Rajiv Gandhi University of Health Sciences, Karnataka

Fellowship Examination – 24-Aug-2021

[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. Enzyme replacement therapy in Lysosomal storage disorders.
2. Clinical features and inheritance pattern in Fanconi pancytopenia.
3. Achondroplasia genetic alteration and therapies.
4. Amniocentesis in prenatal diagnosis.
5. Nuchal translucency ultrasound scan in a fetus.
6. Cherry red sport in the eye.
7. Congenital sensorineural hearing loss an approach to diagnosis.
8. Sturge- Weber syndrome.
9. Primary immune deficiency (PID) inheritance and treatment.
10. Trisomy 13 syndrome.

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