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| **Examination question paper:** | **May 2025** |

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| **Module code:**  **Component number:** | **BM7122**  **003** |
| **Module title:** | **Medical Genetics and Genomics** |
| **Module leader:** | **Dr Sarah Alokozai** |

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| **Date:** | **May 2025** |
| **Duration:** | **1 Hour 30 Minutes** |

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| **Exam type:** | Seen, Closed |
| **Materials supplied:** | **None** |
| **Materials permitted:** | **None** |
| **Warning:** | **Candidates are warned that possession of unauthorised materials in an examination is a serious assessment offence.** |

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| **Instructions to candidates:** | Candidates will be required to answer **Three** questions out of five. |
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|  | **Do not turn page over until instructed** |

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**Question 1**

Thalassaemias are a common genetic disease.

a) Briefly describe the molecular development of α and β thalassaemia.

b) Briefly discuss the potential for gene therapy to treat these diseases.

**30 marks**

**Question 2**

Discuss the genetic basis of inherited metabolic disorders of two inherited disorders including how do mutations in specific genes lead to enzyme deficiencies.

How are these treated and managed?

**30 marks**

**Question 3**

What specific molecular change takes place in SCD (sickle Cell Anaemia), which changes red blood cell morphology.

Why has this mutation persisted in the human population?

**30 marks**

**Question 4**

ENCODE have published data that suggests that around 80% of the genome encodes regulatory RNA and bear crucial DNA regulatory domains.

Discuss how this has affected the interpretation of genome wide association studies (GWAS)?

**30 marks**

**Question 5**

**Tumours result from the dysregulation of the signals that control cell growth.**

Discuss the mechanisms by which oncogenes, tumour suppressor genes and DNA repair genes contribute to cancer progression. In your answer, address the following statements:

a) Explain how **oncogenes** control cell proliferation and describe their role in cancer development.

b) Explain the roles of **caretaker** and **gatekeeper** genes in cancer progression.

**30 marks**

**END OF PAPER**