[LJ 1016] OCTOBER 2016 **Sub. Code: 0402**

FELLOWSHIP IN MEDICAL GENETICS EXAMS
FIRST YEAR
PAPER II – CYTOGENETICS

Q.P. Code :230402

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

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

1. Describe the protocol and practical consideration for first trimester second trimester or combined screening for down syndrome and NTDs.

2. Discuss the principles and practice of non directive genetic counseling relating to reproductive choices presymptomatic, diagnostic, carrier and prenatal testing for families affected by genetic disorder.

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

1. FISH and its use in clinical practice.
2. How do you counsel for preimplantation genetic testing?
3. Briefly describe about the procedure amniocentasis and chorionic villi sampling, its indications and pretest counselling.
4. How do you process a peripheral blood sample for karyotyping?
5. Microdeletion.
6. Describe about the genetic sonogram.
7. What are the numerical chromosomal abnormalities and their clinical presentations?
8. PNDT (prenatal diagnostic technique) Act.
9. Fetal abnormalities identified with trisomy 18, 13 and 21.
10. What are the ethical aspects of medical genetics?

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[LL 1017] OCTOBER 2017 **Sub. Code: 0402**

FELLOWSHIP IN MEDICAL GENETICS EXAMS
FIRST YEAR
PAPER II – CYTOGENETICS

Q.P. Code: 230402

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

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

1. Elaborate on the types of polymorphisms in the human genome.
2. Discuss in detail about next generation sequencing.

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

1. Uni-parental disomy.
2. FISH and its uses in diagnosis of clinical conditions.
3. Sexual differentiation and intersex.
4. Down syndrome and its associated tumours.
5. Amniocentesis and its applications.
6. Genetic abnormalities in neurodegenerative conditions.
7. Methods of mutation analysis.
8. Genetic abnormalities in acute myeloid leukemia.
9. Recent advances in molecular pathology of gliomas.
10. Genetic drift and founder effect.

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