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— **TUITION** —

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BIOLOGY

THE CONTROL OF GENE EXPRESSION

Level & Board	AQA (A-LEVEL)
TOPIC:	GENE MUTATIONS
PAPER TYPE:	SOLUTION - 2
TOTAL QUESTIONS	6
TOTAL MARKS	31

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Gene Mutation - 2

1.

(a)

$$1280 - 560 = 720$$

$$720/4$$

=180 cells killed per unit volume of blood per day

(b)

Similarities

Same/similar pattern/both decreases, stay the same then increase

Number of cells stays the same for same length of time

Differences

Greater/faster decrease in number of healthy cells/more healthy cells killed/healthy cells killed faster

Greater/faster increase in number of healthy cells/more healthy cells replaced/divide/healthy cells replaced/divide faster

(c)

More/too many healthy cells killed

So will take time to place/increase in number

Person may die/have side effects

2.

(a) 250 000

3.

(a) Missense mutations cause a single amino acid change in the protein. Nonsense mutations make a premature "stop" codon. Any codons after that are not translated, and the resulting protein is missing amino acids.

OR

Loss of 3 bases / triplet

Loss of base(s)

(b) In Ellis-van Creveld syndrome, the faulty allele leads to a mutation in the EVC gene, resulting in the production of a protein with one amino acid missing. This missing amino acid disrupts the normal folding of the protein, altering its structure and potentially impairing its function.

OR

Change in tertiary structure / active site

So faulty / non-functional protein / enzyme

4.

(a)

Cell wall not formed

Lower water potential in bacterium;

Water enters via osmosis and causes osmotic lysis

(b) Human cells lack enzyme B

(c) The base sequence changes so the amino acid sequence changes therefore leading to a change in the types and location of bond formation thus a change in the tertiary structure occurs.

OR

Mutation causes a change in base sequence of gene which leads to a change in amino acid sequence

This changes the hydrogen and ionic bonds which changes the tertiary structure

Substrate is no longer complementary to enzyme active site so no enzyme-substrate complexes are formed

5.

(a) 4

(b) Mutations in PAH lead to impaired function of the hepatic enzyme PAH (EC 1.14.16.1), which catalyzes the conversion of the essential amino acid l-phenylalanine (l-Phe) to l-tyrosine (l-Tyr), a precursor of the neurotransmitter's dopamine, noradrenaline and adrenaline.

OR

Change in amino acid / (sequence of) amino acids / primary structure

Change in hydrogen / ionic / disulphide bonds alters tertiary structure / active site of enzyme

Substrate not complementary / cannot bind (to enzyme / active site) / no enzyme- substrate complexes form

6.

(a)

Lack of skin pigment so pale skin

Lack of muscle coordination

(b) Migration and interbreeding

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I am Sorry !!!!!



DR. ASHAR RANA



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