

# Goa University P.O. Goa University, Taleigao Plateau, Goa 403 206 POST GRADUATE DIPLOMA PROGRAMME ON CLINICAL GENETICS AND MEDICAL LABORATORY TECHNIQUES

#### For the Academic Year 2013-2014

Implemented from : 2013-14 (Approved in BOS February 2013 and AC in

)

A brief description of the course:

- **Purpose :** To give the youth broad exposure in the various pathological techniques. To expose the student to the field of Clinical Genetics.
- **Prerequisite** : Science graduate with either Zoology, Microbiology, Biotechnology (6 units / or 3 units) with Chemistry as s subject up to SYBSc.
- Duration: 2 Semesters with Theory and Practical courses (Total courses 8: 4 Theory and 4 Practical courses) followed by compulsory one month Hands on Training in each of the laboratories viz. Biochemistry, Blood Bank & Central Laboratory, Pathology and Microbiology in Goa Medical College /Govt. Hospital and one month training on Clinical Genetics in a laboratory / Institute identified in consultation with Chairman BOS.

**Course fee**: Course fee will be decided by the appropriate authority

**Special feature:** A collaborative teaching program between Departments of Biochemistry, Pathology, Microbiology of Goa Medical College and Zoology Department of Goa University.

The said programme is also offered by some Undergraduate colleges affiliated to Goa university

All the theory as well as practical, Core and Optional courses, will be evaluated by both internal and external examiners (double evaluation)

# **Course Structure**

PAPER CC	DDE COURSE TITLE	CONTACT HOURS		MARKS		PAGE
		LECTURES F	PRACTICALS	THEORY PE	RACTICA	LS
I <sup>ST</sup> SEMESTER						
DLTC01	Clinical Genetics I Techniques	36hrs	15 x 3 hrs	50	50	3
DLTC02	Clinical biochemistry – I	36 hrs	15 x 3 hrs	50	50	4
DLTC03	Clinical Microbiology (General and Systematic)	36 hrs	15 x 3 hrs	50	50	5
DLTC04	Clinical Pathology & Histolo	gy 36 hrs	15 x3 hrs	50	50	7
2 <sup>nd</sup> SEMESTER						
DLTO01	Clinical Genetics II	36 hrs	15 x 3 hrs	50	50	9
DLT O02	Clinical Biochemistry II	36 hrs	15 x 3 hrs	50	50	10
DLTO03	Clinical Parasitology, Mycolo & Virology	ogy 36 hrs	15 x 3 hrs	50	50	11
DLT O04	Hematology & Transfusion Medicine	36 hrs	15 x 3 hrs	50	50	13

List of text and Reference books:

14

# SEMESTER I

# Paper DLTC 01: Clinical Genetics I:

# <u>THEORY</u>

# Module 1:

- Introduction to Human Genetics: growth of human genetics; levels of genetics.
- Structure and composition of the human chromosome: basic structure of DNA; molecular structure and organisation.
- Classification of Human chromosomes: Paris nomenclature / ISCN; methods of studying chromosomes; identification of individual chromosomes; Flow Karyotyping (Quantification on DNA of individual chromosomes); FACS – Fluorescence activated cell sorter.

# Module 2:

- Chromosomal Abnormalities:
- Numerical abnormalities (somies; ploidies; mosaic; chimera; syndromes.)
- Structural: Translocations; Deletions; Duplications; Inversion; isochromosomes; Ring chromosomes; causes for genetic abnormalities- meiotic and mitotic nondisjunction; uniparental disomy; mutations; single gene disorders.

## Module 3:

- Pattern of inheritance: Autosomal Dominant, Autosomal Recessive, X-linked Dominant, X-linked Recessive, Y-linked, sex limited inheritance, sex influenced inheritance, X inactivation, Multifactorial inheritance, mitochondrial inheritance, imprinting
- Pedigree analysis of some genetic disorders: Haemophilia, Color blindness, Duchenne Muscular Dystrophy (DMD), achondroplasia and PKU.

- 1) Specimen procurement and logging for cytogenetic procedure.
- 2) Culture media preparation

4) Identification of Chromosomes.

- 5) Inoculation of Lymphocyte culture/peripheral blood culture.
- 6) Harvesting of Lymphocyte culture to obtain metaphase plates.
- 7) Chromosomal banding technique: GTG Banding.

8) Karyotyping of Human chromosomes (use of Cytovision / any other Karyotyping software is optional: for image capturing, image processing, and analysis).

- 9) Study of Karyotypes: Normal male and female.
- 10) Construction of Pedigree from given data.
- 11) Analysis of pedigree charts to determine the mode of inheritance.

# Paper DLTC 02: Clinical Biochemistry I

# **THEORY**

#### Module 1:

- The scope of biochemistry:
- Chemical organization of the cell.
- Organic and inorganic components of the cell.
- Marker enzymes of the cell.
- Hydrogen ion concentration and buffers: pH
- Blood buffers, regulation of blood pH.
- Acid base metabolism.

#### Module 2:

- Carbohydrate chemistry.
- Protein chemistry.
- Lipid chemistry.

#### Module 3:

- Enzymes:- Definition, classification, factors affecting enzyme action.
- Enzyme inhibition,

- Isoenzymes,
- Regulation of enzyme activity.
- Vitamins.
- Minerals.

# **PRACTICALS:**

- 1. Estimation of pH. Use of pH meter.
- 2. Qualitative and quantitative Carbohydrate chemistry.
- 3. Qualitative and quantitative Protein chemistry.
- 4. Qualitative and quantitative Lipid chemistry.
- 5. Estimation of haemoglobin by cyanmeth haemoglobin
- 6. Estimation of chloride in serum
- 7. Estimation of serum calcium
- 8. Estimation of serum inorganic phosphorus.
- 9. Separation of amino acid and its identification by paper chromatography-
  - Demonstration
- 10. Separation of serum protein by electrophoresis- Demonstration
- 11. Separation of lipid by Thin layer chromatography Demonstration
- 12. Estimation of serum Na+/ K+ ions by Flame photometer- Demonstration.

# Paper DLTC 03: Clinical Microbiology (General and Systematic)

# <u>THEORY</u>

## Module 1:

- Introduction to microbiology- historical prospective, principle of microbiology, microscopes (types and uses)
- Bacteria: Classification, anatomy, reproduction, growth and nutrition.
- Sterilization:- methods employed, both physical and chemical.
- Media used in Microbiology:- Classification, types, constituents, methods of preparation, adjustment of pH, sterilization.

## Module 2:

- Serology:- Antigen, antibody, antigen-antibody reaction.
- Newer methods of diagnosis: PCR, Bactec, Flow cytometry.

#### Module 3:

• Systemic (Individual Bacteria): Diagnosis features (morphology, cultured characters, biochemical reaction,, antigenic characters, pathogenicity and laboratory diagnosis) of *Staphylococcus, Streptococcus, Pneumococcus, Neisseria, Corynebacteria, Clostridia, Escherichia coli, Klebsiella species, Salmonella, Shigella, Proteus, Pseudomonas, Mycobacterium tuberculosis, Treponema pallidum.* 

- 1. Preparation of smears for staining and fixation from samples and culture media (both liquid and solid media).
- 2. Care and use of microscpes (including Fluorescent microscope).
- 3. Staining techniques: (Gram staining, zeihl nelson, Fluorescent method): preparation of satins, procedure, reporting of smears, principle involved.
- 4. Equipments used in sterilization: Description (structure), working principle involved, articles sterilized, advantages and disadvantages.
- 5. Culture media: types, constituents of each media, method of preparation, adjustment of pH, sterilization, uses.
- 6. Culture techniques: different methods of inoculation from clinical samples and bacterial growth from media.
- 7. Preparation of wet mount and motility of organisms.
- 8. Sputum examination: Physical examination, wet preparation, smear examination, concentration techniques for mycobacteria.
- 9. Serology: Widal, VDRL, RPR, ELISA, PCR, Flow cytometry.
- 10. Systemic bacteriology: Practical demonstration of diagnostic features of
  - Gram positive organisms.
  - Gram negative organisms.
  - Anaerobes, spirochetes.

• Mycobacteria.

# Paper DLTC 04: Clinical pathology and histopathology

## **THEORY**

## Module 1:

- Histo-pathological techniques: Fixatives and fixation; Grossing, dehydration, clearing, impregnation and embedding; Microtome knives and types of microtomes, section cutting, errors in cutting, mounting media, decalcification, automation in tissue processing; Frozen section.
- Staining: Theory of staining, dyes and stains; Mordants, differentiation; Haematoxylin and Eosin staining- principles and procedures;
- Special stains-- P.A.S., Verhoeff's, Massons trichrome, Von Giesson, Fat stains and other stains.

#### Module 2:

- Examination of urine: Sample collection; Physical examination and Chemical tests-principles and methods; Reagent strip method
- Microscopic examination- crystals, casts, sediments,
- Stool examination, Pregnancy tests, Semen analysis, Sputum examination.

#### Module 3:

- Cytological techniques: Exfoliative cytology- fixation, pap staining. Cytological processing of fluids. Fine needle aspiration cytology (FNAC) -- procedure, staining of slides, H & E staining and MGG staining. Automation in cytology.
- Examination of CSF and other body fluids-- pleural, peritoneal, synovial fluid.

#### **PRACTICALS:**

1. Grossing and museum techniques.

- 2. Microtomes knives and their sharpening, section cutting, errors in section cutting and mounting.
- 3. Decalification, automation in tissue processing.
- 4. Routine staining techniques-hematoxylin and eosin (H &E) staining.
- 5. Special staining demonstration- P.A.S., Verhoeff's, Massons trichrome, Von Giesson, fat stains.
- 6. Examination of urine- Physical and chemical examination, use of Reagent strips.
- 7. Examination of urine microscopic
- 8. Exfoliative cytology-Fixation, Pap staining procedure.
- 9. Fine needle aspiration cytology (F.N.A.C)- procedure, stains
- 10. Examination of body fluids- pleural, peritoneal and synovial.
- 11. C.S.F. examination.
- 12. Stool examination
- 13. Sputum examination
- 14. Pregnancy tests.
- 15. Semen analysis.

## **SEMESTER II**

# Paper DLTO 01: Clinical Genetics II

# <u>THEORY</u>

### Module 1:

- Molecular genetic techniques used in genetic diagnosis: Blotting techniques Southern, Northern and Western, PCR / RFLP, FISH, DNA sequencing & DNA fingerprinting.
- Genetics of Cancer: introduction, characteristics of cancer cells, origin of cancer cells, genes associated with cancer, environmental causes of cancer, human genome data tailor diagnosis and treatment.
- Dermatoglyphics: Introduction, classification, Flexion creases. Dermatoglyphics in clinical disorders, Clinical application & its advantages and limitations.

## Module 2:

- Reproductive technologies: infertility and subfertility, assisted reproductive technologies (IUI, surrogate motherhood, IVF, GZIT, ZIFT), preimplantation genetic diagnosis.
- Genetics and Society : (i) Human genome project : (ii) Forensic science (iii) DNA finger printing application (iv) Gene therapy (v) Eugenics. vi) Stem cell research.

## Module 3:

- Prenatal Diagnosis: Definition: Various procedures Amniocentesis, Chorionic villus sampling, Ultrasonography and Fetoscopy.
- Genetic Counselling (Stage1: History and Pedigree Construction, Stage 2: Examination, Stage 3: Diagnosis, Stage 4: Counselling; and Stage 5: Follow up).

## **PRACTICALS**

) Introduction to molecular genetic lab: general rules, handling of chemicals, equipments and biological materials; waste disposal.

- 2) Isolation of DNA from human blood.
- 3) Determination of molecular size of DNA.

- 6) Analysis of DNA fingerprints and FISH images.
- 7) Manual DNA sequencing and data analysis.
- 8) Dermatoglyphics: Recording of print of fingertips and palm.
- 9) Amniotic fluid culture: Flask method and Cover slip method.
- 10) Chorionic villi culture: Short term culture
- 11) Chromosomal analysis from product of conception (abortus study)
- 12) Disease suspection by spot tests: Fanconis syndrome, PKU, maple syrup urine

disease, Tryptophanuria.

### Paper DLT O02 Clinical Biochemistry II

#### **THEORY**

#### Module 1

- Carbohydrate metabolism: Clinical aspects of Regulation of Blood sugar and Diabetes
- Protein metabolism: starvation, and protein energy malnutrition
- Lipid metabolism Clinical aspects of lipid profile, artherosclerosis.

#### Module 2 & 3

- Gastric function tests.
- Pancreatic function tests.
- Liver function tests.
- Thyroid function tests.
- Cardiac function test
- Kidney function test

- 1. Chemistry of saliva.
- 2. Chemistry of gastric juices
- 3. Estimation of bilurubin.
- 4. Estimation of glucose in blood. GTT and its interpretatio

- 5. Estimation of serum proteins.
- 6. Estimation of blood urea.
- 7. Estimation of creatinine in blood.
- 8. Estimation of uric acid in blood.
- 9. Normal urine.
- 10. Full urine report.
- 11. Clearance tests Demonstration
- 12. Demonstration of liver function/ cardiac function / kidney function tests.
- 13. Serum lipid profile
- 14. C.S.F. examination.

## Paper DLTO 03: Clinical parasitology, mycology and virology

### **THEORY**

#### Module 1: Parasitology:

- Introduction to parasitology terminologies, definitions, relationships.
- Protozoa: geographic distribution, habitat, morphology, life cycle, pathologenecity, laboratory diagnosis of the following parasites:
  - 1. Entamoeba histolytica
  - 2. Giardia lamblia
  - 3. Trichomonas vaginalis
  - 4. Leishmania donovani
  - 5. Plasmodium
  - 6. Cocoidian parasites causing diarrhea
- Cestodes: On the same line as protozoan parasites for the following:
  - 1. Taenia sagenata
  - 2. Taenia solium
  - 3. Echinococcus granulosus
- Helminths: On the same line as protozoan parasites for the following:
  - 1. Trichuris trichiura
  - 2. Ankylostoma duodenale
  - 3. Ascaris lumbricularis
  - 4. Enterobius vermicularis

#### Module 2: Mycology :

- Introduction to mycology including classification
- Candida albicans and other candida species
- Dermatophytes
- Cryptococcus
- Oppotunistic fungi (Aspergillus, Pencillium, Mucor)

NB: Serial no: ii-v will be on the basis of morphology, cultural characters, biochemical (if any), antigens, pathogenecity and laboratory diagnosis.

#### Module 3: Virology:

- General virology: Definations, classification, properties of viruses, viral replication, cultivation, laboratory diagnosis.
- Systemic virology: On the basis of structure, cultivation, pathogenicity, Laboratory diagnosis of the following viruses:
  - i) Bacteriophage
  - ii) Picomaviruses (Polio viruses)
  - iii) Rhabdoviruses ( Rbies virus)
  - iv) Arboviruses (Dengue, Chikungunya, JE)
  - v) Influenza virus
  - vi) Hepatitis virus
  - vii) HIV
  - viii) Herpes virus

- A) Parasitology
  - 1) Stool examination: gross, microscopic, for adult parasite, segment of Taenia, ova, cysts, and larvae of parasite, etc.
  - 2) Gross and microscopic features (whenever applicable) of intestinal/vaginal protozoa.
  - 3) Laboratory diagnosis of malaria: demonstration of whole parasite, parasite antigen, enzymes, serology, etc.
  - 4) Gross and microscopic features of cestodes: to include adult worms, segment, larvae, eggs.
  - 5) Gross and microscopic features of Helminthes: to include adult worms, eggs, larvae.
- B) Mycology
  - 6) Diagnostic features- practical demonstration of gross and microscopic features ( wet mount, slide culture) and other tests whenever applicable for following: Candida, Cryptococcus, Dermatophyte, Oppurtunistic fungi.
- C) Virology :
  - 7) General virology: types of symmetry, morphology of virus models, cultivation in embryonated egg
  - 8) Laboratory diagnosis of the following viruses: Poliovirus, Rhabdovirus, HIV, Hepatitis.
  - 9) Bacteriophage—structure using a model.

# Paper DLTO 04: Hematology and Transfusion medicine

# THEORY:

## A) Hematology:

- Blood--- composition and function, haemopoesis; RBC'S- structure, function and synthesis; Hemoglobin- structure, function, abnormal haemoglobins; Reticulocytes; Study of peripheral blood smear, parasites in blood.
- Hemolytic disorders—classification, general evidence of hemolytic nature of anaemia (screening tests). Hemolytic workup -- Sickling, Osmotic Fragility tests, Heinz bodies, G-6-P-D screening, Hb electrophoresis, Hb-F estimation.
- White blood corpuscles-- Description, morphology, leucopoiesis, Total WBC count and corrected Total WBC count, leucopenia, leucocytosis. Absolute counts—absolute eosinophil count. Differential WBC count. Leukemia, Leukemoid reaction, special stains for leukemias—PAS, Sudan Black, Myeloperoxidase. Bone marrow examination and iron staining of marrow.
- Platelet structure and function--- The normal hemostatic mechanism. Heamorrhagic disorder due to vascular (capillary) defect and platelet abnormalities. Theory of blood coagulation. Coagulation abnormalities pathogenesis and classification. Laboratory tests and investigations of Bleeding disorders ie. Vascular disorders, platelet disorders, coagulation

#### **B)** Transfusion medicine:

- Blood groups- Introduction, ABO and sub groups, basic genetics, antigen and antibodies. ABO grouping techniques, problems in ABO grouping. , Rh blood group—basic genetics, antigen and antibodies, RH grouping techniques, problem in RH grouping. Other blood group systems and their significance.
- Blood banking--- Selection of a blood donor, blood collection. Complications of blood transfusion. Investigations of a mismatched blood transfusion.
- Blood component separation and therapy, Compatibility testing, Antihuman globulin test.
- Organization and administration of a blood bank, FDA rules, blood safety.

- 1) Use and care of microscopes; blood collection.
- 2) Anticoagulants and study of improved neubaur chamber, erythrocyte count.
- 3) Haemoglobinometry: Sahli's method
- 4) Peripheral Blood smear preparations and staining; differential WBC count
- 5) Peripheral blood smear examination and morphological abnormalities
- 6) Total WBC count
- 7) Reticulocyte count
- Demonstration of Hemolytic workup -- Osmotic fragility test, Heinz bodies, Sickling, G-6-P-D estimation, Hb-electrophoresis, Hb-F estimation.
- 9) Bone marrow examination- staining of smear, iron staining of marrow, Special stains-PAS, Sudan black, Myeloperoxidase
- 10) Platelet count
- 11) BT, CT, CRT
- 12) Demonstration of Prothrombin time, A.P.P.T and FDP estimation
- 13) E.S.R, P.C.V, Blood indices
- 14) ABO grouping and Rh typing
- 15) Demonstration of Coomb's test, Compatibility testing..

#### TEXT / REFERENCE BOOKS:

- 1. Textbook of Medical Laboratory Technology: Ramnik Sood.
- 2. Textbook of Medical Laboratory Technology: Praful Godkar
- 3. Theory and Practice of Histological Techniques : J D Bancroft and M Gamble
- 4. Clinical Diagnosis and Management by Laboratory Methods : J Bernard Henry
- 5. Textbook of Medical Biochemistry : M N Chatterjee & Rana Shinde
- 6. Essentials of Biochemistry : Pankaja Naik
- 7. Text Book of Biochemistry : Vasudevan and Shree Kumari
- 8. Text Book of Biochemistry : A Lehninger
- 9. Text Book of Biochemistry : Deb
- 10. Text Book of Harper's Illustrated Biochemistry : Murray, Granner and Rodwell.
- 11. Hematology of Medical Technologies: Seiverd
- 12. Practical Haematology : John Dacie and S M Lewis
- 13. Compendium of Transfusion Medicine : R N Makroo
- 14. Essentials Of Human Genetics : S.M Bhatnagar, ML Kothari , L A Mehta
- 15. Screening And Diagnosis Of Fetal Malformation- A Practical Guide: A K Debdas
- 16. Human Genetics: A.Gardner and T. Davies
- 17. A Textbook Of Human Genetics : Amita Sarkar
- 18. Medical Genetics At A Glance : Dorian .J. Pritchard & Bruce R. Korf
- 19. Human Genetics Concepts & Application : Ricki Lewis
- 20. Text Book of Microbiology (8<sup>th</sup> ed.) : Ananthanarayan and Panikers
- 21. Concise Microbiology (1st ed.) : C P Baveja and V Baveja
- 22. Medical Parasitology (2<sup>nd</sup> ed.) : C P Baveja and V Baveja
- 23.