionary time, since a fly protein and its distant mammalian relative can still substitute for one another. **A1** almost any mutation to a sequence this central to development is likely to be harmful, so individuals carrying it are strongly selected against and the mutation is rarely passed on.

**B9** HG-Q10 [3]

**B1** external signals include cytokines released by cells of the immune system, hormones, growth factors and nitric oxide, and either the delivery of one of these or, for a growth factor, its withdrawal can initiate apoptosis. **B1** internal signals arise from the cell's own condition, and the example to give is DNA damage that the cell's repair systems cannot correct. **A1** both kinds of signal converge on activating the same enzymes, which then carry out the same ordered sequence of apoptosis regardless of which signal started it.

**B10** HG-Q11 [3]

**B1** the tissue between the developing digits is normally removed by apoptosis, in strips, as the hand plate takes shape. **B1** if the cells of that tissue do not receive, or cannot respond to, the signal to undergo apoptosis, they are not removed and remain in place. **A1** the digits themselves have formed normally and are present in the correct number; only the removal of the tissue joining them has failed.

**B11** HG-Q12

**M1** number per year = number per day × 365. **M1** $1 \times 10^{10} \times 365$. **A1** $3.7 \times 10^{12}$ cells replaced in a year (accept $3.65 \times 10^{12}$). [3]

**B12** HG-Q13

**B1** too little apoptosis lets a cell with DNA damage it cannot repair go on dividing, whereas too much apoptosis destroys cells, such as neurones, that are healthy and often cannot be replaced. **B1** too little apoptosis, combined with unchecked division, is one route to a tumour building up from the damaged cell's descendants, whereas too much apoptosis of neurones contributes to neurodegenerative disease and to the tissue damage seen around a stroke. **A1** both show that apoptosis must be matched to mitosis: neither too little nor too much keeps a tissue at a stable, healthy size. [3]

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