## CfE Higher Biology

## Unit 1 DNA and the Genome

## Key Area 5/6 Structure of the Genome and Mutations Marking Instructions

1.	$\Box$
Ι.	υ

2. A

3. B

4. B

5. D

6. B

7.

a		translocation	1	
b	i	competitive	1	
b	ii	95	1	
b	iii	Drug was effective as white blood count reduced to normal	2	
	Drug works by inhibiting the enzyme produced by Philadelphia chromosome			

8.	(a)		Introns	1	NOT- non-coding regions.
	(b)		1,3,4/2,3,4	1	Must be in correct order and inversions not acceptable.
	(c)	(i)	Shorter protein/fewer amino acids.(1)	2	NOT- non-functional protein. NOT- protein is short.
			Stop codon is produced earlier (in the sequence) (1)		NOT- stop codon is produced alone.
		(ii)	Every amino acid after the mutation is changed/affected.	1	NOT- frame shift mutation alone.

9.	(a)	(i)	Deletion/insertion	1	
		(ii)	Effect on lactase gene: All the codons/base sequences nucleotide sequences/ triplets/ bases/nucleotides after the mutation will change/will move along.	2	NOT - Amino acids produced/made.
			OR		
			All the following codons/base sequences nucleotide sequences/triplets will change.		
			Effect on structure of lactase: All the amino acids after the mutation may change (1)		
	(b)	(i)	Gene is permanently switched on.	1	
			OR		
			More transcription occurred.		
			OR		
			Repressor molecule not produced so operator permanently switches on gene/gene not switched off.		
		(ii)	Founder effect/genetic drift.	1	