National Board of Examinations

Question Paper Name :	DrNB Medical Genetics Paper1
Subject Name :	DrNB Medical Genetics Paper1
Duration :	180
Total Marks :	100
Display Marks:	No
Maximum Instruction Time :	0

Question Number : 1 Question Id : 32718740544 Consider As Subjective : Yes

Please write your answers in the answer booklet within the allotted pages as follows:-

Question Number	Answer to be attempted within	Question Number	Answer to be attempted within
Q. 1	Page 1-5	Q. 6	Page 26-30
Q. 2	Page 6-10	Q. 7	Page 31-35
Q. 3	Page 11-15	Q. 8	Page 36-40
Q. 4	Page 16-20	Q. 9	Page 41-45
Q. 5	Page 21-25	Q. 10	Page 46-50

1. a) Name three X-linked dominant disorders. [3]
b) Draw a four generation pedigree depicting this inheritance pattern for any one of the disorders you have listed. [4]
c) How is this pattern of inheritance different from the X-linked recessive inheritance? [3]

Question Number : 2 Question Id : 32718740545 Consider As Subjective : Yes

a) Define genomic imprinting. [2]
b) Detail the characteristics of this pattern of inheritance. [4]
c) Give a diagrammatic representation of the gene cluster on chromosome 11p15 with mechanism of disease for Beckwith-Weideman syndrome. [4]

Question Number : 3 Question Id : 32718740546 Consider As Subjective : Yes

A family comes for preconception counseling as the grandfather and father are affected with tuberous sclerosis. The grandfather has only hypopigmented macules and ungual fibromas and a pathogenic variant in TSC2 gene. The father has retinal nodular hamartoma, angiofibromas and hypopigmented macules. On evaluation, there is a renal angiomyolipoma. Enumerate the challenges in counseling and name the mechanisms responsible for these challenges. [10]

Question Number : 4 Question Id : 32718740547 Consider As Subjective : Yes

What are supernumerary or marker chromosomes? How will you characterize a marker chromosome? What are the clinical implications and how would you proceed to attribute the phenotype to the marker chromosome? Give two examples of a genetic syndrome due to a marker chromosome. [2+2+4+2]

Question Number : 5 Question Id : 32718740548 Consider As Subjective : Yes

a) Which recommendations are used for gene variant nomenclature? [2]
b) Use this recommendation to describe this sequence variant: Gene – COL1A1 Variant c.2489G>C in exon 36 p.Gly830Ala Zygosity – heterozygous Disease – osteogenesis imperfecta Classification- Likely pathogenic Transcript - NM_000088.4. [4]
c) Can you refine it with scoring system? [2]
d) What are different transcript IDs that can be used? Which method is widely accepted at present? [2]

Question Number : 6 Question Id : 32718740549 Consider As Subjective : Yes

You are given the responsibility of starting a newborn screening program. What is the standard operating procedure you want to establish for newborn screening for congenital hypothyroidism? [10]

Question Number : 7 Question Id : 32718740550 Consider As Subjective : Yes

a) National Sickle Cell Anemia Elimination Mission. [5]
b) Importance, utility and feasibility of such a mission program. [5]

Question Number : 8 Question Id : 32718740551 Consider As Subjective : Yes

What are the strategies for new disease gene identification by next generation sequencing techniques? [10]

Question Number : 9 Question Id : 32718740552 Consider As Subjective : Yes

Deformation, malformation, disruption, dysplasia, sequence and multiple malformation syndrome – define each of these and give two examples for each of them. [4+6]

Question Number : 10 Question Id : 32718740553 Consider As Subjective : Yes

Highlight, with one example each, the situations where a gene disease variant, for a monogenic disease, are missed on exome sequencing. Name the technology and strategy to identify such a variant? [5+5]