National Board of Examinations

Question Paper Name :	DrNB Medical Genetics Paper2
Subject Name :	DrNB Medical Genetics Paper2
Creation Date :	2022-06-25 17:19:59
Duration :	180
Share Answer Key With Delivery Engine :	No
Actual Answer Key :	No

DrNB Medical Genetics Paper2

Group Number :	1
Group Id :	3271871205
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 Paper2

Section Id :	3271871208
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 :	3271871212
Question Shuffling Allowed :	No

Question Number : 1 Question Id : 32718711102 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. Enumerate and describe the various classes of molecular defects in CFTR gene. What are the pleiotropic effects observed in cystic fibrosis? [7+3]

Question Number : 2 Question Id : 32718711103 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

A 6 month old boy, born to a 3rd degree consanguineous couple, presented with cholestasis since neonatal age.Tandem mass spectrometry was suggestive of mildly raised citrulline, normal succinylacetone levels with normal liver function tests. Blood ammonia levels were raised at 2 month age (upto 180 microg/dl) and were subsequently normal on further visits. Ophthalmic evaluation was suggestive of retinal pigmentary epithelial atrophy. A provisional diagnosis of

citrullinemia type 2 was made, however all the features could not be explained by this diagnosis. Exome sequencing delineated two pathogenic homozygous variants in two different genes suggesting a blended phenotype and explained all the features of the above case.

a) Suggest the likely diagnostic possibilities in this case. [5]

b) Discuss difficulties in counselling in situations suggesting a blended phenotype. [5]

Question Number : 3 Question Id : 32718711104 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

a) Newborn screening for congenital hypothyroidism performed on a 3 day old full term baby demonstrates a TSH of 70 mIU/L on dried blood spot testing. Illustrate the further steps to be performed in this neonate. [5]

b) You are called to the nursery to evaluate a newborn baby with Down syndrome. Write your approach and steps in counselling the family. [5]

Question Number : 4 Question Id : 32718711105 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Write an approach to a 4-year-old child with obesity. [10]

Question Number : 5 Question Id : 32718711106 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Explain how a single gene defect can result in defects in multiple enzymes. Give 2 illustrative examples. [4+6]

Question Number : 6 Question Id : 32718711107 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

The diagnostic yield of whole exome sequencing is not 100%. Write the 4 possible explanations. [10]

Question Number : 7 Question Id : 32718711108 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Genotype–phenotype correlation in the FGFR genes. [10]

Question Number : 8 Question Id : 32718711109 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

A 33 year old primigravida visits for genetic counselling for antenatal detection of bilateral pleural effusion, and ascites at 18 weeks of gestation. Her dual marker risk shows high risk for trisomy 21. She also had history of maternal fever at around 16 weeks. Write your approach for this patient. [10]

Question Number : 9 Question Id : 32718711110 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Write an approach to Homocystinuria. [10]

Question Number : 10 Question Id : 32718711111 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

A 6-month-old male developmentally normal baby referred for the evaluation of hepatosplenomegaly. He had history of neonatal cholestasis. There is no history of fever, visual or hearing or joint involvement. Anthropometry examination was unremarkable. He had fading mongolion spots at the back. There was no dysmorphism. Write your approach to diagnosis. [10]