

Rajiv Gandhi University of Health Sciences, Karnataka

Fellowship Examination – 13-Sep-2022

[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. Clinical features, genetics and management of Pompe disease.
2. Management and follow up of a child with Glycogen storage disorder Type 1.
3. Genetic counselling and management of a child with Duchenne muscular dystrophy.
4. Genetic syndromes with hair abnormalities.
5. Long-term IV immunoglobulin maintenance therapy for Primary Immune Deficiency (PID).
6. Genetic counselling and management of children with Sickle Cell anemia.
7. Clinical features and molecular abnormality in Cri-du-chat syndrome.
8. Brief note on genetic anticipation, penetrance and variable expressivity with examples.
9. Approach to a fetus with stippled calcification of bony epiphyses detected during antenatal scan.
10. Brief note on exome sequencing and its clinical utility.

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