[LJ 1016] OCTOBER 2016 **Sub. Code: 0404**

FELLOWSHIP IN MEDICAL GENETICS EXAMS
FIRST YEAR
PAPER IV – GENETIC TECHNIQUES AND BIOSTATISTICS

Q.P. Code :230404

Time : Three hours **Maximum : 100 Marks**

I. Elaborate on: **(2 x 20=40)**

1. What is the progress and major challenges for implementation of next generation sequencing in clinical practice?

2. What is Hardy Weinberg Equilibrium (HWE)? What are the conditions when a population deviates from HWE?

II. Write notes on: **(10 x 6 = 60)**

1. LOD score.
2. Principles for performing enzyme assays for storage disorders.
3. Copy number variation in relation to disease and health.
4. Informed consent.
5. Homozygosity mapping.
6. Compare the techniques of Amniocentesis and Chorionic villus sampling for prenatal diagnosis.
7. Describe different PCR based techniques to identify mutations in single gene disorders.
8. Threshold effect for multi-factorial disorders with reference to congenital malformations.
9. Biochemical markers for Aneuploidy screening.
10. FISH for common microdeletion syndromes.

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[LL 1017] OCTOBER 2017 **Sub. Code: 0404**

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FIRST YEAR
PAPER IV – GENETIC TECHNIQUES AND BIOSTATISTICS

Q.P. Code: 230404

Time: Three hours **Maximum: 100 Marks**

I. Elaborate on: **(2 x 20 = 40)**

1. Discuss the methods used for screening for neural tube defects.

2. Discuss the different types of polymerase chain reactions used in diagnosis.

II. Write notes on: **(10 x 6 = 60)**

1. Gonadal mosaicism.
2. Restriction fragment length polymorphisms.
3. Methods of testing for Fragile X syndrome.
4. Positive and negative predictive values.
5. Pre-implantation genetic diagnosis.
6. Use of mean, median, mode, standard deviation and centiles in analysis.
7. Calculation of genetic risk.
8. Next generation sequencing.
9. DNA microarray testing.
10. Urinary markers in screening for Down syndrome.

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