

QP CODE

Reg. No:

Name:

MAHATMA GANDHI UNIVERSITY, KOTTAYAM
MGU-UGP (HONOURS) END SEMESTER EXAMINATION NOVEMBER 2026
FIFTH SEMESTER
DSE – MG5DSEZGYA00- CLINICAL GENOMICS
(2024 ADMISSION ONWARDS)

Duration: 2 Hours

Maximum Marks: 70

Remember(K), Understand(U), Apply(A), Analyse(An), Evaluate(E), Create(C), Skill(S), Interest(I) and Appreciation(Ap)

Students should attempt at least one question from each course outcome to enhance their overall outcome attainability.

Part A

Multiple Choice Questions

Answer all questions

Each question carries 1 mark

1. Which technique involves direct injection of a single sperm into an oocyte cytoplasm? [K] / [CO1]
(a) IVF (b) ICSI (c) GIFT (d) ZIFT
2. Non-invasive prenatal testing (NIPT) primarily analyses: [U] / [CO1]
(a) Maternal urine hormones (b) Cell-free foetal DNA in maternal blood
(c) Amniotic fluid cells (d) Chorionic villi tissue
3. Down syndrome is caused by: [K] / [CO2]
(a) Trisomy 13 (b) Trisomy 18 (c) Trisomy 21 (d) Monosomy X
4. Neonatal screening for inherited metabolic disorders commonly employs: [U] / [CO2]
(a) Karyotyping (b) Tandem mass spectrometry (c) FISH (d) Southern blotting
5. CYP450 enzymes are primarily involved in: [K] / [CO3]
(a) DNA replication (b) Drug metabolism (c) Protein folding (d) RNA splicing
6. Which epigenetic modification is associated with gene silencing via promoter hypermethylation? [U] / [CO3]
(a) Histone acetylation (b) DNA methylation (c) Alternative splicing (d) Nonsense-mediated decay
7. Genome-Wide Association Studies (GWAS) are primarily used to identify: [K] / [CO3]
(a) Single-gene Mendelian disorders (b) Chromosomal translocations
(c) Common disease-associated variants across the genome (d) Mitochondrial mutations
8. Cystic fibrosis is inherited in which pattern? [K] / [CO4]
(a) Autosomal dominant (b) Autosomal recessive (c) X-linked recessive (d) Mitochondrial
9. Lynch syndrome predominantly increases the risk of which cancer? [U] / [CO4]
(a) Breast cancer (b) Colorectal cancer (c) Lung cancer (d) Prostate cancer
10. Genetic counseling primarily aims to: [U] / [CO5]
(a) Cure genetic disorders (b) Help individuals understand and adapt to genetic risk information
(c) Perform gene therapy (d) Replace clinical diagnosis

[1 x 10 = 10]

Part B

Short Answer Questions

Answer any 14 questions

Each question carries 2 marks

11. Define reproductives. [U] / [CO1]
12. List the types of Preimplantation Genetic Testing (PGT-A, PGT-M, PGT-SR) with one indication each. [U] / [CO1]
13. Differentiate between amniocentesis and chorionic villus sampling (CVS). [An] / [CO1]
14. Define gestational surrogacy and mention one genomic implication. [U] / [CO1]
15. Outline the heel prick test and its significance in neonatal screening. [U] / [CO2]
16. Name any two genes involved in the genetic control of puberty onset. [K] / [CO2]
17. Briefly explain Fragile X syndrome and its genetic basis. [U] / [CO2]
18. Differentiate Turner syndrome and Klinefelter syndrome. [An] / [CO2]
19. Define pharmacogenetics and distinguish it from pharmacodynamics. [U] / [CO3]
20. Explain the significance of Warfarin as a pharmacogenetic case example. [A] / [CO3]

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| 21. | Define genomic imprinting and give one example disorder. | [U] / [CO3] |
| 22. | Differentiate DNA methylation and histone modification as epigenetic mechanisms. | [An] / [CO3] |
| 23. | Briefly explain the role of GWAS in studying TB susceptibility. | [A] / [CO3] |
| 24. | Distinguish between tumor testing and germline testing in cancer genomics. | [U] / [CO4] |
| 25. | Briefly explain whole-exome sequencing and its application in diagnosing Autism Spectrum Disorder. | [A] / [CO4] |
| 26. | List the roles of clinicians, genetic counselors, and lab geneticists in a clinical case study. | [U] / [CO5] |

Part C

Short Essay Questions

Answer any 8 questions

Each question carries 4 marks

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| 27. | Describe the indications and techniques of Preimplantation Genetic Testing (PGT) in assist reproduction, with a note on its ethical and legal aspects. | [E] / [CO1] |
| 28. | Explain the role and scope of genetic counselling in assisted reproductive technologies. | [A] / [CO1] |
| 29. | Discuss the principle, indications, and limitations of non-invasive prenatal testing (NIPT) as compared to invasive diagnostic techniques. | [An] / [CO1] |
| 30. | Analyse the genetic basis of Prader-Willi and Angelman syndromes, and explain how genomic imprinting produces their distinct phenotypes. | [An] / [CO2] |
| 31. | Describe the genetic causes of male and female infertility and discuss how genomic evaluation aids diagnosis. | [A] / [CO2] |
| 32. | Discuss the role of whole-exome/genome sequencing in the evaluation of neurogenetic disorders such as epilepsy and Huntington's disease. | [A] / [CO2] |
| 33. | Explain the concept of pharmacogenomics with reference to CYP450 polymorphisms, using Clopidogrel or Tamoxifen as a case example. | [A] / [CO3] |
| 34. | Discuss the role of epigenetic mechanisms, particularly DNA hypermethylation, in the development and progression of cancers such as BRCA1-associated breast cancer. | [An] / [CO3] |
| 35. | Present a clinical case-based approach to diagnosing Hereditary Breast and Ovarian Cancer, distinguishing tumour testing from germline testing and outlining the genetic counselling implications. | [E] / [CO4] |
| 36. | Analyse the diagnostic and counselling approach for a child with suspected Autism Spectrum Disorder, including the use of CNV analysis and exome sequencing, and discuss the ethical considerations involved. | [An] / [CO5] |

[4 x 8 = 32]