

# 1.5 Regulation of gene expression

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Name: \_\_\_\_\_ Class: \_\_\_\_\_ Date: \_\_\_\_\_

**Total: 23 marks**

**KEY WORDS** enhancer

## Objective

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Reason about enhancers, combinatorial specificity and epigenetic modification of disease.

## 1 Key knowledge and worked examples

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### ■ Specificity is combinatorial

A 6-base motif occurs every  $4^6 = 4096$  bp – ~1.6 M times per strand; only combinations of factors cut false hits multiplicatively ✓. Enhancers act by looping, orientation-independent, from up to 100 kb.

### ■ Worked example: one upstream base

Lactase persistence: one base 14 kb upstream strengthens an enhancer → LCT stays on into adulthood – a zero-protein-change trait ✓.

## 2 Practice

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**2.1** How often does a random 8-base motif occur per strand ( $6.4 \times 10^9$  bp)? [3]

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**2.2** Distinguish promoter from enhancer in one sentence each. [3]

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**2.3** Why is “epigenetic” not “permanent”? [3]

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### 3 Exam-style questions

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#### 3.1 A transcription factor binds a 6-base motif.

Compute the expected genome-wide hit count. Explain why cooperating factors binding adjacent sites restore specificity, quantitatively. State one consequence for interpreting non-coding disease variants. [7]

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#### 3.2 Fictional monozygotic twins, same variant, different severity; one has promoter methylation silencing that allele.

Explain why sequence identity does not guarantee phenotype identity. State what methylation (70–80 % of CpG) does to enhancer/promoter access. Explain why methylation assays sit next to sequencers diagnostically. [7]

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