

Rajiv Gandhi University of Health Sciences, Karnataka

Fellowship Examination – 23-Aug-2024

[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 approach to a floppy infant.
2. Dermatological manifestations in Tuberous sclerosis.
3. Marfan syndrome clinical manifestations and inheritance pattern.
4. Genetic counselling in Turner syndrome.
5. Multiplex ligation – dependent probe amplification (MLPA) its use in clinical diagnosis.
6. Bardet – Biedel syndrome clinical manifestations and inheritance pattern.
7. Give an example of imprinting disorders and write a brief note any one of them.
8. What are the clinical manifestations of chromosomal triploidy and what are the mechanisms that cause it?
9. Clinical manifestations in Chrome 22q microdeletion syndrome.
10. What are the disorders in which Exon skipping (AON) therapy has been attempted?

* * * * *