SRI BALAJI VIDYAPEETH

ACCREDITED BY NAAC WITH 'A' GRADE

DEEMED TO BE UNIVERSITY
DECLARED U/S 3 OF THE UGC ACT, 1956

NIRF - INDIA RANKINGS 2019 : 72 among Universities in India

FELLOWSHIP IN MEDICAL GENETICS

Genetic Unit, Department of Anatomy

SYLLABUS & REGULATIONS

2019-2020 ONWARDS

(As Approved in the Academic Council at the Meeting held on 22.05.2019)

Sri Balaji Vidyapeeth University
Mahatma Gandhi Medical College & Research Institute

GENETIC UNIT, DEPARTMENT OF ANATOMY

FELLOWSHIP IN MEDICAL GENETICS

2019-2020

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FELLOWSHIP IN MEDICAL GENETICS

VISION: Establishing a centre for excellence in Medical Genetics

To promote higher educational programme of global quality and ensure advancement of knowledge in this field through Research publications.

1.**Name of the Department, Institute responsible for running the Course** Genetic Unit, Department of Anatomy, MGMCRI,SBV,Puducherry	**Course Coordinator** Name Dr.AN Uma Phone number >9994040421 e-mail umaan@mgcmcri.ac.in
2.Course Title / Nomenclature FELLOWSHIP IN MEDICAL GENETICS	Is it stand alone course? No Other depts involved **Dept. of Biochemistry** **Course Fee Proposed Rs.**—-_____
3. Target participants/students who are likely to join this course - M.Sc. Nursing - M.Sc. (Pre & Para Medical courses) - M.Sc. M.L.T - Post B.D.S - M.D.S - Post M.B.B.S - M.D /M.S	**Eligibility Criteria** A candidate who has passed any one of the following examinations of the University accepted by the Syndicate of this University as equivalent thereto shall be permitted to appear and qualify for FELLOWSHIP IN MEDICAL GENETICS of this University after a course of study of one year
4. Course Duration One year programme	How many courses do you intend to have in one academic year? One
5.Mention where further details are available about this course Give the web-link from where one can down load the brochure, or place where the printed brochures and application forms are available sbvu.ac.in	

6. Course Need

State in a paragraph how this course leads to opportunities for employment, skill development or career enhancement of students who get enrolled.

The participants will be able toIdentify and diagnose common genetic problems,Offercounseling to families with genetic disorders, Develop interest in research projects related to genetics, Help clinicians in the early diagnosis and intervention in genetic disorders, Knowledge would be of help in setting up a genetic clinic / Counseling cell, Future opportunities in the field of Gene therapy and stem cell research, Become a Scientific officer/Research Scientist in Biological / Drug / Pharmaceutical industries/companies/labs and Basic knowledge of genetics will useful in competitive exams

7. Course Goal(s) (Statement)

- To equip the participants with a strong background in the basic principles of genetics.
- To give additional insights about the modern genetics.
- To help participants to become familiar with the language of genetics and the terminology of molecular biology.
- To provide participants with the ability to solve genetic problems and think analytically.
- To sensitize the participants about the various common genetic conditions in man.
- To emphasize the participants about the significance of molecular diagnostics for various hereditary disorders.
- To give hands on training in cytogenetic Techniques in the Laboratory.
- To involve the participants in short-term research projects.
- To equip the participants with the principles of genetic counseling.

Expected Course Outcomes

The participants will be able to

- Identify and diagnose common genetic problems
- Offer counseling to families with genetic disorders
- Develop interest in research projects related to genetics
- Help clinicians in the early diagnosis and intervention in genetic disorders
- Knowledge would be of help in setting up a genetic clinic / Counseling cell
- Future opportunities in the field of Gene therapy and stem cell research.
- Become a Scientific officer/Research Scientist in Biological / Drug / Pharmaceutical industries/companies/labs
- Basic knowledge of genetics will useful in competitive exams

8. Explain the scheduling structure: Consider the following three options and give your plan

GIVEN IN 10

9. List the teaching learning activities and methods used for instruction, monitoring and mentoring

Lecture
Tutorial
Small Group Teaching
Seminars-
Journal clubs

10. Course schedule and credits

COURSE No:	**Category**	**Course Title**	**Hours / Non-Sem**	**Credit**
MGC 1	Core theory- 1	Cell Biology & Molecular Genetics	80	5
MGC 2	Core theory- 2	Human Genetics	80	5
MGL	Core Lab-1	Experiments	128	4
	Clinical lab	Clinical study	128	4
MGD	Dissertation	Research Project	256	8
MGDE	Discipline elective (to select one)	DE 1- Research Methodology and biostatistics	48	3
		DE 2- Microbial genetics		
MGGE	Generic elective courses (to select one)	GE- 1-Biomedical waste Management	48	3
		GE -2 -Palliative care		
		GE- 3- Yoga		
Total			**768**	**32**

Total Credit for one year duration = 32 Credits

11. Course Evaluation

Internal (formative) assessment- 3 TEST / year

- **Theory**- 50marks
- **Practicals**- 50marks

Summative Assessment

Theory- 2 Papers &Practicals – 1 (80marks each + 20 IA marks)

1.The actual marks in external examination (with a maximum of 80) and in the internal assessment (with a maximum of 20) are added for each paper TO A TOTAL OF 100 MARKS

2. **Internal assessment**: Internal Assessment will be based on formative assessment examinations, log books, records, presentation of seminars and journal clubs and work assessment in laboratory.

3.**RESEARCH PROJECT**: This will be a University internal assessment The students are expected to complete and submit a hard copy of a Human Genetic project within the stipulated period of one year . A Viva –Voce will be conducted. Based on their on their Research Project work and Viva –Voce they will GIVEN 100 MARKS which will find a place in the transcript.

5. If a candidate who has not secured a minimum of 50 percent in external examination shall be deemed to have failed in that course. He/She can repeat the external examination to pass that course till the Regulations are in force. A candidate who has secured a minimum of 50 marks both the papers, the practical examinations and 50 marks in their internal assessment - Project & Viva Voce, prescribed in the Regulations will be considered to have passed the one year FELLOWSHIP IN HUMAN GENETICS Candidates who obtain 75% of marks shall be deemed to have passed the examination FIRST CLASS with Distinction provided they pass all the Examinations prescribed for the Programme at first appearance.

QUESTION PAPER PATTERN

I. Answer ALL questions. (4 X 20 = 80 marks)
Either or

II. Write short notes on any FOUR from the five given in the following (4X 5 = 20)

12. **Course Requirements**

50% theory (internal) and 50% practical's (internal) marks.
80% attendance in theory and practical's

13. **Infrastructure and Physical Facilities available for the course including online resources**
Genetic lab, Anatomy dept
Biochemistry lab for practical sessions
CIDRF for molecular biology practical's

14. **Faculty and Staff in place for the Course**
Genetic lab faculty, Anatomy dept
Biochemistry faculty for theory & for practical sessions
CIDRF faculty for molecular biology theory & practical's

STAFF Requirement – One cytogenetic technologist to cater to the lab procedure is a MUST.

15. Any other information about the unique feature of this course. You may like to highlight the employment potential of the course and tangible benefits

- To give hands on training in cytogenetic Techniques in the Laboratory.
- To involve the participants in short-term research projects.
- To equip the participants with the principles of genetic counseling.

THEORY- FELLOWSHIP IN MEDICAL GENETICS

COURSE SYLLABUS

Paper I : CELL BIOLOGY AND MOLECULAR GENETICS

I.CELL BIOLOGY :

1. Scope of Genetics
2. Microscopy : Use of Microscopes in Cytology, Principles, Magnification, Resolving Power and Handling of Different Microscopes – Compound, Dark field, Bright field, Phase Contrast, Fluorescent, and Electron Microscopes.
3. Ultrastructure of Cell and Cell Organelles .
4. Cell cycle & Cell division :

Mitosis : Essentials of mitosis, plan and variants, Mitotic apparatus, its structure and chemistry ; Cytokinesis, Physiology of dividing cell, Mitotic cycle, Mitotic rhythms, blockage and stimulation of cell division, Significance of Mitosis.

Meiosis : Stages, Synaptonemal complex, Crossing over, Chiasma formation, Molecular mechanism of Crossing over, Variations in meiotic phenomena & Significance of Meiosis ; Cell senescence & Cell death (Apoptosis) : Death of specified cells, Programmed cell death, Mechanism of cell Death & its significance

References :

1. Cell and Molecular Biology 2nd Edition. P. K. Gupta (2003) Rastogi, Meerut.
2. Molecular Biology of the cell – (1989) Bruce Alberts et al. Garland Publications.
3. Cell Biology by Ambrose &Dorotty. M. Esty. ELBS Publications.
4. Cell Biology & Molecular Biology by EDF Robertis& EMF Robertis. Saunder College.
5. Cell Biology by C. B. Powar Himalaya Publication.
6. Instant notes in Microbiology by J. Nicklin et al. 2003. Viva Books.
7. Molecular Cell Biology (2 Edition)-Darnell J.H. Lodih& D. Baltimore. Scicentific American Books, New York.

II. MOLECULAR GENETICS

1. Introduction of Molecular Genetics
2. Chemical basis of Heredity
3. DNA as genetic material
4. Nucleic Acids
5. Replication
6. Transcription
7. Translation
8. Genetic code
9. Gene Action : Protein synthesis, gene expression
10. Mutations affecting structure and synthesis of proteins :
    - HaemoglobinVariants : Sickle cell disease
    - HaptoglobinVariants :Hb Freiburg Hb Marlon.
    - Thalassaemia – hereditary persistence of foetalhaemoglobin.
    - X1-antitrypsin deficiency.
11. Inborn errors of metabolism :
    - Inborn errors of amino acid metabolism
        - Disorders of purine metabolism :
        - Disorders of carbohydrate metabolism.
        - Glycogen storage disease :
        - Disorders of metabolism of lipids and lipoproteins
12. Genetic disorders such as cystic fibrosis; Duchenne& Becker's dystrophy.Genetic registers and the prevention of inherited diseases.
13. Recombinant DNA Technology
    - Basic techniques.
    - Cutting and joining DNA molecules.
    - Gene libraries and C DNA cloning.
    - Recombinant selection and screening.
    - Analysing DNA sequences.
    - The polymerase chain reaction.
    - Changing genes: site directed mutagenesis.
    - The impact of recombinant DNA technology

References:

1. Molecular Biology, Second Ed., D.Friedfelder, Jones and Barlett Inc., 1987
2. Molecular Biology of the Gene, Fourth Ed; Watson JD et al, Addison and Wesley Int. Publ. Group 1987.
3. Cell and Molecular Biology, Elighth Ed. E.D.P. Robertis and E.M.F.D RobertisJr, International Ed. Int. Med. Ltd. 1988.
4. Genes V,BLewin, Oxford University Press, 1994.
5. Text book of Biochemitry with Clinical Correlations (3rd Edn.), T.M. Devlin. 1994.
6. The Thalassaemia Syndromes (Ed.3), by D.J. Weatherall and Clegg., 1981.

7. Principles and Practice of Medical Genetics.A.E.H. Emery and D.L. Rimoin, Vol.II Churchill Livingstone, 1983, Chap.99
8. Genetic Biochemical Disorders, P.F. Benson and A.H. Fenson, Oxford Monographs in Medical Genetics, 1985.
9. Molecular Biology and Biotechnology – Molecular and Cell Biochemistry Series, No. 3, C. Smith and E.J. Wood Chapman and Hall, 1991.
10. Recombinant DNA – Second ed., J.D Watson et.al., Scientific American Books, 1992.
11. Principles of Gene Manipulation, Fifth ed., R.W. Old and S B. Primrose, Blackwell Scientific Publ. 1994.

PAPER II - HUMAN GENETICS

1. Introduction to Human Genetics

2. Human Chromosomes :

- Normal human karyotype : Paris Nomenclature; Flow Karyotyping (Quantification on DNA of individual chromosomes); FACS – Fluorescence activated cell sorter.
- Chromosomal aberration : Numerical : Aneuploidy, Polyploidy (Eg. : Turner, Down &Klinefelter Syndromes).
- Structural : Translocation, Duplication, Inversion, Ring Chromosome and Deletion (Ex. : Cri-du-chat syndrome)
- Others : Mosaic, Chimera [Individual with two cell lines]

3.**Mendelian Traits** :
Strait hair, Curly hair, Widow's peak, Dimpled Cheeks, Mid digital hair, Hitchhicker's thumb, Claspiling of hands, and Hypertrichosis.

4.Genetic Diseases and Inheritance Pattern :

- Autosomal inheritance – Dominant [Eg.: Adult polycystic kidney, Achondroplasia&eurofibromatosis.]
- Autosomal inheritance – Recessive [Eg. : Albinism, Sickle Cell Anemia, Phenyl Ketonuria]
- X-linked : Recessive {Eg. : Duchenne muscular dystrophy – DMD}
- Y-linked inheritance [Holandric – Eg. Testes determining factor]
- Multifactorial inheritance (Eg. : Congenital malformations – Cleft lip & palate, Rheum atoid arthritis and Diabetes.

5.Genomic imprinting – relevance to human diseases.

6.Cancer Genetic :
Regulation of mitotic cell cycle in eukaryotes and intercellular communication in multi cellular eukaryotes.Properties of cancer cells. Proto oncogences, Oncogenes, Cellular oncogenes, Tumor suppressor genes, Viral oncogenes. Chromosomal abnormalities associated with the specific malignancies-CML, APL, ALL, ANLL, CLL & Retinoblastoma.

7.Pedigree studies :
Symbols used in pedigree analysis. Pedigree analysis of important genetic diseases like Haemophilia, Color blindness, DuchenneMusculat Dystrophy (DMD),

8.Immunogenetics :
Genetics of normal immune system. Inherited immunodeficiency, Eg. X-linked agammaglobulinaemia. Major Histocompatibility Complex – Study of Twins (MHC), HLA disease associations. Tranplantation, graft –versus-host disease.
Pharmacogenetic – definition, gene loci influencing drug metabolism and pharmacogenetic interactions.

9.Dermatoglyhics:
Introduction, classification, Flexion creases.Dermatoglphics in clinical disorders.Clinical application & its advantages and limitations.

10.Prenatal Diagnosis and Genetic Counseling:
Definition : Various procedures used such as Amniocentesis, Chorionic villus sampling, Ultrasonograp[hy and Fetoscopy.

11.Eugenics :
Positive and Negative, Euthenics, Eugenics and Genetic Counseling

Stage 1 : History and Pedigree Construction, Stage 2 :Examination, Stage 3 : Diagnosis,
Stage 4 :Counselingand Stage 5 : Follow up

12. Genetics and Society :
(i) Human genome project : (ii) Forensic science ; (iii) DNA finger printing ; (iv) Human health care(Growth, hormone, Insulin, Interferon) and (v) Gene therapy

References :

1.Essentials of Human Genetics by S.M. Bhatnagaretal (1999) IV edition. Orient Longman.
2.Human Genetics : Concepts and Applications by Lewis R (2001) McGrawHi;; Boston.
3.Mendelian inheritance in Man : Catalogs of Autosomal recessive, and x-linked phenotypes.[12teditions–1998] by McKusick, V.A. Johns Hopkins university press, Baltimore.
4.Principles and Practive of Medical Genetics, by Emery, A.E.H and D.L. Rimoin (Eds_ (1990-2nd edition) Churchill Livingstone, Edinburgh.
5.Molecular Basis of Inherited Diseases, (6 th Edition-1989) by Scriver, C.R. A.L. Beudit, W.S. Sty abnd D. Valle (Eds0 McGrawHill, New York.)
6.Human Genetics byS.D. Gangane (2nd edition-Reprint 2001), B.L Churchill Livingstone Pvt. Ltd., New Delhi.
7.Genetics in Medicine by M.W. Thompson et al, 5 th Edition, W.B. Sounders Company, London
8.Genetic basis of common diseases by R. A. King et al, Oxford University Press. Mendelian inheritance in Man by Mc. Kusick V.A. (1998), 12 th Edition, John Hopsins University Press, Baltilmore.

PRACTICALS- FELLOWSHIP IN MEDICAL GENETICS

1.Microcopy : Handling of dissection, stereo and compound microscopes.

2.Demonstration of Phase contrast, Bright field, Dark Field, Fluorescence at CIDRF, MGMCRI, Puducherry and Electron microscopes [Visit to Dr. ARLM PGIBMS, Genetic dept., Taramani, Chennai]

3.Observation of mitotic stages in permanent slide and temporary squash preparation of onion root up for mitosis

4.Study of Karyotypes I :
Normal karyotyping in Humans Male(46 XY) and female (46, XX),G banded metaphase plate.

5.Study of Karyotypes II :
Abnormal Karyotypes – Down syndrome (Autosomal). Turner syndrome and Klinefelter syndrome (sex chromosomal)

6.Sex chromatin :
Buccal smear study and staining methods for Barr bodies
Blood smear study of drum sticks in Neutrophils

7.Blood cell counting using Haemocytometer

8.Pedigree analysis :
Symbols used in autosomal recessive disorder autosomal dominant disorder, sex chromosomal (x & Y linked)disorders.

9.Dermatoglyphics :
Recording of print of fingertips and plam. Classify ridges on the Finger tips arch, loop, and whorl. Paim print – area dem ark as hypothenar, thenar& inter digitlaread. Record presence or absence of Simian crease.

10.DNA extraction and Polymerase chain reaction (PCR)
1. Principles
2. Explanation
3. Demonstration

DISCIPLINE ELECTIVES

RESEARCH METHODOLOGY & BIOSTATISTICS RESEARCH METHODOLOGY

Unit 1. Introduction to Research: General (10 hours) Definition, need Kinds and purposes of Research: Diagnostic, Descriptive, Exploratory, Explanatory Research approaches Significance of research& importance Criteria of good research Research process : components Types of Research: Quantitative, Qualitative, Basic and appliedGeneral Topics Guidelines for Research ICMR, WHO, Nursing Research Ethics – Animal ethics; Human ethics Biosafety : Good Lab Practices, Scientific integrity and code of conduct; Plagiarism

Unit 2. Literature survey; Proposal writing (10 hours) Types of Literature search – use of library, books& journals – Medlines, internet, getting patents and article reprints as a source of literature survey Review of Literature– Formulation of Hypothesis Identification and selection of research problems, preparation of research proposal,synopsis. Research Proposal writing IP users, Plagiarism

Unit 3. Research Design;Study design (10 hours) Basic Concepts of Research Design& selection of research design Classification and types : Experimental, Pre-experimental, Quasi-Experimentaldesigns and Non - experimental Historical design, Descriptive design, case control, cohort, cross sectional,longitudinal

Unit 4. Data Collection Techniques and Interpretation (8 hours) Types of Data . Data Collection methods: Interview; Observation; Questionnaire Developing tools – Validity (internal& external), Reliability of the tools.

Unit 5. Research Reporting Scientific Writing: (10 hours) General structure of scientific reports :- IMRAD; Different types of scientificdocuments - journal articles, books, thesis, conference and project reports Components of a research paper - abstract, key words, main text, illustrations,supporting information; Publication process, copyright transfer. Open access terms Thesis: Structure and Content; Style manuals with examples (Harvard, Vancouver, APA, MLA); Citation styles:reference writing Evaluation of research reports/papers- Criteria: novelty, originally, adequacy ofinformation, responsibility, limitations, etc.

MICROBIAL GENETICS (TOTAL HOURS -48)

UNIT -I : Mendelian Genetics

Introduction to Genetics; terminology; symbols , Mendel's experiments - monohybrid cross; Dominance,Recessive, Codominance, Semidominance Lethal alleles; Complementation analysis
Dihybrid ratios; Principles of segregation; Independent assortment; Trihybrid ratios Epistasis and its types; Multiple alleles Laws of probability; Chi-square analysis and problems
Chromosomes and Inheritance .Structural organization of eukaryotic chromosome
Cell Cycle; Mitosis and Meiosis, Meiosis and Mendel's principles, Giant chromosomes: polytene and lampbrush ,Extranuclear inheritance – Mitochondrial inheritance. Extranuclear inheritance – Chloroplast inheritance.

UNIT –II : Morgan's discovery of sex linkage in Drosophila
Inheritance of sex linked genes in Drosophila ,Changes in Chromosome Structure and Chromosomal deletions in Drosophila , Chromosomal duplications in Drosophila ,Mechanisms of chromosomal inversions; Chromosomal inversion in Drosophila , Mechanisms of chromosomal translocations; Chromosomal translocation in Drosophila , Position effects of chromosome rearrangements , Nondisjunction and aneuploidy in Drosophila
Polyploidy in plants and animals

Unit III: Introduction to Microbiology

Basic of microbial existence: History of Microbiology
Microscopy: Bright, Dark field, Florescence, Phase contrast, and Scanning Electron Microscope, Transmission Electron Microscope
Microscopic examination of microorganisms
Morphology and fine structure of Bacteria -Cell wall, Flagella,
Pili, Fimbriae, Capsules, Slime layer, Endospores, Cysts,
Cytoplasmic inclusions.

Unit IV: Microbial Taxonomy and Classification

Taxonomy ranks, Classification systems
Major Characteristics used in Taxonomy, Major divisions

Bergey's Manual of Systemic Bacteriology: (Archaea, Proteobacteria, Low G+C Gram Positive bacteria, High G+C Gram Positive bacteria, Planctomycetes, Spirochetes, Bacteriodetes and Fusobacteria)
Classification of fungi and viruses

Unit V: Microbial Growth, Nutrition and Pure Culture

- Different types of media
- Growth kinetics and methods to quantitate bacterial growth
- Influence of environmental factors on growth
- Control of growth-physical, chemical methods ,antibiotics
- Isolation and preservation of microorganisms

Unit VI: Bacterial Genetics

- Conjugation, sex factors
- High frequency recombination
- Transduction (Generalized, Specialized)
- Bacterial transformation
- Mutation types, Repair mechanism, Selection of mutants

Unit VII: Genetics of Bacteriophage

- Bacteriophages Classification,
- Phage T4 – structure, gene expression and genomeorganization
- Lamda phage replication, lytic and lysogenic cycles
- Mechanisms of repressor synthesis and its control, autoregulation, one step growth curve
- Importance of bacteriophages

MODEL QUESTION PAPER
FELLOWSHIP IN MEDICAL GENTICS

Paper I : CELL BIOLOGY AND MOLECULAR GENETICS

Time : 3 Hrs. Max. : 100

I. Answer ALL questions. (4 X 20 =80 marks)

1. Explain RNA transcription and processing and Describe electron transport chain in mitochondria.
Or
2. Explain the different stages in transcription. Add a note on regulation of transcription and detail the role of enzymes in DNA replication.

3. Write a note on tumor suppressor genes with examples and discuss in detail lac operon system and its regulatory components.
Or
4. Give a brief definition of a gene library. What is the essential difference between a genomic library and a cDNA library? List the major advantages/limitations on the use of each.

5. Write in detail about molecular markers and their importance.
Or
6. Write about primer designing in PCR. Which DNA polymerase is used in PCR?

7. Restriction endonucleases
Or
8. Palindromic sequences

II. Write short notes on any FOUR of the following (4X 5 = 20)

i. Golgi bodies.
ii. Cytoplasm.
iii. Micro tubules
iv. Endocytosis
v. tRNA
vi. Cell junctions.
vii. Oncogenes
viii. AIDS

MODEL QUESTION PAPER

FELLOWSHIP IN MEDICAL GENETICS

PAPER II - HUMAN GENETICS
Time : 3 Hrs. Max. : 100

I. Answer ALL questions. (4 X 20 = 80 marks)

1. Write an essay on Mendel's Laws of Inheritance with suitable examples.
Or
2. Explain Polygenic Inheritance with suitable examples.

3. Give an account of Pedigree Analysis and its significance in family studies.
Or
4. Define and discuss the concept of Hardy-Weinberg Law with reference to simple Mendelian inheritance.

5. Describe the various methods of Genetic Counseling.
Or
6. Explain any two methods of Prenatal Diagnosis.

7. Write about different human chromosome banding techniques.
Or
8. Give a brief account on origin of numerical chromosomal abnormalities.

II.. Write short notes on any FOUR of the following – (4 X 5 = 20 marks)

a. X-linked Inheritance
b. Sex influenced characters
c. Lethal genes
d. Mutations
e. Genotype and Phenotype
f. Inbreeding Co-efficient
g. Amniocentesis
h. ά – fetoprotein