IB · SL · Biology

1 hour 10 questions

Structured Questions

Mutations & Gene Editing

Gene Mutations / Consequences of Gene Mutations / Mutations & Genetic Variation

Easy (2 questions)	/15
Medium (1 question)	/3
Hard (7 questions)	/56
Total Marks	**/74**

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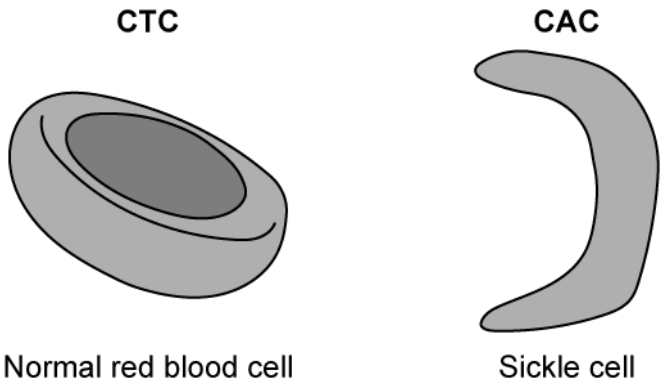
Easy Questions

1 (a) Sickle cell anaemia is a genetic disease where a DNA triplet base in the gene coding for alpha-globin in haemoglobin changes from CTC to CAC. This mutation results in sickle shaped red blood cells that can cause a range of different symptoms.The diagram below shows the DNA base triplet change and the resulting change in the structure of red blood cells.



(i) Identify the type of mutation that is illustrated in the diagram.

[1]

(ii) State a reason for your answer in part i).

[1]

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(2 marks)

(b) List **two** consequences of the change in shape of the red blood cells in a person suffering from sickle cell anaemia.

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(2 marks)

(c) Some mutations, such as those causing sickle cell anaemia, may be inherited by offspring.

Describe how mutations can be inherited by offspring.

(2 marks)

(d) Gene mutations lead to the formation of new alleles in a population.

Define the term 'allele'.

(1 mark)

2 (i) Define the term mutation.

[2]

(ii) List **four** factors that can increase the rate of mutation within cells. Describe the possible consequence of exposure to that factor.

[6]

(8 marks)

Medium Questions

1 Outline how a mutation leads to the development of Down syndrome.

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(3 marks)

Hard Questions

1 (a) *One mark is available for clarity of communication throughout this question.*

Compare and contrast the X and Y chromosome.

(4 marks)

(b) The sex of a foetus is determined by the father.

Explain this statement.

(3 marks)

(c) Outline how a substitution mutation can alter the amino acid sequence of a polypeptide by using sickle cell anaemia as an example.

(7 marks)

2 (a) Wilson's disease is a condition caused by a mutation of gene ATP7B located on chromosome 13, which codes for an ion transport enzyme. This enzyme is responsible for transporting copper ions (Cu^{2+}) into bile so that it can be removed from the body through the digestive tract. There are several mutations which may lead to Wilson's disease; one of these mutations involves the replacement of the amino acid histidine by glutamine.

Describe the type of mutation that could have led to this disease.

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(2 marks)

(b) Based on the information provided in part a), explain the effect this mutation would have on the transport of copper ions.

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(2 marks)

(c) In most cases of Wilson's disease, a sufferer must have two copies of the mutated ATP7B allele before the disease is present.

Suggest what this may indicate about the dominance of the ATP7B allele.

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(1 mark)

(d) People suffering from Wilson's disease have high levels of free copper in their bloodstream which have been shown to cause damage to the cell membranes of red blood cells.

Explain the consequences of this to the sufferer.

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(2 marks)

3 (a) Compare and contrast the X and Y chromosomes.

(4 marks)

(b) The sex of a foetus is determined by the father.

Explain this statement.

(3 marks)

4 (a) The five codons in the diagram at part **a)** are near the start of the sequence coding for a polypeptide. A mutation led to the deletion of one of the bases from codon 3.

Explain the possible consequences of this mutation.

(4 marks)

(b) Guanine (G) in codon 4 changed to adenine (A) due to a mutation.

(i) Describe the effect this mutation would have on the amino acid sequence in the diagram of part a).

[1]

(ii) Explain your answer.

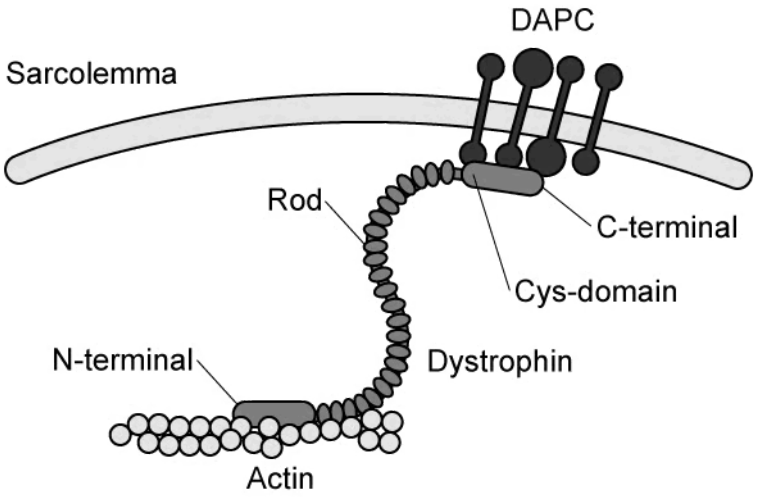
[1]

(2 marks)

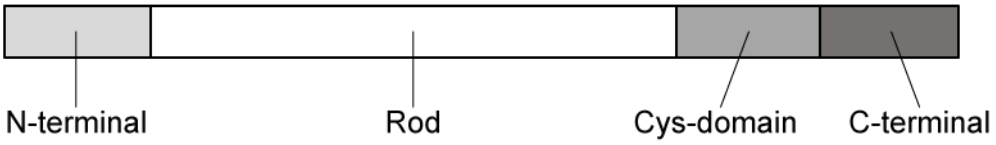
5 (a) Duchenne muscular dystrophy (DMD) is a genetic disorder that leads to the degeneration of muscle tissue over time due to changes in a protein called dystrophin.

Dystrophin is a rod-shaped protein that acts as a link connecting actin filaments in muscle fibres to the extracellular matrix by attaching to a protein complex (DAPC) located in the sarcolemma.

Dystrophin is coded for by the DMD gene, and the complete protein consists of four domains (N-terminal, Rod, Cys-domain and C-terminal), as shown in the diagram below.



The following diagram shows the regions of the DMD gene that codes for the different domains of dystrophin.



One of the causes of Duchenne muscular dystrophy is a substitution mutation that leads to the formation of a stop codon in the rod domain of the DMD gene.

Explain the impact this mutation would have on the resulting dystrophin protein by using the information in the diagrams.

(3 marks)

(b) After transcription of the DMD gene, the pre-mRNA measures about 2.1 megabases (Mb) while the mature mRNA consists of about 14 kilobases (kb). Note that 1 Mb = 10^3 kb.

Calculate the percentage decrease in size of the mRNA molecule after modification. Show your working and give your answer to three significant figures.

(2 marks)

6 (a) Hereditary transthyretin (hATTR) amyloidosis is an inherited condition that is caused by a mutation of a gene that codes for the blood protein transthyretin.

This mutation results in the protein forming clumps in different areas of the body, such as the cardiovascular system, digestive system and around nerve fibres.

Certain drugs that are designed to bind to mRNA molecules are used as treatment for this condition.

Suggest why these drugs could be used as a treatment for hATTR.

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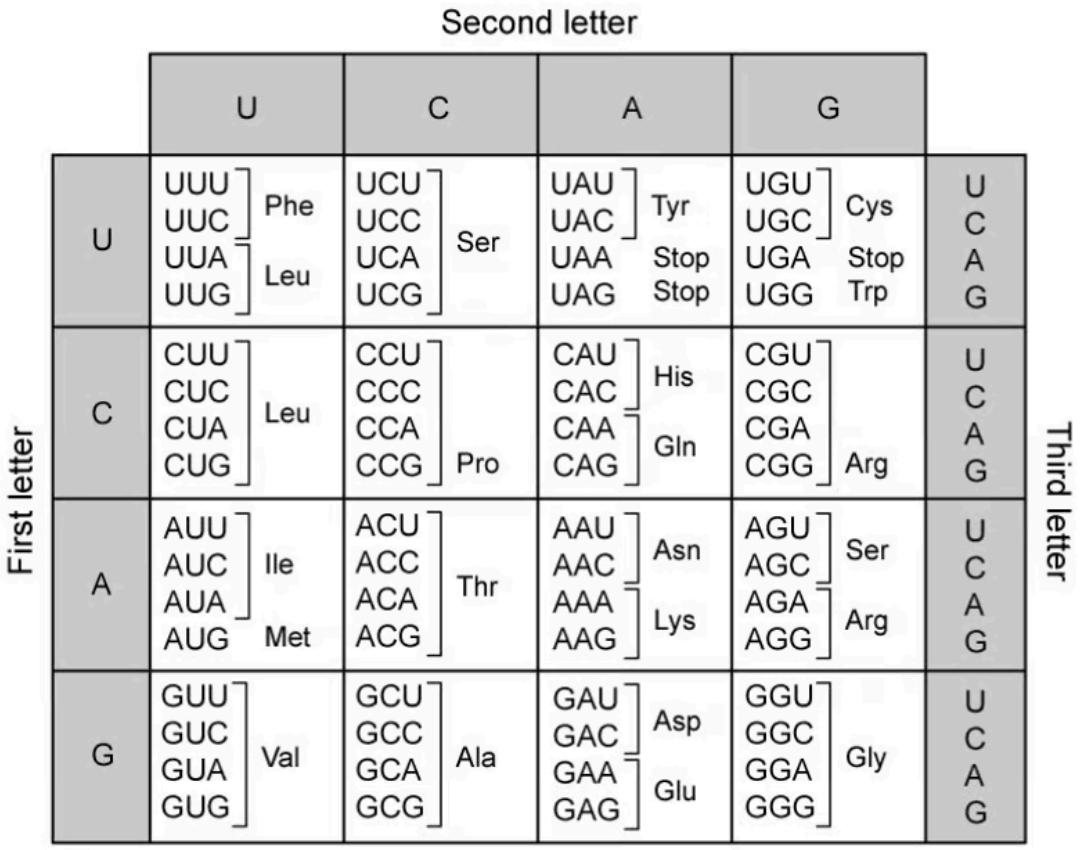
(3 marks)

The gene that codes for transthyretin is known as the TTR gene. The following diagram shows a section of this gene in a normal individual and someone suffering from hATTR.

Normal TTR gene	GGTCCGATTAACCACTTA
Mutated TTR gene	GGTCCGATTAACTACTTA

(b) The table below shows the genetic code and the amino acids that it codes for.



Use the information in the diagram and table to describe the effect the mutation would have on transthyretin.

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(2 marks)

(c) Mitochondrial diseases (MD) are a group of genetic disorders where body cells cannot aerobically respire properly.

One example of an MD is caused by the mutation of a mitochondrial gene that codes for a tRNA molecule. The mutation leads to the replacement of a guanine base with adenine in the anticodon of the tRNA molecule. This results in the formation of a non-functional protein in the mitochondrion.

Suggest how the change in the anticodon of a tRNA molecule leads to an MD.

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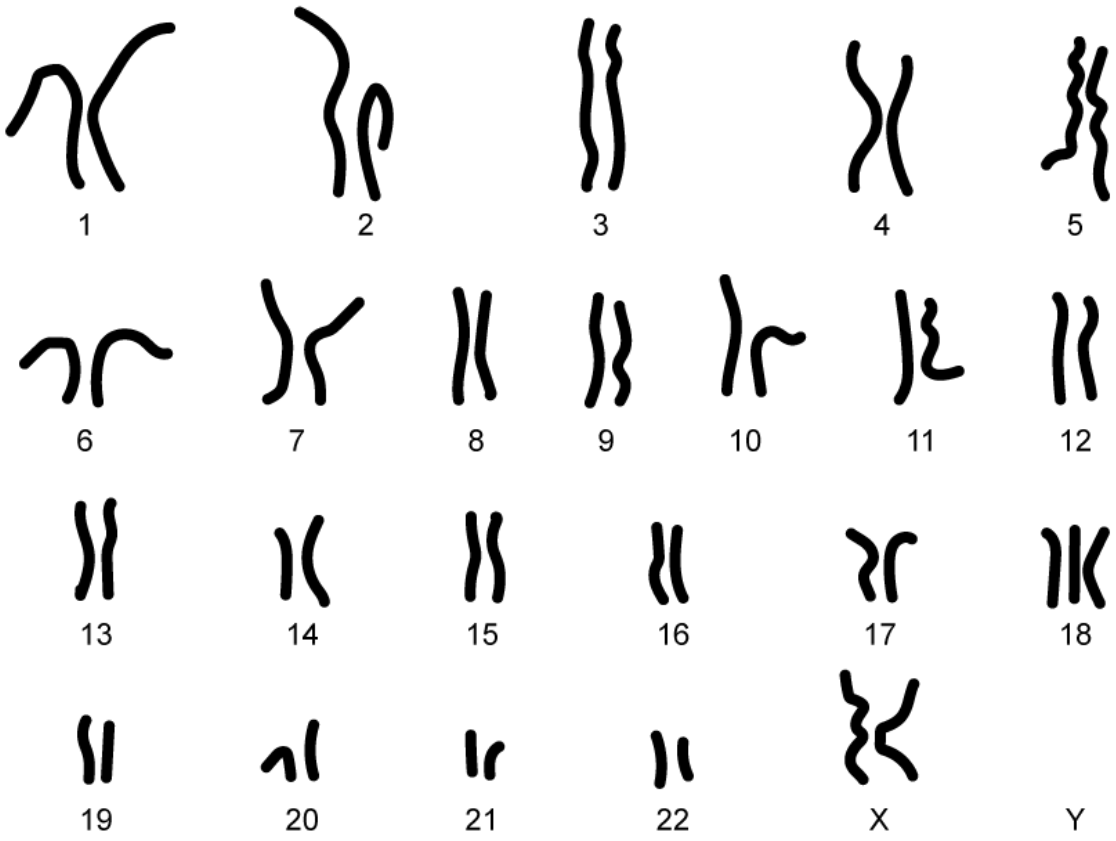
(3 marks)

(d) Explain the role of ATP in translation.

(1 mark)

7 (a) Edwards syndrome is a rare but serious condition that influences birth weight and development. Death rates during infancy are high. The image below shows the karyogram of an individual with Edwards syndrome.



(i) Use the karyogram to suggest the cause of Edwards syndrome.

[1]

(ii) Describe the events that have led to the feature noted in part i).

[2]

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(3 marks)

(b) Edwards syndrome affects every cell in the body.

Explain why this is the case.

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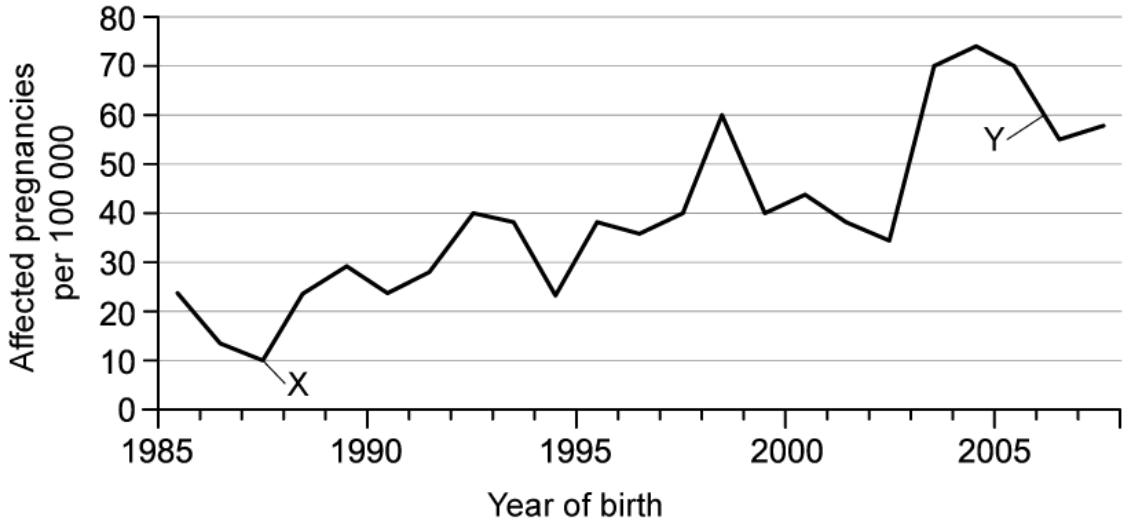
(2 marks)

(c) The graph below shows the number of pregnancies affected by Edwards syndrome between 1985 and 2008.



(i) In the year marked **X** there were 1 100 000 pregnancies, and in the year marked **Y** there were 700 000 pregnancies.

Calculate the number of affected pregnancies at the times marked **X** and **Y**.

[2]

(ii) Suggest **one** reason for the difference in affected pregnancies between times **X** and **Y**.

[1]

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(3 marks)