National Board of Examinations

Question Paper Name :	DrNB Medical Genetics Paper2
Subject Name :	DrNB Medical Genetics Paper2
Creation Date :	2021-08-25 22:35:05
Duration :	180
Total Marks :	100
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DrNB Medical Genetics Paper2

Group Number :	I
Group Id:	327187477
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

DrNB Medical Genetics Paper2

Section 1a :	32/18/480
Section Number :	1
Section type:	Offline

Mandatory or Optional : Mandatory

Number of Questions: 10

Number of Questions to be attempted: 10

Section Marks: 100

Enable Mark as Answered Mark for Review and

Clear Response:

Yes

Sub-Section Number: 1

Sub-Section Id: 327187484

Question Shuffling Allowed: No

Question Number: 1 Question Id: 3271873291 Question Type: SUBJECTIVE

Correct Marks: 10

Diagnosis and management of Gaucher disease. [5+5]

Question Number: 2 Question Id: 3271873292 Question Type: SUBJECTIVE

Correct Marks: 10

Discuss an approach to a case of prenatally detected aplasia of corpus callosum at 22 weeks of gestation. List the possible disorders and approach to genetic testing and counseling. [5+5]

Question Number: 3 Question Id: 3271873293 Question Type: SUBJECTIVE

Correct Marks: 10

Discuss disorders with increased bone density. [10]

Question Number: 4 Question Id: 3271873294 Question Type: SUBJECTIVE

Correct Marks: 10

Discuss the differential diagnosis for a 2 year old child presenting with 2 episodes of hypoglycemia. List tests used to identify the causes in which hypoglycemia is a predominant feature. Enlist steps in emergency management of this case. [5+3+2]

Question Number: 5 Question Id: 3271873295 Question Type: SUBJECTIVE

Correct Marks: 10

Free floating DNA in plasma and its role in diagnosis. [5+5]

Question Number: 6 Question Id: 3271873296 Question Type: SUBJECTIVE

Correct Marks: 10

Successful gene therapies for monogenic disorders. [10]

Question Number: 7 Question Id: 3271873297 Question Type: SUBJECTIVE

Correct Marks: 10

A family with consanguinity has a child with renal tubular acidosis and deafness. The child is receiving satisfactory treatment for renal tubular acidosis and has been showing improved growth. The disorders is inherited in an autosomal recessive fashion. Discuss the points to be covered in genetic counseling and issues related to prenatal diagnosis of a treatable disorder with this phenotype. [5+5]

Question Number: 8 Question Id: 3271873298 Question Type: SUBJECTIVE

Correct Marks: 10

Discuss genetic diagnosis of a 35 year old woman with carcinoma breast and issues to be discussed in genetic counseling. [4+6]

Question Number: 9 Question Id: 3271873299 Question Type: SUBJECTIVE

Correct Marks: 10

Discuss issues related to secondary findings in genetic testing based on Next Generation sequencing. Give important groups of actionable genes. [5+5]

Question Number: 10 Question Id: 3271873300 Question Type: SUBJECTIVE

Correct Marks: 10

Discuss approach to a 2 month old boy presenting with hypotonia. List the possible differential diagnosis, their clinical clues and diagnostic tests to be performed with this clinical presentation. [5+5]