CfE Higher Biology

Unit 1 DNA and the Genome

Key Area 5/6 Structure of the Genome and Mutations

1. Which line in the table below shows features of the human genome?

	Contains base sequences that regulate transcription	*Contains base sequences transcribed to RNA but never translated*	*Contains base sequences from which primary transcripts are produced*
A	✗	✓	✗
B	✗	✗	✓
C	✓	✓	✗
D	✓	✓	✓

2. Types of single gene mutation are given in the list below.

1 substitution

2 insertion

3 deletion

Which of these would affect only one amino acid in the polypeptide produced?

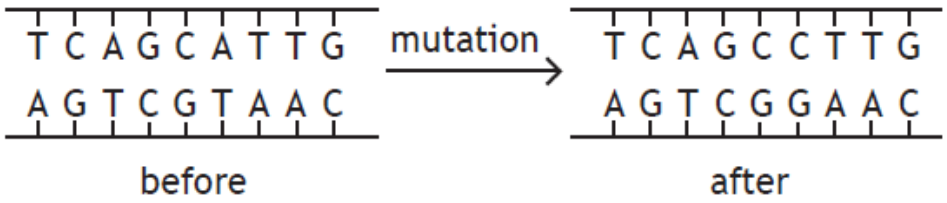
A 1 only

B 2 only

C 3 only

D 2 and 3 only

3. The diagram below shows part of a DNA molecule before and after a mutation.



The type of mutation shown is

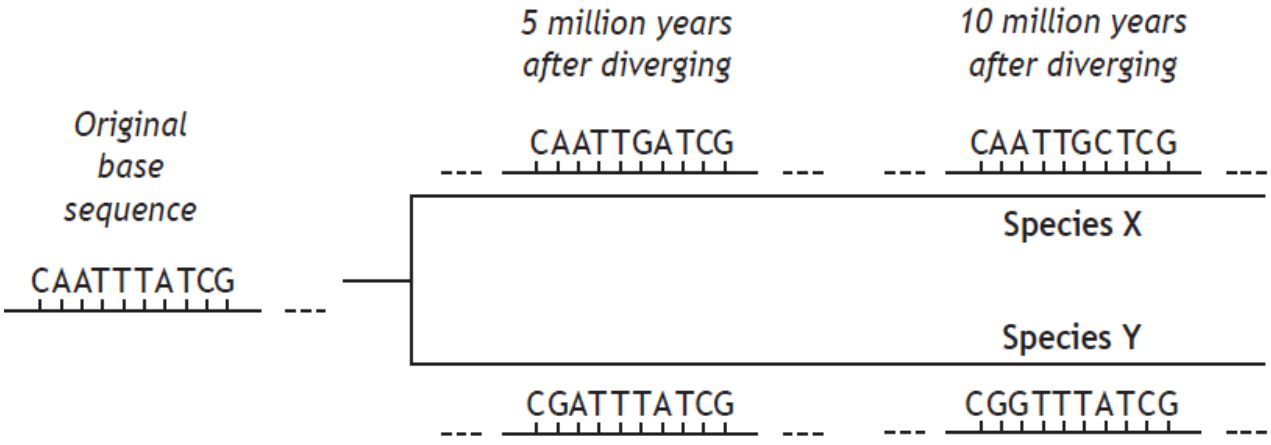
A deletion

B substitution

C insertion

D inversion.

4. Over millions of years of evolution, mutations occur at a broadly constant rate within a gene. This allows genes to be used as molecular clocks. The diagram below shows how the base sequence in part of a gene changed as two evolutionary lineages diverged from an original base sequence. The base sequence in the gene has changed at a rate of 1 base per 5 million years as shown.



Assuming this rate of mutation continued, by how many bases would this part of the gene differ in Species X compared with Species Y 20 million years after diverging from the original base sequence?

A 4

B 8

C 16

D 20

5. The statements refer to DNA sequences in the chromosomes of eukaryotic species.

1 code for protein

2 regulate transcription

3 are transcribed but not translated

Which statements describe the DNA sequences which make up the genome?

A 1 only

B 1 and 2 only

C 1 and 3 only

D 1, 2 and 3

6. The table contains information about the relative genome sizes and number of genes found in a variety of organisms.

Organism	Size of genome (million base pairs)	Number of genes
Human	3080	30 000
Mouse	2600	25 307
Fruit fly	120	13 601
Yeast	12	6294
Mosquito	278	13 688
Nematode worm	97	19 873
Thale cress	125	25 000

What conclusion can be drawn from the data in the table?

A The larger the genome, the fewer genes it contains.

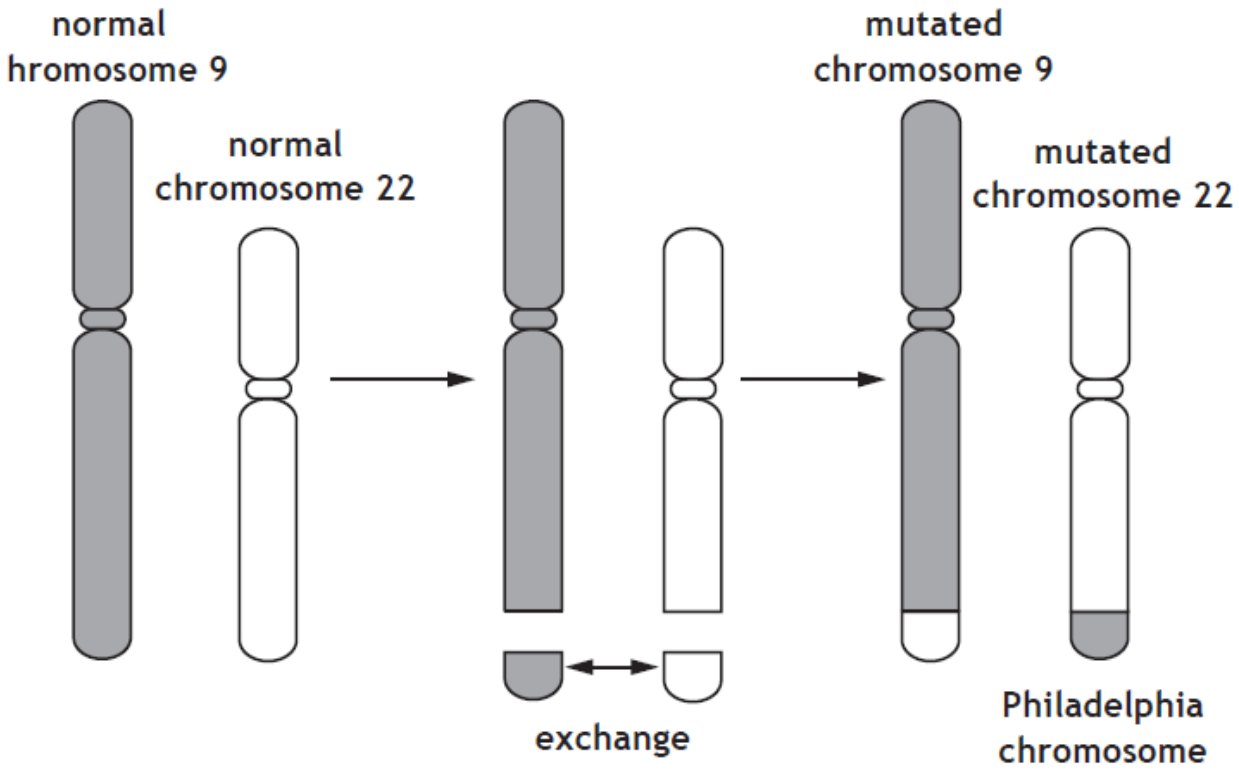
B There is no relationship between genome size and number of genes.

C The larger the genome, the more genes it contains.

D The smaller the genome, the more genes it contains.

7. A chromosome mutation in humans can result in the formation of the Philadelphia chromosome, which is associated with a form of leukaemia.

The stages leading to the formation of a Philadelphia chromosome are shown in the diagram below.



(a) Name the type of chromosome mutation, shown in the diagram, which results in the formation of a Philadelphia chromosome. 1

(b) (i) The presence of a Philadelphia chromosome causes a form of leukaemia through the over-production of an enzyme.

A drug has been used to successfully treat this form of leukaemia by blocking the active site of the enzyme.

Name the type of enzyme inhibition shown by this drug. 1

7.

(b) (continued)

(ii) White blood cell counts in humans normally range from 5000 to 10 000 cells per µl of blood.

The table below shows the white blood cell counts from a patient with leukaemia before and after treatment with this drug.

	Number of white blood cells (per µl blood)
Before treatment	150 000
After treatment	7500

Calculate the percentage decrease in the number of white blood cells after treatment with this drug. 1

Space for calculation

_____ %

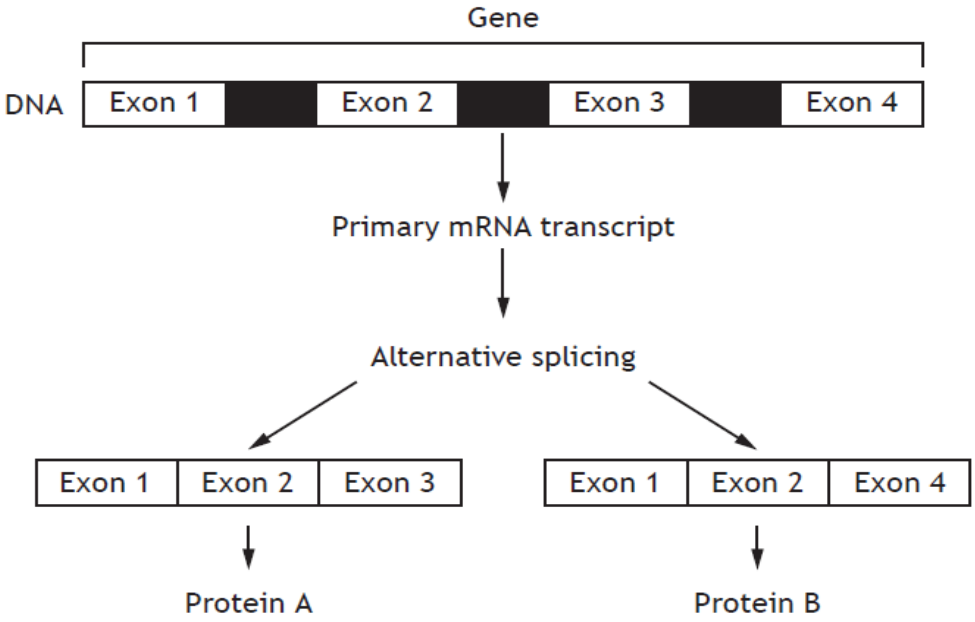
(iii) Explain how the results suggest that the type of leukaemia in this patient was a result of the presence of a Philadelphia chromosome. 2

_____

_____

_____

8. The diagram illustrates steps in the transcription and translation of a gene.



(a) Name the regions always removed from a primary mRNA transcript. **1**

_____

(b) Insert numbers in the boxes below to show the three exons in the gene shown above which could be translated to produce a protein which is different from proteins A and B. **1**

Exon _____	Exon _____	Exon _____

(c) Single gene mutations can occur which may affect the structure of the proteins produced.

(i) Describe the effect of a nonsense mutation on Protein A and give a reason for your answer. **2**

Description _____

_____

Reason _____

_____

(ii) A deletion mutation occurred in Exon 2.

Explain why this would have a major effect on the structure of proteins A and B. **1**

_____

_____

_____

9. Congenital lactase deficiency in humans is caused by very low activity of the enzyme lactase, resulting in individuals being unable to digest lactose in milk. This is caused by a number of different mutations in the lactase gene.

(a) One of the mutations involved causes a frame-shift mutation in the lactase gene.

(i) Name a gene mutation which causes a frame-shift. 1

_____

(ii) Describe the effect of this frame-shift mutation on the lactase gene and on the structure of lactase. 2

Effect on lactase gene _____

_____

_____

Effect on structure of lactase _____

_____

_____

(b) (i) Some mutations occur in the sequences which regulate the transcription of the lactase gene.

Suggest why this may lead to more lactase enzyme being produced. 1

_____

_____

_____

(ii) Mutations in this gene are more common in Finland than in other parts of Europe. This is thought to be due to a small number of individuals who settled in Finland many generations ago.

State the term which describes this change in gene frequency when a small population of individuals breaks away from a larger population. 1

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