

National Board of Examinations

Question Paper Name : DrNB Medical Genetics Paper2
Subject Name : DrNB Medical Genetics Paper2
Duration : 180
Total Marks : 100
Display Marks: No

Maximum Instruction Time : 0

Question Number : 1 Question Id : 32718740554 Consider As Subjective : Yes

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
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Q. 5	Page 21-25	Q. 10	Page 46-50

1. The lady is 39 year old and her husband is 42 year old. She is married to her first cousin. Their first child, 12 year old, has congenital adrenal hyperplasia and harbors a homozygous pathogenic variant in CYP21A2 gene. She has come for preconception counseling for their next pregnancy, which they consider very precious in view of the parental ages. State specifically pointwise, your counselling and risk estimation for this family. [10]

Question Number : 2 Question Id : 32718740555 Consider As Subjective : Yes

Write the approach to a 5-year-old child with macrocephaly and developmental delay. [10]

Question Number : 3 Question Id : 32718740556 Consider As Subjective : Yes

Amniocentesis was performed for a positive aneuploidy screen for trisomy 21. The karyotype report confirms that the fetus has Klinefelter syndrome: 47, XXY. How will you counsel this family? [10]

Question Number : 4 Question Id : 32718740557 Consider As Subjective : Yes

Explain the indications, utility and caveats of "Liver transplantation" for inherited diseases. [10]

Question Number : 5 Question Id : 32718740558 Consider As Subjective : Yes

Draw the organization of the alpha and beta globin gene clusters. Describe the genotype and phenotype of alpha thalassemia. [5+5]

Question Number : 6 Question Id : 32718740559 Consider As Subjective : Yes

The second trimester anomaly scan in a low risk primigravida reports short long bones in the upper and lower limbs. What can be the causes? What are the markers in the ultrasound for lethal skeletal dysplasia and what should be the approach for confirmation of the type of lethal skeletal dysplasia? [5+5]

Question Number : 7 Question Id : 32718740560 Consider As Subjective : Yes

A 5-day-old male newborn was brought to the NICU with lethargy after a period of appropriate feeding. He has an ammonia of 750 micromol/L and a normal arterial blood gas analysis. There is a family history of a death of a sibling at 10 days of age with similar complaints. Describe the investigations to reach a diagnosis and the management of the neonate in the nursery and counseling issues for the family. [10]

Question Number : 8 Question Id : 32718740561 Consider As Subjective : Yes

Describe the management options of a 4-month-old child with spinal muscular atrophy type 1. [10]

Question Number : 9 Question Id : 32718740562 Consider As Subjective : Yes

A third degree consanguineous couple are found to be carriers of both phenylketonuria and beta thalassemia. They are referred for preconception genetic counselling in view of their carrier screening report – how will you counsel them? They are prepared to treat phenylketonuria if the neonate is affected – how will you manage this clinical scenario? [7+3]

Question Number : 10 Question Id : 32718740563 Consider As Subjective : Yes

Enumerate syndromes with primary craniosynostosis. Provide salient features to differentiate them clinically. [10]