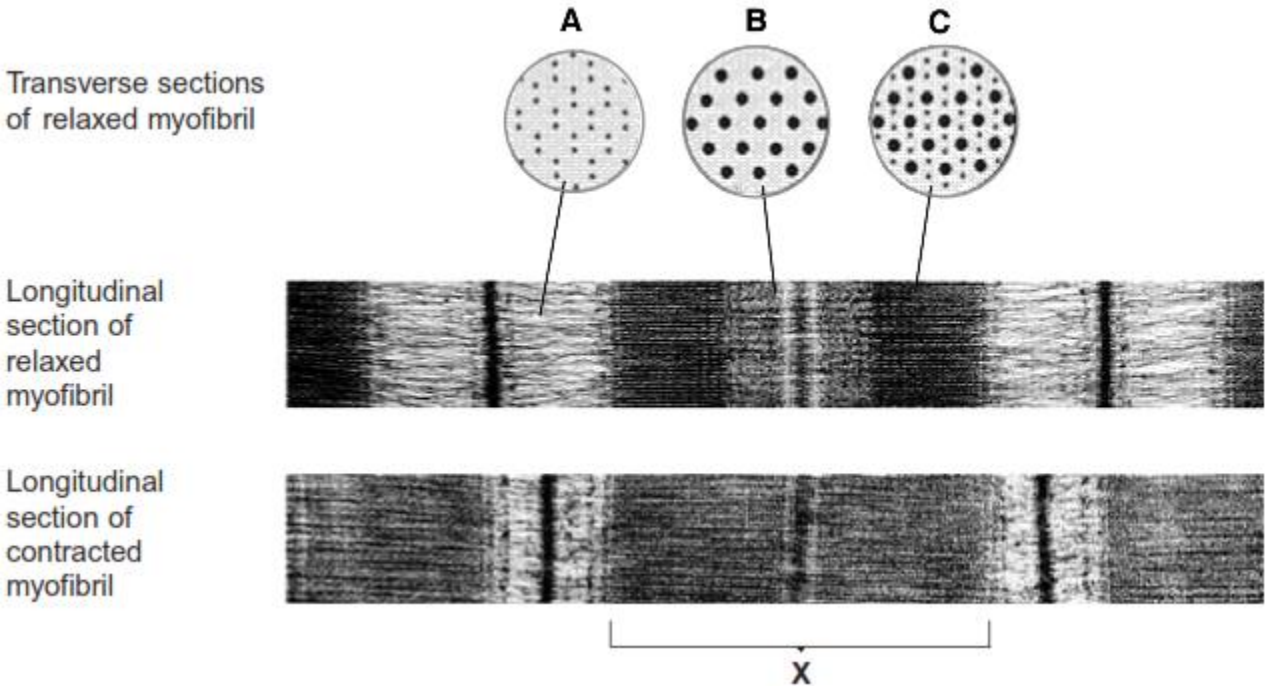
Name of the Student:_____
Max. Marks : 25 Marks **Time : 25 Minutes**

Q1.

Figure 1 shows sections through relaxed and contracted myofibrils of a skeletal muscle. The transverse sections are diagrams. The longitudinal sections are electron micrographs.

Figure 1



(a) (i) The electron micrographs are magnified 40 000 times.
Calculate the length of band **X** in micrometres.
Show your working.

Length of band **X** = _____ µm

(2)

(ii) Explain the difference in appearance between transverse sections **A** and **C** in **Figure 1**.

_____

_____

(1)

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(b) Explain what leads to the differences in appearance between the relaxed myofibril and the contracted myofibril.

_____

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(*Extra space*) _____

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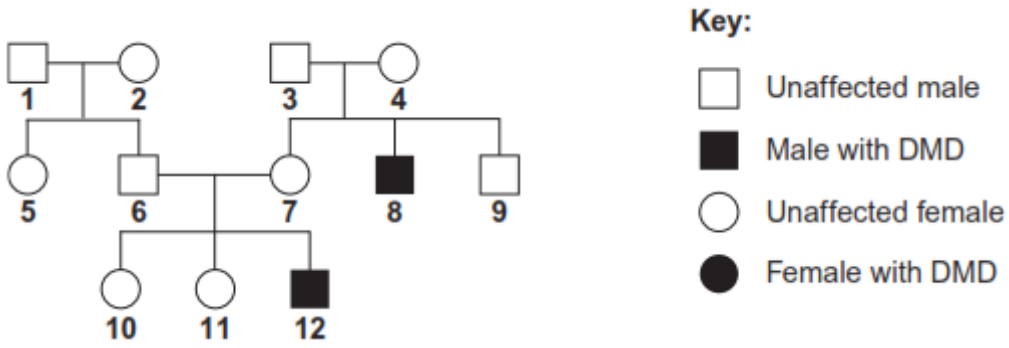
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(4)

(c) Duchenne muscular dystrophy (DMD) is a condition caused by the recessive allele of a sex-linked gene. A couple have a son with DMD. They want to know the probability that they could produce another child with DMD. They consulted a genetic counsellor who produced a diagram showing the inheritance of DMD in this family.
This is shown in **Figure 2**.

Figure 2



The couple who sought genetic counselling are persons **6** and **7**.

(i) Give the evidence to show that DMD is caused by a recessive allele.

_____

_____

(1)

(ii) Give the numbers of **two** people in **Figure 2** who are definitely carriers of muscular dystrophy.

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_____

(1)

(iii) Complete the genetic diagram to find the probability that the next child of couple **6** and **7** will be a son with muscular dystrophy. Use the following symbols:

$\mathbf{X}^{D}$ = normal X chromosome
$\mathbf{X}^{d}$ = X chromosome carrying the allele for muscular dystrophy
Y = normal Y chromosome

	6	**7**
Parental phenotypes	Unaffected	Unaffected
Parental genotypes	_____	_____
Gametes	_____	_____

Offspring genotypes _____

Offspring phenotypes _____

Probability of having a son with DMD _____

(4)

(d) DMD is caused by a deletion mutation in the gene for a muscle protein called dystrophin. A deletion is where part of the DNA sequence of a gene is lost. People in different families may inherit mutations in different regions of this gene.

Scientists isolated the dystrophin gene from DNA samples taken from children **10**, **11** and **12**. They cut the gene into fragments using an enzyme. The scientists then used two DNA probes to identify the presence or absence of two of these fragments, called **F** and **G**. This allowed them to find the number of copies of each fragment in the DNA of a single cell from each child.

The table shows their results.

Child	**Number of copies of gene fragment per cell**	
	F	**G**
10 (unaffected girl)	2	1
11 (unaffected girl)	2	2
12 (boy with DMD)	1	0

(i) The number of copies of gene fragments **F** and **G** shows that person **12** has DMD. Explain how.

_____

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_____

(1)

(ii) The number of copies of gene fragments **F** and **G** shows that person **12** is male. Explain how.

_____

_____

_____

_____

(2)

(iii) The genetic counsellor examined the scientists' results. He concluded that person **10** is a carrier of DMD but her sister, **11**, is not.

Describe and explain the evidence for this in the table.

_____

_____

_____

_____

_____

_____

(*Extra space*) _____

_____

_____

(3)

(e) Person **12** took part in a trial of a new technique to help people with DMD.

Doctors took muscle cells from person **12**'s father and grew them in tissue culture.

They suspended samples of the cultured cells in salt solution and injected them into a muscle in person **12**'s left leg. They injected an equal volume of salt solution into the corresponding muscle in his right leg. Person **12** was given drugs to suppress his immune system throughout the trial.

Four weeks later, the doctors removed a muscle sample from near the injection site in each leg. They treated these samples with fluorescent antibodies. These antibodies were specific for the polypeptide coded for by gene fragment **G** of the dystrophin gene.

The results are shown in the table.

Location and treatment	**Percentage of muscle fibres labelled with antibody**
Left leg - injected	6.8

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with cultured cells suspended in salt solution	
Right leg - injected with salt solution	0.0

(i) Why was it necessary to treat person **12** with drugs to suppress his immune system?

_____

_____

(1)

(ii) Explain why salt solution was injected into one leg and cultured cells suspended in salt solution into the other.

_____

_____

_____

(1)

(iii) This technique is at an early stage in its development. The doctors suggested that further investigations need to be carried out to assess its usefulness for treating people with DMD.

Explain why they made this suggestion.

_____

_____

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(*Extra space*) _____

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(4)
(Total 25 marks)

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