CHEMISTRY ONLINE
TUITION

Phone: +442081445350

www.chemistryonlinetuition.com

Email:asherrana@chemistryonlinetuition.com

BIOLOGY

THE CONTROL OF GENE EXPRESSION

Level & Board	AQA (A-LEVEL)
TOPIC:	GENE MUTATIONS
PAPER TYPE:	QUESTION PAPER - 1
TOTAL QUESTIONS	6
TOTAL MARKS	25

Gene mutations - 1

1.

A class of hereditary illnesses is known as sickle cell disease (SCD). Red blood cells in SCD patients have a sickle form to them. One kind of SCD can result from a mutation in a single base substitution. The beta polypeptide chains in hemoglobin undergo structural alteration as a result of this mutation.

(a) Describe how this polypeptide's structure changes as a result of a single base substitution.

Don't include in your response any information on transcription and translation. **(3)**

2.

Within mitochondria lies a little circular DNA molecule known as mitochondrial DNA (mtDNA). It has 37 genes, a regulatory region, and 16 569 nucleotides in length.

Sports scientists looked into the possibility that a mutation in the mtDNA regulatory region in male humans might be linked to a longer exercise tolerance.

• Thymine was present at nucleotide position 16 519 in the males in Group T.

• Group C's male members possessed a mutation that produced cytosine at nucleotide position 16 519.

(a) Group T and Group C control areas were of same length.

At nucleotide position 16 519, identify the kind of gene mutation that is most likely to have happened. **(2)**

3.

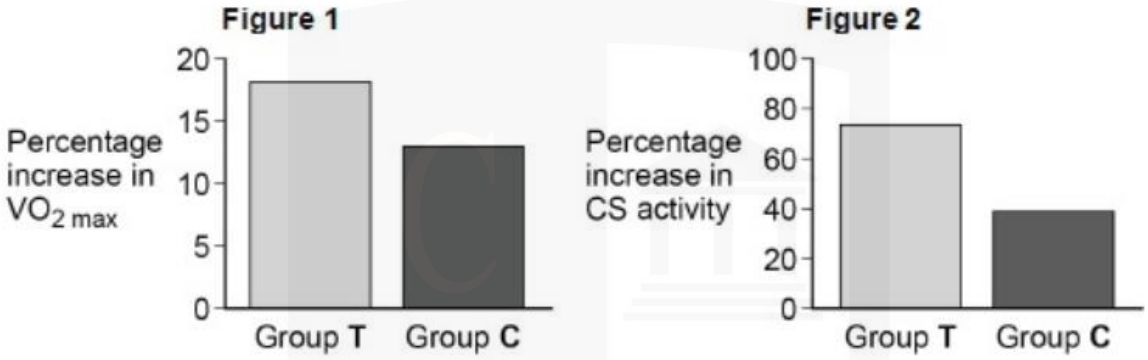
Groups T and C underwent an identical eight-week training program. The ensuing measures were obtained both at the beginning and the conclusion of the eight-week program.

1. VO_2 max, or maximal oxygen uptake, is measured.

2. The activity of the enzyme citrate synthase (CS), which is a component of the Krebs cycle.

The percentage rise in each measurement for both groups was then determined by the scientists.

Their findings are shown in Figures 1 and 2.



(a) A student concluded from Figure 1 and Figure 2 that training has a positive effect on VO_2 max and CS activity.

Evaluate the student's conclusion**. (3)**

(b) The non-coding portion of the mitochondrial DNA (mtDNA) is known as the regulatory region. This area promotes the synthesis of mitochondrial messenger RNA and mtDNA.

Using the information provided, propose two scenarios in which the variations shown in Figure 2 could result from the mutation at nucleotide position 16-519. **(2)**

4.

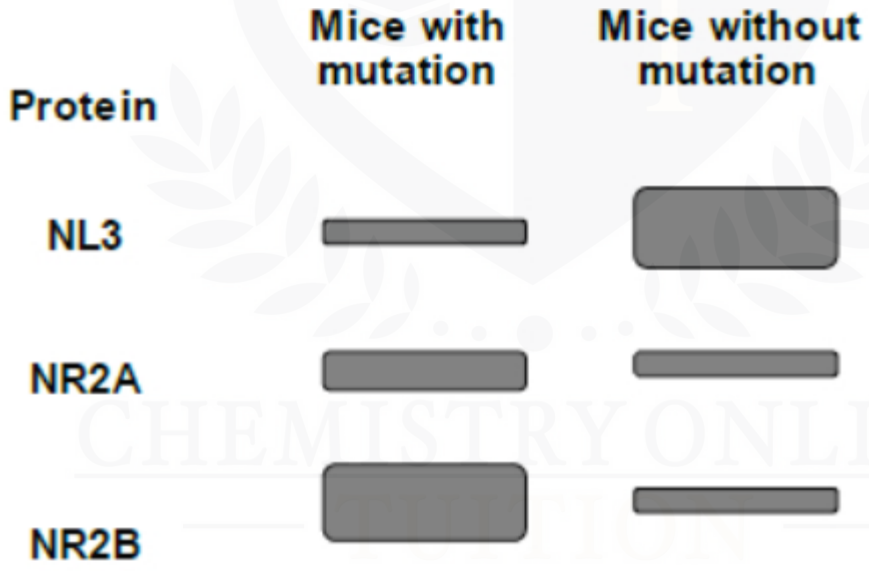
There is a connection between some autism spectrum disorders (ASDs) and a neuroligin-3 gene mutation. This gene produces the NL3 protein, which is present in synapses.

Researchers looked into the consequences of a mutation in mice that affects NL3. Both mice with and without the mutation's brains were obtained by them.

For every kind of mouse, they obtained a solution made up of all the proteins from synapses in a particular area of the brain; used gel electrophoresis to separate the proteins; and used three different labelled antibodies to identify and quantify the amount of three proteins in the solution.

Parts of a postsynaptic membrane receptor comprise the three proteins.

The scientists' findings are depicted in the diagram below. Every band indicates the existence of a protein. The amount of protein present is indicated by the size of a band.



(a) These mice's NL3 mutation was caused by a replacement in the neuroligin-3 gene.

Describe substitution mutation? **(2)**

(b) What information does this data provide regarding how the mutation affects the proteins? **(2)**

I am Sorry !!!!!

(c) These proteins are a component of a receptor that is present in hippocampal synapses. A high NR2B to NR2A protein ratio in this receptor has been linked to memory function.

Make a suggestion on the potential effects of the NL3 protein mutation on a mouse based on the facts provided. **(2)**

5.

(a) Describe the relationship between the functions of DNA and its structure.

(6)

6.

Researchers looked at the three genes C, D, and E that regulate cell division. They investigated how these genes' alterations affected a person's chance of getting lung cancer.

The genes C, D, and E from lung cancer patients and healthy individuals were examined by the investigators.

• The symbol N was used to represent a person who carried a normal allele for a gene.

• A person used the symbol M if they carried two mutant alleles for a given gene.

They computed the risk of lung cancer for individuals with various combinations of the N and M gene alleles using their data. When the risk number is 1.00, there is no elevated risk. The scientists' findings are displayed in the table below.

Gene C	Gene D	Gene E	Risk of developing lung cancer
N	N	N	1.00
M	N	N	1.30
N	N	M	1.78
N	M	N	1.45

N = at least one copy of the normal allele is present
M = two copies of the mutant allele are present

(a) In terms of the relative contribution of the mutant alleles of genes C, D, and E to raising the risk of lung cancer, what do these data suggest? Give an explanation for your response. **(3)**

DR. ASHAR RANA

CHEMISTRY ONLINE TUITION

Phone: +442081445350
www.chemistryonlinetuition.com
Email: asherrana@chemistryonlinetuition.com

- Founder & CEO of Chemistry Online Tuition Ltd.
- Tutoring students in UK and worldwide since 2008
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CONTACT INFORMATION FOR **CHEMISTRY ONLINE TUITION**

- UK Contact: 02081445350
- International Phone/WhatsApp: 00442081445350
- Website: www.chemistryonlinetuition.com
- Email: asherrana@chemistryonlinetuition.com
- Address: 210-Old Brompton Road, London SW5 OBS, UK