UNIVERSITI KUALA LUMPUR
INSTITUTE OF MEDICAL SCIENCE TECHNOLOGY

FINAL EXAMINATION

OCTOBER 2025 SEMESTER

COURSE CODE	:	HDB10203
COURSE TITLE	:	HUMAN GENETICS
PROGRAMME NAME	:	BACHELOR OF BIOMEDICAL SCIENCE (HONOURS)
DATE	:	31 JANUARY 2026
TIME	:	9:00AM - 12:00PM
DURATION	:	3 HOURS

INSTRUCTIONS TO CANDIDATES

1. Please read the instructions given in the question paper CAREFULLY.
2. This question paper is printed on both sides of the paper.
3. This question paper consist of TWO sections.
4. Answer ALL questions for Section A.
5. Section B consist of four questions. Answer THREE (3) questions only.
6. Please write your answer on the answer booklet provided.
7. Please answer all questions in English only.
8. Please answer MCQ/EMQ questions using OMR sheet. ☐ *Tick if applicable*
9. Refer to the attached Formula/ Appendies. ☐ *Tick if applicable*

THERE ARE 17 PAGES OF QUESTIONS INCLUDING THIS PAGE

Sat. A.

SECTION A (Total: 40 marks)

Answer ALL questions.

Please use the answer booklet provided.

1. During transcription, RNA polymerase synthesizes the RNA strand by reading the DNA template strand from the _____ direction, and the new nucleotides are always added to the _____ end of the RNA.

   A. 3' to 5'; 5'

   B. 5' to 3'; 3'

   C. 5' to 3'; 5'

   D. 3' to 5'; 3'

2. In eukaryotes, which combination of components is essential to form the transcription initiation complex at the start site of a gene?

   I. snRNP
   II. Promoter
   III. TATA box
   IV. RNA polymerase

   A. I, II, III and IV

   B. II and III only

   C. II, III and IV only

   D. I, II and III only

3. The underlying genetic cause of sickle-cell disease is a _____ mutation.

   A. nonsense

   B. frameshift

   C. point

   D. nondisjunction

4. Linkage between two genes is strongly suggested when _____.

   A. they do not assort independently in meiosis

   B. a single gene influences two different traits

   C. they function together to produce one trait

   D. they are always found together in a gamete

5. Which type of mutation occurs when a single base change results in a codon that signals the termination of translation?

   A. Missense

   B. Neutral

   C. Frameshift

   D. Nonsense

6. Select the primary function of proto-oncogenes in a normal, healthy cell.

   A. They provide defense against oncogenic viruses

   B. They are genes that help regulate and promote normal cell division

   C. They are produced as a consequence of cell division

   D. They are genes that protect the cell from environmental carcinogens

7. Two sister chromatids differ from each other due to variations in _____.

   A. gene identity and order

   B. staining appearance

   C. centromere location

   D. exact nucleotide sequence

8. The inheritance pattern of an X-linked trait from a father to his children is such that _____ it.

   A. sons will always inherit

   B. daughters will always inherit

   C. sons have a 50% chance of inheriting

   D. daughters have a 50% chance of inheriting

9. Mendel's F1 hybrids resembled only one parent because _____.

   A. multiple genes interacted

   B. their traits blended

   C. one allele masked the other

   D. both alleles affected the phenotype equally

10. Place the following steps of DNA replication in the correct sequence.

    I. Covalent bonds form between adjacent nucleotides
    II. The double helix is unwound, separating the two strands
    III. Nucleotides align in a complementary fashion on the template strands
    IV. Two new DNA molecules are formed, each with one old and one new strand

    A. III → II → I → IV

    B. II → III → I → IV

    C. II → I → III → IV

    D. III → I → II → IV

11. Which of the following genetic alterations may activate a proto-oncogene, turning it into an oncogene?
    I. Deletion of the entire proto-oncogene
    II. An increase in the number of copies of the gene
    III. Movement of the gene to a new chromosomal location
    IV. A point mutation that makes the gene's product overactive

    A. I, II and III only

    B. I, II and IV only

    C. I, III and IV only

    D. II, III and IV only

12. The primary roles of tumor-suppressor genes include _____.

    A. triggering apoptosis in healthy cells

    B. promoting cellular differentiation only

    C. accelerating the cell cycle

    D. halting cell division and repairing DNA damage

13. Which sampling methods supply fetal cells for karyotype analysis?
    I. Fetoscopy
    II. Amniocentesis
    III. CVS (chorionic villus sampling)
    IV. Percutaneous umbilical blood sampling

    A. II and III only

    B. I, II, and III only

    C. I and II only

    D. I, II, III and IV

14. The genetic variation in a typical natural population is such that for most genes, there are _____ alleles present.

A. only dominant

B. only one or two

C. only recessive

D. multiple different

15. Transcription of the DNA template strand *5'-AACGTAACG-3'* would produce an mRNA with the sequence of _____.

A. *5'-AACGTAACG-3'*

B. *5'-UUGCAUUGC-3'*

C. *5'-TTGCATTGC-3'*

D. *5'-CGUUACGUU-3'*

16. An individual with a hereditary predisposition to cancer typically inherits _____ .

A. one somatic mutation

B. one germline mutation

C. two somatic mutations

D. one germline mutation and acquires a second somatic mutation

17. Telomeres function like the _____.

A. engine of a car

B. pages of a book

C. zipper on a jacket

D. plastic tips on shoelaces that prevent fraying

18. In a cross *AaBbCc* X *AaBbCc*, determine the probability of producing the genotype *AABBCC.*

A. 1/32

B. 1/4

C. 1/16

D. 1/64

19. If a DNA sample contains 38% cytosine bases, calculate the percentage of adenine bases.

A. 24%

B. 38%

C. 76%

D. 12%

20. For an autosomal dominant disorder like Huntington's disease, if one parent is heterozygous and the other is homozygously normal, the risk to any child is _____.

A. 100%

B. 50%

C. 75%

D. 25%

21. Which alteration to the phrase "*THE BIG CAT*" is analogous to a frameshift mutation caused by an insertion?

A. *THB IGC AT*

B. *CAT BIG THE*

C. *THE BAT CAT*

D. *THE BIG GCAT*

22. A haplotype refers to _____.

A. the physical location of a gene on a chromosome

B. the entire set of genes in an individual

C. a single, unique allele in a population

D. a specific combination of alleles (SNPs) on a single chromosome that are inherited together

23. Genes and chromosomes share all the following properties which includes _____.

I. they segregate during meiosis
II. their number is reduced during meiosis
III. they are present in pairs in diploid cells
IV. their homologous pairs physically synapse during prophase of mitosis

A. I, III and IV only

B. I, II and III only

C. II, III and IV only

D. I, II, III and IV

24. Choose the correct statement that describes tumor-inducing retroviruses.

A. Their oncogenes are identical to the host's proto-oncogenes and contain introns

B. They replicate in the host cytoplasm without entering the nucleus

C. They do not require reverse transcriptase for their life cycle

D. They can integrate into the host genome and alter the expression of cellular genes

25. When cell cycle regulators like *c-ras* and *c-myc* become overactive or overexpressed, the cell becomes _____.

A. predisposed to developing cancer

B. cancerous immediately

C. resistant to cancer

D. undergoes cell-cycle arrest to allow DNA repair

26. A change in the DNA sequence can lead to a different _____ being inserted into a polypeptide chain during translation.

A. RNA

B. nucleotide

C. codon

D. amino acid

27. For a gene with two alleles, *C* and *c*, the allele frequencies are p=0.4 and q=0.6. Assuming Hardy-Weinberg equilibrium, the frequency of the heterozygous genotype is _____.

A. 0.16

B. 0.24

C. 0.48

D. 0.60

28. Identify a normal example of mosaicism found in all human males.

A. Gonadal mosaicism

B. X-inactivation mosaicism

C. Somatic mosaicism

D. Y-chromosome mosaicism

29. Identify the TRUE statements regarding somatic gene therapy in patients.
    I. Germline cells are not altered by the treatment
    II. The procedure is typically simpler than an organ transplant
    III. The therapy targets only the affected individual's body cells

    A. I and III only

    B. II and III only

    C. I and II only

    D. I, II and III

30. Which of the following is an important mechanism of epigenetic regulation?

    A. Point mutation in a gene's coding region

    B. DNA methylation and histone modification

    C. Deletion of a chromosome segment

    D. Translocation of genes between chromosomes

31. The finding that in any DNA sample, the amount of adenine equals thymine, and the amount of guanine equals cytosine is known as _____.

    A. the law of segregation

    B. Chargaff's rules

    C. the central dogma

    D. the one gene-one enzyme hypothesis

32. On the lagging strand during DNA replication, the enzyme that removes RNA primers and fills in the gaps with DNA is _____.

    A. Ligase

    B. Helicase

    C. Primase

    D. DNA polymerase I

33. The nitrogenous base adenine is a component of _____.

A. starch and DNA

B. proteins and ATP

C. DNA and RNA

D. ATP, DNA, and RNA

34. Which feature found in mature eukaryotic mRNA is absent in prokaryotic mRNA?

I. Introns
II. Cytosine
III. A poly-A tail
IV. A 5' methyl-guanosine cap

A. II and III only

B. I and II only

C. II and IV only

D. III and IV only

35. Which type of genetic cross is most likely to yield a 9:3:3:1 phenotypic ratio in the offspring?

A. Tetrahybrid cross

B. Test cross

C. Dihybrid cross

D. Monohybrid cross

36. During transcription, which RNA base complements the nucleotide marked * in the DNA template strand.
Refer Below - Figure1 : DNA template strand .

Figure 1: DNA template strand

A. Uracil

B. Guanine

C. Cytosine

D. Thymine

37. The ABO blood group system, where three alleles (*IA, IB, i)* determine the blood type, is an example of _____.

A. epistasis

B. codominance

C. pleiotropy

D. multiple allelism

38. When a single gene affects multiple distinct traits in an organism, this effect is called _____.

A. epistasis

B. pleiotropy

C. polygenic inheritance

D. incomplete dominance

39. If a family has six boys in a row, calculate the probability that their next child will be a girl.

    A. 1/2

    B. 1

    C. 1/6

    D. 0

40. The centromere of a metacentric chromosome is located _____.

    A. close to one end

    B. near the center

    C. at the very end

    D. at two separate points

SECTION B (Total: 60 marks)

Answer THREE (3) questions only.

Please use the answer booklet provided.

Question 1

Phenylketonuria (PKU) is a metabolic disorder inherited in an autosomal recessive manner. A man and a woman, who are both phenotypically normal, have an infant son diagnosed with PKU. The couple plans to have two more children. Based on this scenario, answer all the following questions:

(a) Illustrate the family's genetic history by drawing a pedigree. The pedigree must include the parents, their affected son, and their two future children (daughters) who are phenotypically normal. Assign the correct genotypes to all individuals in your diagram.

(6 marks)

(b) Using Punnett square, illustrate how the parents' genotypes may produce a child with PKU. Indicate the genotypic ratio for this inheritance pattern.

(5 marks)

(c) Determine the chance that both future daughters will be unaffected by PKU. Describe your calculation clearly.

(5 marks)

(d) Calculate the probability that both daughters will only inherit the normal allele and be completely free of the PKU allele.

(4 marks)

Question 2

The pedigree diagram below illustrates the inheritance of a specific genetic trait known as 'X' across three generations. '1', '2' and '3' indicate individuals who express the trait.
Refer Below - Figure2 : Pedigree of X disease .

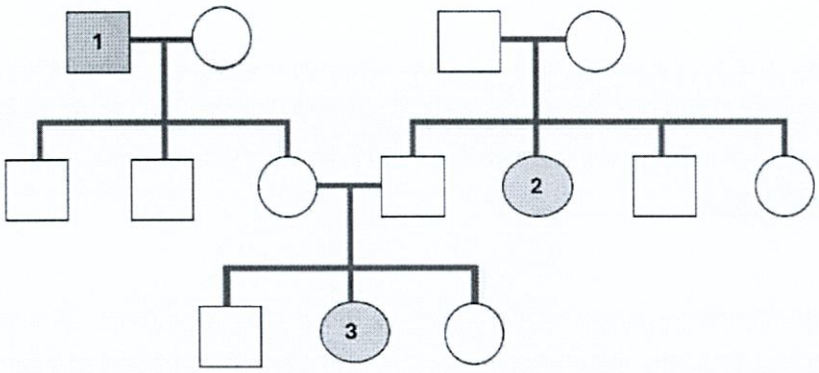

Figure 2: Pedigree of X disease

(a) Based on the pattern of inheritance observed in the pedigree, identify the most likely mode of inheritance of the 'X' disease. Justify your answer with TWO (2) distinct evidence from the pedigree.

(5 marks)

(b) Using standard allele symbols (e.g., *A/a*), state the genotypes for individual '1', '2' and '3'. Justify your answers.

(5 marks)

(c) The unaffected male married an affected woman from the second generation in the pedigree, and they have had two children. Their first child is affected with disease 'X', and their second child is unaffected. Calculate the probability that their child will be affected. Show your calculation using a Punnett square or a clear genetic cross.

(5 marks)

(d) The affected female in the second generation and her unaffected husband decide to have another child. Calculate the probability that this child will be unaffected and not a carrier. Show your calculation.

(5 marks)

Question 3

A research team discovered a new protein called *TPX1*. In healthy colon tissue, *TPX1* is expressed at high levels, but in colon cancer cell lines and metastatic samples, its expression is significantly reduced. Genetic analysis reveals a point mutation in the *TPX1* gene in cancer cells. Further experiments show that cells with *TPX1* mutations proliferate faster and are less responsive to DNA damage.

(a) Based on the findings, explain the likely role of *TPX1* in preventing colon cancer. Include at least SIX (6) points in your answer.

(12 marks)

(b) Classify the *TPX1* gene into an appropriate gene group.

(2 marks)

(c) State ONE (1) function of the type of gene mentioned in (b).

(2 marks)

(d) Explain how a single mutated allele of *TPX1* may contribute to cancer formation and appear to act dominantly.

(4 marks)

Question 4

(a) Define somatic gene therapy and germline gene therapy.

(4 marks)

(b) State TWO (2) advantages and TWO (2) disadvantages of using viral vectors in gene therapy.

(8 marks)

(c) Describe TWO (2) non-viral gene delivery methods and explain ONE (1) advantage of each method.

(6 marks)

(d) Explain ONE (1) clinical application of gene therapy.

(2 marks)

END OF EXAMINATION PAPER