National Board of Examinations

Question Paper Name :	DrNB Medical Genetics Paper1
Subject Name :	DrNB Medical Genetics Paper1
Creation Date :	2022-06-25 17:18:18
Duration :	180
Share Answer Key With Delivery Engine :	No
Actual Answer Key :	No

DrNB Medical Genetics Paper1

Group Number :	1
Group Id :	3271871204
Group Maximum Duration :	0
Group Minimum Duration :	180
Show Attended Group? :	No
Edit Attended Group? :	No
Break time :	0
Group Marks :	100
Is this Group for Examiner? :	No
Examiner permission :	Cant View
Show Progress Bar? :	No

DrNB Medical Genetics Paper1

Section Id :	3271871207
Section Number :	1

Section type :	Offline
Mandatory or Optional :	Mandatory
Number of Questions to be attempted :	10
Section Marks :	100
Enable Mark as Answered Mark for Review and Clear Response :	Yes
Maximum Instruction Time :	0
Sub-Section Number :	1
Sub-Section Id :	3271871211
Question Shuffling Allowed :	No

Question Number : 1 Question Id : 32718711092 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

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. Describe single nucleotide variants, structural variants and their types. [2+2+6]

Question Number : 2 Question Id : 32718711093 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

What are mitochondrial disorders? Draw a typical pedigree for a mitochondrial disorder. Explain heteroplasmy using a diagram. [3+3+4]

Question Number : 3 Question Id : 32718711094 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Discuss briefly isochromosomes, its mechanism and provide two examples. [3+3+4]

Question Number : 4 Question Id : 32718711095 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Explain Uniparental disomy, its types and detection methods. [2+4+4]

Question Number : 5 Question Id : 32718711096 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

What is RNA splicing? Explain alternative splicing and its types using diagrams. [3+3+4]

Question Number : 6 Question Id : 32718711097 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

a) Explain locus and allelic heterogeneity. [5]

b) Elucidate the difference between the two with examples. [5]

Question Number : 7 Question Id : 32718711098 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Describe mosaicism and its different types. Give two examples. What are the various approaches to detect mosaicism? [4+2+4]

Question Number : 8 Question Id : 32718711099 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Explain identity-by-descent and homozygosity mapping of recessive disorders with examples. [5+5]

Question Number : 9 Question Id : 32718711100 Question Type : SUBJECTIVE Consider As

Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Explain three major mechanisms of disease in autosomal dominant disorders using one example for each. [4+3+3]

Question Number : 10 Question Id : 32718711101 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

What are genetic markers? List their uses in clinical practice. [5+5]