

M.Sc Semester-2 Examination

409

Human Genetics

April-2024

[Max. Marks : 70]

Time : 2-30 Hours]

Q-I	1	What is Mosaicism? Explain the types and causes giving examples.	(14)
	2	Discuss Uniparental Disomy in detail and briefly explain any two associated disorders.	
	OR		
	1	Write an account on any 3 disorders inherited in an Autosomal Dominant manner.	
	2	Explain the phenomenon of Genomic Imprinting, citing the related disorders.	
Q-II	1	Explain various types of diabetes.	(14)
	2	Write a note on "Congenital cataract".	
	OR		
	1	Describe any one mitochondrial disorder in detail.	
	2	Write a note on "Macular degeneration".	
Q-III	1	Explain briefly: Importance of biochemical tests in diagnosis.	(14)
	2	Give a note on "Calibration" in a pathology laboratory.	
	OR		
	1	What are the key roles of QA & QC in UV-visible spectrophotometer?	
	2	Explain briefly: Importance of molecular diagnosis with an example.	
Q-IV	1	What is Thalassemia? Write down different types of Thalassemia in detail.	(14)
	2	What are the different types of Albinism, explaining their genetics and features	
	OR		
	1	Write a note on genetical causes of female infertility.	
	2	What is hermaphroditism? Write a detailed account.	
Q-V	Answer any SEVEN out of TWELVE.		(14)
	1	Define: Frequency of occurrence; Lifetime Risk of a disorder.	02
	2	Distinguish between Penetrance and Expressivity.	02
	3	What is a Phenocopy? Give one example.	02
	4	What is aniridia? Mention any two mutations responsible for aniridia.	02
	5	Enlist any four factors associated with obesity.	02
	6	Mention any four factors associated with high incidence of diabetes in Gujarat.	02
	7	Explain "Dementia".	02
	8	How is chorea characterized?	02
	9	What is a screening test?	02
	10	Write a note on genetical causes of female infertility.	02
	11	What is the inheritance pattern of Tuberous Sclerosis.	02
	12	Which gene is altered in Marfan syndrome.	02