

Mutations

5. Types of single gene mutation are given in the list.

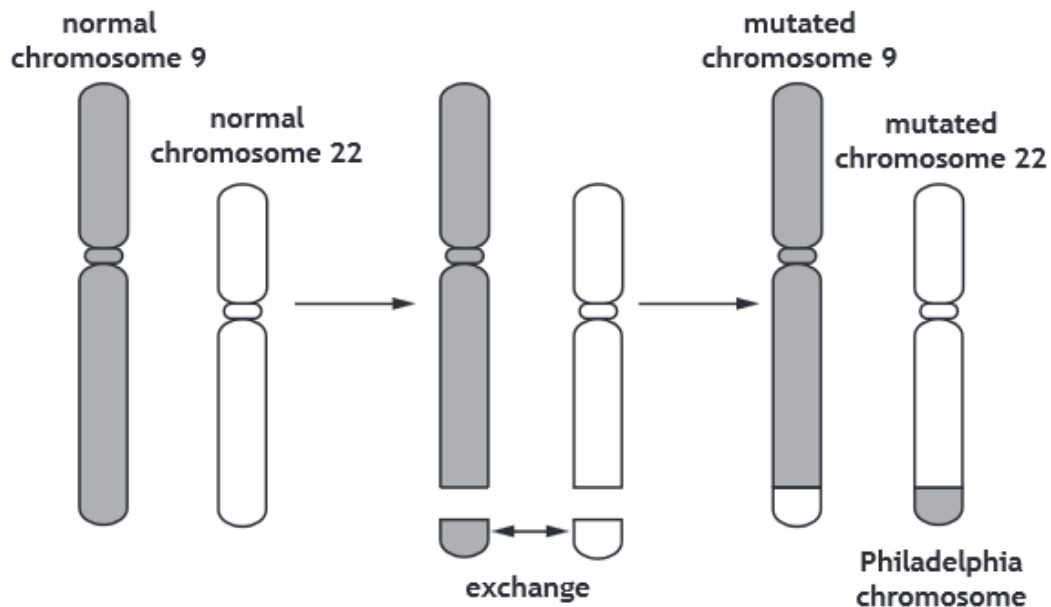
- 1 substitution
- 2 insertion
- 3 deletion

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

- A 1 only
- B 2 only
- C 3 only
- D 2 and 3 only

2. 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.



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

12. 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

(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
