Reg. No.										

MANIPAL ACADEMY OF HIGHER EDUCATION

DM (MEDICAL GENETICS) DEGREE EXAMINATION – JULY 2022

SUBJECT: PAPER I

Monday, July 18, 2022

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

✍ **Answer all the following questions.**

1. Define reduced penetrance. Discuss the clinical significance.

2. What is translocation? What are the different types of translocations? Discuss the clinical significance of a balanced translocation.

3. Write briefly on X chromosome inactivation and clinical relevance.

4. How is the human genome organised?

5. What is a gain of function mutation? What are the features of a gain of function mutation? Illustrate with examples.

6. What is a pedigree? What are the different symbols used in pedigree drawing? Discuss the utility of pedigree in clinic.

7. Write briefly on the molecular pathology of mitochondrial disorders.

8. Discuss genotype-phenotype correlation.

9. What are oncogenes? Discuss protooncogenes and tumour suppressor genes with examples.

10. What is mosaicism? What are the clinical consequences?

(10 marks × 10 = 100 marks)

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MANIPAL ACADEMY OF HIGHER EDUCATION

DM (MEDICAL GENETICS) DEGREE EXAMINATION – JULY 2022

SUBJECT: PAPER II

Tuesday, July 19, 2022

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

✍ **Answer all the following questions.**

1. Write briefly on approach to fetal ventriculomegaly.
2. Enumerate the utility of fetal autopsy.
3. Define polydactyly. Write briefly on the types and genetic approach to polydactyly.
4. Discuss the approach to diagnosis of DNA repair disorders.
5. Elaborate the principles of genetic counseling.
6. Discuss the etiology, genetics and approach to congenital cataract
7. A newborn has arthrogryposis multiplex congenita. Elaborate on the genetic evaluation.
8. Genetic approach to primary immune deficiency diseases
9. Discuss the diagnosis and management of spinal muscular atrophy.
10. Define phenocopies. Discuss the impact of some of the common teratogens.

(10 marks × 10 = 100 marks)

Reg. No.										

MANIPAL ACADEMY OF HIGHER EDUCATION

DM (MEDICAL GENETICS) DEGREE EXAMINATION – JULY 2022

SUBJECT: PAPER III

Wednesday, July 20, 2022

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

✍ **Answer all the following questions.**

1. Discuss homozygosity mapping as a tool for a clinical geneticist.

2. Discuss the different types of variants and nomenclature.

3. Enumerate the different variant databases used in clinical genetics.

4. What is whole genome sequencing? What are the indications, advantages, and disadvantages of whole genome sequencing?

5. Define variant of uncertain significance. How do you approach a patient with a variant of uncertain significance?

6. What are the different techniques used for diagnosis of triplet repeat disorders?

7. Discuss the advantages and disadvantages of chromosomal microarray in comparison to karyotyping.

8. Discuss the relevance of newborn screening.

9. What is noninvasive prenatal screening? Discuss the technology. What are the advantages and disadvantages?

10. Discuss the various techniques in invasive prenatal testing.

(10 marks × 10 = 100 marks)

Reg. No.										

MANIPAL ACADEMY OF HIGHER EDUCATION

DM (MEDICAL GENETICS) DEGREE EXAMINATION – JULY 2022

SUBJECT: PAPER IV

Thursday, July 21, 2022

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

Answer all the Following questions.

1. Discuss the recent advances in the therapeutics of Duchenne muscular dystrophy.
2. Describe antisense oligonucleotide (ASO) therapies and their current status.
3. Discuss the various treatment modalities for lysosomal storage disorders.
4. Discuss the current applications of hematopoietic stem cell transplantation.
5. Compare the various in-vivo model systems for investigation of genetic diseases.
6. Discuss the current role of artificial intelligence in genetic diagnoses.
7. Discuss personalized medicine with examples.
8. Discuss the technique of optical genome mapping and its applications.
9. Describe the various modalities for fetal therapy.
10. Discuss the current recommendations for carrier screening for genetic disorders.

(10 marks × 10 = 100 marks)