Student Name: _____

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BIOLOGY 2016

Unit 4
Key Topic Test 2 – Mutations

Recommended writing time*: 45 minutes
Total number of marks available: 45 marks

QUESTION BOOK

* The recommended writing time is a guide to the time students should take to complete this test. Teachers may wish to alter this time and can do so at their own discretion.

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Conditions and restrictions

- Students are permitted to bring into the room for this test: pens, pencils, highlighters, erasers, sharpeners and rulers.
- Students are NOT permitted to bring into the room for this test: blank sheets of paper and/or white out liquid/tape.
- No calculator is permitted in this test.

Materials supplied

- Question and answer book of 14 pages.

Instructions

- Print your name in the space provided on the top of the front page.
- All written responses must be in English.

Students are NOT permitted to bring mobile phones and/or any other unauthorised electronic communication devices into the room for this test.

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SECTION A – Multiple-choice questions

Instructions for Section A

Select the response that is **most correct** for the question. A correct answer scores 1, an incorrect answer scores 0. Marks are not deducted for incorrect answers. If more than 1 answer is completed for any question, no mark will be given.

Question 1

Which of the following is true of mutations?

A. They occur at a very fast rate in species that are becoming extinct.

B. They give rise to greater amounts of genetic variation within a population

C. Decrease in frequency in populations that are changing rapidly

D. Are a beneficial component of populations

Question 2

Correctly identify the substitution mutation in the following examples.

A. Fragile-X syndrome individuals have larger amounts of CGG repeats present on their X chromosome

B. Tay-Sachs individuals have a whole section of DNA that is not utilised due to a change in genetic code.

C. A nucleotide is added to the genetic sequence of an individual to result in Huntington's disease

D. Sickle cell anaemia is caused by a change in codon from GAG to GTG

Question 3

Which of the following mutations would be the least likely to be passed onto offspring?

A. A female with the mutation copulates with a male without the mutation.

B. A female is exposed to high doses of radiation that causes a mutation in germline cells

C. A nucleotide is added to the genetic sequence of an individual to result in Huntington's disease, a genetic disorder.

D. A known mutagenic agent found in ground water was consumed by individuals in a community from birth and some individuals have had issues with producing healthy offspring.

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Question 4

Which of the following would best describe a point mutation that leads to two different variations of a particular gene?

A. A single nucleotide polymorphism

B. A female is exposed to high doses of radiation that causes a mutation in germline cells

C. A nucleotide is added to the genetic sequence of an individual to result in Huntington's disease

D. A point mutation results in two different alleles being expressed at the same time

Please use the following information to answer questions 5 and 6

The following chromosome diagrams show particular genes labelled from A to L in alphabetical order.

A. 

B. 

C. 

D. 

Question 5

Which of the following diagrams best represents a duplication mutation?

A. A

B. B

C. C

D. D

Question 6

Two of the diagrams represent the same type of mutation. Which two diagrams are these?

A. A and B
B. A and D
C. B and C
D. C and D

Question 7

Which of the following is incorrect of all types of mutations?

A. They all involve changes to a genetic sequence
B. They are more likely to result in a change that is detrimental to the cells function
C. They do not increase the variation in a gene pool
D. The majority of new mutations found in organisms are spontaneous.

Question 8

The scientific testing of how mutations arise is integral to developing new understandings in the evolution of species and the changes to populations over time. Which of the following organisms would be the best to use in a genetic study on variation changes over time due to mutations within a single population.

A. Bacteria
B. Humans
C. Drosophila (flies)
D. Grass seeds

Please use the following information to answer questions 9 and 10

A mutation involving a deletion of two codons was found in a plant with a particular enzyme condition. The plant was no longer able to produce viable flowers that allowed reproduction.

Question 9

How many bases were removed due to the deletion?

A. 2
B. 3
C. 6
D. 12

Question 10

The deletion would have the greatest effect if it was found in which of the following positions?

A. The middle of the gene
B. The end of an exon
C. The end of an coding sequence
D. The start of an intron

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Question 11

The human genome is littered with many tandem repeats of nucleotide bases. Fragile X Syndrome has above normal repetition of CGG repeats at the *FRAXA* site on the X chromosome leading to around 1000 copies being produced in comparison to a normal X chromosome having around 40 copies. Fragile X syndrome would best be described as:

A. A mutation involving replication and relocation of chromosome segments
B. An expanded segment of trinucleotide repeats on the X chromosome
C. A deletion of nucleotide bases on the sex chromosomes that leads to a frameshift mutation
D. A point mutation that is masked by the repeats of the nucleotide bases as it still forms the same protein

Question 12

Blue eyes is widely acknowledged among the scientific community as a mutation that has shaped the evolution of humans that we see today. In which of the following is this mutation most likely to be shown?

A. Two recessive individuals for blue eyes reproduce
B. Two heterozygous individuals for blue eyes reproduce
C. Two homozygous individuals for brown eyes reproduce
D. A heterozygous individual and a homozygous recessive individual reproduce

Question 13

Identify which of the following is not an example of a mutagenic agent.

A. Increased UV radiation during DNA replication
B. Alkylating agents that mutate DNA during both replication and resting phases
C. Denaturation of DNA strands during meiosis that re-join at a different position
D. Carcinogenic compounds found in soot.

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SECTION B- Short-answer questions

Instructions for Section B

Answer all questions in the spaces provided.

Question 1 (12 marks)

Sickle cell anaemia is a disease that occurs due to mutations present in an individual's DNA code. The change to the code results in different amino acid sequence being produced and haemoglobin produced to have a malformed shape. Individuals with the disease have various symptoms due to the inability of the haemoglobin to effectively carry oxygen to the rest of the body. Whilst the disease can be fatal, these individuals do have a selective advantage in that they cannot acquire the mosquito borne virus malaria.

The following sequence of DNA was obtained from an individual without sickle cell anaemia

DNA	ATG	GTG	CAC	CTG	CCT	CCT	GAG	GAG	AAG	TCT	GCC	GTT	ACT
mRNA													
Amino acid													

The following sequence of DNA was obtained from an individual with sickle cell anaemia.

DNA	ATG	GTG	CAC	CTG	CCT	CCT	GTG	GAG	AAG	TCT	GCC	GTT	ACT
mRNA													
Amino acid													

a. Using the amino acid table provided below, write the correct mRNA and amino acid sequence for both individuals above.

4 marks

	U		C		A		G		
	code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid	
U	UUU	phe	UCU	ser	UAU	tyr	UGU	cys	U
	UUC		UCC		UAC		UGC		C
	UUA	leu	UCA		UAA	STOP	UGA	STOP	A
	UUG		UCG		UAG	STOP	UGG	trp	G
C	CUU	leu	CCU	pro	CAU	his	CGU	arg	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	gln	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	ile	ACU	thr	AAU	asn	AGU	ser	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	lys	AGA	arg	A
	AUG	met	ACG		AAG		AGG		G
G	GUU	val	GCU	ala	GAU	asp	GGU	gly	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	glu	GGA		A
	GUG		GCG		GAG		GGG		G

b. Identify the type of mutation found in individuals with sickle cell anaemia.

_____

1 mark

c. Explain why this same type of mutation identified in part b sometimes leads to no change in the protein produced in other circumstances.

_____

_____

_____

2 marks

d. What is the name given to the type of mutation outlined in part c?

_____

1 mark

e. Explain how the sickle cell anaemia mutation contributes to the process of evolution.

_____

_____

_____

1 mark

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f. Outline why mutations are an important factor in the evolution of a species?

2 marks

g. Given that sickle cell anaemia is a mutation that has been passed from one generation to the next as a recessive condition. What type of cells must have been affected by the mutation originally?

1 mark

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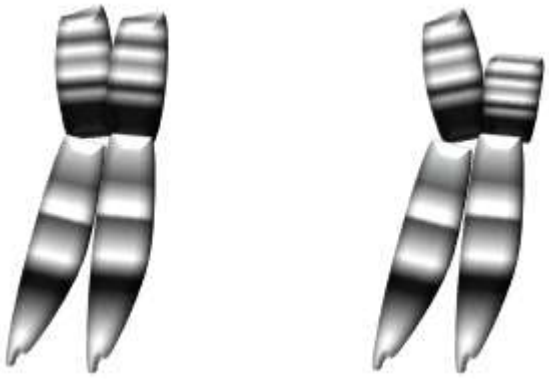
Question 2 (8 marks)

Cri du chat syndrome is a chromosomal disorder effecting chromosome number 5. It has a range of symptoms that vary from individual to individual that include intellectual disability, speech delay and motor skill impairments. There is no cure for this condition, however, various physical treatments are able to assist the quality of life for those affected.

The following is a diagrammatic representation of the effect of the chromosome at a microscopic level.



a. Identify the type of mutation found in individuals with Cru du chat syndrome.

_____

1 mark

b. Outline why the effect of the mutation that leads to Cru du chat syndrome results in further changes to the production of amino acids found in the same region as the mutation.

_____

_____

_____

2 marks

c. Give an example of another condition that involves mutation of chromosomes and identify what type of mutation is present.

_____

_____

_____

2 marks

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d. Would it be expected that individuals with Cru du chat syndrome who are able to reproduce, would also have offspring that also have the condition? Explain

1 mark

e. How might undergoing genetic testing at an early age assist individuals living with symptoms of Cri du chat syndrome?

2 marks

Question 3 (10 marks)

Scientists investigating the embryonic development in *Drosophila* (flies) undertook experiments that involved determining the cause of mutant flies that exhibited unusual developmental defects. These flies often had body parts located in abnormal positions or additional appendages such as legs. The genes studied controlled the segmentation of the fly into its appropriate order, head, abdomen then anterior end. It was found that *homeotic* genes are responsible for the correct formation of body segments with appropriate appendages. The mutant versions of these genes can result in the wrong location of appendage position.

a. The homeotic genes have a 60 amino acid region that assists in the control of non-mutant drosophila.

i. How many codons are required to produce this region?

_____

1 mark

ii. How many bases are required to produce this region?

_____

1 mark

The following amino acid sequence was found in a non-mutant drosophila.

It was found that a point mutation in the second amino acid in the sequence above was responsible for the position of the hind legs on the abdomen.

b. Using the amino acid table attached outline all possible codons for the production of this amino acid.

_____

1 mark

c. It was found that the middle base for this codon was replaced with an A.

i. Name the type of point mutation in this scenario.

_____

1 mark

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ii. Outline the possible consequences for the polypeptide created by this sequence.

_____

_____

_____

2 marks

iii. Name two other types of point mutations.

_____

1 mark

d. It was determined that whilst these drosophila were not viable in producing their own offspring, the mutation arose in the parental gametes.

i. What type of mutation must have occurred in the parent?

_____

1 mark

ii. What are the possible agents for the mutation occurring?

_____

1 mark

iii. In which process did the mutation originally occur in?

_____

1 mark

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Question 4

Bowel polyps are an indicator of a precancerous condition that if unattended can lead to bowel cancer. They are found within the intestine and some types can be associated with a single dominant gene. It was found that 10 out of 30 individuals from a small farming town were found to have these precancerous polyps within their lower intestines at some point in their lives.

Explain why there may not have been any inquiry into possible mutagenic agents within the small farming town and what further investigations could take place to out rule a mutagenic agent being the cause.

2 marks

END OF KEY TOPIC TEST

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