JYOTI NIVAS COLLEGE AUTONOMOUS
SYLLABUS FOR 2018 BATCH AND THEREAFTER

Programme: B.Sc. **Semester: V**

GENETICS PAPER VI

HUMAN GENETICS

Course Code: 18VGT6 **No. of Hours: 45**

COURSE OBJECTIVES:

To define the broad scope of human genetics, its relevance in human health andDiseases

2. To appreciate the contribution of Human Genetics to society.

3. To understand the ethical, legal and social implications of Genetic research.
4. To further improve critical and analytical thinking

LEARNING OUTCOMES:

- Will get an understanding on the human structural chromosomal anomalies and the techniques used
- Will obtain knowledge on the various autosomal, sex linked, epigenetic and multifactorial human genetic diseases and the pattern of inheritance
- Students will learn diagnosis of genetic disorder through non-invasive, invasive, pedigree and dermatoglyphics analyses
- They will also have an understanding on the role of genetics on human society

UNIT – I STUDY OF HUMAN CHROMOSOMES 08 HRS

Flow Cytometry – Basic introduction and its applications (Sperm sexing, Pre natal

diagnosis) **03 HRS**

Flow cytometer and FACS – Fluorescence activated cell sorter Flow karyotyping- a brief account

Structural chromosomal abnormalities: **02 HRS**

Deletion –Smith-Magenis syndrome

Translocation - Burkitt's lymphoma, Philadelphia translocation

Inversion - Chromosome 9, 11

Numerical chromosomal abnormalities: **2 HRS**

Aneuploidy in man - Autosomal aneuploidy syndromes- trisomy 21, trisomy 18, trisomy13 Sex chromosome aneuploidy syndromes - Turner, Klinefelter, triple X XYY, Mosaicism (Turner's syndrome) and Chimeras **01 HR**

UNIT – II MENDELIAN TRAITS AND GENETIC DISEASES 13 HRS

Straight hair, Curly hair, Widow's peak, Dimpled cheeks, Mid digital hair, Hitchhiker's thumb, Clasping of hands, Attached ear lobes, Tasters and Non-tasters and

Hypertrichosis **02 HRS**

Single gene disorders: **06 HRS**

Autosomal Dominant (AD) – Eg: Neurofibromatosis

Autosomal Recessive (AR) - Eg: Phenylketonuria,Albinism

X- Linked Dominant (X-LD) - Eg: Vitamin D resistant rickets

X- Linked Recessive (X-LR) - Eg: Ducenne Muscular Dystrophy

Y-Linked inheritance: Holandric -Eg: Testes determining factor.

Lysosomal storage disorder: lipid and glycogen storage diseases; TaySach's disease

Triplet repeat disorder: Fragile X syndrome **01 HR**

Epigenetic disorder: Angelman syndrome **01 HR**

Multifactorial disorders - Eg: Congenital malformations –Diabetes, Neural tube defect, Rheumatoid arthritis **02 HRS**

Mitochondrial disorder: Eg: Leber Hereditary Optic Neuropathy **01 HR**

UNIT – III DIAGNOSIS OF GENETIC DISEASES **13 HRS**

Pedigree studies and analysis: **04 HRS**

Symbols used in pedigree analysis, Pedigree construction; Pedigree analysis of important genetic diseases like Hypercholesterolemia, Cystic fibrosis, DuchenneMuscular Dystrophy.

Problems on pedigree analysis

Dermatoglyphics analysis: **04 HRS**

Introduction, Classification, Palmar patterns and Fingertip patterns, Flexion creases.Methods of recording dermatoglyphics.Dermatoglyphics in clinical disorders; itsadvantages and limitations

Detection of Chromosomal and Genetic Disorders by Investigations: **02 HRS**

Newborn screening and carrier screening; Karyotyping Molecular techniques – FISH,GISH, CGH, mutational analysis

Detection by Prenatal Diagnosis: **03 HRS**

Introduction and Definition Pre-implantation screening and diagnosis Non-invasivetechniques - Ultrasonography, maternal serum screening Invasive techniques - Chorionic villi sampling, Amniocentesis, Cordocentesis, Fetoscopy

UNIT - IV GENETICS AND SOCIETY **11 HRS**

Eugenics: **02 HRS**

Positive and Negative Eugenics, Euthenics, Euphenics

Genetic Counselling: **02 HRS**

Stage 1: History and Pedigree Construction;

Stage 2: Examination;

Stage 3: Diagnosis;

Stage 4: Counselling;

Stage 5: Follow up.

Gene therapy – types, viral and non-viral gene transfer, ex vivo and in vivo gene therapy, single gene therapy (eg. ADA SCID) and multiple factor therapy (eg. cancer), problems and ethical issues **3 HRS**

Stem cell therapy – properties of stem cells, embryonic and adult stem cells, bone marrow transplant and peripheral blood stem cell therapy, applications and ethical

Concerns **3 HRS**

Human Genome Project: Content of genome; benefits of human genome research; ethical, legal and social implications **01 HR**

GENETICS V SEMESTER PRACTICAL VI

DURATION: 3 HOURS / UNIT **NO. OF UNITS: 15**

1. **Study of Mendelian Traits:** **2 UNITS**

Straight hair (recessive), Curly hair, Widow's peak, Dimpled cheeks, Mid digital hair, Hitchhiker's thumb, Clasping of hands, and Hypertrichosis, Attached ear lobe

2. Study of Karyotypes: **3 UNITS**

Normal karyotyping in Humans – male (46, XY) and female (46 XX), G banded metaphase plate.

Abnormal karyotypes –Down syndrome, Turner syndrome and Klinefelter syndrome Cri-du-chat syndrome

3. Sex chromatin: **2 UNITS**

Observation of Buccal smear to study for Barr bodies.

Observation of blood smear to study drum sticks in Neutrophils

4. **Blood cell counting** using Haemocytometer **2 UNITS**
5. **Pedigree analysis** **2 UNITS**

Symbols used in autosomal recessive disorder, autosomal dominant disorder, sex chromosomal (X & Y linked) disorders

6. Dermatoglyphics: **2 UNITS**

Recording of print of fingertips and palm. Classify ridges on the finger tips - arch, loop and whorl. Palm print – area demark as hypothenar, thenar& inter digital areas. Ridge counting and ATD angle calculation.

Practical tests/repetition **2 UNITS**
Note: 13 Practical + 2 units for practical tests/repetition

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8. HUMAN CYTOGENETICS, Rooney D.E. (2001), 3rd edition, Oxford University Press, London.
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