Reg. No.										

MANIPAL ACADEMY OF HIGHER EDUCATION

DM (MEDICAL GENETICS) DEGREE EXAMINATION – DECEMBER 2023

SUBJECT: PAPER I

Monday, December 11, 2023

Time: 14:00 – 17:00 Hrs. Max. Marks: 100

Answer all the following questions.

1. Describe and illustrate the mitochondrial genome and briefly explain the mitochondrial genome variants and their implications

2. Elaborate on the genetic basis of cancer

3. Describe the processes of transcription and translation

4. Describe imprinting, various imprinted regions in the genome and imprinting disorders

5. Describe and define microdeletion syndromes with examples and discuss the mechanism underlying these disorders

6. Discuss inversions and their behavior in mitosis and the clinical implications

7. Describe the various recommendation guidelines and databases for interpretation of pathogenicity of copy number variants

8. Describe the various types of RNA and their functions

9. Discuss oligogenic and multifactorial inheritance patterns

10. Describe dynamic mutations with examples

(10 marks × 10 = 100 marks)

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MANIPAL ACADEMY OF HIGHER EDUCATION

DM (MEDICAL GENETICS) DEGREE EXAMINATION – DECEMBER 2023

SUBJECT: PAPER II

Tuesday, December 12, 2023

Time: 14:00 – 17:00 Hrs. Max. Marks: 100

✍ **Answer all the following questions.**

1. Write briefly on approach to echogenic cardiac focus in a fetus
2. Discuss the genetic approach to hearing loss
3. Write briefly on hereditary cancer predisposition syndromes
4. Discuss the genetic approach to diagnosis of isolated intellectual disability
5. Elaborate the genetic conditions with overgrowth
6. Discuss the etiology, genetics, and approach to congenital ichthyosis
7. Enumerate the clinical features, genetic diagnosis and management of neurofibromatosis type 1
8. What are the principles involved in breaking bad news?
9. Discuss the diagnosis and approach to adult-onset ataxia
10. Define non-immune fetal hydrops. Discuss the etiology and approach to non-immune hydrops

(10 marks × 10 = 100 marks)

Reg. No.										

MANIPAL ACADEMY OF HIGHER EDUCATION

DM (MEDICAL GENETICS) DEGREE EXAMINATION – DECEMBER 2023

SUBJECT: PAPER III

Wednesday, December 13, 2023

Time: 14:00 – 17:00 Hrs. Max. Marks: 100

✍ **Answer all the following questions.**

1. Describe the testing strategy for diagnosing a simplex case of intellectual disability and dysmorphism

2. Discuss the various methods used for diagnosis of dynamic mutations or repeat disorders

3. Discuss the current applications of karyotype and chromosomal microarray

4. Discuss preimplantation genetic diagnoses

5. Discuss newborn screening

6. Discuss the various techniques used for RNA and protein analysis

7. Discuss the principle of multiplex ligation-dependent probe amplification and it's uses

8. Discuss and compare exome and genome sequencing for diagnoses of rare genetic disorders

9. Laboratory testing for premature ovarian failure

10. Discuss principle, technique and utility of Quantitative fluorescent PCR.

(10 marks × 10 = 100 marks)

Reg. No.										

MANIPAL ACADEMY OF HIGHER EDUCATION

DM (MEDICAL GENETICS) DEGREE EXAMINATION – DECEMBER 2023

SUBJECT: PAPER IV

Thursday, December 14, 2023

Time: 14:00 – 17:00 Hrs. Max. Marks: 100

Answer all the following questions.

1. Discuss the recent perspectives in gene therapy for treatment of beta thalassemia

2. What are stem cells? Discuss the indications (genetic disorders) for stem cell transplantation. Enumerate the utility of umbilical cord blood as a source of stem cells.

3. Discuss the recent ACMG practice resources on screening for autosomal recessive and X linked conditions during pregnancy and preconception.

4. Write briefly on gene silencing by RNA interference.

5. Discuss the recent advances in treatment of Duchenne muscular dystrophy.

6. What is fetal therapy? Discuss the different modalities and their current status.

7. Discuss the utility, advantages and disadvantages of targeted gene panel testing by next generation sequencing technique

8. Write briefly on the advances in the diagnosis of triplet repeat disorders.

9. What are secondary findings? Discuss the domains of evaluation to be addressed when a secondary finding is identified.

10. What are pseudogenes? Discuss the implications in diagnosis of rare diseases.

(10 marks × 10 = 100 marks)