File Ref.No.34937/GA - IV - J1/2013/CU

UNIVERSITY OF CALICUT

Abstract

Faculty of Science- Scheme and Syllabus for Post Graduate Diploma Course in Clinical Genetics- Approved- implemented- Orders issued.

G & A - IV - J

U.O.No. 10798/2017/Admn Dated, Calicut University.P.O, 26.08.2017

Read:-1. Minutes of the Board of Studies in Genetics held on 28.06.2017- item No.1

2. Minutes of the Faculty of Science held on 10.07.2017- item No.17

3. Extract of the item No.II.H of the minutes of the meeting of the LXXVI meeting of the Academic Council held on 17.07.2017.

4. Orders of the Vice Chancellor in the file of 191466/GA IV/J1/2013/CU dated 27.07.2017

ORDER

Vide paper read first above, the Board of Studies in Genetics has resolved to approve the Scheme and Syllabus for Post Graduate Diploma Course in Clinical Genetics.

Vide paper read second above, the Faculty of Science has resolved to approve the Minutes of the meeting of the Board of Studies in Genetics held on 28.06.2017.

Vide paper read third above, the LXXVI meeting of the Academic Council resolved to approve the minutes of the meeting of the Faculty of Science held on 10-07-2017 and the minutes of the meetings of various Boards of Studies coming under the Faculty.

Vide paper read fourth above, the Vice Chancellor has accorded sanction to implement the resolutions of the Academic Council.

Accordingly orders are issued to implement the Scheme and Syllabus for Post Graduate Diploma Course in Clinical Genetics.

Orders are issued accordingly.

(The Scheme and Syllabus for Post Graduate Diploma Course in Clinical Genetics is appended)

Ajitha P.P

Joint Registrar

To

Examination Branch.

Forwarded / By Order

Section Officer

UNIVERSITY OF CALICUT

Scheme and Syllabus for

POSTGRADUATE DIPLOMA COURSE

CLINICAL GENETICS

TYPE OF COURSE

Post Graduate (PG) Diploma

SUBJECT

Clinical Genetics

MEDIUM OF INSTRUCTION

English

DURATION

The program will be of 12 months duration in 2 semesters (6 months duration for each semester)

BACKGROUND

Owing to the availability of genetic information from Human Genome Project and the translation of this information for clinical use, there is an increasing demand for disease diagnosis based on genetic tests. Genetic testing identifies alterations in chromosomes or genes. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine an individual's chance of developing or passing on a genetic disorder. Precise genetic tests are available for several diseases for which an accurate diagnosis is not possible using conventional methods. More than 2,000 genetic tests are currently in use, and more are being developed. Information about causative genes, disease diagnosis and available treatment options makes disease diagnosis, treatment and management easier for numerous single gene and complex genetic disorders as well as several birth defects.

Several of the modern genetic tests are not very well known even among the clinicians. More important, the implications of the results must be discussed with the patient and his/her relatives in a professional manner, since it could have tremendous emotional impact on families. Keeping this in mind a one-year PG Diploma Course on "Clinical Genetics" is framed. The course is designed to reflect the increasing impact of Genomics in unraveling the molecular and genetic underpinnings of human diseases. This will be a service-oriented course with theory, practical and hospital-based components.

OBJECTIVES

1. To develop awareness on the core concepts of Human Genetics thus bridging the gap between scientific and medical fraternity in understanding recent advances in the field of Medical Genetics
2. To provide information on the role of genetic factors in the pathogenesis of diseases
3. To impart knowledge and practical skills on conventional techniques and modern cutting edge technologies in the fields of Molecular Genetics, Cytogenetics, Clinical Genetics and Biochemical Genetics, which in turn will be useful for precise diagnosis, proper management and genetic counseling of various disorders
4. To develop awareness on the principles and components of genetic counseling, and on the ethical, legal and social issues involved associated with genetic counseling
5. To develop genetic investigators and skilled personnel in this sector
6. The course will be conducted in a clinical setting to ensure maximum real time clinical experience

SCOPE

1. Employment opportunities in academic, research and medical institutions, industries, diagnostic centers and government health schemes
2. Doctors who enroll for the course will get a deeper understanding of the core concepts of Medical Genetics, so that they can continue their practice duly prescribing genetic tests, interpreting related reports and serving as genetic counselors
3. Clinical geneticists can create awareness among medical professionals, healthcare staff, policy makers and the public about the genetic contribution to health and disease

ELIGIBILITY FOR ADMISSION

M. Sc. in Life Sciences/Applied Biology or B. Tech in Biotechnology/ Genetic Engineering/ Bioinformatics, or MBBS, with 55% marks in aggregate from a recognized university.

MODE OF SELECTION

Selection will be on the basis of entrance examination held by the respective centre.

COURSE OUTLINE

The course will be organized as a one year regular program of the university. There will be 4 core papers with 150 marks each (75 marks for theory and 75 marks for practicals) and project worth 150 marks at the end of the program. The total marks will be 750.

Evaluation of each core paper will be done in 2 parts- continuous internal assessment and external evaluation. 15 marks will be set apart for the 1st component and 60 marks for the 2nd component. The faculty concerned will be in charge of internal assessment.

Distribution of internal evaluation marks for each theory paper will be as follows:

Theory exams	5 marks (minimum 2 exams)
Assignment	2.5 marks
Seminar	2.5 marks
Attendance	5 marks
Total	15 marks

Seminar marks will be awarded on the basis of content (50% of total mark) and presentation (50% of total mark).

Distribution of internal evaluation marks for each practical will be as follows:

Practical exam	7.5 marks
Record	2.5 marks
Viva voce	5 marks

There will be a course-end University Examination for theory papers which will be held at a notified examination centre by the University. Question pattern for University theory exams (3 hrs duration) will be as follows:

Type of question	**No. of questions to be answered**	**No. of choice questions**	**Marks per question**	**Total marks**
Essay	2	3	10	20
Short Essay	2	3	5	10
Brief answers	10	12	2	20
Short answer	10	0	1	10

External practical exams will be of 4 hrs duration, 60 marks which will be distributed as follows:

Major Experiment	20 marks
Minor Experiment	10 marks
Protocol	10 marks
Record	10 marks
Viva voce	10 marks

Duration of the project work will be 180 hrs. Students are encouraged to take up projects in their field of interest at different centers in India. Dissertations are to be submitted at the time of practical exam duly signed by supervising guide and countersigned by Head of the Dept. One hundred marks are for the dissertation and 50 marks for the viva voce

(Total Project -150 marks)

Criteria for the evaluation of dissertations	**Marks**
1. Introduction, review of literature etc.	5
2. Objectives and relevance of the study	15
3. Methodology	15
4. Results	15
5. Discussion and interpretation	15
6. Conclusions	15
7. Involvement of the student	5
8. Style and neatness of the dissertation	5
9. References	10

Criteria for the Viva-voce	
Presentation of project work-	**Marks**
(POWER POINT Presentation)	
1. Quality and correctness of slides	10
2. Clarity of presentation	20
3. Communication skill	5
4. Answers to questions	15

COURSE REQUIREMENTS

Students should attend the lecture classes and practical sessions without fail and should submit assignments based on extra reading material and practical work within the stipulated deadlines. The minimum attendance for appearing in the course-end University Examination is 75%. The Head of the institution where the course is being offered shall certify to the completion of the course requirements of the students before they are admitted to the examination. However, condonation of 10% shortage in attendance is permitted as per provisions. Such students should apply and obtain orders from the University for the same before the commencement of examinations.

PASS MINIMUM

A candidate shall be declared to have passed the course if he/she obtains not less than 35% of marks in each paper and 40% of the aggregate marks. Candidates failing to secure the minimum marks need to reappear only for that paper.

CLASSIFICATION OF SUCCESSFUL CANDIDATES

Successful candidates in examinations shall be classified as follows,

a. Distinction: Those who obtain 80% and above of the aggregate marks

b. First Class: Those who obtain 60% marks and above, but less than 75% of the aggregate marks

c. Second Class: Those who obtain 50% and above, but less than 60% of the aggregate marks

d. Third Class: Those who obtain 40% and above, but less than 50% of the aggregate marks

NO. OF SEATS PROPOSED

A total of 12 seats

Members Board of Studies in Genetics

1. **Dr. C. D. Sebastian** (Chairman), Department of Zoology, University of Calicut
2. **Dr. P. R. Varghese**, Research Coordinator, , Jubilee Centre for Medical Research, Jubilee Mission Medical College, Thrissur
3. **Dr. Jyothi P. V.**, Associate Professor, Department of Botany, MES Ponnani College, Ponnani, Malappuram
4. **Dr. Jyothilekshmi P**., Associate Professor, Department of Botany, NSS College, Nenmara.
5. **Dr. K. C. Chitra**, Assistant Professor, Department of Zoology, University of Calicut
6. **Dr. Sunoj Kumar P.**, Assistant Professor, Department of Botany, University of Calicut
7. **Dr. Rajesh K. P**., Assistant Professor, Department of Botany, ZG College, Kozhikode.
8. **Dr. Abdul Hameed C**., Associate Professor, MES KVM College, Valancheri
9. **Dr. Vivek**, Assistant Professor, Department of Botany, Govt. SNGS College, Pattambi.
10. **Dr. Bosco Lawrence**, Assistant Professor, Department of Botany, Govt. Arts College, Trivandrum

SYLLABUS

SUBJECTS OF STUDY AND SCHEME OF EXAMINATION

Paper Code	Paper Title	Hours		Marks		
				External	Internal	Total
SEMESTER I						
PGDCG T01	Medical Genetics – Theory	90	-	60	15	75
PGDCG T02	Molecular Diagnostics, Bioinformatics – Theory	90	-	60	15	75
PGDCG P01	Medical Genetics – Practical	-	90	60	15	75
PGDCG P02	Molecular Diagnostics, Bioinformatics – Practical	-	90	60	15	75
SEMESTER II						
PGDCG T03	Human Cytogenetics – Theory	90	-	60	15	75
PGDCG T04	Genetic Counseling – Theory	90	-	60	15	75
PGDCG P03	Human Cytogenetics – Practical	-	90	60	15	75
PGDCG P04	Genetic Counseling – Practical	-	90	60	15	75
PGDCG D01	Project work	-	180	-	-	150
	TOTAL	**360**	**540**	**-**	**-**	**750**

PGDCG T01
MEDICAL GENETICS (Theory)

Total: 90 hrs

OBJECTIVES

1. To understand the basic principles of Genetics, Pharmacogenetics and Population Genetics
2. To provide information on pedigree analysis in disease conditions
3. To develop awareness on the genetic background of common disease conditions

UNIT 1 **20 hrs**

- Modes of inheritance; non-traditional modes of inheritance- mosaicism, genomic imprinting, uniparental disomy, mitochondrial inheritance
- Exception to Mendel's principles- incomplete dominance, codominance, multiple alleles, polygenic traits, threshold traits, epistasis, pleiotropy, environmental effects on gene expression, sex-linked and sex-limited traits
- DNA mutation- types, mutagens, molecular mechanism, lethal mutation
- Online Mendelian Inheritance in Man (OMIM) catalog of human genes and genetic disorders

UNIT 2 **20 hrs**

- Pedigree analysis- pedigree symbols, pedigree construction, family study; complications in pedigree analysis- variable expressivity, heterogeneity, penetrance, anticipation, epigenetics, mosaicism
- Mapping and identification of disease genes- linkage equilibrium and disequilibrium; linkage analysis and LOD score; genetic association study
- Human Genome Project

UNIT 3 **15 hrs**

- Monogenic disorders, sex-linked inherited disorders; polygenic disorders, complex disorders, multifactorial disorders, metabolic disorders, syndromes, congenital malformations
- Hematological disorders- haemoglobinopathies, sickle cell anemia, hemophilia; muscular disorders- Duchenne muscular dystrophy, Becker’s muscular dystrophy, spinal muscular atrophy, myotonic dystrophy; neurological and neuropsychiatric disorders- autism, schizophrenia, bipolar disorder, major depressive disorder; skeletal disorders- osteogenesis imperfecta, rheumatoid arthritis; skin disorders- albinism; eye disorders- retinitis pigmentosa; inborn errors of metabolism

- Pharmacogenetics: Basic concepts; Drug metabolism; Altered drug response and genetic factors; Application of molecular diagnostics and Pharmacogenetics in personalized medicine

UNIT 4 **15 hrs**

- Developmental and reproductive genetics: Genetic and environmental susceptibility for reproductive disorders
- Reproductive disorders- Male infertility, endometriosis, recurrent early pregnancy loss, polycystic ovarian disorder
- Assisted reproductive technologies (ART)- IUI, IVF, ICSI, ZIFT, GIFT
- Pre-implantation genetic diagnosis

UNIT 5 **20 hrs**

- Cancer genetics: Genetic basis of cancer- neoplasms, tumorigenesis, apoptosis
- Oncogenes; tumor suppressor genes; protooncogenes; DNA repair mechanisms; telomeres and telomerases; genomic instability and cancer- mutation rates in normal and neoplastic cells
- DNA damaging agents (environmental, physical, chemical, biological)

PGDCG P01
MEDICAL GENETICS (Practicum)

Total: 90 hrs

1. Extraction of genomic DNA from blood/tissue samples (phenol extraction, salting out)
2. Extraction of total RNA from lymphocytes/tissue samples
3. Quantification of nucleic acids
4. OMIM database
5. Data mining
6. Clinical case studies (monogenic disorders, sex-linked inherited disorders, polygenic disorders, complex disorders, multifactorial disorders, metabolic disorders, syndromes, congenital malformations) at ICCONS and nearby hospitals
7. Pedigree analysis in disease conditions
8. Visit to hospitals offering ART
9. Visit to hospitals/institutes involved in cancer research

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3. Griffiths AJF, Wessler SR, Carroll SB, Doebley J. 2015. An Introduction to Genetic Analysis, 11th Ed.,W H Freeman & Co, NY
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10. Nussbaum RL, McInnes RR, Willand HF. 2016. Thompson & Thompson Genetics in Medicine, 8th Ed., Elsevier, PA
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12. Kumar D, Weatherall D. 2008. Genomics and Clinical Medicine, Oxford University Press, NY
13. Baraitser M, Winter R. 1983. A Colour Atlas of Clinical Genetics, Sheridan House Inc., MD
14. Snyder M. 2016. Genomics & Personalized Medicine, Oxford University Press, NY

PGDCG T02
MOLECULAR DIAGNOSTICS AND BIOINFORMATICS (Theory)

Total: 90 hrs

OBJECTIVES

1. To develop awareness on molecular diagnostic techniques and molecular markers
2. To provide information on biological databases and Bioinformatics tools

UNIT 1 **20 hrs**

- Sample collection- method of collection, transport and processing of samples
- Molecular techniques: Polymerase chain reaction (PCR), gel electrophoresis, DNA sequencing, microarray, next generation sequencing (NGS), DNA fingerprinting, gene expression profiling, RNA sequencing, blotting techniques, cloning

UNIT 2 **15 hrs**

- Molecular markers: Restriction fragment length polymorphism (RFLP), PCR-RFLP, allele-specific PCR, amplification-refractory mutation system (ARMS)-PCR, multiplex PCR, amplified fragment length polymorphism (AFLP), random amplified polymorphic DNA (RAPD), single nucleotide polymorphism (SNP), multiplex ligation-dependent probe amplification (MLPA), short tandem repeats (STR), variable number of tandem repeats (VNTR)
- SNPs in diagnostics

UNIT 3 **15 hrs**

- Neonatal and prenatal disease diagnostics
- Molecular diagnosis for early detection of genetic disorders; gender identification using amelogenin gene locus; amplification of Y chromosome-specific short tandem repeats; analysis of mitochondrial DNA for maternal inheritance

UNIT 4 **20 hrs**

- Disease identification and genetic tests- Thalassemia, Fanconi anemia, sickle cell anemia, fragile-X syndrome, Alzheimer's disease, Duchenne Muscular Dystrophy/Becker's Muscular Dystrophy, Huntington's disease
- Allelic susceptibility test for multifactorial disorders- neural tube defect, cleft lip and palate, cardiovascular disorder, male infertility
- HLA typing and tissue transplantation matching

UNIT 5 **20 hrs**

- Introduction to Bioinformatics- applications, gene, genome, genomics
- Sequence Analysis- Nucleotide sequence analysis; homology sequence analysis- BLAST, PASTA, pair-wise sequence analysis; multiple sequences- CLUSTALW; phylogenetic analysis
- Analysis of NGS data; meta-analysis
- Genome annotations
- Biological Databases- Importance, primary and secondary databases, nucleotide and proteome databases, database querying software; Webtools
- Submitting sequences to databases- BankIt, Sequin
- Applications of genome analysis and genomics

PGDCG P02

MOLECULAR DIAGNOSTICS AND BIOINFORMATICS (Practicum)

Total: 90 hrs

1. Agarose gel electrophoresis (DNA, RNA)
2. Agarose gel staining
3. PCR
4. Multiplex PCR
5. RFLP
6. PCR-RFLP
7. RAPD
8. Detection of SNPs
9. Detection of STRs
10. Gender identification using amelogenin gene locus
11. Genetic tests for single gene disorders (DNA sequencing-automated)
12. Genetic tests for polygenic disorders (Next Generation Sequencing- targeted sequencing)
13. DNA fingerprinting
14. Western blot
15. Plasmid extraction
16. Preparation of competent cells (calcium chloride)
17. Transformation (heat shock)
18. Ligation
19. Cloning
20. Selection of transformants (antibiotic screening) and clones (blue white screening)
21. Bioinformatics (UCSC Genome Browser, in silico PCR, primer design, BLAST, RefSeq, nucleotide databases, proteome databases, nucleotide sequence analysis, proteomic sequence analysis; pair-wise sequence analysis; multiple sequence analysis; NGS data analysis; meta-analysis)
22. Submitting sequences to databases

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5. Higgins D, Taylor W. 2000. Bioinformatics Sequence, Structure and databanks, Oxford University Press, NY

6. Attwood TK, Parry-Smith DJ. 1999. Introduction to Bioinformatics, Pearson Education Limited, England
7. Krane DE, Raymer ML. 2005. Fundamental concepts of Bioinformatics, Pearson Education Limited, England

PGDCG T03
HUMAN CYTOGENETICS (Theory)

Total: 90 hrs

OBJECTIVES

1. To understand the fundamental concepts of Cytogenetics
2. To provide information on cytogenetic techniques
3. To develop awareness on the chromosomal diagnostics

UNIT 1 **30 hrs**

- Molecular organization of eukaryotic chromosomes; higher order of eukaryotic chromosomes; nucleosome structure; chromosomal protein; chromomere, kinetochores, centromeres and telomeres; heterochromatin and euchromatin; X-chromosome inactivation
- Supernumerary chromosomes- occurrence, role during meiosis and mitosis, evolutionary significance
- Microscopy- bright-field, phase-contrast, polarization, fluorescence, confocal; microphotography; video processing and image processing

UNIT 2 **10 hrs**

- Basics of cell culture- equipment, culture media, safety, contamination, cell observation, factors affecting cell growth in culture, passaging, transfection, subculturing
- Techniques of cell culture- short term lymphocyte, primary and secondary cell cultures, maintenance of cell lines, harvesting of cells for chromosomal analysis
- Cryopreservation

UNIT 3 **15 hrs**

- Techniques of chromosome analysis; chromosome preparation from cultured lymphocytes, cell lines and solid tumors
- Karyotyping- spectral karyotyping, digital/virtual karyotyping
- Banding- C-banding, G-banding, R-banding, Q- banding, M- banding and NOR banding, chromosome banding patterns and ISCN nomenclature
- Fluorescence in situ hybridization (FISH)- M-FISH, Q-FISH; comparative genomic hybridization (CGH); comet assay; cytokinesis-blocked micronucleus assay (CBMA)

UNIT 4 **15 hrs**

- Chromosomal aberrations: Numerical- polyploidy, aneuploidy, autosomal, sex-chromosomal; Structural- deletion, duplication, translocation, inversion; isochromosome; ring chromosome
- Chromosomal abnormalities in cancer- Philadelphia chromosome, acute lymphoid leukemia, chronic myeloid leukemia

UNIT 5 **20 hrs**

- Clinical cytogenetics: chromosomal diagnostics; heritable chromosomal abnormalities; incidence of chromosome aberrations; disorders of autosomes; disorders of sex chromosomes; disorders of sexual differentiation; chromosome breakage syndromes

PGDCG P03
HUMAN CYTOGENETICS (Practicum)

Total: 90 hrs

1. Microscopy
2. Techniques of cell culture
3. Cryopreservation
4. Harvesting of cells for chromosomal analysis
5. Chromosome preparation for harvesting
6. Banding techniques
7. Karyotyping
8. Numerical aberrations (Down syndrome)
9. Structural aberrations
10. Sex chromatin (buccal mucosa)
11. FISH

REFERENCES

1. Bell S, Morris K. 2009. An Introduction to Microscopy, CRC Press, Taylor And Francis Group, FL
2. Gardner EJ, Simmons MJ, Snustad DP. 1991. Principles of Genetics, John Wiley & Sons Inc., NY
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PGDCG T04
GENETIC COUNSELING (Theory)

Total: 90 hrs

OBJECTIVES

1. To understand the principles and components of genetic counseling
2. To develop awareness on the treatment and management of genetic disorders
3. To provide knowledge on the ethical and social issues involved in genetic counseling

UNIT 1 **15 hrs**

- Principles of genetic counseling; causes and factors for seeking counseling; ethical and legal issues in genetic counseling
- Risk evaluation- Mendelian risk, empirical risk

UNIT 2 **20 hrs**

- Genetic screening: Prenatal testing, preimplantation testing, newborn screening, diagnostic testing, carrier testing, predictive testing, presymptomatic testing
- Non-invasive tests- triple test, ultrasonography
- Invasive tests- amniocentesis, chorionic villi sampling, fetal blood sampling

UNIT 3 **20 hrs**

- Treatment and management of genetic disorders
- Gene therapy- principles and strategies, classical gene therapy; therapeutics based on targeted inhibition of gene expression, mutation corrections in vivo, exon skipping
- Molecular medicine; molecular therapeutic approaches

UNIT 4 **20 hrs**

- Biology and genetics of stem cells; molecular circuitry of pluripotency and nuclear reprogramming
- Stem cells and niches- mechanism that promotes stem cell maintenance through life, mechanisms of asymmetric cell divisions
- Stem cell therapy for cancer; prospects for stem cell therapy
- Ethical and social considerations of stem cell research

UNIT 5 **15 hrs**

- Psychology and medical ethics- Medicine as a social institution, nature of bioethics, patient-physician relationship, informed consent, decision making for incompetent patients, right to know (telling patient the truth), confidentiality, psychological basis of counseling

PGDCG P04
GENETIC COUNSELING (Practicum)

Total: 90 hrs

1. Clinical case studies
2. Pedigree analysis
3. Risk calculation
4. Mock genetic counseling sessions
5. Visit to hospitals involved in non-invasive and invasive genetic screening procedures

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1. Nussbaum RL, McInnes RR, Willand HF. 2016. Thompson & Thompson Genetics in Medicine, 8th Ed., Elsevier, PA
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Model Question PaPers

PGDCG T01
MEDICAL GENETICS (Theory)

Duration: 3 hrs **Max Marks: 60**

I. Answer any TWO. Each question carries 10 marks

1. Explain the non-traditional modes of inheritance with examples
2. Explain the various complications involved in pedigree analysis
3. Write an essay on assisted reproductive technologies

II. Answer any TWO. Each question carries 5 marks

4. Molecular mechanism of DNA mutations
5. Online Mendelian Inheritance in Man (OMIM)
6. Human Genome Project

III. Answer any TEN. Each question carries 2 marks

7. Mosaicism
8. Genomic imprinting
9. Uniparental disomy
10. Mitochondrial inheritance
11. Polygenic traits
12. Environmental effects on gene expression
13. Types of DNA mutation
14. Linkage disequilibrium
15. Congenital malformations
16. Duchenne muscular dystrophy
17. Pre-implantation genetic diagnosis
18. DNA damaging agents

IV. Answer ALL. Each question carries 1 marks

19. Threshold traits
20. Epistasis
21. Pleiotropy
22. Sex-limited traits
23. LOD score
24. Genetic association study
25. Complex disorders
26. Personalized medicine
27. Endometriosis
28. Telomeres

PGDCG T02
MOLECULAR DIAGNOSTICS AND BIOINFORMATICS

Duration: 3 hrs **Maximum Marks: 60**

I. Answer any TWO. Each question carries 10 marks

1. Write an essay on neonatal and prenatal disease diagnostics
2. Write an essay on HLA typing and tissue transplantation matching
3. Write an essay on biological databases

II. Answer any TWO. Each question carries 5 marks

4. Maternal inheritance
5. Bioinformatics tools for nucleotide sequence analysis
6. Blotting techniques

III. Answer any TEN. Each question carries 2 marks

7. Electrophoresis
8. Microarray
9. DNA fingerprinting
10. RNA sequencing
11. Cloning
12. Allele-specific PCR
13. Amplified fragment length polymorphism
14. Variable number of tandem repeats
15. Genetic test for sickle cell anemia
16. Male infertility
17. CLUSTALW
18. Online tools for submitting sequences to databases

IV. Answer ALL. Each question carries 1 marks

19. PCR-RFLP
20. Multiplex PCR
21. Random amplified polymorphic DNA
22. Single nucleotide polymorphism
23. Multiplex ligation-dependent probe amplification
24. Short tandem repeats
25. Amelogenin gene
26. Fragile X syndrome
27. Huntington's disease
28. Meta-analysis

PGDCG T03
HUMAN CYTOGENETICS

Duration: 3 hrs **Maximum marks: 60**

I. Answer any TWO. Each question carries 10 marks

1. Write an essay on molecular organization of eukaryotic chromosomes
2. Write an essay on chromosomal abnormalities in cancer
3. Write an essay on chromosome banding patterns, ISCN Human Cytogenetic Nomenclature and banding techniques

II. Answer any TWO. Each question carries 5 marks

4. X-chromosome inactivation
5. Supernumerary chromosomes
6. Chromosome breakage syndromes

III. Answer any TEN. Each question carries 2 marks

7. Chromosomal protein
8. Chromomere
9. Microphotography
10. Cell culture media
11. Passaging
12. Transfection
13. Karyotyping
14. G-banding
15. Fluorescence banding
16. Fluorescence in situ hybridization
17. Aneuploidy
18. Chromosomal abnormalities in acute lymphoid leukemia

IV. Answer ALL. Each question carries 1 marks

19. Nucleosomes
20. Centromeres
21. Heterochromatin and euchromatin
22. Comet assay
23. Cytokinesis-blocked micronucleus assay
24. Comparative genomic hybridization
25. Isochromosome
26. Ring chromosome
27. Philadelphia chromosome
28. Disorders of sex chromosomes

PGDCG T04
GENETIC COUNSELING

Duration: 3 hrs **Maximum Marks: 60**

I. Answer any TWO. Each question carries 10 marks

1. Write an essay on the principles and components of genetic counseling
2. Write an essay on the strategies for genetic screening
3. Write an essay on the ethical and social considerations of stem cell research

II. Answer any TWO. Each question carries 5 marks

4. Ethical issues in genetic counseling
5. Risk evaluation in genetic counseling
6. Principles of gene therapy

III. Answer any TEN. Each question carries 2 marks

7. Causes and factors for seeking genetic counseling
8. Empirical risk
9. Preimplantation testing
10. Carrier testing
11. Predictive testing
12. Mutation corrections in vivo
13. Stem cells
14. Pluripotency and nuclear reprogramming
15. Stem cell therapy for cancer
16. Bioethics
17. Patient-physician relationship
18. Psychological basis of counseling

IV. Answer ALL. Each question carries 1 marks

19. Triple test
20. Amniocentesis
21. Chorionic villi sampling
22. Targeted inhibition of gene expression
23. Exon skipping
24. Molecular medicine
25. Pluripotency
26. Informed consent
27. Decision making for incompetent patients
28. Confidentiality