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(UNIVERSITY OF CHOICE)

MASINDE MULIRO UNIVERSITY OF SCIENCE AND TECHNOLOGY (MMUST)

MAIN CAMPUS

UNIVERSITY EXAMINATIONS
2022/2023 ACADEMIC YEAR

THIRD YEAR SECOND SEMESTER MAIN EXAMINATIONS

FOR THE DEGREE
OF
BACHELOR OF SCIENCE MEDICAL BIOTECHNOLOGY

COURSE CODE: BMB 321

COURSE TITLE: MEDICAL GENETICS

DATE: 26TH APRIL 2023 **TIME: 8.00 – 10.00AM**

INSTRUCTIONS TO CANDIDATES

This paper is divided into three sections, **A B** and **C,** carrying respectively: Multiple Choice Questions (**MCQs**), Short Answer Questions (**SAQs**) and Long Answer Questions (**LAQs**). **Answer all questions. DO NOT WRITE ON THE QUESTION PAPER.**

TIME: 2 Hours

MMUST observes ZERO tolerance to examination cheating

This Paper Consists of 5 Printed Pages. Please Turn Over

D. Sickle cell anemia

10. Which is the infectious genetic disease:

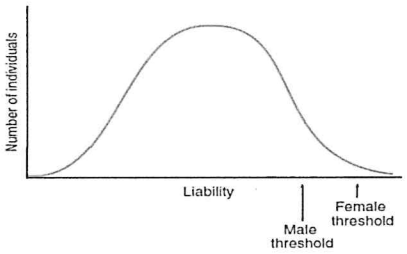
A. Leber's hereditary optic neuropathy (LHON)

B. Phocomelia

C. Albinism

D. Prion diseases

11. Pyloric stenosis is five times more common in males than in females in certain Japanese populations. The liability curve for the development of this condition in that population is shown below: Within this population, which of the following is most at risk for the development of disease?



A. The daughters of affected fathers

B. The daughters of affected mothers

C. The sons of affected fathers

D. The sons of affected mothers

12. In studying a large number of families with a small deletion in a specific chromosome region, it is noted that the disease phenotype is distinctly different when the deletion is inherited from the mother as opposed to the father. What is the most likely explanation?

A. Imprinting

B. Mitochondrial inheritance

C. Sex-dependent penetrance

D. X-linked dominant inheritance

13. Waardenburg syndrome is an autosomal dominant disorder in which patients may exhibit a variety of clinical features, including patches of prematurely grey hair, white eyelashes, a broad nasal root, and moderate to severe hearing impairment. Occasionally, affected individuals display two eyes of different colours and a cleft lip and/or palate. Patients who possess a mutation in the *PAX3* gene on chromosome 2 can present with all of these disparate signs and symptoms. Which of the following characteristics of genetic traits is illustrated by this example?

A. Anticipation

B. Pleiotropy

C. Incomplete penetrance

D. Locus heterogeneity

14. A 25-year-old woman has mild expression of haemophilia A. A genetic diagnosis reveals that she is a heterozygous carrier of a mutation in the X-linked factor VIII gene. What is the most likely explanation for mild expression of the disease in this individual?

A. A high proportion of the X chromosomes carrying the mutation are active in this woman

B. Her father is affected, and her mother is a heterozygous carrier

C. Nonsense mutation causing truncated protein

D. One of her X chromosomes carries the SRY gene

15. A 6-year-old boy has a family history of mental retardation and has developmental delay and some unusual facial features. He is being evaluated for possible fragile X syndrome. Which of the following would be most useful in helping establish the diagnosis?

A Genetic test for a trinucleotide repeat expansion in the fragile X gene