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| **Examination question paper:** | **July 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:** | **July 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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Answer **Three** questions out of five.

**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:**

“*Many human traits and diseases are polygenic*”. With respect to this statement, briefly answer the following questions:

a) What does polygenic mean?

b) What is a quantitative trait locus QTL?

c) Does the environment play a role and if so how?

d) What is the use of twin studies in genetics?

e) If heritability has a value near 1.0, how can this be interpreted?

**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. Include in your answer, the following statements:

a) Clarify the mechanism by which **BRCA1** and **BRCA2** mutations lead to breast cancer, incorporating their role in DNA repair.

b) Describe how tumours can originate from adult tissues, and explain how they bypass the state of **quiescence** (cell cycle arrest).

**30 marks**

**END OF PAPER**