

National Board of Examinations

Question Paper Name :	DrNB Medical Genetics Paper2
Subject Name :	DrNB Medical Genetics Paper2
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Actual Answer Key :	No

DrNB Medical Genetics Paper2

Group Number :	1
Group Id :	327187790
Group Maximum Duration :	0
Group Minimum Duration :	180
Show Attended Group? :	No
Edit Attended Group? :	No
Group Marks :	100
Is this Group for Examiner? :	No
Examiner permission :	Cant View
Show Progress Bar? :	No

DrNB Medical Genetics Paper2

Section Id :	327187793
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 :	327187797
Question Shuffling Allowed :	No
Is Section Default? :	null

Question Number : 1 Question Id : 3271876982 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

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. How will you counsel a pregnant woman detected to have dilated pelvi-calyceal system on sonography at 18 weeks gestation. Which tests will you consider to detect any genetic abnormality in the fetus? [7+3]

Question Number : 2 Question Id : 3271876983 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Correct Marks : 10

Which conditions are considered for presymptomatic testing. Give three examples and discuss ethical issues in such scenarios. [4+6]

Question Number : 3 Question Id : 3271876984 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Correct Marks : 10

A 25-year-old male comes with history of adenocarcinoma of colon. Which genes are implicated in such situation? How will you approach to reach the diagnosis and provide genetic counselling?

[3+4+3]

Question Number : 4 Question Id : 3271876985 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Correct Marks : 10

A family consulted you for risk calculation for hyperlipidemia in a 12-year-old boy with family history of same in grandfather. Discuss the approach to be followed for risk calculation. Write notes on management of this condition. [7+3]

Question Number : 5 Question Id : 3271876986 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Correct Marks : 10

Write down management of antenatally detected Congenital High Airway Obstruction Syndrome (CHAOS) at 30 weeks and discuss the role of a multidisciplinary team? What are the challenges faced when counselling such a couple in a perinatal setting? [7+3]

Question Number : 6 Question Id : 3271876987 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Correct Marks : 10

Write down steps in counseling of a family with Trisomy 21 detected in the surrogate mother at 21 weeks gestation? [10]

Question Number : 7 Question Id : 3271876988 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Correct Marks : 10

Enumerate determinants of Quality of Life (QoL) for rare disabling genetic conditions giving two disease examples from a pediatric and an adult group. Discuss your role as a Genetic expert? [7+3]

Question Number : 8 Question Id : 3271876989 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Correct Marks : 10

A 15-year-old girl presented with primary amenorrhea with Sexual Maturity Rating of stage II. Her pelvic sonography showed a uterus of 2 X 2 cm and bilateral ovaries of 1.4 x 1 cm size. Her Follicle Stimulating Hormone level (FSH) was 80 IU/ml and Karyotype was 46, XY. How will you approach this child and counsel the family? [5+5]

Question Number : 9 Question Id : 3271876990 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Correct Marks : 10

A healthy term neonate presents with encephalopathy at 5 days of life. His basic profile reveals an ammonia level of 580 ug/dl. His lactate levels are also raised 3.2 mmol/L. Discuss further tests to arrive at a diagnosis in this case. Enlist steps in the management of this case. [6+4]

Question Number : 10 Question Id : 3271876991 Question Type : SUBJECTIVE Consider As Subjective : Yes Calculator : None Response Time : N.A Think Time : N.A Minimum Instruction Time : 0

Correct Marks : 10

Write down challenges in classifying skeletal dysplasias. Mention salient features of recent Nosology and classification of skeletal dysplasia (2019). What are challenges in genetic diagnosis

of Osteogenesis Imperfecta? [3+4+3]