

2023 Bi H2 Q2

Section: DNA and the Genome

Topic: Gene Expression

Question Summary

The doublesex gene in fruit flies determines sex by producing different mature mRNA transcripts from the same primary transcript. You are asked to: (1) name process A, (2) explain why a certain transcript cannot be formed, and (3) describe the effect of a splice-site mutation.

Worked Solution

(a)(i) Name process A

The same primary mRNA transcript can be spliced in different ways to include or exclude certain exons. This process is called alternative RNA splicing.

■ Answer: Alternative RNA splicing.

(a)(ii) Why the transcript shown cannot be produced

In the diagram, the exons appear out of order — Exon 2 comes before Exon 1. This cannot occur because exons are always joined in their correct genomic order during splicing. The splicing machinery (spliceosome) only removes introns, not rearranges exon order.

■ Answer: The spliceosome cannot join exons in the wrong order; exons must remain in sequence.

(b) Effect of a splice-site mutation

- An intron may be retained or an exon may be missed out, changing the mRNA sequence.
- The protein produced will then be altered — it may be shorter, longer, or non-functional.
- Such changes can disrupt the protein's structure or prevent it from folding correctly.

■ Answer Summary: Mutation changes the mRNA sequence and results in an altered or faulty protein.

Revision Tips

- Alternative splicing allows one gene to code for several proteins by using different exon combinations.
- Splice sites mark the boundaries between introns and exons.
- A mutation at a splice site can cause introns to remain, exons to be skipped, or changes to the reading frame.
- Remember: 'Alternative splicing = one gene, many proteins.'

