DIPLOMA IN HUMAN GENETICS DISORDERS

(Non-Semester)

(With effect from the academic year 2013-14)

Eligibility for the Course

Candidates for admission to Diploma in Human Genetics Disorder could posses a Bachelors degree in Zoology, Botany, Chemistry, Biochemistry, Microbiology Biotechnology/Environmental/ Animal/plant Food sciences, Dietetics & Nutrition, Bioinformatics, BE in Chemical Engineering & Biotechnology; B.Tech in Biotechnology & Bioinformatics/Nanotechnology; BDS; MBBS; B.Sc in Agri/Agri Biotechnology;B.V.Sc., B.F.Sc., .Pharm and BPT.

Duration of the Course

One year Diploma in Human Genetics Disorder course non-semester for One Year duration

Examination

All the theory paper are of 3hours duration each for maximum of 100 marks with passing minimum of 35 marks Practical examinations are also for 3 hours duration for a maximum of 100 marks and passing minimum of 35 marks.

Question Paper Pattern

Maximum marks: 100 Time: 3 hours

Part A (5 x 3 = 15)

Five short answer questions (One question from each unit)

Part B (5 x 8 = 40)

Paragraph questions (Total questions 8, out of which answers are to be given for any five questions; Part C (3x 15 = 45)

Total questions 5, out of which answers are to be given for any Three questions;

S.No	Theory & Practicals	Maximum Marks	Minimum Marks
1.	Basics in Human Molecular Genetics	100	35

2.	Genetic Disorders and Patterns of Inheritance	100	35
3.	Methods of Testing Genetic Disorders and Counseling	100	35
P1	Laboratory Course in Screening Human Genetic Disorders	100	35

Paper I – Basics in Human Molecular Genetics

Unit-1

Introduction to Genetics - Mendelism - Mendel and his experiments - Chromosomal basis of segregation and independent assortment - Chi-square test and its application in analysis of genetic data - Extensions of Mendelism- Linkage and crossing over.

Unit-2

Allelic variation and gene function - Dominance relationships - Dominant and recessive mutations - Multiple allelism - sterile and lethal mutations - Effect of the environment on phenotype development- Penetrance and expressivity

Unit-3

Sex determinatin in Human- Gene interactions and modifying genes- Pleiotropy - Polygenic inheritance - Cytoplasmic inheritance and Maternal effects – Population Genetics – Hardy Weinberg Lab – Allele frequency

Unit-4

Structure of DNA and RNA- Basics in Transcription, Translation – Arrangement of genes in Chromosomes – Chromosome Architecture – Chromatin and epigenetic modifications in gene regulations.

Unit-5

Human Genome sequence – Genome-wide analysis methods, basics, advantages and applications – Human Genetics and genome databases

Book:

1. Gardner *et. al.,* (1991). Principles of Genetics. John Wiley.

References:

2. Brooker (1999). Genetics – Analysis and Principles. Benjamin/Cumings.
3. Griffiths *et. al.*, (2002). Modern genetic Analysis. Freeman.
4. Griffiths *et. al.*, (2004). An Introduction to Genetic Analysis. Freeman.
5. Strickberger (1985). Genetics. Mcmillan.

Paper II – Genetic Disorders and Patterns of Inheritance

Unit - 1

Inheritance of traits - Basic methods of analysis - Y chromosome and mitochondrial or maternal inheritance - Chromosomal disorder - single gene disorders – multi-factorial or polygenic disorders - spontaneous mutations - Rare modes of inheritance – Mosaicism and chimerism - Uniparental disomy - Male lethality, X-inactivation

Unit - 2

Basics in pedigree interpretation – Presentation of molecular genetic data in pedigrees - Complications to the basic pedigree patterns - Monogenic traits - Autosomal inheritance: dominant, recessive - Sex-linked inheritance - Sex-limited and sex-influenced traits - nonpenetrance - expressivity, pleiotropy - late onset - dominance problems, anticipation, genetic heterogeneity

Unit - 3

Risk assessment - Allele frequency in population - Consanguinity and its effects - Complex traits - monozygotic and dizygotic twins and adoption studies - Polygenic inheritance of continuous (quantitative) traits - discontinuous (dichotomous) traits- normal growth charts - Dysmorphology - threshold model, liability and recurrence risk - Calculation of carrier incidence - Concept of heterozygote advantage

Unit - 4

Characteristics and modes of inheritance for selected genetic disorders: Down Syndrome, Turner syndrome, Muscular Dystrophy, Retinoblastoma, Retinitis pigmentosa, Cystic Fibrosis, Neurofibroblast, Marfan syndrome, Colour Blindness – Physiology, and biochemistry of selected human genetic disorders - Genetic susceptibility in multi-factorial disorders - diabetes mellitus, obesity

Unit -5

Estimation of genetic components of multi-factorial traits: empiric risk, heritability, coefficient of relationship - Common syndromes due to numerical chromosome changes - Common syndromes due to structural alterations - translocations, duplications, deletions, microdeletion, fragile sites - chromosome abnormalities in cancer

Book:

1. Genetics: patterns of inheritance, 2002, Launchpad Multimedia.

References:

2. Quality Issues In Clinical Genetic Services, 2010, Ulf Kristoffersson, Springer
3. The Genetic Basis of Common Diseases, 2002,Oxford University Press
4. Genetic Disorders of the Indian Subcontinent, 2004, Dhavendra Kumar, Springer
5. Human Heredity: Principles & Issues, 2010, Michael R. Cummings, Cengage Learning.
6. Genetic disorders sourcebook, 2001, Kathy Massimini, Omnigraphics
7. Emery's Elements of Medical Genetics, 2011, Peter D Turnpenny, Elsevier Health Sciences

Paper III - **Methods of Testing Genetic Disorders and Counselling**

Unit -1

Screening and diagnosis, invasive and non-invasive methods - Prenatal, neonatal, postnatal and perinatal diagnosis – embryo, foetal sampling methods, aminocentesis, fetoscopy, CVS, cordocentesis, maternal blood sampling - Karyotyping, DNA investigation, Biochemical, immunological and enzymatic analysis - Presymptomatic and predictive testing - Genetic Registers - Population screening, carrier screening, Two step screening - Advantages and disadvantages of testing

Unit -2

Counselling: Aspects – components - Prospective and retrospective genetic counselling - Genetic counsellor - Preconceptional genetic counselling - Psychological counselling - Scenario based decision - Person centred approach

Unit -3

Management and Remedial measures for genetic disorders - Conventional approach - Application of recombinant methods - Multi - speciality approach - foetal medicine - foetal vaccination - recurrent loss of pregnancy - Foetal therapy - Antenatal and postnatal surgical anomalies - Management of inherited metabolic disorders in India - The role of prenatal pathologists - Gene therapy - hormonal replacement therapy.

Unit -4

Population education - individual - families – society - Awareness programme - policy markers - community influences -Training of medical/para medical/health care providers - Pharmacogenetics and ecogenetics

Unit -5

Ethical and Legal issues – Eugenics – Euthenics – Positive and negative euthenics - Euphenics – ELSI.

Book:

1. Preventive Genetics by Gogate, 2006, Jaypee Brothers, Medical Publishers
2. Basic Human Genetics, 2006, V. Kapur, R. K. Suri, Jaypee Brothers Publishers

References:

1. Genetic Diagnosis of Endocrine Disorders, 2010, Roy E. Weiss, Samuel Refetoff, Academic Press.
2. Genetic Disorders and the Fetus: Diagnosis, Prevention and Treatment, 2011, Aubrey Milunsky, Jeff Milunsky, John Wiley & Sons
3. The Encyclopedia of Genetic Disorders and Birth Defects, 2009, James Wynbrandt, Mark D. Ludman, Infobase Publishing.
4. Genes, Chromosomes, and Disease: From Simple Traits, to Complex Traits, to Personalized Medicine, 2011, Nicholas Wright Gillham, FT Press.

Paper IV – Laboratory in Screening Human Genetic Disorders

1. Karyotyping
2. Genomic DNA preparation from various sources – Blood and Buccal swap.
3. DNA Quantitation and Quality analysis
4. DNA analysis by Agarose Gel Electrophoresis
5. Finger printing in triradial analysis
6. Genotyping using PCR – PCR evaluation – Post PCR sample preparation – DNA Elution
7. Genotyping by DNA sequencing
8. SNP typing
9. Screening for selected Genetic Disorders by PCR based methods
10. Online Mendelian Inheritance in Man (OMIM)
11. Bioinformatic approaches in human genetic disorders

References:

1. Molecular Cloning, 2012, Michael Richard Green, Joseph Sambrook, Cold Spring Harbor Laboratory Press
2. Human Molecular Biology Laboratory Manual, 2003, Stefan Surzycki, Wiley