

Biology OCR A -Exam question s-answers on Cellular control chapter 19

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**1** The development of lactose tolerance is thought to have spread over approximately 20 000 years, which in evolutionary terms is very quick.

Explain why the percentage of adults with the ability to digest lactose increased at such a rate.

**2** Outline why the majority of mutations do not have an influence on phenotype. (2 marks)

**3** Discuss why beneficial mutations are rare and suggest a process that underpins beneficial mutations. (4 marks)

**1** Ability to digest lactose is a beneficial characteristic (1); drinking milk prevented starvation (1); reduced osteoporosis (1); directional selection (1).

**2** Mutation is random (1); majority of DNA is non-coding; mutations more likely to occur in non-coding regions (1); mutations in non-coding regions do not affect phenotype (1);

**3** Majority of mutations are silent (1); *idea that* random change to protein structure is more likely to reduce function (1); *idea that* beneficial mutations increase chances of survival if environment changes (1); surviving organisms reproduce and pass new alleles to offspring (1); leading to evolution (1).

- 1** The lac operon is often referred to as being 'leaky' meaning that it is still transcribed to a limited extent even in the absence of lactose.
- a** Using your knowledge of how the lac operon works, explain why this is necessary. (3 marks)
- b** Suggest the functions of  $\beta$ -galactosidase and lactose permease synthesised by the lac operon. (3 marks)
- 2** Another example of gene regulation in prokaryotes is the trp operon. This operon codes for the production of tryptophan, an essential amino acid for the bacterium *E. coli*. When tryptophan is available in the environment the structural genes in the trp operon are not expressed. Suggest a mechanism for the genetic regulation of this operon. (5 marks)
- 3** Using your knowledge of enzymes, explain how enzyme cofactors could play a role in gene regulation. (4 marks)

**1a** enzyme coded for by lac operon enables lactose to enter bacteria (1); lactose binds to repressor protein; (repressor) protein changes shape (1); transcription no longer blocked (1); enzymes needed to metabolise lactose are synthesised (1) (3 max).

**b**  $\beta$ -galactosidase catalyses the hydrolysis of lactose (1); to galactose and lactose (1); lactose permease enables the entry of lactose into cells (1).

**2** Tryptophan binds to repressor protein (1); shape of repressor protein changes (1); repressor protein binds to promoter (1); blocks RNA polymerase from binding (1); transcription prevented (1); of genes coding for enzymes responsible for tryptophan synthesis (1) (5 max).

**3** Cofactors bind to proteins that regulate transcription (1); changes binding of proteins to control elements (1); rate of transcription changed (1); RNA polymerase activated (1).

**1** Explain, with reference to their body shape, why human beings are referred to as bilaterally symmetrical but jellyfish are radially symmetrical. (3 marks)

**2** The Hox gene Pax6 is necessary for the normal development of the retina in humans. A mutation in this gene can lead to blindness. Pax6 mutations also cause blindness in mice and fruit flies. Describe how scientists could have tested the idea that Pax6 plays a role in eye development in all three species. (5 marks)

**3** Consider the statement.

*All Hox genes are homeobox genes but not all homeobox genes are Hox genes.*

Discuss the validity of this statement. (5 marks)

**1** Bilateral symmetry is along (single plane through) central axis (1); e.g., two arms, two legs (1); radial symmetry is along a plane at any angle through central axis (1); e.g., tentacles around central axis (1).

**2** Isolate (Pax6) gene from one species (1); detail e.g., PCR, restriction enzymes (1); test in different, tissue/species (1); example of positive result e.g., eyes develop on legs (1); switch gene off early in development and eyes will not develop (1); DNA sequencing and compare genes from different species (1) (max 5).

**3** Statement is valid (1); Hox genes are one form of homeobox gene (1); present in vertebrates (1); in Hox clusters (1); other forms of homeobox gene present in other clusters (1).

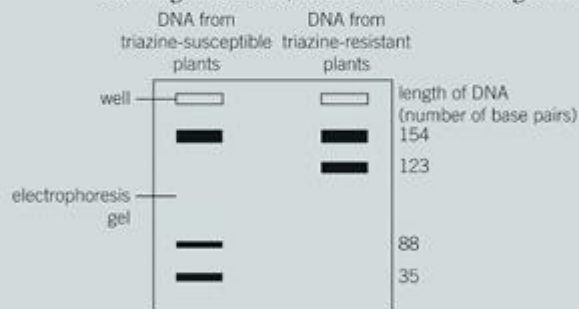
- 1** The common groundsel, *Senecio vulgaris*, is a weed that is often found in large numbers on cultivated land. It was the first plant species to develop resistance to triazine herbicides. This resistance is the result of a gene mutation in the chloroplast DNA.

Since its first appearance, triazine resistance has spread very rapidly in groundsel populations.

- a** Explain the rapid spread of herbicide resistance in a weed such as groundsel. (5 marks)

- b** DNA was extracted from the chloroplasts of triazine-susceptible and triazine-resistant groundsel plants. Equivalent lengths of DNA, including the site of the mutation, were isolated from each extract and then treated with the restriction enzyme *MaeI* prior to electrophoresis.

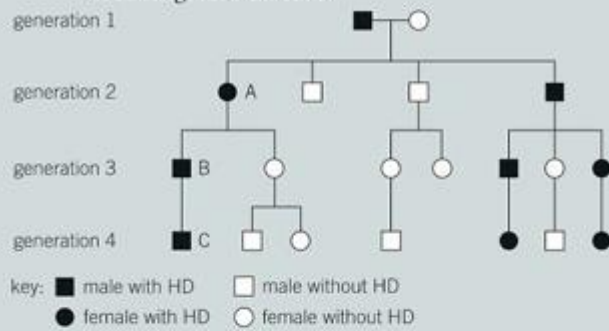
The resulting electrophoresis gel, after staining the DNA, is shown in the diagram.



- (i) State, giving a reason, whether the mutation giving resistance to triazine is a deletion, a substitution or an addition. (2 marks)
- (ii) Explain the difference in banding pattern between DNA from triazine-susceptible and triazine-resistant plants. (2 marks)
- (iii) Suggest one way in which the mutation could give resistance to triazine. (2 marks)

1 (a)	<p>Herbicide, is selective agent / exerts selective pressure;  natural selection;  resistants have, selective advantage / AW; e.g. competitive advantage  resistants survive / susceptibles die;  (more) resistants (reproduce and) pass, mutation / allele / trait, to offspring;  increasing frequency of, mutation / allele (in population); R gene  (common weed so) sprayed in many different places / AW;  (common weed so) large number of seeds / rapid spread / many generations in a year;  (weed so likely to have) good dispersal mechanism / described;  ref to, large number of loops of DNA in chloroplasts / large number of chloroplasts, so greater chance of, replication error / mutation;</p>
1 (b) (i)	<p>Substitution;  no missing base pair / <math>88 + 35 = 123\text{bp}</math> / <math>88 + 35 + 154 = 123 + 154 \text{ bp}</math>  (277bp);</p>
1 (b) (ii)	<p>(mutation) removes / no, target site;  for the restriction enzyme;  so 123 bp fragment is not cut in two (88 + 35);</p>
1 (b) (iii)	<p>Mutation A <i>ecf</i> from (i)</p> <p>ref to (mutation) altering DNA triplet code;  (so) codes for different amino acid;  (so) primary / tertiary, structure of protein different;  or codes for premature stop triplet;  incomplete / no, protein produced;  AVP;</p> <p><i>effect in terms of gene product / protein / enzyme / transcription factor / AW</i>  (altered) protein no longer, binds / inactivated by, triazine / AW; A fits  (altered) protein no longer allows triazine through membrane;  (altered) protein inactivates triazine;  (altered) enzyme breaks down triazine; A denatures  AVP; e.g. ref to different metabolic pathway (so triazine not effective)</p>

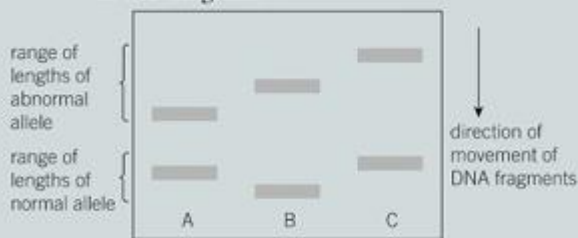
**2 a** The diagram shows a family's history of Huntington's disease.



Explain how the diagram provides evidence that Huntington's disease results from the inheritance of an autosomal dominant allele. (3 marks)

- b** Genetic screening for Huntington's disease can be carried out, using a process similar to genetic fingerprinting, to find the length of a repeated triplet, or 'stutter', in an allele.
- c** DNA from individuals A, B and C from the family shown in the diagram was analysed.

The resulting banding patterns are shown in the diagram.



Explain why the DNA of the following bands in the diagram are not the same length:

- (i) the three normal alleles (1 mark)
- (ii) the three abnormal alleles. (1 mark)

2 (a)	<p><i>Autosomal / not sex-linked, because approximately equal numbers of male (5) and female (4) sufferers; appears in every generation / no alternate generation pattern; male passes trait to son; Dominant / cannot be recessive, because</i></p>
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only inherited from sufferer; ORA  
 approximately equal numbers of sufferers and non-sufferers in each generation/ora expect smaller numbers if recessive;  
 e.g. 2 sufferers and 2 non-sufferers in offspring of sufferers in generation 2  
 ;  
 e.g. 3 sufferers and 2 non-sufferers in offspring of sufferers in generation 3

2 (b) (i)	cuts DNA into, pieces / fragments / AW; at specific sites; close to, but not in stutter; detail of site; e.g. 4-6 base pairs, palindromic
2 (b) (ii)	negatively charged; detail; e.g. phosphate groups attracted to anode; smaller fragments travel further (towards anode) / ORA; smaller fragments have less impedance / AW / ORA;
2 (c) (i)	inherited from three different individuals/ each parent (unaffected parents of A, B and C);
2 (c) (ii)	length of stutter increases in each generation/ C longer than A/different sizes of stutter:

**3** Homeobox genes show astonishing similarity across widely different species of animal, from fruit flies, which are insects, to mice and humans, which are mammals. The sequences of these genes have remained relatively unchanged throughout evolutionary history and the same genes control embryonic development in flies and mammals.

**a** State what is meant by a homeobox gene. (2 marks)

**b** Homeobox genes show 'astonishing similarity across widely different species of animal'.

Explain why there has been very little change by mutation in these genes.

(2 marks)

**c** Frogs reproduce by laying eggs in water, each egg develops into a tadpole, which has external gills to extract oxygen from the water, and a tail to help it swim. The tadpole gradually changes into an adult frog as it grows. During this time its gills and tail disappear.

List two cellular processes that must occur during the development of a tadpole into a frog. (2 marks)

**d** Name another kingdom of organisms, other than animals, that have similar homeotic genes. (1 mark)

3 (a)	homeotic / regulatory, (gene); contains, 180 bp / homeobox, sequence; that codes for homeodomain (on protein); (gene product) binds to DNA; initiates transcription / switch genes, on / off; control of, development / body plan;	2 max
3 (b)	these genes very important; mutation would, have big effects / alter body plan; many other genes would be affected / knock-on effects; mutation likely to be, lethal / selected against;	2 max
3 (c)	protein synthesis / transcription and translation; respiration; DNA replication; mitosis; cytokinesis; apoptosis; differentiation / gene switching;	2 max

4 An enhancer of a regulatory gene responsible for limb development in mammals, called the sonic hedgehog gene, is located in the intron of a neighbouring gene. This regulatory gene was first investigated using *Drosophila* – genetically modified flies. Without this gene the fruit flies grew small projections (known as denticles) all over their bodies.

Point mutations in the enhancer sequence for this gene result in polydactyly, extra fingers or toes.





The photograph and X-ray show a child's foot with polydactyl – a deformity in which more than the usual number of digits are present. The condition is genetic in origin, and in most cases causes no harm. The extra digits are often underdeveloped (those at lower left in this case), and if removed surgically soon after birth cause no long term complications.

- a** Explain how an enhancer works. (2 marks)
- b** State the meaning of the term intron. (1 mark)
- c** Describe what is meant by a point mutation. (2 marks)
- d** Explain why the sonic hedgehog gene is an example of a Hox gene. (2 marks)
- e** The sonic hedgehog gene is usually expressed in cells close to tissue that eventually develops into the small fingers or toes.  
Suggest how different rates of transcription of this gene leads to the formation of fingers with different sizes and shapes. (3 marks)

4 (a)	Increase expression of gene; bind transcription factors / aid binding of RNA polymerase to promotor;	2
4 (b)	Section of non-coding DNA.	1
4 (c)	Change in single, base / nucleotide; e.g. substitution;	2
4 (d)	Regulatory gene; involved in body development; hedgehog is a mammal;	2 max
4 (e)	<i>Idea that</i> rate of transcription determines enzyme production; enzymes required for protein synthesis; enzymes required for respiration; energy / proteins, required for growth;	3 max