

# Rajiv Gandhi University of Health Sciences, Karnataka

## Fellowship Examination – 25-Aug-2021

**[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. Approach to a child with short stature.
2. Fragile X syndrome genetic inheritance pattern.
3. Neurofibromatosis type I manifestations and inheritance pattern.
4. Genetic counselling in Trisomy 21 Down Syndrome.
5. Newer therapies for Spino – Muscular Atrophy (SMA).
6. Fluorescence in situ hybridization (FISH) uses in clinical diagnosis.
7. Prader – Willi syndrome clinical manifestations and type of genetic defect causing it.
8. X-linked inheritance pattern.
9. Counselling in a couple where one partner has a balanced chromosomal translocation.
10. Polycystic kidney disease types and inheritance patterns.

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