SYLLABUS

POST-GRADUATE DIPLOMA in

CHROMOSOMAL & MOLECULAR DIAGNOSTICS

(PGDCMD)

(Annual System)

A 1-year interdisciplinary diploma course for post-graduates in science subjects or graduates in medicine or technology inclined towards vocational training in diagnostics of chromosomal and genetic disorders.

Department/School/Centre: Centre for Genetic Disorders, Faculty of Science

Eligibility: Post-graduation in any stream of science/agriculture or graduation in medicine/technology with at least 50% aggregate marks. Biology at +2 level essential

Seats: 10 seats; 2 supernumerary for sponsored candidates

Fee: Rs 30,000.00 over and above the regular fee charged by the University

Sponsored candidates: Rs 50,000.00 over and above the institutional fee

Admission: On the basis of marks obtained at +2 and higher degrees, and interview

Total Marks: 1000 (Theory: 500; Lab Exercises: 500)

Course	Theory	Marks	Laboratory work	Marks
CMDT-1	Basic Human Genetics and Genomics	100	CMDL-1	100

			(based on CMDT-1)	
CMDT-2	Human Cytogenetics	100	CMDL-2 *(based on CMDT-2)*	100
CMDT-3	Clinical Genetics & Genetic Counselling	100	CMDL-3 *(based on CMDT-3)*	100
CMDT-4	Biochemistry & Instrumentation Recombinant DNA Technology & Immunology	100 (50+50)	CMDL-4 *(based on CMDT-4)*	100
CMDT-5	A. Physiology & Pathology B. Developmental & Reproductive Genetics	100 (50+50)	CMDL-5 *(based on CMDT-5)*	100

1. Candidate will be examined for each Theory and Lab exercise at the end of the session for which 70% of the total marks will be allocated.

30% of the total marks will be allocated to the evaluation of candidate's performance during the session through tests (2 in number: 20% of the total marks) and assignment (1 in number: 10% of the total) in each paper.

Theory

CMDT-1 Basic Human Genetics and Genomics

26 lectures

1. DNA as the genetic material, genetic code, flow of genetic information, mutation

2. Principles of inheritance and probability rules

3. Pedigree construction & family study

4. Complications in pedigree analysis (variable expressivity, heterogeneity, penetrance, anticipation, epigenetics, mosaicism)

Non-Mendelian inheritance

5.1 Polygenic inheritance

5.2 Multifactorial trait

5.3 Threshold trait

5.4 Genetic Susceptibility & risk factors

6. Mitochondrial genome and disorders

7. Human genome, genome organization, annotations and databases, markers (microsatellites, SNPs) 8. Pharmacogenomics, Ecogenomics, Metabolomics, Teratogenetics

9. Mapping and identification of disease genes (linkage analysis, LOD score, association study)

10. Allele frequency in population, estimation of carrier frequency

CMDT-2 Human Cytogenetics

26 Lectures

1. Organization of cell and cell cycle

2. Cell division (Mitosis, Meiosis)

3. Chromatin structure and chromosome organization

4. Chromosomal basis of inheritance, sex chromosome, X-chromosome inactivation

5. Basics of cell culture

6. Techniques of cell cultures (short term lymphocyte, primary and secondary cell cultures, maintenance of cell lines)

7. Techniques of chromosome analysis

Chromosome preparation from cultured lymphocytes, cell lines and solid tumors

Karyotyping, C-,G-banding and fluorescence banding, nomenclatures of bandings

In-situ hybridization techniques

Meiotic chromosomes

8. Chromosomal anomalies and disorders

8.1 Numerical (polyploidy, aneuploidy, autosomal, sex- chromosomal)

8.2 Structural (deletion, duplication, translocation, inversion, isochromosome, ring chromosome)

8.3 Chromosomal abnormalities in cancer

CMDT-3 Clinical Genetics & Genetic Counselling

26 lectures

1. Genetic Disorders

1.1 Classification of genetic disorders

1.2 Single gene Disorders (Cystic Fibrosis, Marfan's syndrome)

1.3 Multifactorial disorders (Diabetes, Atherosclerosis, Schizophrenia)

2. Molecular Techniques

2.1 PCR-RFLP

2.2 ARMS-PCR

2.3 Multiplex-PCR

2.4 SSCP, CSGE, DGGE, DHPLC

2.5 MALDI-TOF

2.6 DNA Sequencing

3. Disease identification and Genetic tests for following disorders

3.1 Thalassemia, Fanconi anemia, Sickle Cell anemia, Fragile-X syndrome, Alzheimer's disease

3.2 Duchenne Muscular Dystrophy/Becker's Muscular Dystrophy, Huntington's disease

3.3 Allelic susceptibility test for multifactorial disorders (Neural Tube Defect, Cleft Lip and Palate, Cardio Vascular Disorder, Male infertility)

3.4 Molecular basis of cancer

4. Genetic Counseling

4.1 Principles of genetic counseling

4.2 Causes and factors for seeking counseling

4.3 Dysmorphology

4.3 Ethical and legal issues in genetic counseling

4.4 Risk evaluation (Mendelian risk, empirical risk)

4.5 Prenatal and pre-implantation diagnosis.

Non-invasive: Triple test, Ultrsonography (USG)

Invasive: Amniocentesis (AC), chorionic villi sampling (CVS), Fetal blood sampling (FBS)

Population screening for genetic disorders

Treatment and management of genetic disorders

CMDT-4 Biochemistry & Instrumentation and Molecular Biology & Immunology

26 lectures

Section A: Biochemistry & Instrumentation **13 Lectures**

Introduction to Nucleic Acids, Proteins, Carbohydrates, Lipids, Vitamins, Minerals

Instrumentation (pH-meter, spectrophotometer, centrifugation, ELISA, radioactivity detection, Mass spectrometry, High performance liquid chromatography)

Biochemical tests: sugar, albumin, urea, protein, globulin, vitamin

Biochemistry and diagnostic tests of following diseases

Duchenne Muscular Dystrophy (DMD) (Creatine phosphokinase-CPK)

Phenylketonuria-PKU (phenylketone)

G6PD deficiency syndrome (G6PD)

Mucopolysaccharidosis

Endocrine disorders related to thyroid and reproduction (TSH, T3, T4, Estradiol, Testosterone, LH, FSH)

Section B: Recombinant DNA Technology & Immunology **13 Lectures**

1. Recombinant DNA Technology

1.1 Bacterial culture

1.2 Restriction enzymes

1.3 Vectors

1.4 Transformation and transfection

2. Immunology

2.1 Overview of immune system

2.2 Antigens and antibodies

2.3 Antigen-antibody interactions

2.4 Major Histocompatibility Complex (MHC), HLA typing

2.5 Immunotherapy

CMDT-5 Physiology & Pathology and Developmental & Reproductive Genetics 26 lectures

Section A: Physiology & Pathology

13 Lectures

1. Blood (formation, composition, function and pathology of blood disorders (haemoglobinopathies, sickle cell anemia, hemophilia)

2. Muscle disorders (Duchenne muscular dystrophy-DMD, Becker's muscular dystrophy-BMD, spinal muscular atrophy-SMA)

3. Bone disorders (Osteogenesis imperfecta, Rheumatoid arthritis)

4. Skin disorder (Albinism)

5. Eye disorder (Retinitis pigmentosa)

6. Tests at cellular level: TLC, DLC, platelets

7. Molecular Pathology – principles & application

Infectious diseases and Pathogen (Tuberculosis-*M. tuberculi*, Peptic ulcer-*H. pylori*, Cervical Cancer-HPV, AIDS-HIV, Hepatitis- Hepatitis virus, Rubella- Rubella virus)

Homocysteinuria

Nutritional deficiency and disease

Genetic susceptibility to infectious diseases

Section B-Developmental & Reproductive Genetics

13 lectures

Overview of human development and developmental disorders (DiGeorge syndrome, neural tube defect, cleft lip and palate, microcephaly)

Sex Determination

Hormonal basis of sexual differentiation

Reproductive organs and gonads

Reproductive Disorders: Genetic and environmental susceptibility for reproductive disorders (Male infertility), endometriosis, recurrent early pregnancy loss, polycystic ovarian disorder

Assisted Reproductive technologies (ART)

Ethical and legal issues

Laboratory Exercises

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CMDL-1

(Based on CMDT-1)

26 Lectures

Good laboratory practice

1. Universal safety precaution, importance of personal hygiene, disposal of organic waste washing, cleaning of glass ware, sterilization of glassware and lab ware

2. Drawing blood, preparation of blood film and identification of cells

3. Learning professional English, use of OMIM

4. Use of computers, word processing and retrieval of data

Retrieval of information from patients, informed consent, preparation of reports

Tissue culture

1. Cleaning, sterilization and maintenance of culture chamber, cleaning and maintenance of laminar flow hood, preparation of culture medium

2. Collection and storage of samples (blood, biopsy material), culturing lymphocytes, stem cells (from limbal tissue and articular cartilage (chondrocytes), chorionic villi, preparation of cell lines from blood cells

Basic Human Genetics

1. Preparation of pedigree, recording of physical feature (facial morbidity, gonadal abnormality), other physical and physiological abnormality

2. Dermatoglyphics, understanding the prescription and test reports, preparation of report

3. Risk calculation and prediction of recurrence

Detection of patterns of inheritance by PCR-RFLP and microsatellite markers

CMDL-2

(Based on CMDT-2)

Human Cytogenetics

26 lectures

1. Microscopy

2. Metaphase chromosome preparations from bone marrow of mouse, rat, human

3. Chromosome preparation from lymphocyte culture

4. G-banding, C-banding, R-banding

5. Karyotyping

6. Fluorescence *in-situ* Hybridization (FISH)

7. Meiosis in mouse testis

8. Sex chromatin (buccal mucosa, hair bud)

9. Comet assay

10. Micronuclei assay

11. Chromosome preparation from chorionic villi, stem cells, cell line

12. Sister Chromatid Exchange (SCE)

CMDL-3

(Based on CMDT-3)

Clinical Genetics and Genetic Counselling

26 lectures

1. Detection of mutation using ARMS-PCR (e.g.; Thalassemia) and microsatellite markers (e.g.; fragile-X syndrome)

Tests for genetic disorders

Overview of genetic diagnostics

SNP (e.g.; cleft lip and palate)

STS (e.g.; male infertility: Y-chromosome microdeletion)

Triplet repeat polymorphism (e.g.; Huntington's disease)

Molecular markers for tumor detection

Bcr-abl (RT-PCR)

BRCA1 (PCR)

Her2new (FISH)

3. Genetic counseling (pedigree analysis in disease conditions, risk calculation)

4. Haplotyping of risk alleles in multifactorial disorders

Prenatal diagnosis of Thalassemia

CMDL-4

(Based on CMDT-4)

Biochemistry & Instrumentation and Recombinant DNA Technology & Immunology

26 lectures

Biochemistry & Instrumentation

Weighing, preparation of distilled and purified water, preparation of solutions and stains, pH-meter, spectrophotometer, centrifugation, ELISA, microscope

Biochemical tests for sugar, albumin, Creatine phosphokinase-CPK, glucose 6 phosphate dehydrogenase-G6PD

Recombinant DNA Technology & Immunology

E. coli culturing, transformation

Cloning

Extraction of DNA from plasmid and restriction enzyme mapping of plasmid DNA

Extraction of DNA (from human blood, other tissues) and RNA, quantitation, Southern and Nothern blotting

cDNA preparation and RT-PCR

Polymerase chain reaction (PCR)

DNA sequencing (deomontration)

DNA extraction / PCR from semen, hair bud, mucosa

Immunogenetics (HLA typing)

ELISA, SDS-PAGE, Western blotting

CMDL-5

(Based on CMDT-5)

26 lectures

A. Physiology & Pathology

1. Total leucocyte count (TLC), differential leucocyte count (DLC), general blood picture (GBP), hematocrit, hemoglobin (Hb) content and electrophoretic resolution of Hb in human blood, platelets

2. DNA based detection of *H. pylori*, *M. Tuberculi*, *M. lepri*

3. Assay of homocysteine, vitamin-B-12, folic acid

Developmental and Reproductive Genetics

Models for human developmental stages

Hormone assay for thyroid (TSH, T3, T4) and sexual disorders (testosterone, dihydrotestosteron, estradiol, FSH, LH by RIA), semen analysis, Y-chromosome microdeletion