

Rajiv Gandhi University of Health Sciences, Karnataka

Fellowship Examination – 07-Sep-2023

[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. Diagnosis and management of a child with infantile onset Pompe disease.
2. Genetics of sickle cell anemia and newer therapies.
3. Ellis van Creveld syndrome.
4. Chorionic villous sampling its application in prenatal diagnosis.
5. What are some of the uses of a fetal MRI and when can it be done in pregnancy?
6. What are the implications of stippled calcification of epiphyses in a fetus?
7. Name three genetic syndromes associated with retinitis pigmentosa along with their inheritance pattern.
8. What are the conditions in which hemi-vertebrae may be seen? List the clinical features in any one of them.
9. Role of bone marrow transplantation in genetic disorders.
10. Organic acidemias.

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