Rajiv Gandhi University of Health Sciences, Karnataka

Fellowship Examination – 08-Sep-2023

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

PAEDIATRI GENETICS – PAPER-II

QP Code: 4142

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 recent advances in the management of Prader Willi syndrome.
2. Discuss genetic testing in a child of with Achondroplasia and write a brief note on the newer advances in the treatment of Achondroplasia.
3. Ethical issues in prenatal diagnosis.
4. Clinical features and genetic counselling in Edward syndrome.
5. Amniotic band sequence.
6. Approach to a child with congenital bilateral profound hearing loss.
7. Substrate reduction therapy (SRT).
8. Role of Hematopoetic stem cell transplantation in genetic disorders.
9. Clinical feature and genetics of ataxia telangiectasia.
10. Genetics and management of osteogenesis imperfecta.

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