

Rajiv Gandhi University of Health Sciences, Karnataka

Fellowship Examination – 22-Aug-2024

[Time: 3 Hours]**[Max. Marks: 100]**

PAEDIATRIC GENETICS – PAPER-I

QP Code: 4141

Your answers should be specific to the questions asked
Draw neat labeled diagrams wherever necessary

Answer All The Questions**10 X 10 = 100 Marks**

1. Evaluation and management of a child with Gaucher disease.
2. Genetics of beta thalassemia and counselling
3. Osteogenesis imperfect genetic abnormalities and clinical manifestations.
4. Cell free fetal DNA and its use in prenatal diagnosis
5. Preimplantation genetic diagnosis (PGD).
6. Short femur in a fetus, its implications and recurrence.
7. An approach to congenital cataract.
8. Name some of the syndromes associate with radial ray defect and describe any one.
9. Phenylketonuria the biochemical basis and its management.
10. Sensorineural hearing loss (SNHL) and the commonest gene associated with non-syndromic SNHL.

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