(*University of Choice*)

MASINDE MULIRO UNIVERSITY OF SCIENCE AND TECHNOLOGY (MMUST)

MAIN CAMPUS

UNIVERSITY EXAMINATIONS
2018/2019 ACADEMIC YEAR

THIRD YEAR SECOND TRIMESTER EXAMINATIONS

FOR THE DEGREE OF BACHELOR OF SCIENCE MEDICAL BIOTECHNOLOGY
MAIN EXAM

COURSE CODE: **BMB 321**

COURSE TITLE: **MEDICAL GENETICS**

DATE: 23RD MAY 2019 **TIME:** 3.00 -5.00 PM

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**).

TIME: 2 Hours

MMUST observes ZERO tolerance to examination cheating

This Paper Consists of 5 Printed Pages. Please Turn Over.

SECTION A: MULTIPLE CHOICE QUESTIONS (20 MARKS)

1. Which is X-linked recessive inheritance
A. Down syndrome is an example
B. Retinoblastoma is an example
C. Show female to male transmission
D. Are determined by both environmental and genetic factors

2. Which disease is a kind of mitochondrial disorder?
A. Parkinson disease
B. MELAS
C. Huntington's disease
D. Turners syndrome

3. Which gene does not involve in the cause of diabetes mellitus:
A. DRD3 gene
B. HNF1alpha
C. Glucokinase
D. Mitochondrial DNA

4. Klinefelter Syndrome is
A. The cause is a mutation affection the long arm of X chromosome
B. It is commonly associated with congenital heart disease
C. The cause is mitochondrial DNA damage
D. Sexually underdeveloped

5. Which is polygene disorder:
A. Schizophrenia
B. Cri du Chat syndrome
C. Klinefelter Syndrome
D. Cystic fibrosis

6. Rentino-blastoma is the tumor with
A. Single gene disorder
B. Multifactorial disorder
C. Related with chromosome translocation
D. p53 mutation

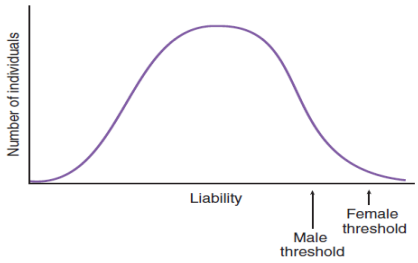
7. Which one is not the environmental cause for birth defect:
A. Virus
B. Alcohol use in pregnancy
C. Folic acid
D. Toxoplasma

8. Characteristics of mitochondrial diseases:
A. Threshold effect
B. X-linked
C. Inherited from the father
D. Only female can be affected

9. Which is the disorder with autoregressive model:
A. Meningocele
B. KSS syndrome
C. Parkinson disease
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

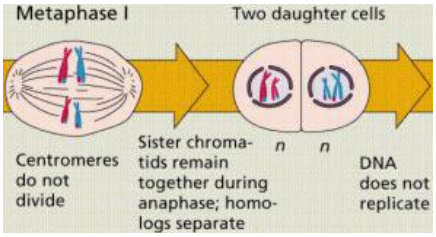
14. A 25-year-old woman has mild expression of haemophilia A. A genetic diagnosis reveals that she is a heterozygous carrier of a mutation in the X-linked factor VIII gene. What is the most likely explanation for mild expression of the disease in this individual?
A. A high proportion of the X chromosomes 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
B IQ test
C Karyotype of the child's chromosomes
D Karyotype of the father's chromosomes

16. Endonuclease activation and chromatin fragmentation are characteristic features of eukaryotic cell death by apoptosis. Which of the following chromosome structures would most likely be degraded first in an apoptotic cell?
A. Barr body
B. 10-nm fiber
C. 30-nm fiber
D. Centromere

17. The cells that have no nucleus are known as:
A. Eukaryotic cells
B. Proliferative cells
C. Prokaryotic cells
D. Animalcules

18. Study the following illustration of a cell division stage where the chromosomes have gathered at the equator and answer the question to follow: In which organ of the human body did this process take place?



A. Liver
B. Spleen
C. Ovarium
D. Bone marrow

Question 19 and 20
A. ATGCAA...→ ATGTAA
B. ATGAAA...→ GTGAAA
C. TATAAG...→ TCTAAG
D. CTTAAG...→ GTTAAG
The options above represent mutations in the DNA with base changes indicated in boldface type. For each mutation described in the questions below, choose the most closely related sequence change in the options above.
19. Nonsense mutation.
20. Mutation decreasing the initiation of transcription.

SECTION B: SHORT ANSWER QUESTIONS (40 MARKS)

1. Describe mitochondrial inheritance. [8 Marks]
2. a) List any four diseases that result from a delayed age onset. [4 Marks]
   b) Briefly describe imprinting in Prader-Willi syndrome. [4 Marks]
3. a) Describe chromosomal nomenclature. [4 marks]
   b) Briefly describe X-inactivation. [4 Marks]
4. Discuss briefly advances in molecular cytogenetics. [8 Marks]
5. Describe how recurrent risks for multifactorial diseases is assessed. [8 Marks]

SECTION C: LONG ANSWER QUESTIONS (40 MARKS)

1. Citing relevant examples, give a detailed account of numerical chromosomal abnormalities. [20 Marks]

2. Discuss polymorphic markers and linkage analysis. [20 Marks]