

National Board of Examinations

Question Paper Name :	DrNB Medical Genetics Paper2
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DrNB Medical Genetics Paper2

Group Number :	1
Group Id :	3271872842
Group Maximum Duration :	0
Group Minimum Duration :	180
Show Attended Group? :	No
Edit Attended Group? :	No
Break time :	0
Group Marks :	100

DrNB Medical Genetics Paper2

Section Id :	3271872845
Section Number :	1
Section type :	Offline
Mandatory or Optional :	Mandatory
Number of Questions :	10
Number of Questions to be attempted :	10
Section Marks :	100
Maximum Instruction Time :	0
Sub-Section Number :	1
Sub-Section Id :	3271872849
Question Shuffling Allowed :	No

Question Number : 1 Question Id : 32718729704 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

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) What do you understand by carrier screening and expanded carrier screening? [3]
- b) What are the anticipated challenges? [3]
- c) List a few conditions and justify for including them in the screening program for India. [4]

Question Number : 2 Question Id : 32718729705 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

Illustrate a geneticist's approach to autism spectrum disorders. [10]

Question Number : 3 Question Id : 32718729706 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

A 28-year-old lady comes for counseling for a previous termination with hypoplastic left heart syndrome. In her second pregnancy, fetus had right aortic arch, left ductus origin with left subclavian artery with thymic hypoplasia. What will be your approach towards this patient? [10]

Question Number : 4 Question Id : 32718729707 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

Discuss PIK3CA related overgrowth syndromes and its classification. What are the treatment options available? [6+4]

Question Number : 5 Question Id : 32718729708 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

A 5-year-old female with normal development is admitted in emergency for severe anemia and epistaxis. There is no fever. She has poor vision and moderate hepatosplenomegaly. Her investigations were unremarkable except hemoglobin of 5 g/dl and platelet count of 50000 per cumm.

- a) What are the likely differentials? [4]
- b) Write management approach for this child. [6]

Question Number : 6 Question Id : 32718729709 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

A 32-year-old lady comes for genetic counseling at 14 weeks' gestation for previous child diagnosed as monogenic obesity by Exome sequencing at 2 years of age. Report showed a variant of unknown significance in LEPR. Her combined risk shows a risk of 1 in 100 for trisomy 21. Discuss the approach for this family. [10]

Question Number : 7 Question Id : 32718729710 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

A one-month old baby was brought with newborn screening report showing phenylalanine levels of 3.6 mg/dl. How will you approach this baby for further management? [10]

Question Number : 8 Question Id : 32718729711 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

Write a note on cobalamin related disorders, their diagnosis and management. [3+3+4]

Question Number : 9 Question Id : 32718729712 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

Discuss the management of Hemophilia A and B. [5+5]

Question Number : 10 Question Id : 32718729713 Question Type : SUBJECTIVE Consider As Subjective : Yes

Correct Marks : 10

Explain birth defects and importance of birth defect registries. [6+4]