Faculty of Biomedical Science

JSS ACADEMY OF HIGHER EDUCATION & RESEARCH
(DEEMED TO BE UNIVERSITY) MYSURU

JSS Academy of Higher Education & Research
(Deemed to be University)
Re-Accredited "A+" Grade by NAAC
Sri Shivarathreeshwara Nagara Mysuru - 570015, Karnataka

Regulation & Syllabus

MEDICAL GENETICS AND GENOMICS
M.Sc MG&G
2022

MSc

REGULATIONS AND CURRICULUM

M.Sc. MEDICAL GENETICS AND GENOMICS

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MSc PROGRAM (MEDICAL GENETICS AND GENOMICS – MSc MG&G)

About the regulations and Commencement

- The following regulation is made for the **MSc program (Medical Genetics and Genomics).**
- It intends to identify and foster the unique capability of each student, and provide them flexibility in learning, and conceptual understanding of subjects. It emphasizes practical work/research-oriented learning.
- The regulation presented is student-centric and aims to provide holistic and multidisciplinary education suitable for the present 21st generation.
- These regulations will come into force from the Academic year 2022-23

About the program:

- The program consists of 2 years (4 semesters) of MSc including a research thesis (4 months) and an industry internship (2 months) leading to a master's degree.
- It is a credit-based program.

Total credits for the program: 98 credits (For MSc degree)

Year of implementation: 2022-2023

Total intake: 15

Program vision and mission

- To educate young minds through teaching driven by curiosity and research.
- To impart advanced knowledge in genetics and to expose students to the emerging field of genomic medicine.

Program Goal

- To enable students to acquire knowledge and understanding of molecules, molecular mechanisms, and current theories of human molecular genetics.
- To train individuals and nurture, them to emerge as skilled laboratory experts or industrial technical experts and nurture their research attitude.
- To engage students with genetic counsellors and improve their communication skills and enable them to counsel the patients about genetic diseases.

Program outcome

At the end of the program students should be able to -

- PO1. Acquire the basic and specialized knowledge in medical genetics and understand the applications of Cytogenetics and Biochemical genetics.
- PO2. Understand the complexity of the human genome and the molecular mechanisms at the system level and how the body functions in health and diseases.
- PO3. Familiarize with complex and rare genetic disorders and establish themselves as Medical Geneticist professionals.
- PO4. Understand the working principles of lab equipment used for genetic testing and take up scientists jobs at molecular diagnostics and hospitals
- PO5. Understand the genetic component of immune-mediated traits and update the concepts about the regulation of immune responses and build a basis for an interdisciplinary idea for translational research.
- PO6. Comprehend psychological and ethical issues; familial and social implications of genetic contributions to health and human disease and take up genetic counselling jobs

after completing appropriate exams.

- PO7. Students can pursue higher studies (Ph.D.) in life sciences
- PO8. Students can pursue a career in academia or industry based on their interest

Pedagogy

- The syllabus is designed to encourage critical thinking and problem-solving ability in students. The classroom lectures include discussions, presentations and encourage self-learning.
- Students are encouraged to develop reasoning abilities and learn through questioning. Faculties are to provide all the necessary fundamentals so that students can develop the art of questioning.
- Learning through experimentation- Most of the core subjects have practical lab work to provide hands-on experience.
- Faculties will be free to follow suitable pedagogy to impart an education that is best suited for the courses. Emphasis is given to learning which suits the Indian context and aligns with our country, context, and culture.
- A combination of an online and in-person mode of teaching is encouraged for effective learning. This Flexibility allows students to listen to lectures/seminars and talks by experts from different geographical locations.

Eligibility for admission:

- A candidate seeking admission to the program must have passed BSc with at least one subject of biological science or BAMS or MBBS or BHMS or BPT of BPharm from a recognised university.

Medium of instruction:

- Instructions and examinations shall be in English

Definition of Keywords:

- **Academic Year:** Two consecutive semesters (an odd and an even).
- **Semester:** Each semester will consist of over 15-16 weeks of academic work.
- **Program:** A program leading to the award of a Degree, diploma, or Certificate.
- **Credit:** A unit by which the course work is measured. It determines the number of hours of instruction required per week in a semester.
- **Choice Based Credit System (CBCS):** The CBCS provides options for students to select subjects from the prescribed open elective, discipline elective, ability, and skill enhancement language, soft skill subjects, etc.
- **Course:** is referred to as 'papers' is a component of a program. It includes subjects comprising lectures/ tutorials/laboratory work/ project work/ Internships /viva/ seminars/ term papers /assignments/ presentations / self-study or a combination of some of these.
- **Credit-Based Semester System (CBSS):** Under the CBSS, the requirement for awarding a degree /diploma /certificate is prescribed in terms of the number of credits to be earned.
- **Grade Point:** Numerical weight allotted to each letter grade on a 10-point scale.
- **Credit Point:** A product of grade points and the number of credits for a course.
- **Letter Grade:** It is an index of the performance of students in a said course. Grades are denoted by the letters O, A, B, C, D, E and Fail.
- **Semester Grade Point Average (SGPA):** It is a measure of performance of work done in a semester. Defined as the ratio of total credit points secured by a student in various

courses registered in a semester and the full course credits taken during that semester. It shall be expressed up to two decimal places.

- **Cumulative Grade Point Average (CGPA):** It measures the overall cumulative performance of a student in all the semesters of the program. The CGPA is the ratio of total credit points secured by a student in various courses in all the semesters and the sum of the total credits of all courses in all the semesters. It is expressed up to two decimal places.
- **Transcript or Grade Card or Marks Card:** Based on the grades earned, a graded transcript shall be issued to all the registered students after every semester. The grade transcript will display the course details (title, number of credits, grade secured).

Research component during the program

- Students will carry out research in their 4th semester along with an industrial internship.
- Students must take up 2 months of internship during the 4th semester.

Credit awarded for the program

- Each subject taught in the program carries a defined number of credits. The credits are based on the teaching mode i.e. number of contact hours for lectures, tutorials, and laboratory practicals.
- 1 hour of theory/tutorial teaching per week is equal to 1 Credit
- 2 hours of laboratory practicals/laboratory postings per week is equal to 1 Credit.
- Grading calculations – 1 credit is equal to 25 marks in a semester. Therefore, a 4- or 5-credit course will have a maximum of 100 marks, a 2-credit course would cover a maximum of 50 marks. The proportion of marks earned in a course and the credits given to that course is used to calculate the Semester Grade Point Average (SGPA) or Cumulative Grade Point Average (CGPA).

Subjects of study (Table 1)

- Subjects include Discipline specific courses (DSC), discipline-specific elective (DSE) courses, open elective (OE) courses, ability enhancement courses (AEC), skill enhancement courses (SEC), and value-based activity (VBA), laboratory practicals, internships and research project work.

Definitions

- **Discipline Specific Course** (DSC) is a core course, which should be compulsorily studied by a student as a core requirement of the program.
- **Elective Courses** can be chosen from a pool of courses. It may be very specific, specialized, advanced, or supportive to the discipline/subject of study, which provides an extended scope, or enables exposure to some other discipline/subject/domain or nurtures the student's proficiency/skill.
- **Discipline Specific Elective** (DSE) is offered under the main discipline/subject of study.
- **Open Elective** (OE) is chosen from an unrelated discipline/subject to seek exposure beyond discipline/subject.
- **Ability Enhancement Courses** (AEC) may be of two types: Ability Enhancement Compulsory Courses (AECC) and Skill Enhancement Courses (SEC).
- **AECC** courses are mandatory courses based upon the content that leads to knowledge enhancement viz., Environmental Studies, Indian Constitution, and English/ /Communication skills.
- **SEC** courses are aimed at providing hands-on training, competencies, skills, etc.

- **Value-Based/Activity Based** (VBA) courses are intended to enhance the employability of the students. The courses will help to bridge the gap between the skill requirements of the employer or industry and the competency of the students.
- **Massive Open Online Courses** (MOOCs) are online courses, which are available on the SWAYAM (Study Web of Active-Learning for Young Aspiring Minds) platform of the Government of India. In case a student selects MOOCs instead of the interdisciplinary course, the credit earned therefrom will be considered for grading and ranking. The credits earned under the SWAYAM platform are also transferable from one institution to another. The student is eligible to take additional courses under MOOCs if he/she opts to earn extra credits, however, these would be considered as additional SWAYAM/ MOOCs and will not be considered either for grading or ranking.

Table 1: Program design and Subject of study

SEM	Discipline Core (DSC) Credits L+T+P	Discipline-specific/Open elective subject (Credits) L+T+P	Skill enhancement courses (SEC)/Ability enhancement courses (AEC) Languages (Credits) L+T+P	Value-based activity/ course (Credits) L+T+P	Total Credits
1	Fundamentals of Genetics and Human Cytogenetics (DSC-1) 3+1+2 Cell & Molecular Biology (DSC-2) 3+1+2 Biochemistry & Metabolic Disorders (DSC-3) 3+1+2		Essentials of Immunology (AEC-1) (2+0+0) Use of model organisms to study human diseases (AEC-2) (2+0+0)		22
2	Biostatistics & Bioinformatics (DSC-4) 3+1+2 Human Evolutionary and Population Genetics (DSC-5) 3+1+2 Human Molecular Genetics (DSC-6) 3+1+2	Proteomics OR Microbial Genetics (DSE-1) 2+0+0 Humanities OR Health Economics (OE-2) 2+0+0	Fundamentals of Epigenetics (AEC-3) 2+0+0 Research Methodology (AEC-4) 2+0+0		26
3	Genetics of Rare Diseases (DSC-7) 3+1+2 Genetics of Complex Diseases (DSC-8) 3+1+2 Genetic Counselling (DSC-9) 3+1+2	Soft Skills OR IPR (OE-3) 2+0+0 Pharmacogenetics OR Nutrigenomics (DSE-4) 2+0+0	Infertility and Assisted Reproductive Technology (AEC-5) 2+0+0 Scientific Writing (AEC-6) 2+0+0		26
4			**Masters Research Project 0+0+18**	Industry Internship (VBA-1) 0+0+6	24
MSc in Medical Genetics & Genomics (98 credits)					

Tutorials/Seminars/Journal clubs

- Students are expected to actively participate in departmental seminars and journal clubs.
- Seminars for the subjects may be conducted by the subject faculty at his/her convenience.
- A record should be maintained for each student and the list of seminars and papers presented in the journal club by each student should be maintained in the department.

Industry/Academia internship

- Students will be placed in reputed industries identified and approved by the JSSAHER for 2 months. Students must maintain the logbook which has to be certified by the concerned authority in the industry on daily basis. At the end of the internship, the student is expected to attain additional competencies and should submit an internship report which will be certified by the industry authorities along with their feedback.

Research project report/thesis

- Every student is required to carry out work on a selected research project under the guidance of a recognized post-graduate teacher in their respective subjects. The thesis shall be submitted to the controller of examination of the JSSAHER 15 days before the end of the 4th semester to be evaluated by the examiners.
- Every student will be given an introductory course in research methodology, biostatistics, and scientific writing before they embark on the research project. He/she will be taught how a research project can be planned and implemented. He/she must also acquire basic knowledge of statistical methods and their applications.
- The project is aimed to train a postgraduate student in research methods and techniques. It includes identification of a problem, formulation of a hypothesis, search and review of literature, getting acquainted with recent advances, designing of a research study, collection of data, critical analysis, and comparison of results, and drawing conclusions.
- Every student shall submit a synopsis in the prescribed format containing particulars of the proposed project work in the first four weeks of the 3rd semester.
- The synopsis shall be sent through the proper channel. Such a synopsis will be reviewed, and the project topic will be registered by the JSS Academy of Higher Education & Research. No change in the research topic or guide shall be made without prior approval of the JSSAHER. A co-guide may be included provided the work requires a substantial contribution from a sister department or another institution recognized for teaching/ training by the JSS Academy of Higher Education & Research.
- The candidates shall report the progress of the research work to the concerned guide periodically and obtain clearance for the continuation of the project work. The **Report/ Thesis** should be written under the following headings

i. Introduction
ii. Aims of Objectives of the study
iii. Review of Literature
iv. Material and Methods
v. Results
vi. Discussion
vii. Conclusion
viii. Summary
ix. References
x. Tables
xi. Annexure

- Four copies of the Report/Thesis thus prepared shall be submitted 15 days before the end

of the 4th semester or before the dates notified by the JSS Academy of Higher Education & Research.

Attendance

- Candidates should have attended at least 80% of the total number of classes conducted in a semester, from the date of commencement of the term to the last working day, as notified by the Deemed to be University, in each of the subjects prescribed for that semester, separately in theory and practical, to be eligible to appear for the Deemed to be University examinations. Otherwise, the candidate shall not be allowed to appear for the exam.

Maintenance of Logbook and Practical record

- A diary showing each day's work has to be maintained by the candidate, which shall be scrutinized by the Head of the Department every month. A list of the seminars and journal reviews that have been attended and presented by the student must be maintained which should be scrutinized by the Head of the Department.
- A practical record must be maintained by every candidate and duly scrutinized and certified by the Head of the Department and submitted to the external examiner during the end-of-semester examination.

Assessment and Evaluation

The whole purpose of assessment and evaluation is to constantly improve the quality of students. It also acts as an indicator of faculties ability to teach effectively. Following are the guidelines

- As far as possible the assessments should provide constructive feedback to the candidates.
- In addition to the regular theory examinations, assessments will involve the active participation of students in seminars, presentations, project participation, and peer assessments.
- The examination questions may include, open-ended concepts, multiple choices answer to trigger out-of-box thinking of students.

Process of evaluation – Theory

1. The evaluation will be done based on
    - formative assessment/internal assessment (IA) and
    - summative assessment/end-semester examination.
2. Weightage provided for the above evaluation mode will be in a 30:70 pattern for IA and end-of-semester examination respectively.

    Total Marks for each course = 100%
    - Internal assessment (IA 1) = 15% marks
    - Internal assessment (IA 2) = 15% marks
    - Semester End Examination = 70% marks.

Internal assessment

- The Department will conduct IA tests, each semester. IA shall assess the student's performance on a continuous basis. The tests may include written papers, assignments, seminars, practicals, and viva voce. Marks obtained in each test will be maintained by the Head of the Department and sent to the controller of examination, JSSAHER. The candidates who have failed the end-semester examination shall be given an internal assessment improvement test and the best marks shall be submitted to the controller of examination.

- Internal assessment IA-1 will be scheduled after completing 50% of the syllabus of the course within the stipulated time. The assessment is for 15% of the total marks.
- Internal assessment IA-2 will be scheduled after completing the remaining 50% of the syllabus of the course within the stipulated time. The assessment is for 15% of the total marks.
- Evaluated papers shall be discussed with the students for their improvement. IA marks shall be communicated to the controller of examination and shall be put on the notice board. IA marks may be provided separately in the final transcript.

Industry/Academia internship evaluation

- The industry/academia internship report will be submitted to the Head of the Department at JSS Medical College. Based on the logbook, the report and the feedback from the industry marks will be allotted a maximum mark of 150 (6 credits) for the report.

Research thesis evaluation

- Master's degree (450 marks, 18 credits) - Marks shall be awarded for the thesis writing and a project ORAL PRESENTATION (50:50 pattern). Two examiners shall be present for the project presentation (**One internal and One from an external university)**

End semester examination

- The end-of-semester examination shall be conducted by the university during the 20th – 22nd week of the semester. The assessment is for the remaining 70% of the total marks.
- If a student fails to attend take IA-1 and IA-2 on a scheduled date, it shall be deemed that the student has dropped the test. However, in case of genuine reasons such as health emergencies, the student may appeal to the Program Coordinator for a re-test before the commencement of the concerned semester end examinations
- The examination consists of both theory and practical at the end of every semester as prescribed in the schemes of examination.
- Practical shall be conducted by two examiners (Both internal examiners, nominated by the university)
- The student should submit record books for practical examination signed by the respective faculty for evaluation.

Examination conducted by JSSAHER

- Theory – 3 hours of paper, 70 marks, 4 credits in each course.
- The JSSAHER shall conduct an examination for the core course at the end of each semester. The candidates, who satisfy the requirement of attendance and internal assessment, shall be eligible to appear for the JSSAHER examination. The head of the institution shall verify the same before forwarding the applications to the JSSAHER within the stipulated time along with the prescribed fee.

Examination conduction at the Department level

- Theory – 1.5 hours of paper, 50 marks, 2 credits in each subject. For the scientific writing & presentation course, the student must submit the assignment given to him which will be evaluated for 50 marks.
- Examination for Allied subjects and Elective subjects shall be conducted by the college and the marks obtained shall be submitted to the JSSAHER along with the IA marks of the core subjects at least 15 days before the commencement of the JSSAHER examination. The marks of non-core subjects shall be incorporated into the marks card issued by the

JSSAHER.

Criteria for appointment of examiners:

- Examiners shall be appointed by the JSSAHER to conduct the end-semester JSSAHER examinations, from the panel of examiners approved by the Board of Studies. For Practical examinations, there shall be two internal examiners. Theory papers shall be valued by both examiners. Postgraduate teachers with MD/MS/Ph.D. degrees with 3 years of experience shall be appointed as examiners.

Scheme of Examination:

The distribution of subjects, number of teaching hours, credits, and marks distribution for each semester's theory and practical courses and examinations are shown in Table 2.

Table 2: Marks distribution for each semester's theory and practical courses and examinations

SEMESTER 1								
Sl. No.	**Study Components and Code**	**Title of the Paper**	**Teaching hours**		**Examination**			**Total Credit**
			Hrs/ Week	**Total Hours**	**IA**	**Theory/ Practical**	**Max. Marks**	
1.	**DSC-1**	Fundamentals of Genetics & Human Cytogenetics	4	60	30	70	**100**	**4**
2.	**DSC-2**	Cell and Molecular Biology	4	60	30	70	**100**	**4**
3.	**DSC-3**	Biochemistry and metabolic disorder	4	60	30	70	**100**	**4**
4.	**AEC-1**	Essentials of Immunology	2	30	--	50	**50**	**2**
5.	**AEC-2**	Use of model organisms to study human diseases	2	30	--	50	**50**	**2**
6.	**Practical-1**	Genetics and Cytogenetics	2	60	100 marks practicals 50 marks viva		**150**	**2**
7.		Cell and Molecular Biology	2	60			**2**	2
8.		Biochemistry	2	60			**2**	2
	Total Marks and Credits						**550**	**22**
SEMESTER 2								
1.	**DSC-4**	Biostatistics & Bioinformatics	4	60	30	70	**100**	**4**
2.	**DSC-5**	Human Evolutionary & Population Genetics	4	60	30	70	**100**	**4**
3.	**DSC-6**	Human Molecular Genetics	4	60	30	70	**100**	**4**

4.	**AEC-3**	Fundamentals of Epigenetics	2	30	--	50	**50**	**2**
5.	**AEC-4**	Research Methodology	2	30	--	50	**50**	**2**
6.	**Practical-2**	Biostatistics & Bioinformatics	2	60	100 marks practicals 50 marks viva		**150**	**2**
7.		Population genetics	2	60				**2**
8.		Human Molecular Genetics	2	60				**2**
9.	**DSE-1**	Proteomics/Microbial genetics	2	30	--	50	**50**	**2**
10.	**OE-1**	Humanities/Health economics	2	30	--	50	**50**	**2**
	Total Marks and Credits						**650**	**26**
SEMESTER 3								
1.	**DSC-7**	Genetics of rare diseases	4	60	30	70	**100**	4
2.	**DSC-8**	Genetics of complex diseases	4	60	30	70	**100**	4
3.	**DSC-9**	Genetic counselling	4	60	30	70	**100**	4
4.	**AEC-5**	Infertility & Assisted Reproductive Technology	2	30	--	50	**50**	**2**
5.	**AEC-6**	Scientific writing	2	30	--	50	**50**	**2**
6.	**Practical-7**	Molecular Diagnostics for rare diseases	2	60	100 marks practicals 50 marks viva		**150**	**2**
7.		Molecular Diagnostics for complex diseases	2	60				**2**
8.		Genetic Counselling	2	60				**2**
9.	**DSE-2**	Pharmacogenetics/ Nutrigenomics	2	30	--	50	**50**	**2**
10.	**OE-2**	Soft skills/Intellectual Property Rights	2	30	--	50	**50**	**2**
	Total Marks and Credits						**650**	**26**
SEMESTER 4								
	VBA-1	Industry internship	6	2 months			**150**	**6**
	VBA-2	Master's research thesis	18	4 months			**450**	**18**
	Total Marks and Credits						**600**	**24**

Question paper pattern for end-of-semester examinations of Discipline Specific Course conducted by JSSAHER – Theory (70 marks) is shown in Table 3

Table 3: Question paper pattern of Discipline-specific course

I	Long Answers	2 x 10 marks	= 20
II	Short Essay	7 x 5 marks	= 35
III	Short Answer	5 x 3 marks	= 15
	Total =		70 marks

Question paper pattern for end-of-semester examination of Discipline Specific Elective/Open Elective/Ability Enhancement/Value-based courses conducted at the Department level – Theory (50 marks) is shown in Table 4

Table 4: Question paper pattern of non-Discipline-specific course

I	Long Answers	1 x 10 marks	= 10
II	Short Essay	5 x 5 marks	= 25
III	Short Answer	5 x 3 marks	= 15
	Total =		50 marks

The minimum requirement for a PASS

- DSC papers: Students are declared to have passed in a subject if they secure a minimum of 40% in the end semester examination and an aggregate of 50% of marks in the JSSAHER examination and internal assessment added together. Theory & practicals shall be considered separately for evaluation. If a student passes in the practical examination but fails in a theory paper, such a student is exempted from reappearing for practical but shall have to appear in the subsequent examination for the theory paper in which the candidate has failed OR vice versa.
- AEC, DSE, and OE papers: The minimum prescribed marks for a pass shall be 35% of the maximum marks prescribed for a subject.
- Rank allocation – The students who pass all the semester examinations in the first attempts are eligible for ranks provided they secure at least a CGPA of 6.00. Ranks are not allowed for those students who clear their examination in parts.

The student will be eligible for securing a final degree only upon successful completion of all the subjects in the lower semester's examinations.

Grading of performances

- Based on the performances, each student shall be awarded a letter grade for each course. The letter grades and their corresponding grade points are given in Table 5.

Table 5: Grade description

Percentage of Marks obtained	Letter Grade	Grade Point	Result/grade description
90.00 – 100	O	10	Outstanding
80.00 – 89.99	A	9	Excellent
70.00 – 79.99	B	8	Good
60.00 – 69.99	C	7	Fair
50.00 – 59.99	D	6	Satisfactory
40.00 – 49.99	E	5	Average

Less than 40	F	Below 4	Fail
Absent	AB	0	Fail

Illustration of SGPA and CGPA (Tables 6 and 7)

The Semester Grade Point Average (SGPA)

The performance of a student in a semester is indicated by a number called Semester Grade Point Average (SGPA). The SGPA is the weighted average of the grade points obtained in all the courses by the student during the semester. The Credit Points (CP) shall then be calculated as the product of the grade points earned and the credits for the course. For example, if a student takes five courses (Theory/Practical) in a semester with credits C1, C2, C3, C4, and C5, and the student's grade points in these courses are G1, G2, G3, G4, and G5, respectively and then students' SGPA is equal to:

$$SGPA = \frac{C1G1 + C2G2 + C3G3 + C4G4 + C5G5}{C1 + C2 + C3 + C4 + C5}$$

The SGPA is calculated to two decimal points.

Cumulative Grade Point Average (CGPA)

The CGPA is calculated with the SGPA of all the 8 semesters for BSc Honors and 10 semesters for integrated MSc. CGPA is calculated up to two decimal points and is indicated in the final grade report card/final transcript showing the grades of all semesters and their courses. The CGPA shall reflect the failed status in case of F grade(s), till the course(s) is/are passed. When the course(s) is/are passed by obtaining a pass grade on subsequent examination(s) the CGPA shall only reflect the new grade and not the fail grades earned earlier. The CGPA is calculated as:

$$CGPA = \frac{C1S1 + C2S2 + C3S3 + C4S4}{C1 + C2 + C3 + C4}$$

where C1, C2, C3, & C4 is the total number of credits for semesters 1, 2, 3, & 4 and S1, S2, S3, & S4 is the SGPA of semesters 1, 2, 3, & 4

Table 6: An Illustration of Calculation of Semester Grade Point Average (SGPA): I Semester (Typical)

Courses/Papers	DSC1	DSC2	DSC3	AEC1	AEC2	P1	Total
Maximum Marks	100	100	100	50	50	150	550
Marks Obtained	77	74	62	38	39	108	398
% Marks Obtained	77	74	62	76	78	72	-
Grade Points Earned (G)	8	8	7	8	8	8	-
Letter grade	B	B	C	B	B	B	-
Credits for the Course (C)	4	4	4	2	2	6	22
Credit Points CP (G x C)	32	32	28	16	16	48	172

SGPA = Total CP / Total Credits = 172 / 22 = 7.81

Table 7: Calculation of Cumulative Grade Point Average (CGPA) for the Integrated Master's Degree: Illustration

Semester	1	2	3	4	Total
Maximum Marks	550	650	650	600	2,450

Marks Obtained	398	480	464	484	1,826
Semester GPA	7.81	7.38	7.14	8.06	30.39
Semester Credits (C)	22	26	26	24	98
Semester Credit Points (CP) (SGPA x C)	172	192	186	193	743

Cumulative Grade Point Average (CGPA) = Total of Semester CP / Total Credits for the program = 743 /98= **7.58**
Classification of Result: **First Class with Distinction**

Declaration of class
The class shall be awarded based on CGPA as follows:

- First Class with Distinction = CGPA of 7.50 and above
- First Class = CGPA of 6.00 to 7.49
- Second Class = CGPA of 5.00 to 5.99
- Pass Class = CGPA of 4.00 to 4.99

Carryover system

- A student can move on to the higher semester even if he/she fails the lower semester examinations. However, at the end of the final semester, the candidate should clear all the exams including lower semester examinations to be awarded a result/degree.

Award of Ranks/Medals

- Ranks and Medals shall be awarded on the basis of final CGPA. However, candidates who fail in one or more subject during the course shall not be eligible for award of ranks.

Revaluation and Re-totaling of answer papers

- There is no provision for revaluation of the answer papers in any examination. However, the candidates can apply for re-totalling by paying prescribed fee.

Maximum duration for completion of course

- A candidate shall complete MSc degree within four years from the date of admission, failing which candidate shall re-register for the course.

SEMESTER 1

DSC- 1 FUNDAMENTALS OF GENETICS & HUMAN CYTOGENETICS

(60 hours, LTP – 3+1+1)

Course Objectives

Students should be able to

- CO1: Understand the basics of genetics and basic features of cellular reproduction.
- CO2: Conceptualize classical Mendelian genetics and understand the exceptions.
- CO3: Perform Karyotype technique for human chromosomes
- CO4: Perform FISH technique.

Unit 1

The Science of Genetics **2 hrs**

- The personal genome, three great milestones in genetics, DNA as the genetic material, genetics and evolution, levels of genetic analysis, genetics in the world: applications of genetics to human endeavours.

Basic principles of inheritance **2 hrs**

- Mendel's study of heredity, applications of Mendel's principles, testing genetic hypotheses, mendelian principles in human genetics.

Extensions of Mendelism **4 hrs**

- Allelic variation and gene function, incomplete dominance and codominance, multiple alleles, allelic series, testing gene mutations for allelism, variation among the effects of mutations, genes function to produce polypeptides, why are some mutations dominant and others recessive?

Gene action: from genotype to phenotype **2 hrs**

- Influence of the environment, environmental effects on the expression of human genes, penetrance and expressivity, gene interactions, epistasis, pleiotropy.

Inbreeding: another look at pedigrees **1 hrs**

- The effects of inbreeding, genetic analysis of inbreeding, measuring genetic relationships.

Unit 2

The Chromosomal Basis of Mendelism **3 hrs**

- Sex, chromosomes, and genes, chromosome number, sex chromosomes, the chromosome theory of heredity, experimental evidence linking the inheritance of genes to chromosomes, nondisjunction as proof of the chromosome theory, the chromosomal basis of Mendel's principles of segregation and independent assortment.

Sex-Linked Genes in Humans **2 hrs**

- Hemophilia, an X-linked blood-clotting disorder, color blindness, an X-linked vision disorder, genes on the human y chromosome, genes on both the X and Y chromosomes.

Sex Chromosomes and Sex Determination **2 hrs**

- Sex determination in drosophila, sex determination in other animals, dosage compensation of X-linked genes, hyperactivation of X-linked genes in male drosophila, inactivation of X-linked genes in female mammals.

Pedigree analysis **1 hrs**

- Penetrance and Expressivity, Family Tree, Dominant Inheritance, Recessive Inheritance, Sex-Linked Inheritance.

Unit 3

Linkage, Crossing Over, and Chromosome Mapping in Eukaryotes **2 hrs**

- Early evidence for linkage and recombination, crossing over as the physical basis of recombination, evidence that crossing over causes recombination, chiasmata and the time of crossing over.

Chromosome Mapping **3 hrs**

- Crossing over as a measure of genetic distance, recombination mapping with a two-point testcross, recombination mapping with a three-point testcross, interference and the coefficient of coincidence, recombination frequency and genetic map distance.

Cytogenetic Mapping **3 hrs**

- Localizing genes using deletions and duplications, genetic distance and physical distance, linkage analysis in humans, detecting linkage with molecular markers.

Recombination and Evolution **2 hrs**

- Evolutionary significance of recombination, suppression of recombination by inversions.

Probability and Statistics **2 hrs**

- Probability, Types of Probabilities, Combining Probabilities, Use of Rules, Statistics, Hypothesis Testing, Chi-Square, Failing to Reject Hypotheses.

Unit 4 – Introduction to Cytogenetics and Clinical Cytogenetics **(5 Hrs)**

1. **Banding techniques:** History of human Cytogenetics, morphology of human chromosomes, (Q- banding, G- banding, C banding, silver staining for nucleolus organizer region (NOR), R-banding, sister chromatid exchange (SCE))
2. **Chromosome band nomenclature:** Identification and definition of chromosome landmarks, regions, bands and sub-bands, high resolution banding (HRB), Karyotyping and its importance.
3. **Chromosomal abnormalities:** deletion, duplication, insertions, translocations and inversions, Isochromosomes, ring chromosomes, Autosomal abnormalities, uniparental disomy.

Unit 5 – International System for Human Cytogenetic Nomenclature (ISCN) and Quality Assurance **(2Hrs)**

1. **Chromosomal nomenclature:** General principles, specification of breakpoints, symbols, designating structural chromosome aberrations by breakpoints and band composition.
2. **ISCN:** Interpretation and reporting of normal/abnormal reports

Unit 6 – Introduction to Cancer Cytogenetics **(15 Hrs)**

1. **Cancer diagnosis:** Analysis and interpretation of results, quality assurance
2. **Clones and clonal evolution**: definition of a clone, clone size, mainline, stemline, sideline, clonal evolution, composite karyotype, unrelated clones, modal number, constitutional karyotype.
3. **Chromosome markers in Lymphomas and leukemias**: (CML, AML, APML, ALL, CLL,

MPN, Multiple myeloma, Myelodysplastic syndromes etc.,) and solid tumors (Sarcomas and carcinomas).

4. **Introduction to Molecular Cytogenetics:** History of molecular cytogenetics, molecular techniques, advantages and applications in clinical Cytogenetics.
5. **Fluorescence in situ Hybridization (FISH):** Principles, procedure, labeling of DNA with Direct and Indirect methods.
6. **FISH probes:** Alpha satellite, telomeric, NOR specific, chromosome specific paint probes, unique sequence specific, repetitive sequence etc., applications in clinical diagnosis.

Unit 7 – Application of FISH in Prenatal Diagnosis and confirming micro deletion syndromes (6 Hrs)

1. **Diagnosis through FISH:** Prenatal and Postnatal diagnosis of microdeletion syndromes - Prader-Willi, Angelman, Williams, DiGeorge etc., using FISH probes
2. **Application of and types of probes in Cancer Diagnosis:** Principles and procedure, FISH probes for markers Leukemia and solid tumors. probes ,Tissue FISH, RNA in situ hybridization

Unit 8 – Advanced Molecular Cytogenetic Techniques (2 Hrs)

1. **Types of FISH and their applications:** Primed in situ labeling (PRINS), comparative genomic hybridization (CGH), Spectral karyotyping (SKY), multicolor FISH (mFISH) and multicolor banding (mBAND), Fiber FISH, Chromosomal Microarray (CMA) etc., using appropriate examples.

Practicals

1. **Basics of Microscopy –** Principle and types of microscopes, Handling and mounting
2. **GTG banding –** culturing, harvesting and chromosome preparation using peripheral blood lymphocytes (PBL)
3. **Karyotyping using karyotype workstation**–Identification of chromosomes under the microscope, use of software in capturing G-banded metaphases, chromosome analysis and karyotyping.
4. **Fluorescent insitu hybridization (FISH)**: Tissue/cell preparation, Probe Hybridization, Post hybridization and Analysis.
5. **Sister chromatid exchange (SCE)** – Culturing PBL with BrdU, Harvesting, slide preparation, staining and analysis.
6. **Quality Assurance**: Interpretation and Reporting of normal and abnormal reports using International System for Human Cytogenetic Nomenclature (ISCN 2020) for karyotyping and FISH.

Recommended books

1. S Gersen and MB Keagel (2013) The Principles of Clinical Cytogenetics, Springer Science
2. S Heim, F Mitelman (2015) Cancer Cytogenetics: Chromosomal and Molecular Genetic Aberrations of Tumor Cells, Fourth edition, John Willey and Sons Publications
3. Hollander A (Editor) 1971-76 Chemical mutagens: Principles and Methods of their detection. Vols. 1-3, Plenum Press New York.
4. Verma R. S. (Editor) 1988. Heterochromatin: Molecular and Structural aspects. Cambridge University Press. Cambridge.
5. International System for Human Cytogenetic Nomenclature (ISCN) 2020, Karger Publications

6. Marilyn S. Arsham (2017) The AGT Cytogenetics Laboratory Manual, Wiley-Blackwell
7. Principles of genetics, D. Peter Snustad, Michael J. Simmons.
8. Principles of Genetics, Tamarin.
9. Introduction to Genetic Analysis, Anthony J. F. Griffiths, John Doebley, Catherine Peichel, David A. Wassarman

SEMESTER 1

DSC-2 BIOCHEMISTRY AND METABOLIC DISORDER

(60 Hours, LTP – 3+1+2)

Course Objective

Students should be able to

- CO1: Understand the fundamental biochemical principles, such as the structure and function of biomolecules.
- CO2: Understand the basic concepts of enzymes, vitamins and minerals along with their role in metabolizing macromolecules and metabolic disorders.
- CO3: Estimate biochemical compounds
- CO4: Demonstrate usage of equipment's for analysing biochemical compounds

Amino Acids: Structure and Properties **2 hrs**

- Classification based on structure, Classification based on side chain characters, Classification based on metabolic fate, Classification based on nutritional requirement
- Properties of amino acid, Reactions of amino acid: Decarboxylation, Amide formation, Transamination, Transmethylation.
- Amino acid derivatives of importance, Peptide bond formation, Colour reactions of amino acids and proteins

Proteins: Structure and Function **3 hrs**

- Structure of proteins: primary, secondary, tertiary and quaternary structures, Primary structure of insulin.
- Structure-function relationship, protein structure determination,
- Physical properties of protein, iso-electric pH, Precipitation reactions of proteins, Denaturation of proteins.
- Classification of proteins: Based on function, composition & solubility, based on shape, based on nutritional value
- Biologically important peptides

Chemistry of Carbohydrates **2 hrs**

- Functions of carbohydrate, Classification of sugars, Stereoisomers
- Monosaccharides: Glucose, Fructose, Mannose, Galactose, , Epimers, Reactions of monosaccharides: Benedict's reaction, Osazone,
- Glycosides, amino sugars, Deoxy sugars.
- Disaccharides: Sucrose, Lactose, Maltose.
- Polysaccharides: Starch, Glycogen, Cellulose, heteroglycans, mucopolysaccharides

Enzymology **4 hrs**

- Classification of enzymes, Co-enzymes, Mode of action of enzymes,
- Michaelis menten theory, Michaelis constant, Km value, Vmax,
- Factors influencing enzyme activity,
- Enzyme activation, Enzyme inhibition: Competitive inhibition, Noncompetitive inhibition, uncompetitive inhibition, suicide inhibition,
- Allosteric inhibition, Key enzymes, Feedback inhibition, Covalent modification,Repression, Induction. Specificityo f enzymes, Iso-enzymes.

Chemistry of Lipids **3 hrs**

- Functions of lipids
- Classification of lipids, Classification of fatty acids
- Saturated fatty acids, Unsaturated fatty acids, trans fatty acids, Properties of fatty acids
- Neutral fats or triglycerols, properties of triacylglycerols, hydrolysis of triaclyglycerols,
- Saponification, iodine number, rancidity
- Phospholipids, Liposomes, Phosphotidly choline, cephalin, Phosphotidly inositol, Phosphotidlyglycerol, shingolipids, shingomyelin, non-phosphorylated lipids, cholesterol, prostaglandins

Vitamins **5 hrs**

- Fat Soluble Vitamins (A, D, E, K): Chemistry, biochemical function, sources, recommended daily allowance, absorption, and deficiency manifestation
- Water Soluble Vitamins (Thiamine, Riboflavin, Niacin, Pyridoxine, Pantothenic acid, Biotin,
- Folic acid, Vitamin B12 and Ascorbic acid): Chemistry, biochemical function, sources, recommended daily allowance, absorption, and deficiency manifestation

Biological Oxidation and Electron Transport **2 hrs**

- Primary, secondary and tertiary metabolism, Redox potential, Biological oxidation, Enzymes of biological oxidation, High energy compounds
- Organization of electron transport chain, NADH shuttle, Malate aspartate shuttle, Flow of electrons, Oxidative phosphorylation, Chemi-osmotic theory, ATP synthase, Inhibitors of ATP synthesis, Uncouplers of oxidative phosphorylation.

Metabolism

Carbohydrate metabolism **8 hrs**

- Stages of metabolism, types of metabolic pathways
- Digestion and absorption of carbohydrates and their clinical applications, glucose transporters.
- Embden-Meyerhof pathway- glycolysis, Cori's cycle, BPG shunt, Fate of pyruvate.
- Gluconeogenesis, Glucose-alanine cycle, Glycogenolysis, Glycogen synthesis, Glycogen storage diseases.
- Hexose monophosphate shunt pathway, Glucose-6-phosphate dehydrogenase deficiency
- Glucuronic acid pathway, Essential pentosuria, Polyol pathway
- Fructose metabolism, Hereditary fructose intolerance,Fructosuria
- Galactose metabolism, Galactosemia,
- Metabolism of alcohol, Amino sugars, Glycoproteins,
- Mucopolysaccharidoses, Inborn errors associated with carbohydrate metabolism.
- Regulation of blood sugar

Fatty Acids metabolism **6 hrs**

- Digestion and absorption of lipids and their clinical applications
- β-oxidation of fatty acids- even and odd-numbered saturated and unsaturated fatty acids, oxidation of odd chain fatty acid, Alpha oxidation, Omega oxidation, Organic acidurias.
- De novo synthesis of fatty acids
- Synthesis of triglycerides, Metabolism of adipose tissue, Liver adipose tissue axis

- Fatty liver, Lipotropic factors
- Ketone bodies, Ketogenesis, Ketolysis, Ketosis
- Structure and function of cholesterol, Biosynthesis of cholesterol, Plasma lipids- classification of lipoproteins, apolipoproteins
- Chylomicrons- metabolism and functions
- VLDL, LDL, HDL, Lp(a), Free fatty acid, Non-esterified fatty acids, Bile salts, Steroid hormones- metabolism
- Polyunsaturated fatty acids, Desaturation of fatty acids, Essential fatty acids, Eicosanoids-
- Prostanoids & Leukotrienes, Very long chain fatty acids, Synthesis of Compound Lipids, Phosphatidyl choline, Sphingomyelin, Lipid storage diseases.

Amino Acid Metabolism **6 hrs**

- Digestion of proteins and Absorption of amino acids and their clinical applications, Meister cycle, Intracellular protein degradation, Cathepsins, Ubiquitin pathway, Proteasomes, Inter-organ transport of amino acids, Glucose Alanine cycle;
- Formation of ammonia- Transamination, Oxidative deamination, Nonoxidative deamination
- Detoxification of ammonia
- Urea cycle, Disorders of urea cycle, Hepatic coma, Blood urea.
- Metabolism of Simple, Hydroxy and Sulfur Containing Amino Acid - Glycine, Serine, Methionine, Cysteine and their clinical significance
- Metabolism of Acidic, Basic and Branched Chain Amino Acids -Glutamic Acids, Aspartic Acid, Lysine, Arginine, Nitric Oxide, Histidine, Valine, Leucine, Isoleucine and their clinical significance
- Metabolism of Aromatic Amino Acids and Amino Acidurias- Phenylalanine, Tyrosine, Tryptophan, Proline and their clinical significance
- Citric Acid Cycle - Citric acid cycle reactions, significance, Amphibolic role, Regulation

Nucleotides Chemistry and Metabolism **4 Hrs**

- Composition of nucleotides: Purine bases, Pyrimidine bases, Nucleosides, Nucleotides
- Biosynthesis of purine nucleotides- Denovo and Salvage pathway, Regulation of synthesis,
- Degradation of purines, Uric acid, Gout, Secondary hyperuricemia,Lesch-Nyhan syndrome
- Synthesis of pyrimidine nucleotide, Regulation, Orotic aciduria, Deoxyribonucleotide formation, Degradation of pyrimidine.

Heme Synthesis and Breakdown **2 hrs**

- Structure of heme, Biosynthesis of heme, disorders of heme synthesis, Catabolism of heme,
- Hyperbilirubinemias- Congenital hyperbilirubinemia, Hemolytic jaundice, Hepatocellular jaundice, Obstructive jaundice

Hemoglobin **3 hrs**

- Structure of hemoglobin, Transport of gases,Oxygen dissociation curve, Hemoglobin interaction, Effect of 2,3-BPG, Isohydric transport of carbon dioxide, Chloride shift, Fetal hemoglobin, Hemoglobin derivatives, Carboxy hemoglobin, Met-hemoglobin,
- Hemoglobin variants- haemoglobinopathies: Sickle cell hemoglobin (HbS),HbE, HbC, HbD,
- Thalassemias- Beta and alpha thalassemia, Myoglobin, Anemias.

Free Radicals and Anti-Oxidants **1 hr**

- Free radicals, Reactive oxygen species, Generation, Damage, Free radical scavenger systems,
- Inflammation, Respiratory diseases, Retrolental fibroplasia, Reperfusion injury, Atherosclerosis, Skin
- diseases, Age-related diseases, Lipid peroxidation, Initiation, propagation and termination phases, Preventive anti-oxidants, Chain breaking anti-oxidants.

Plasma Proteins **1 hr**

- Serum electrophoretic pattern in normal and abnormal states
- Albumin- functions and clinical significance
- Hypergammaglobulins
- Transport proteins, Polymorphism
- Acute phase proteins- Ceruloplasmin, Alpha-1-anti-trypsin, Alpha-2- macroglobulin,
- Negative acute phase proteins
- Clotting factors, Anticoagulants, abnormalities in coagulation

Acid-Base Balance and pH **2 hrs**

- Acids and bases, Henderson-Hasselbalch equation, Buffers, Buffer capacity,
- Regulation of acid base balance-n Buffers of body fluids, Respiratory regulation of pH, Renal regulation of pH,
- Disturbances in acid base balance, Anion gap, Metabolic acidosis, Metabolic alkalosis, Respiratory acidosis, Respiratory alkalosis.

Electrolyte and Water Balance **2 hr**

- Body water compartments, Donnan membrane equilibrium, Osmolality, Electrolyte concentration of body fluid compartments
- Regulation of sodium and water balance, Renin-angiotensin system,
- Assessment, Disturbances, Isotonic contraction, Hypotonic contraction, Hypertonic contraction, Isotonic expansion,Hypotonic expansion, Hypertonic expansion
- Clinical applications- Hypernatremia, Hyponatremia, Hypokalemia, Hyperkalemia, Hyperchloremia, Hypochloremia.

Mineral Metabolism and Abnormalities **4 hrs**

- Sources, daily requirement, functions, regulations and clinical applications of Calcium, Phosphorus, Magnesium, Sulphur, Iron, Iodine, Zinc, Fluoride, Selenium, Manganese, Molybdenum, Cobalt, Nickel, Chromium, Lithium.

Practical's

- Estimation of blood glucose by glucose oxidase method (Single standard)
- Estimation of serum total protein by biuret method (Single standard)
- Estimation of blood uric acid by uricase method (Single standard)
- Estimation of blood urea by blood urea nitrogen (Single standard)
- Estimation of serum creatinine by Jaffe's method (Single standard)
- Estimation of serum total cholesterol by Zak's method (Single standard)
- Estimation of serum HDL by PTA method (Single standard)

- Estimation of serum bilirubin by modified Jendrassik and Grof's method (Single standard)
- Estimation of blood glucose using glucometer
- Estimation of reducing sugar, protein using dipstick method
- Demonstration of Autoanalysers.
- Demonstration and Interpretation of ABG
- Demonstration and Interpretation of HPLC pattern for haemoglobinopathies
- Demonstration and Interpretation of electrophoretic pattern of serum protein
- Demonstration and Interpretation of IFE
- Demonstration and Interpretation of chromatograms- carbohydrate and proteins

Recommended books - Recent edition

1. Textbook of Biochemistry -D.M.Vasudevan
2. Biochemistry - Pankaja Naik
3. Textbook of Biochemistry-RafiM.D
4. Clinical Biochemistry - Principles and Practice - Praful. B. Godkar
5. Textbook of Biochemistry - Chatterjea and Shinde
6. Harpers Biochemistry
7. Lippincott's Illustrated review of Biochemistry
8. Clinical Biochemistry-Michael L. Bishop
9. Practical Clinical Biochemistry-HaroldVarley

SEMESTER 1

DSC-3 CELL AND MOLECULAR BIOLOGY

(60 hours, LTP – 3+1+0)

Course Objectives

Students should be able to

- CO1: Understand fundamentals of cell structure and function of different organelles
- CO2: Conceptualize cell-cell interaction mechanisms underlying various cellular functions
- CO3: Perform hands-on mammalian cell culture techniques
- CO4: Perform molecular techniques involved in RNA isolation and real time PCR.

Unit 1: Fundamentals of cell biology **(10hr)**

- The cell theory- fundamental units of life, Eukaryotic and prokaryotic cells, structure and function of cell components- cell membrane and fluid mosaic model, cell wall, golgi apparatus, endoplasmic reticulum, nucleus, lysosomes, mitochondria **(4hr)**. Fluidity of membranes- membrane proteins, permeability of lipid bilayer protein-protein transport-passive diffusion, active transport, ATP driven pumps in transport, ABC transport system and sorting, pinocytosis, phagocytosis, receptor mediated endocytosis, Electron transport chain and proton pump **(6hr)**

Unit 2: Cell- cell communications **(20hr)**

- Signalling, paracrine, juxtacrine and endocrine factors, tight and Gap junctions, cadherins and cell adhesion, integrins and cell adhesion, morphogen gradients in development, cell surface receptors and internal receptors, signal transduction**(6hr)**
- Cell Signalling – G protein coupled cell surface receptors, enzyme couple cell surface receptors, signalling pathways dependent on latent gene regulatory proteins (Hedgehog and Wnt signalling) **(6hr)**
- Cytoskeletal filaments - microtubes and filament, centrosomes and centrioles, actin cytoskeleton, molecular motors. Eukaryotic cell cycle – overview of cell cycle, different phases, mitosis and meiosis and cell cycle regulation and apoptosis **(8hr)**

Unit 3: Fundamentals of molecular biology **(20hr)**

- Structure and function of genetic materials, DNA, RNA, Chromosome and its packaging as chromatin, organization and regulation of chromatin, Prokaryotic and Eukaryotic DNA replication, Conservative and semi-conservative DNA replication, DNA repair and recombination- Types of DNA repair- Mismatch repair, base excision repair, nucleotide excision repair, homologous and site specific recombination **(6hr)** Mitochondrial DNA and associated disorders. Central dogma of molecular biology- DNA to protein, transcription, post transcriptional modification, translation, post-translational modification, Differential gene expression – anatomy of gene- exons, introns, promoters, activation and repressor mechanisms of gene expression, generation of transgenic cell lines and organisms (**14hr)**

Unit 4: Techniques in molecular biology **(10hr)**

- Nucleic acids isolation, protein isolation and analysis, gene expression studies- PCR analysis, Basic principles of gene cloning- Vectors and bacteriophages introduction of DNA to living cells, cloning vectors for eukaryotes, application of gene cloning in medicine.

Practicals

1. Mammalian cell culturing (Thawing, passaging, trypsinization, cell counting and replating and freezing)
2. Transfection using lipofectamine in mammalian cells
3. Wound healing assays using cancerous cell lines
4. Cell cycle analysis using PI staining by flow cytometry
5. Immunofluorescence staining in mammalian fixed cells
6. MTT assay for cell proliferation studies
7. RNA isolation, cDNA preparation and PCR to analyse gene expression
8. Preparation of cell lysates from cancer cell line; estimation of total protein (Lowry/Bradford); analysis of protein of interest by western blotting.
9. Restriction digestion analysis of DNA using enzymes

Recommending reading:

1. Molecular Biology of the Cell: B. Alberts, A. Johnson, J. Lewis, M. Raff, K. Roberts and P. Walter (2007) 5 edition, Garland Science
2. Campbell Biology: Jane B. Reece, Lisa A. Urry et al, Tenth edition
3. Molecular Biology of the Gene: J.D. Watson, T.A. Baker, S.P. Bell, A.A.F. Gann, M. Levine and R.M. Losick (2007) 7 edition, Benjamin Cummings

SEMESTER 1

AEC 1 – Essentials in Immunology

(30 hours, LTP – 2+0+0)

Course Objectives

Student should be able to

- CO1: Understand the process and mechanisms underlying innate and adaptive immunity phagocytosis and natural killer cells in innate body defense.
- CO2: Describe the roles of different types of T cells, B cells and APCs and MHC in immune system

Unit I – Organs and cells of the Immune system (1 Hr)

- Primary lymphoid organs, Secondary lymphoid organs, B lymphocytes, T lymphocytes, mononuclear phagocytes, granulocytes, mast cells, dendritic cells, natural killer cells.

Unit II – Innate immunity (2 Hrs)

- Anatomical barriers, inflammation, anti-microbial peptides, acute phase proteins, toll like receptors

Unit III – B cell immunity (4 Hrs)

- Development of B cells, B cell activation, proliferation & differentiation, structure and Functions of Immunoglobulins

Unit IV – Complement system (2 Hrs)

- Components of the classical, alternate, lectin pathways, complement cascade, biological consequences of complement cascade, regulation of the complement system

Unit V – Antibody diversity (2 Hrs)

- Organization of immunoglobulin genes, mechanism of gene rearrangements, class switching, regulation of immunoglobulin genes

Unit VI – The major histocompatibility complex and antigen presentation (3 Hrs)

- General organization of the MHC genes, cellular expression of MHC genes, MHC and immune responsiveness, MHC antigens and antigen processing

Unit VII – T cell immunity (4 Hrs)

- Development of T cells and their education, T cell activation, proliferation & differentiation, T cell types, T cell receptor and co receptors, signaling in T cells, helper and cytotoxic T cells, cytokines

Unit VIII – Hypersensitivity (2 Hrs)

- IgE antibody-mediated hypersensitivity, antibody-mediated cytotoxicity, immune complex mediated hypersensitivity, delayed type hypersensitivity

Unit IX – Autoimmunity (2 Hrs)

- Organ specific autoimmune diseases, systemic autoimmune diseases, mechanisms of autoimmunity, treatment of autoimmune diseases

Unit XI – Immunodeficiency disorders **(2 Hrs)**

- Primary immunodeficiency diseases of lymphoid cells, myeloid cells and granulocytes, complement components, acquired immunodeficiency diseases

Unit XII – Cancer and immune system **(2 Hr)**

- Tumor evasion of the immune system, cancer immunotherapy

Unit XIII – Vaccines **(2 Hrs)**

- Active and passive immunization, vaccine designs, subunit vaccines, DNA vaccines

Recommended books

1. T J Kindt, B A Osborne, R Goldsby (2006) Kuby Immunology, W H Freeman publication, 6th Edition
2. W E Paul (2003) Fundamental Immunology, Lipincott Williams and Wilkins publication, 4th Edition
3. P J Delves, S J Martin, D R Burton, I M Roitt (2017) Roitt's Essential Immunology, Wiley Blackwell publication, 13th edition.

SEMESTER 1

AEC-2 Use of model organisms to study human diseases

(30 hours, LTP – 2+0+0)

Learning objectives:

- The student will be able to Understand and explain the use and value of model organisms such as Planaria, Drosophila, C. elegans, and Zebrafish in the analysis and elucidation of biological principles and describe the hierarchy of testing models.
- Basic Master culturing, genetic manipulation, and phenotypic analysis techniques in handling genetic model organisms. This course is aimed at an interactive learning experience for the students and will consist of seminars and discussions led by invited speakers who use model organisms in their research. Students will also make presentations and write essays defending a particular choice of a model organism.

Unit I – Introduction **(2 hours)**

Use of model organisms to study human diseases, History, Characteristic of ideal model organism, different Applications of model organisms in the research field, about genomes of model organisms (Planaria, C. elegans, Drosophila melanogaster, Zebrafish, laboratory mouse,), genomes & genetic variation, comparison of different genomes. Advantages, disadvantages & Limitations of model organisms in the research field.

Unit 2 – Planaria as a model organism **(5 hours)**

Introduction to Planaria – Background, Overview of common Brown Planaria (*Dugesia sp*)- Anatomy, Life cycle, ease of culture & economy. Advantages & Limitations. As a system for Regenerative studies- Stem Cell & Neoblasts Cell function and the complexity of tissue regeneration, wound healing.

Unit 3 – *Caenorhabditis elegans* as model organism **(10 hours)**

Introduction to C. elegans – Background, Overview of C. elegans - Anatomy, Life cycle, Nematode handling, and husbandry, economy, Advantages & Limitations; C. elegans as a system to study human diseases – ease of performing cellular and genetic studies, conservation in cellular pathways; use of the system for RNA interference studies – gene silencing by double-stranded RNA, used to identify genes that switch on cell death in cancer cells (such as leukemia), facilitate cellular differentiation studies, drug screening.

Unit 4 – Drosophila as a model organism **(10 hours)**

Introduction to Drosophila – Background, Overview of Drosophila - Anatomy, Drosophila husbandry, Life cycle, ease of performing cellular and genetic studies, Advantages & Limitations of Drosophila as a model organism for genetic analysis; Generation of Transgenic Drosophila – formidable range of genetic tools available, conservation of cellular pathways and cellular mechanisms; Germ-line transformation and selection of vectors, Application of P-element based vectors in a transgenic generation; Drosophila model for human genetic disorders like Genetic control of early embryonic development, neurodegenerative disorders (e.g. Parkinson's, Huntington's, Alzheimer's diseases, etc.), triplet repeat expansion diseases, Fragile X syndrome, use of the system in drug screening.

Unit 5 – Zebrafish as a model organism **(8 hours)**

Introduction to Zebrafish – Background, Overview of Zebrafish - Anatomy, Life cycle, ease of culture & economy. Overall advantages and disadvantages of the zebra fish D. rerio as a system to study human diseases, performing cellular and genetic studies, formidable range of

genetic tools available, conservation of cellular pathways and cellular mechanisms; advantages of vertebrate models, similarity in organ systems across different vertebrates; using vertebrate orthologous genetic mutant models to study hematological diseases such as sideroblastic anemia, polycythemia, and porphyria; T-cell leukemia models, Melanomas, heart defects resembling human dilated cardiomyopathies (DCMs), modeling Duchenne muscular dystrophy, Polycystic kidney disease (PKD), etc.; use in drug screening.

Unit 6 –Laboratory mouse as a model organism (10 hours)

Introduction – Background, Overview of Mouse model - Anatomy, Life cycle & economy, advantages and disadvantages of using mammalian systems, similarity in physiology and organ function with humans, high genetic conservation; use as models to study – diseases that affect cellular pathways and cellular mechanisms, metabolic disorders, and diabetes, tumor formation, Studying cellular mouse models containing transplanted human cells or the human orthologues of specific genes; modeling cancers; genetic disorders such as hearing-loss disorders, and memory; uses in drug testing and treatment of early onset cancers such as acute promyelocytic leukemia (APL), the role of the protein Leptin in controlling obesity; ethical issues concerning the use of mice, and primates.

Unit 7 – Techniques to generate transgenic animals (15 hours)

mutagenesis – X-ray and chemical mutagenesis, insertional mutagenesis, transposon mutagenesis, site-direct mutagenesis, and P-element in drosophila; Mitotic recombination, generation, and analysis of somatic clones, generation of germline clones, conditional and/or targeted expression/ ablation of genes/transcripts (e.g. UAS/GAL4 system); Construction of a transgene, introduction of a foreign gene into the animal model – pronuclear microinjection methods, embryonic stem cell transformation; RNA interference-based screening of gene function & mechanism, knock-in & knockout models, CRISPR/CAS-9 techniques, Genetic mosaic techniques – FLP-FRT recombination, Cre-Lox recombination.

Recommended Books

1. Developmental Biology by Gilbert S., F. Sinauer
2. The Biological Resources of Model Organisms by Robert L. Jarret, Kevin McCluskey
3. Molecular and cellular aspects of planarian regeneration, cell & developmental biology (Vol. 10, No. 4, pp. 377-383).

SEMESTER 2

DSC-4 Biostatistics & Bioinformatics

(60 hours, LTP – 3+1+2)

Part A – Biostatistics

Course Objectives

Students should be able to:

- CO1: Recognize the importance of data collection and its role in determining scope of inference. Interpret statistical results correctly.
- CO2: Understand and critique data based claims, power of data and other basic terminologies
- CO3: Apply appropriate statistical methods for analyzing one or two variables
- CO4: Use tools to perform descriptive and inferential data analysis for one or two variables.

Syllabus:

Unit – I: **2 Hours**

Introduction: Introduction to Biostatistics; levels of measurement – nominal, ordinal, interval and ratio scales; Types of Data- quantitative and qualitative

Unit –II: **2 Hours**

Descriptive statistics – central tendency, dispersion, skewness and kurtosis.

Unit – II: **3 Hours**

Sampling: Probability and non-probability; simple random, stratified, systematic, cluster and multistage sampling; sampling and non – sampling errors

Unit III: **3 Hours**

Sample size estimation: Sample size determination for estimation : sample size determination for estimation of mean, estimation of proportion, comparing two means and comparing two proportions.

Unit – IV: **5 Hours**

Hypothesis testing: formulation and types; null hypothesis, alternate hypothesis, type I and type II errors, level of significance, power of the test, p –value , concept of standard error and confidence interval . Concept of Probability “probability distribution – normal, poisson, binomial

Unit – V: **3 Hours**

Epidemiological studies: Rates – Prevalence and incidence; types – Prospective and retrospective studies; Diagnostic Efficiency Statistics (Sensitivity, specificity, predictive values); Risk Estimation – odds ratio and survival analysis.

Unit – V: **4 Hours**

Tests of significance – Parametric tests: requirements, “t” test, normal z – test , and “F’ test including post – hoc tests, one – way and two-way analysis of variance, analysis of covariance, repeated measures analysis of variance, simple linear correlation and regression.

Unit – VI: **3 Hours**

Test of significance – Non – parametric tests: Assumptions; one – sample tests (sign test, McNemar test); two – sample test (Mann whitney U test, Wilcoxon rank sum test); k –sample tests (Kruskal wallies test, and Friedman test) and chi-square test.

Unit – VII: **5 Hours**

Multivariate analysis: Introduction, Multiple regression, logistic regression, factor analysis, cluster analysis,

Recommended books:

1. B.L (2007). Qualitative Research: Methods for the social sciences (6th ed.) New york: Pearson education.
2. Daniel, W.W. (2005). Biostatistics: a foundation for analysis in health sciences (8th ed.) New York: John wiley and Sons.
3. Dillon, W.R. & Goldstein, M. (1984). Multivariate analysis: Methods & Applications. New York: John Wiley & Sons.
4. Hassart, T.H (1991). Understanding Biostatistics. ST. Louis: Mosby year Book.
5. Kerlinger, F.N. (1995). Foundations of Behavioral research. New York: Holt Rineheart & Winston.
6. Kothari, C.R. (2003) Research Methodology. New Delhi: Wishwa Prakshna.
7. Siegal, S. & castellan, N.J (1988). Non – parametric statistics for the behavioral sciences. McGraw Hill: New Delhi

Part B – Bioinformatics

Course objectives

Students should be able to

- CO1: Understand various applications in the field of Bioinformatics in Genetics and Genomics.
- CO2: Learn about different databases used for the retrieval of information on several biologically significant biomolecules such as DNA, RNA and proteins.
- CO3: Apply Bioinformatics tools and advanced methods relevant to the execution of genome, transcriptome and phylogenetic analysis.

Unit I – Basics in Bioinformatics

Introduction to Bioinformatics **(3 hrs)**

- Definition, history and application of Bioinformatics; Genomic research: Genome projects, an overview of activities in bioinformatics with emphasis on the types of information in modern biology and the need for (computational biology and bioinformatics) databases and software; Contribution of Bioinformatics in Genomics; commonly used computer programming languages in Bioinformatics

Unit II – Databases used in Bioinformatics

Commonly used Databases **(4 hrs)**

- General concepts and organization of biological databases; an overview of database types; common databases and their utilities (NCBI, EBI, ExPasy, Entrez, ENSEMBL, SRS system,

Genecards); Sequence databases (NCBI: Gene, Nucleotide, Genbank, dbEST, STS, UniGene); using DNA sequence databases for PCR primer designing and restriction site analysis (NEBcutter, Primer3plus, Primer Blast); Primary Protein structure databases (SwissProt/Uniprot, EMBL, PIR, PDB, HPRD, KEGG, etc.); Secondary (Derived) Protein Databases (Prosite, Pfam, SCOP, CATH, DSSP, FSSP, RNAbase); Information resources – Pubmed, OMIM; Significance of data analysis at various levels: genome vs. exome vs transcriptome and proteome.

Unit III – Similarity and Sequence Alignment

Similarity Searching and Sequence Alignment **(5 hrs)**

- common methods used in sequence analysis and alignment; basic concepts of sequence similarity (similarity, identity, homology, definitions of homologues, orthologues and paralogs, scoring, Gap cost, Linear and affine Gap Penalty); basics of scoring system and matrices (PAM, BIOSUM, GONNET, ClustalW and ClustalX); pair-wise sequence alignment: Brute Force method, Dot matrix method; Global Alignment (Needleman- Wunsch); Local Alignment (Smith-Waterman) using Dynamic programming; BLAST and its variants, PSI-BLAST, MSA, comparison of operation and applications of BLAST and MSA, and interpretation of results.

Unit IV – Sequence Analysis

Basic concepts **(2 hrs)**

- importance of sequence analysis; sequence file formats: fasta, genbank, embl, Swiss-prot, pdb, nbrf, pir and multiple sequences formats (Aln, Mega, Pileup, Phylip etc.); common databases used to retrieve gene, mRNA and protein sequences; concepts in sequence pattern and profiles (DNA and RNA motif analysis, relevant databases and softwares).

Human genome analysis **(3 hrs)**

- genetic variations - types and their significance; understanding the relationship between mutations, SNPs, insertions or deletions (indels), copy number variations (CNVs) and alleles; SNP and other variant databases and analysis of reference and sample sequences using databases (dbSNP, Clinvar, dbVar, and Cosmic); methods for detection of genetic variations: PCR-RFLP, Sanger Sequencing, Genome wide association studies (GWAS), Next-Generation Sequencing (NGS) approaches; genome versus exome analysis; concept of metagenomics: types of metagenomics approaches and their relative significances; significance of microbiomes to human health; case studies.

Transcriptome analysis **(4 hrs)**

- databases used for obtaining transcriptomic data; microarray technology - introduction, principle, applications, microarray databases and data analysis; discussions on RNA-seq analysis; miRNA analysis; ChIP-seq technology and related data analysis; DNA motif analysis.

Bioinformatics for Proteins **(3 hrs)**

- review of protein structures and domains; techniques and databases for protein-structures, gene ontologies, protein-interactions and pathways; domain analysis; significance of interaction analysis and systems biology; concepts in Structural Bioinformatics – protein structure visualization tools (RasMol, Pymol), homology modelling, drug discovery, docking and drug design.

Computational Methods for Phylogenetic analysis **(4 hrs)**

- phylogenetic analysis with reference to nucleic acids and proteins, and its significance; Phylogenetics prediction methods: Basics, molecular clock, Substitution Models of evolution, Tree reconstruction methods (Distance based, character based method, statistical), Bootstrapping; software and Programmes for comparative genome analysis (sequence comparison); phylogenetic analysis software; molecular structure drawing tool.

Unit V – Applications of Bioinformatics

Applications **(2 hrs)**

- applications of Computational Biology/Bioinformatics in Biotechnology, Molecular Biology, human health, Neurobiology, drug designing, Environment Sciences, Agricultural and Veterinary Sciences

Practical:

1. Retrieval of nucleotide and protein sequences from suitable databanks
2. Online tools for similarity search between nucleotide and protein sequences
3. Online tools for PCR primer designing
4. Online tool for designing RFLP study protocol
5. Tools for finding conserved sequences
6. ORF finding using bioinformatics tools
7. Tools for visualization of genomic sequences
8. Tools for visualization of protein and nucleic acid structure
9. Linux and its commands used in Bioinformatics analysis

Recommended books:

1. N. Gautham (2006) Bioinformatics: Databases and Algorithms; Alpha Science.
2. J. Bedell, I. Korf and M. Yandell (2003) BLAST; O'Reilly Press.
3. J. M. Keith (2008) Bioinformatics Vol. 1, Data, sequence analysis & evolution; Humana Press.
4. R. Durbin (1998) Biological sequence analysis; Cambridge University Press.
5. David, W Mount (2004) Bioinformatics: Sequence and genome analysis, Cold Spring Harbour Press.
6. R. M. Holmes (2007) A cell biologists' guide to modelling and bioinformatics; Wiley Interscience.
7. Bryan Bergeron, Bioinformatics Computing, Publisher: Prentice Hall PTR.
8. A D Baxevanis and B.F. Francis Ouellette (2002) Bioinformatics a practical guide to analysis of genes and protein, Wiley Interscience.
9. Primrose SB, Twyman RM, (2002) Principles of Genome analysis and genomics, Blackwell Science.
10. M.R. Barnes, I.C. Gray (2002) Bioinformatics for Geneticists, John Wiley & Sons, New York
11. Rastogi SC, Mendiratta N, Rastogi P (2016) Bioinformatics Concepts Skills and Application 2nd Ed., CBS Publishers and Distributors, New Delhi
12. Dov Stekel (2003) Microarray Bioinformatics, Cambridge University Press
13. Knudsen S. (2004) Analysis of DNA Microarray data, 2nd Ed., John Wiley & Sons, New York

14. Baldi P. and Brunak S. Bioinformatics (2001) The Machine Learning Approach, The MIT Press, USA
15. Des Higgins and Willie Taylor, Bioinformatics: Sequence, structure and databanks, Oxford University Press

Databases:

1. NCBI Genome Browser and databases: http://www.ncbi.nlm.nih.gov/
2. UCSC Genome Browser: http://genome.ucsc.edu/
3. Ensemble Genome Browser: http://www.ncbi.nlm.nih.gov/
4. Protein Catalogue ExPASy: http://www.expasy.org/
5. Protein Catalogue - Uniprot: http://www.uniprot.org/; RCSB PDB: https://www.rcsb.org/
6. Online course: ExPASy: http://www.expasy.org/
7. Web portal of multiple sources: wws.startbioinfo.com

SEMESTER 2

DSC-5 HUMAN EVOLUTIONARY & POPULATION GENETICS

(60 hours, LTP – 3+1+2)

Course objectives

Student should be able to

- CO1: Understand the evolutionary concepts and analytical tools that are used to interpret diversity.
- CO2: To demonstrates the wider applications of an evolutionary approach for our understanding of phenotypic variation, the genetics of diseases both simple and complex, and the identification of individuals.
- CO3: Analyse and interpret results from different studies in the evolution
- CO4: Calculate allelic and genotypic frequencies in a population

Unit 1

An Introduction to Human Evolutionary Genetics **1 hrs**

- Understanding evolutionary history to understand human biology, an understanding of evolutionary history that shapes our expectations about the future, and understanding the chronology of events from different scientific approaches. The ethics of studying human populations.

Human Genome Variations **3 hrs**

- Single Nucleotide Polymorphisms (SNPs) in the Nuclear Genome, Sequence Variation in Mitochondrial DNA**,** Variation in Tandemly Repeated DNA Sequences, Transposable Element Insertions, Structural Variation in The Genome, The Effects of Age and Sex on Mutation Rate, The Effects of Recombination on Genome Variation

Unit 2

Finding and Assaying Genome Diversity **4 hrs**

- Find your DNA, The Polymerase Chain Reaction (PCR), Sanger Sequencing, the Human Reference Sequence, and SNP Discovery, Next-Generation Sequencing, SNP Typing: Low -, Medium-, and High -Throughput Methods for Assaying Variation, Databases of Sequence Variation, Discovering and Assaying Variation at Microsatellites, Discovering and Assaying Structural Variation on Different Scales, methods to determine haplotypes from genotypes.

Making Inferences from Diversity **3 hrs**

- Measuring Genetic Distance, Phylogenetics, Coalescent Approaches to Reconstructing Population History, Dating Evolutionary Events Using Genetic Data, Effect of selection.

Unit 3

What Genetic Changes Have Made Us Human **3 hrs**

- Morphological and Behavioral Changes En Route to Homo Sapiens, Genetic Uniqueness of Humans and Hominins, Genetic Basis of Phenotypic Differences Between Apes and Humans

Origin of Modern Humans **4 hrs**

- Evidence from Fossils and Morphology, Evidence from Archaeology and Linguistics hypotheses to Explain The Origin of Modern Humans, Evidence from the Genetics of Present-Day Populations, Evidence from Ancient DNA.

Unit 4

Understanding the Past, Present, and Future of Phenotypic Variation **2 hrs**

- Normal and Pathogenic Variation in an Evolutionary Context, Known Variation in Human Phenotypes, Skin Pigmentation as an Adaptation to Ultraviolet Light, Life at High Altitude and Adaptation to Hypoxia, Variation in the Sense of Taste, Adapting to a Changing Diet by Digesting Milk and Starch, The Future of Human Evolution

Evolutionary Insights into Simple Genetics Diseases **3 hrs**

- Genetic Disease and Mutation–Selection Balance, Genetic Drift, Founder Effects, and Consanguinity, Evolutionary Causes of Genomic Disorders, Genetic Diseases and Selection by Malaria

Evolution and Complex Diseases **3 hrs**

- Defining Complex Disease, The Global Distribution of Complex Diseases, Identifying Alleles Involved in Complex Disease, What Complex Disease Alleles do we Expect to Find in the Population? Genetic Influence on Variable Response to Drugs

Identity and Identification **2 hrs**

- Individual Identification, What DNA Can Tell Us About John or Jane Doe, Deducing Family and Genealogical Relationships, The Personal Genomics Revolution

Haplogroup Nomenclature **2 hrs**

- The Mitochondrial Genome, The Y Chromosome

Unit 5

Genetic, and anthropological background **3 hrs**

- The scope of population genetics, Genetics background, Mendel's laws, Alleles, genotypes and phenotypes, how do we assess human genetic diversity? the anthropological connection, what is a population? Anthropology and population genetics, a short history of population genetics.

Hardy–Weinberg equilibrium **7 hrs**

- Genotype and allele frequencies, computing genotype frequencies, computing allele frequencies, what is Hardy–Weinberg equilibrium, The mathematics of Hardy–Weinberg equilibrium, what does equilibrium mean? Assumptions of Hardy-Weinberg equilibrium, using Hardy–Weinberg equilibrium, detecting deviations from Hardy-Weinberg equilibrium, Hardy–Weinberg equilibrium and dominant alleles, extensions of Hardy–Weinberg equilibrium, Linkage disequilibrium, more than two alleles, X-linked genes, Hardy–Weinberg equilibrium and evolution

Unit 6

Inbreeding **3 hrs**

- Quantifying inbreeding, genealogies and inbreeding, types of inbreeding, the inbreeding coefficient, mean inbreeding, Population genetics and inbreeding, the impact of inbreeding on genotype frequencies, why inbreeding does not change allele frequencies, the medical impact of inbreeding, inbreeding in human populations, rates of inbreeding in human population, examples of inbreeding studies using genealogical data, surname analysis, potential-mates analysis,

Mutation **2 hrs**

- The nature & types of mutations, evolutionary impact of mutation, rates of mutation, Models of mutation, simple mutation model, reverse mutation, number of new mutations in a generation, fate of mutant alleles, Mutational history and anthropological questions, mutation and haplogroup trees.

Genetic drift **4 hrs**

- What is genetic drift? Genetic sampling, simulation of genetic drift, outcome of genetic drift, Genetic drift and population size, how does population size affect genetic drift, effective population size, Effects on genetic variation, measuring genetic variation, decay of genetic variation over time, Mutation and genetic drift, fate of a mutant allele, equilibrium between mutation and genetic drift.

Unit 7

Models of natural selection **4 hrs**

- How does natural selection work? Absolute and relative fitness, simulation of natural selection, A general model of natural selection, Types of natural selection, selection against the recessive homozygote, selection against dominant alleles, selection with codominant alleles, selection against heterozygote, selection for the heterozygote, other aspects of selection, selection and mutation, selection and genetic drift, selection and inbreeding, natural selection and quantitative traits.

Natural selection in human populations **4 hrs**

- Case studies of natural selection in human populations, Hemoglobin S and malaria, Duffy blood group and malaria, CCR5-Δ32 allele and disease resistance, lactase persistence and the evolution of human diet, genetic adaptation to high-altitude population, evolution of human skin color, are humans still evolving? How do we detect recent selection, the future.

Unit 8

Gene flow **3 hrs**

- The evolutionary impact of gene flow, introducing new alleles, reducing genetic differences between population, Models of gene flow, the island model, Two-way gene flow, Kin-structured migration, Gene flow and genetic drift, measuring genetic variation between populations, equilibrium between gene flow and genetic drift, isolation by distance, migration matrix analysis.

Practicals

- Construct a Phylogenetic tree for the given specimens according to their evolutionary pattern.
- Perform the Clustal W analysis and comment on the divergence pattern.
- Essential bioinformatic tool to study the rate of human evolution.
- Analyze and interpret results from different studies in the evolution.
- Calculate the allele frequencies from the observed population genotype numbers.
- Calculate the genotype frequencies from the observed genotype numbers.
- Apply the Hardy-Weinberg principle to calculate the expected genotype frequencies from the allele frequencies in the population.
- Use Chi-Square test to determine if the observed and expected genotype frequencies are significantly different from each other or not.
- Problems with models of mutation – calculating the expected frequencies of any allele in

generation *t.*

- To study the influence of genetic drift in a population using a Bioinformatics tool (PopG)

Recommended books

1. Human Evolutionary Genetics by Mark Jobling, Edward Hollox, Toomas Kivisild, Chris Tyler-Smith.
2. Human Gene Evolution by David N. Cooper.
3. Theoretical Evolutionary Genetics by Joseph Felsenstein.
4. Human Evolutionary Genetics by Isaac Jenkins.
5. Human Population Genetics John H. Relethford John Wiley & Sons, Inc., Publication.
6. The Genetics of Human Populations by Luigi Luca Cavalli-Sforza, Walter Fred Bodmer.
7. Principles of Population Genetics (Daniel L. Hartl, Andrew G. Clark)
8. Population Genetics (Matthew Hamilton)

SEMESTER 2

DSC-6 HUMAN MOLECULAR GENETICS

(60 hours, LTP – 3+1+2)

Course objectives

Student should be able to

- CO1: Understand fundamental concepts to analyse human genome and genetic manipulation
- CO2: analyse and interpret DNA sequences through molecular techniques from PCR to Sanger sequencing
- CO3: Understand genes and genetic variation involved in monogenic and complex disorders.

Unit-1: Basic principles of nucleic acid and gene expression and analysis (10 hours)

Introduction to central dogma of molecular biology

The process of human genome sequencing- generation of genomic DNA library, strategies for genome sequencing- whole-genome shotgun and hierarchical shotgun sequencing, RFLP, microsatellite DNA polymorphism, high and low density DNA markers for genome mapping, mapping with somatic cell hybrid panels, construction of clone contigs by STS content mapping, DNA cloning using YAC and BAC, the basic method of gene expression analysis-RT-PCR, sequence assembly in complex genomes, high-throughput gene expression analyses-microarrays, profiling global protein expression using mass spectrometry, protein separation and annotation.

Unit-2: Principles of genetic manipulation of mammalian cells (15 hours)

Construction of immortalized cell lines and immortalized euploid cell lines, the concept of genome editing using homologous recombination, site-specific recombination, role of site-specific endonucleases-Zinc finger nucleases, TALENs (transcription activator-like effector nucleases)and its role in gene therapy, CRISPR-Cas system, gene silencing using RNAi, RISC, RITS, siRNA and methods of inducing RNAi into animal cells, RNAi therapy for treating genetic disorders.

Principles of transgene expression in mammalian cells-tetracyclin-regulated expression, tamoxifen-regulated expression, transgenic animals- pronuclear microinjection method of germ-line transgenesis.

Unit-3: Genome architecture (15 hours)

Characteristics of human nuclear and mitochondrial genome, replication and transmission of mtDNA, limited autonomy of mitochondrial DNA, variant mitochondrial genetic code, transfer of mtDNA sequences into nuclear genome, human nuclear genome and heterochromatic DNA, mtDNA disorders, the concept of short and long noncoding RNA, heterochromatin DNA transcription, the concept of retrotransposons and its types- LINEs, SINEs, LTR, SVA repeats, Alu repeats, the concept of transposon repeats.

Unit-4: Mapping and identifying genes for monogenic disorders and complex disorders (20 hours)

Method of positional cloning by mapping to identify disease genes- recombinants identification by genotyping parents, recognizing recombinants in human pedigrees, mapping human disease using genetic markers, LOD-score calculation and analysis, haplotype sharing and autozygosity- autozygosity mapping using SNP arrays, Whole-exome and whole-genome sequencing to identify the cause of the monogenic condition, strategies for exome-based disease gene identification.

Multifactorial Traits or Complex Disorders

Polygenic theory of quantitative traits; partitioning of variance, heritability; polygenic theory of discontinuous characters; genetic component assessment in families: risk-ratio, twin and adoption studies; mapping of complex traits by parametric linkage analysis in near-Mendelian families and affected sib-pair analysis; non-parametric linkage analysis: association-mapping studies: role of linkage disequilibrium in association studies, odds ratio and chi2 test, tag-SNPs, genome-wide association studies (GWAS), Transmission disequilibrium test (TDT); identifying susceptibility variation through association studies, common disease-common variant hypothesis and mutation selection hypothesis.

Practicals:

1. Isolation and Estimation of DNA: Isolation of DNA from human cells; DNA quantification and agarose gel electrophoresis.
2. Primer design: Guidelines for primer designing; tools for designing primers- NCBI primer BLAST and MFEprimer; Primer design for genes to be used for PCR and sequencing.
3. Polymerase chain reaction (PCR): Amplification of DNA sequences from genomic DNA/ plasmid DNA/cDNA; optimization of conditions, eg. MgCl2 concentration and annealing temperature.
4. Types of PCRs and its multiple applications; Gradient PCR, Multiplex PCR, ARMS PCR, DNA gel electrophoresis of PCR amplified products
5. Purification of PCR amplified products.
6. Sanger sequencing of gene exons for mutation detection.
7. Molecular cloning: Preparation of competent cells, transformation of E coli DH5α, preparation of plasmid DNA; restriction enzyme digestion, analysis of products through agarose gel electrophoresis; gel elution of restriction enzyme digested fragments; ligation, transformation and analysis of clones.

Recommended books:

1. Human Molecular Genetics by Tom Strachan, Andrew Read, 5th edition.
2. Brown T. A. 2007, Genomes 3. Garland Science Publishing, New York.
3. Dunham, I., 2003. Genome Mapping and sequencing. Horizon Scientific

SEMESTER 2

DSE-1a Proteomics

(30 hours, LTP – 2+0+0)

Learning objectives

- The student will be able to understand the basic concept of proteomics
- The student will be able to describe the technique used to study proteomics
- The student will be able to understand the applications of proteomics in diagnosis and therapy

Unit I **(7 hours)**

Public protein databases and interfaces, Protein structural and functional databases Uniprot, Swiss Prot and String Bioinformatics and proteomic technologies, Placing proteins in pathways using Reactome, Ab initio protein structure prediction:

Unit II **(8 hours)**

Proteomic technologies Protein Microarrays, Construction, applications. Advantages and limitations. Transcriptomes and analysis; SAGE, DNA Microarray technology; Next Generation Sequencing. Analytical proteomics tools (1-D & 2-D gel electrophoresis); Mass spectrometry and analysis (ESI, MALDI and Hybrid), LC/MS-MS; Applications of mass spectrometry (PMF and PTMs)

Unit III **(8 hours)**

Computational methods, Structure prediction from sequence, Deriving function from sequence, Application of structural proteomics, Merits and demerits of structural proteomics techniques. Comparison of various technologies used in structural proteomics.

Unit IV **(7 hours)**

Interactomes and Proteomic interactions (Y2H approaches, Co-IP); Proteome- wide interaction maps; Protein structure determinations and Structural proteomics tools (experimental and computational); Concepts of protein engineering.

Recommended books

1. Twyman, R.M. (2004) Principles of Proteomics. Bios Scientific Publisher, Oxford.
2. Kraj, A. & Silberring J. (2008) Introduction to Proteomics. Ed. Wiley, UK.
3. Lovrik, J. (2011) Introducing Proteomics: From concepts to sample separation, mass spectroetry and data analysis. Ed. Wiley-Blackwell, UK.
4. D.C. Liebler, (2002) Introduction to Proteomics: Tools for the New Biology, Humana Press.
5. R.M. Twyman, (2004) Principles of Proteomics, Bios Scientific Pub.
6. Timothy D. Veenstra, John R.Yates III (2006)Proteomics for Biological Discovery, John-Wiley & Sons, Hoboken, New Jersey, USA.
7. R. Hubert, (2006) Protein Biochemistry and Proteomics (The Experimenter Series), Academic Press.
8. Reiner Westermeier, Tom Naven, Hans Rudolf Höpker (2008) Proteomics in Practice: A Guide to Successful Experimental Design, Wiley-Blackwell.
9. N Saraswathy, P Ramalingam (2011) Concepts and Techniques in Genomics and Proteomics. E Book
10. Naveen Mistra (2010)Introduction to proteomics E Book

SEMESTER 2

DSE-1b Microbial Genetics

(30 hours, LTP – 2+0+0)

Learning objectives

- To understand the DNA metabolism, DNA manipulation techniques and its applications in molecular biology.
- To understand the interaction of microorganisms with the environment and mechanisms of bacteria in establishing the disease states.
- To understand the genetic exchange mechanisms in bacteria.

Unit 1: Basic concepts of Bacterial DNA Metabolism **10 hrs**

DNA replication in prokaryotes- semiconservative synthesis, replication model- initiation elongation, termination, Types of metabolic pathways and multi enzyme complexes. Introduction to specific organisms – E coli, B. Subtilis, Plasmid R K2.

Genetics of bacteriophages – T4 infectious cycle, phage structure, phage genomic structure, Process of infection – adsorption, injection, transcription to translation. T odd coliphages.

Bacteriophages- lytic and lysogenic cycle, phage integration and regulation. Different types of restriction modification systems. Mobile DNA elements.

Unit 2: Genetic response in bacteria **10 hrs**

Bacterial quorum sensing, biofilm formation, antibiotic/biocide resistance, endospore formation in bacteria- morphological changes, gene expression changes and process of sporulation, stress shock response in E-Coli and Bacillus subtilis. Introduction to Myxococcus xanthus, Genetic analysis of gliding, starvation induced fruiting body formation and sporulation in Myxooccus xanthus. Introduction to agrobacterium- The vir regulon, conjugation model of T-DNA transfer

Unit 3: Genetic exchange **10 hrs**

Transposons, integrons, retrotransposons. Bacterial transformation – natural competence and artificial competence, DNA uptake and integration, electroporation, biolistic transformation. Conjugation by E-coli F factor, prokaryotic and eukaryotic plasmids, generalized transduction process.

Recommended books

1. Modern microbial genetics, edited by Uldis N Streips, Ronald E Yasbin, A John Wiley and Sons INC, Publication.
2. Microbial Genetics, edited by Stanley R. Maloy, John E. Cronan, Jr., David Freifelder

SEMESTER 2

OE-1a Humanities

(30 hours, LTP – 2+0+0)

Learning objectives

- The student will be able to analyze the politics and narrative forms of health-related texts.
- To recognize and understand the histories of medical discourses, to understand and analyze bioethical dilemmas in contemporary biomedicine.
- To interpret cultural representations of health, illness, and well-being.

Unit I

How does the transition from student to professional (professionalization) occur: objectification of the body, responsibility vs. inexperience, instruction in "professionalism" vs. the hidden curriculum Interaction between professional and personal life

Unit II

Perspectives on personal-professional and patient-physician boundaries
Narrative and empathy

Unit III

What is "normal": defining disease; social construction of disability; race and race-based medicine Difference, rejection, Otherness Medical uncertainty

Unit VI

Illness as exile Socioeconomic marginalization and illness

Recommended books

- Michael Wesch, (2018), The Art of Being Human, reateSpace Independent Publishing Platform; 1 edition.
- Coulehan, Jack. (2006) "You Say Self Interest, I Say Altruism." In Professionalism in Medicine: Critical Perspectives, eds. Delese Wear & Julie M. Aultman (New York: Springer) pp. 103-128

SEMESTER 2

OE-1b Health economics

(30 hours, LTP – 2 + 0 + 0)

Learning objectives

- The student will be able to demonstrate advanced knowledge of how medical knowledge, individual and organizational competence and the resources of healthcare and society can best be utilized for improving people's health
- Demonstrate advanced knowledge about structure and governance as well as developments within medical and social care work
- Demonstrate advanced knowledge and understanding of how health systems function and health policy is created and implemented

Theory **30 hrs**

Unit I

Introduction to Health Economics, The economic way of thinking about health, Health measurement, determinants and long run trends, economic evaluation of healthcare

Unit II

Economic Models of Health, Health and Socioeconomic Status, cost containment measures in genetics

Unit III

Health Insurance, Indian scenario and social security, third party administrators

Unit IV

National health programs and screening programs, planning and designing of genetic laboratories, equipment management in genetic laboratory, legal and statutary compliances in Medical genetics.

Unit V

Medical audit, clinical audit

Recommended books

1. Sherman Folland, Allen C. Goodman, and Miron Stano (2017) The Economics of Health and Health Care, 8th ed. New York, NY: Routledge.
2. Jay Bhattacharya, Timothy Hyde, Peter Tu (2013) Health Economics, Palgrave Macmillan

SEMESTER 2

AEC-3 FUNDAMENTALS OF EPIGENETICS

(30 hours, LTP – 2+0+0)

Learning Objectives:

1. The subject covers fundamental of epigenetics
2. It aims to cover concepts underlying transcriptional regulation of genes by epigenetic factors
3. Topics covers various techniques used to study epigenetic marks

Unit 1: Basic concepts in epigenetics (8hr)

Definition and importance, brief introduction to transcription, transcription initiation, gene structure (promoters and enhancers) – heterochromatin and euchromatin,
Nucleosome structure and chromatin architecture, chromatin remodelling and role of SWI/SNF complex, Epigenetic modifications of DNA– DNA methylation- CpG islands, activity of DNA methyltransferases, DNA hydroxymethylation (5hmc), concept of epigenetic memory

Unit 2: Post translational modification of histones (10hr)

Post translational modification of histone tails, acetylation and methylation of histone residues, Histone modifying enzymes – methyltransferases (SET domain family members), acetyltransferases, and demethylases and deacetylases. Sumoylation and ubiquitination of histones and enzymes involved. Functioning of Polycomb and trithrox family proteins, functioning of bromodomain family proteins, histone variants

Unit 3: Epigenetic regulation of transcription (12hr)

Epigenetic control of transcription- in myogenesis and β-globin, Hox genes, Non coding RNAs and microRNA in transcriptional regulation. Epigenetic control in cellular differentiation, development, concept of bivalent domains in stem cells
Epigenomic reprogramming during somatic nuclear transfer technology and induced pluripotency Epigenetic mechanisms in stem cell plasticity. Techniques used to study DNA methylation – bisulphite conversion and methylation specific PCR. Understanding DNA methylation through restriction digestion analysis. Chormatin immunoprecipitation (ChIP) and ChIP sequencing.

Recommended books

- Epigenetics by Lyle Armstrong, 2014 by Garland Science, Taylor & Francis Group, LLC
- Bradley E. Bernstein Alexander MeissnerEric S. Lander Cell 128, 669–681, February 23, 2007

SEMESTER 2

AEC-4 RESEARCH METHODOLOGY

(30 hours, LTP – 2 + 0 + 0)

Learning objectives

- The subject covers key points to be considered while designing and executing a research project.
- The topics helps students to frame result oriented objectives and aims in their research.
- To conduct research with ethics and organization of research data.

Unit 1 **(10 hrs)**

Foundations of Research: Meaning, Objectives, Motivation, Utility. Concept of theory, empiricism, deductive and inductive theory. Characteristics of scientific method – Understanding the language of research – Concept, Construct, Definition, Variable. Research Process. Problem Identification & Formulation – Research Question – Investigation Question – Measurement Issues – Hypothesis – Qualities of a good Hypothesis – Null Hypothesis & Alternative Hypothesis. Hypothesis Testing – Logic & Importance

Unit 2 **(10hrs)**

Research Design: Concept and Importance in Research – Features of a good research design – Exploratory Research Design – concept, types and uses, Descriptive Research Designs – concept, types and uses. Experimental Design: Concept of Independent & Dependent variables. Qualitative and Quantitative Research: Qualitative research – Quantitative research.

Unit 3 **(10hrs)**

Interpretation of Data and Paper Writing – Layout of a Research Paper, Impact factor of Journals, When and where to publish? Ethical issues related to publishing, Plagiarism and Self-Plagiarism. Use of Encyclopedias, Research Guides, Handbook etc., Academic Databases for Health research. Use of tools / techniques for Research: methods to search required information effectively, Reference Management Software like Zotero/Mendeley, Software for paper formatting like LaTeX/MS Office, Software for detection of Plagiarism

Recommended books

1. David E Gray (2017) Doing Research in the Real World 2nd Edition, SAGE publications Ltd
2. Donald H. McBurney, Theresa L. White (2009) Research Methods 7th Edition, Wadsworth/ Cengage Learning

SEMESTER 3

DSC-7 GENETICS OF RARE DISEASES

(60 hours, LTP – 3 + 1 + 1)

Course Objectives:

Students should be able to

- CO1: Understand the socio-economic burden of rare disorders
- CO2: Understand the inheritance pattern and genetic defect in different rare disorders
- CO3: Perform screening test for inborn errors of metabolism

Unit 1 **(15 hrs)**

Definition of rare disease, Incidence and prevalence of rare diseases globally and in India, Indian scenario of rare disease, socio-medical issues regarding diagnosis and treatment.

List of rare disorders based on carbohydrate, amino acid, lipid, and nucleotide metabolism. Peroxisomal. Lysosomal and mitochondrial disorders, single gene disorders and disorders of coagulation and bleeding.

Introduction to Inborn Errors of Metabolism- definition, types, lab investigations and diagnosis

Peroxisomal Disorders: X-linked adrenoleukodystrophy, Zellweger spectrum syndrome, neonatal adrenoleukodystrophy, infantile Refsum disease incidence, prevalence, inheritance pattern, biochemical & pathological basis, different types, genetic defect, gene mutation, function of the gene, lab diagnosis and treatment options available

Mitochondrial disorders: MELAS, Leigh syndrome and Mitochondrial myopathies- incidence, prevalence, inheritance pattern, biochemical & pathological basis, different types, genetic defect, gene mutation, function of the gene, lab diagnosis and treatment options available

Unit 2 **15 hrs**

Mucopolysaccharidosis (Lysosomal Storage Disorders) - incidence, prevalence, inheritance pattern, biochemical & pathological basis, different types, genetic defect, gene mutation, function of the gene, lab diagnosis and treatment options available

Organic-Acid Disorders: maple syrup urine disease methylmalonic aciduria, propionic aciduria and isovaleric aciduria - incidence, prevalence, inheritance pattern, biochemical & pathological basis, different types, genetic defect, gene mutation, function of the gene, lab diagnosis and treatment options available

Unit 3 **15 hrs**

Mucolipidosis (Lysosomal Storage Disorders) - incidence, prevalence, inheritance pattern, biochemical & pathological basis, different types, genetic defect, gene mutation, function of the gene, lab diagnosis and treatment options available

Disorders of Copper Metabolism- Wilson *disease*, Menkes *disease* : incidence, prevalence, inheritance pattern, biochemical & pathological basis, different types, genetic defect, gene mutation, function of the gene, lab diagnosis and treatment options available

Unit 4 **15 hrs**

Disorders of coagulation and bleeding**:** Factor VIII, Factor IX, Afibrinogenemia, Other disorders of coagulation, Wiskott Aldrich syndrome and others - incidence, prevalence, inheritance pattern, biochemical & pathological basis, different types, genetic defect, gene mutation, function of the gene, lab diagnosis and treatment options available

Single-Gene Disorders**:** Sickle Cell Disease, Huntington Disease, Myotonic Dystrophy, Hereditary Motor and Sensory Neuropathy, Neurofibromatosis, Marfan Syndrome, Cystic Fibrosis, Inherited Cardiac Arrhythmias and Cardiomyopathies, Spinal Muscular Atrophy, Duchenne Muscular Dystrophy, Hemophilia, Collagen disorders: Osteogenesis imperfects

and Ehlers Danlos syndrome- incidence, prevalence, inheritance pattern, biochemical & pathological basis, different types, genetic defect, gene mutation, function of the gene, lab diagnosis and treatment options available

Practicals:

1. Screening tests for Inborn errors of metabolism
    - Ferric Chloride Test
    - Cyanide Nitroprusside test
    - Guthrie's Test
    - Murexide test for Galactosemia
2. LCMS to detect abnormal metabolites for Mucolipidoses and Mucopolysaccharidoses
3. Paper/Thin layer Chromatography for carbohydrates
4. Newborn Screening

References:

- https://ordindia.in/about-rd/rare-disease-in-india/
- https://rarediseases.org/for-patients-and-families/information-resources/rare-disease-information/
- Rare Diseases and Orphan Drugs: Keys to Understanding and Treating the Common Diseases by Jules J Berman.
- Genomics of Rare Diseases: Understanding Disease Genetics Using Genomic Approaches 1st Edition - June 12, 2021 Editors: Claudia Gonzaga-Jauregui, James R. Lupski

SEMESTER 3

DSC-8 GENETICS OF COMPLEX DISEASES

(60 hours, LTP – 3+1+2)

Course objectives

Student should be able to

- CO1: understand complex diseases with Mendelian and chromosomal diseases, various plans and strategies to investigate complex diseases.
- CO2: To understand the statistical analysis in complex diseases.
- CO3: perform biochemical investigations for complex diseases

Unit 1

Defining Complex Disease **8 hrs**

Chromosomal Diseases, Mendelian Diseases, Variation in The Mitochondrial Genome and Associated Diseases, *De Novo* Mutations and Human Diseases, Three Different Types of Complex Disease, Alzheimer's Disease a Monogenic Complex Disease, HSCR – An Oligogenic Complex Disease, Crohn's Disease is Mostly a Polygenic Complex Disease, Applying Disease Models to Populations.

How to Investigate Complex Disease Genetics **7 hrs**

Planning Stage 1: Gathering the Basic Knowledge, Planning Stage 2: Choosing a Strategy, Good and Bad Practice, New Technologies and the Future.

Unit 2

Why Investigate Complex Disease Genetics? **6 hrs**

Disease Diagnosis, Patient Treatment/Management and Care, Disease Pathogenesis of ankylosing spondylitis, Rheumatoid arthritis, Bipolar disease, coronary artery disease.

Statistical Analysis in Complex Disease: Study Planning and Data Handling **9 hrs**

Linkage Analysis, The Basic Statistical Concepts of Association Analysis and their Application in Study Design, Statistical Error, Power, and *P* Values, The Basic Statistical Considerations for Analysis of Case Control Association Studies and their Application to Data Collection and Analysis, How to Interpret a GWAS

Unit 3

The Major Histocompatibility Complex **8 hrs**

Histocompatibility, The Extended Human MHC MAP, Immune Function of HLA Class I and Class II, HLA Class I and Disease, HLA Class II and Disease, Comparing the Hla Associations of the Three Liver Diseases, Non-HLA MHC Genes and Disease, A Single Gene or a Risk Portfolio, How to Compare and Critically Evaluate Contrasting Studies.

Cancer as a Complex Disease: Genetic Factors Affecting Cancer Susceptibility and Cancer Treatment **7 hrs**

Defining Cancer, Cancer as a Complex Disease, Genetic Risk Factors for Particular Cancers Detected by GWAS, General Cancer Risk Loci Detected by GWAS, Previously Established Cancer Risk Factors Confirmed by GWAS, Individualizing Drug Treatment Based on Tumor Genotype.

Unit 4

Genetic Studies on Susceptibility to Diabetes **7 hrs**

Diabetes Mellitus, Genetics of T1D, Early Genetic Studies in T1D, GWAS Studies in T1D, Early

Genetics of T2D, GWAS Studies in Type T2D, The Future of Genetics in T2D, Genetics of Monogenic Diabetes.

Sequencing Technology and the Future of Complex Disease Genetics 8 hrs
DNA Sequencing: The Past, Present, and Future, The Future of NGS in Clinical Practice and Research, Whole-Genome Versus Exome Sequencing, The Next Generations of Genome/ Exome-Wide Association Studies, Epigenetics: A Complimentary Strategy in Complex Disease Studies, Metagenomics and the Bacterial Genome, Major Ongoing International Genome Projects, Systems Biology.

Practical's

- Biochemical investigations for complex diseases – Lipid profile, liver function test, renal function test, hormones and tumor markers.
- Identifying the Molecular basis of complex disorders using real-time PCR and next generation sequencing.
- DNA isolation and bisulfite conversion of DNA for methylation studies
- DNA methylation analysis using MSP QPCR
- DNA methylation analysis using restriction digestion
- Chromatin immunoprecipitation assays
- Isolation of histones and western blot for histone, global methylation and acetylation status of histones in mammalian cells

Recommended books

1. Genetics of complex disease/Peter Donaldson, Ann Daly, Luca Ermini, Debra Bevitt.
2. Current topics in human genetics: studies in complex diseases, co-editors-in-chief, Hong-Wen Deng and Hui Shen; associate editors, Yong-Jun Liu, Hai Hu.
3. Genes and Common Diseases, Alan Wright & Nicholas Hastie.
4. Genetic Analysis of Complex Disease, Jonathan L. Haine.

SEMESTER 3
DSC-9 GENETIC COUNSELING
(60 hours, LTP – 3+1+2)

Course Objectives
Students should be able to

- CO1. To know what a referral to a medical genetics service entails and to understand the importance of genetic investigations when such a diagnosis is suspected.
- CO2. Co-ordinate with clinical geneticists or genetic counsellors.
- CO3. To comprehend genetic basis of diseases
- CO4. To provide a basic level of genetic information and appropriate emotional support to the patients and to patients' families.

Unit 1
General aspects of Genetic Counselling **5 hrs**
Introduction, Genetic Counselling in Mendelian Disorders, Common Disorders and Genetic Counselling, Chromosome Abnormalities, Molecular Genetics and Genetic Counselling, Dysmorphology and Genetic Syndromes, Carrier Testing and Genetic Prediction, Prenatal Diagnosis and Reproductive Aspects of Medical Genetics, Special Issues in Genetic Counselling, The Genetic Counselling Clinic.

Unit 2
Genetic Counselling: Specific organ systems Part 1 **25 hrs**
Pedigree, inheritance pattern and molecular basis of Neuromuscular Disorders – Eg: Muscular dystrophies, Congenital myopathies, Metabolic myopathies, Myotonic dystrophy, Myasthenia gravis, Spinal muscular atrophies, Motor neurone disease, Charcot–Marie–Tooth disease.

Pedigree, inheritance pattern and molecular basis of Central Nervous System Disorders – Eg: Huntington's disease, Parkinson's disease, Multiple sclerosis, Neurofibromatosis, Von Hippel–Lindau syndrome, Tuberous sclerosis, Epilepsy, Cerebral aneurysms and stroke, Migraine, Cerebral palsy, Neural tube defects, Hydrocephalus.

Pedigree, inheritance pattern and molecular basis of Disorders of Mental Function – Eg: The dementias, Mental retardation, Behavioural disorders.

Pedigree, inheritance pattern and molecular basis of Disorders of Bone and Connective Tissue – Primary bone dysplasias, Osteopetrosis, Limb defects, Connective tissue disorders, Arthritis and arthropathies.

Pedigree, inheritance pattern and molecular basis of Oral and Craniofacial Disorders – The teeth, Cleft lip and palate.

Pedigree, inheritance pattern and molecular basis of The Eye – Choroidoretinal degenerations, Nystagmus, Colour vision, Leber's optic atrophy, Corneal dystrophies, Retinal detachment, Retinoblastoma, Cataract, Glaucoma, Refractive errors, Heterochromia of the iris.

Pedigree, inheritance pattern and molecular basis of Deafness – Severe congenital sensorineural deafness, The external ear, Ménière's disease.

Unit 3

Genetic Counselling: Specific organ systems Part 2 **25 hrs**

Pedigree, inheritance pattern and molecular basis of Cardiovascular and Respiratory Disorders – Congenital heart disease, Cardiomyopathies, Coronary heart disease, Aneurysms.
Cystic fibrosis, Asthma and atopy, Emphysema and chronic obstructive pulmonary disease, Sarcoidosis.

Pedigree, inheritance pattern and molecular basis of The Gastrointestinal Tract – Oesophageal atresia, Diaphragmatic hernia, Infantile pyloric stenosis, Omphalocele, Inflammatory bowel disease, Familial adenomatous polyposis and colon cancer, Hirschsprung's disease, Anal atresia.

Pedigree, inheritance pattern and molecular basis of Inborn Errors of Metabolism – Amino acid disorders, Galactosaemia, Sphingolipidoses, Glycogen storage diseases, Hyperlipidaemias, The porphyrias, Fatty acid metabolic defects and sudden infant death.

Pedigree, inheritance pattern and molecular basis of Disorders of Blood and Immune Function – Disorders of haemoglobin structure and synthesis, Immune deficiency disease, Genetic aspects of infectious disease, Haemophilia.

Unit 4

Genetic Counselling in Context: The broader picture **5 hrs**

Communication in genetic counselling, Population aspects of genetic counselling and genetic screening, Genetics, society and the future.

Practicals

- Genetic counselling clinic postings

Recommended books

1. Harper's Practical Genetic Counselling, Eighth Edition.
2. Genetic Counseling Research A Practical Guide (Ian MacFarlane)

SEMESTER 3

OE-2a Soft Skills

(30 hours, LTP – 2+0+0)

Learning objectives

- To give each student a realistic perspective of work and work expectations
- To help formulate problem solving skills, to guide students in making appropriate and responsible decisions
- To create a desire to fulfill individual goals, and to educate students about unproductive thinking, self-defeating emotional impulses, and self- defeating behaviors

Unit I

Definition of soft skills, Soft skills and Hard Skills, Advantage of Soft Skills,
Real life scenarios, Measurement of soft skills.
Self Discovery, Definition of Self, Identification of Strengths and weakness of self, Setting goals, Personal beliefs, values and ethics.

Unit II

Mindsets: Types of Mindsets, Developing a learning and Growth mindset,
Developing a positive outlook towards life, Increasing emotional and Spiritual intelligence.
People skills, Types of people - passive, assertive and aggressive people, Developing assertive personality, dealing with aggressive and submissive people.

Unit III

Communication Skills: Definition of Communication, Verbal and Nonverbal communication, Telephone and internet communication, Common mistakes in communication.
Interpersonal skills: Listening skills, Understanding body language, polite communication and people friendly attitude.

Unit IV

Time management: Importance of punctuality, Efficient time handling,
Avoiding leakage of time and procrastination
Stress Management: Definition of Stress, Positive and negative stress. Handling major projects through effective delegation.

Unit V

Organizational behavior: Definition of an organization, Understanding the rules and regulations of an organization, Creating an ideal working Environment.
Professional attitude-Definition and developing an effective professional attitude.
Leadership Skills: Developing a positive attitude, Presentation and public speaking skills, effective handling of the team and sub ordinates. Recognizing and encouraging talents in Sub ordinates.

Recommended books

- Barun Mitra (2016), Personality Development and Soft Skills, 2nd edition, Oxford University Press
- Alex K (2014), Soft Skills Paperback, S Chand & Company
- Peggy Klaus (2008) The Hard Truth About Soft Skills: Workplace Lessons Smart People Wish They'd Learned Sooner 1st edition, HarperBusiness.

- Sanjay Kumar, Pushp Lata (2018) Communication Skills Paperback 1st edition, Oxford University Press
- John Hayes (1994), Interpersonal Skills: Goal Directed Behavior at Work, Routledge.
- Gurdeep Singh Gujral (2013) Leadership Qualities for Effective Leaders, VIJ Books (India) Pty Ltd.

SEMESTER 3

OE-2b INTELLECTUAL PROPERTY RIGHTS

(30 hours, LTP – 2+0+0)

Learning objectives

- The student will be able to understand the fundamental aspects of Intellectual property Rights
- The students will be introduced to all aspects of the IPR Acts and will be familiarized with the remedies and licensing regime associated with each kind of intellectual property

1. Introduction to Intellectual Property Rights
2. Fundamentals of Patent Law

   Criteria of Patentability, Invention, Novelty, Utility, Inventive step/ Non-obviousness, Non-patentable Inventions and Drafting of patent specification: patent specification, provisional specification, complete specification and Patent procedure in India, Patent infringement
3. Concept of Trademark

   What is a trademark?; Rights of trademark?; What kind of signs can be used as trademarks?; types of trademark function does a trademark perform; How is a trademark protected?; How is a trademark registered?; How long is a registered trademark protected for?; How extensive is trademark protection?
4. Geographical Indications and industrial Design: What is a geographical indication? How is a geographical indication protected? Why protect geographical indications?

   Industrial Designs: What is an industrial design? How can industrial designs be protected? What kind of protection is provided by industrial designs? How long does the protection last?; Why protect industrial designs?
5. Copy right law: Definition, Rights of the Copyright Owner, Term of Copyright, Assignment and Licensing of Copyright, Rights of the Performers and Broadcasting Organisations,

   and Overview of Biotechnology and Intellectual Property

Recommended books

1. T. M Murray and M.J. Mehlman, (2000) Encyclopedia of Ethical, Legal and Policy issues in Biotechnology, John Wiley & Sons
2. P.N. Cheremisinoff, R.P. Ouellette and R.M.Bartholomew, (1985) Biotechnology Applications and Research, Technomic Publishing Co., Inc. USA,
3. D.Balasubramaniam, C.F.A.Bryce,K. Dharmalingam, J. Green and K. Jayaraman, (2002) Concepts in Biotechnology, University Press (Orient Longman Ltd.)
4. Bourgagaize, Jewell and Buiser, (2000) Biotechnology: Demystifying the Concepts, Wesley Longman, USA.
5. Ajit Parulekar and Sarita D' Souza, (2006) Indian Patents Law – Legal & Business Implications; Macmillan India ltd.
6. B.L.Wadehra; (2000) Law Relating to Patents, Trade Marks, Copyright, Designs & Geographical Indications; Universal law Publishing Pvt. Ltd., India
7. P. Narayanan;(2010) Law of Copyright and Industrial Designs; Eastern law House, Delhi.
8. Dr. Kalyan C. Kankanala, Arun Narasani and Vinita Radhakrishnan, Indian Patent Law and Practice, OUP Publications, ISBN: 0-19-806774-7 978-0-19-8066740.
9. P. Narayanan, Patent Law, Eastern Law House, 4th edition, 2006, ISBN: 81-7177-1785.

10. Thomas G. Field, Introduction to Intellectual Property, California Academic Press, 2003 edition, ISBN: 0-089089-236-9
11. Basudurga das, the Constitutional Law of India, (8th edition. Vol.3, 2008) Lexis Nexis Butter Worths Wadhwa, Nagpur.
12. Constitutional Law of India , Dr. J. N.Pandey
13. B.L Wadhera- Intellectual Property
14. WIPO - Reading Material on Intellectual Property Law
15. Brainbridge, David – Cases and Materials in Intellectual Property Law
16. Cornish W.R - Cases and Materials in Intellectual Property Law
17. Dr.S.K Singh- Intellectual Property Rights Laws

SEMESTER 3

DSE-2a Pharmacogenetics

(30 hours, LTP – 2+0+0)

Learning objectives

- To understand the basic principles of pharmacogenetics including factors relevant to drug disposition and the role of pharmacodynamics.
- To have an overview of pharmacogenetics in many important therapeutic areas.
- To know the ethical and related issues in implementing pharmacogenetics into clinical practice.

Unit 1: Pharmacogenetics: Relationship To Pharmacokinetics And Pharmacodynamics

Pharmacogenetics in Drug Metabolism: Role of Phase I Enzymes, Pharmacogenetics of Phase II Drug Metabolizing Enzymes, Pharmacogenetics of Drug Transporters, Pharmacogenetics of Drug Targets.

Unit 2: Pharmacogenetics: Therapeutic Areas

Cardiovascular Pharmacogenetics, Pharmacogenetics in Psychiatry, Pharmacogenetics in Cancer, Pharmacogenetics of Asthma and COPD, Pharmacogenetics of Adverse Drug Reactions, Pharmacogenomics of Inflammatory Bowel Diseases, Pharmacogenetics of Pain Medication.

Unit 3: Pharmacogenetics: Implementation in Clinical Practice

Ethical and Social Issues in Pharmacogenomics Testing, High-Throughput Genotyping Technologies for Pharmacogenetics, Developments in Analyses in Pharmacogenetic Datasets.

Unit 4: Pharmacogenetics: industry and regulatory affairs

Applications of Pharmacogenetics in Pharmaceutical Research and Development, Role of Pharmacogenetics in Registration Processes, Pharmacogenetics: Possibilities and Pitfalls.

Recommended books

- Pharmacogenetics and individualized therapy / edited by Anke-Hilse Mailand-van der Zee, Ann K. Daly.
- Pharmacogenomics: The Search for Individualized Therapies. Edited by J. Licinio and M.-L. Wong.

SEMESTER 2

DSE-2b Nutrigenomics

(30 hours, LTP – 2+0+0)

Learning objectives

- The subject discusses how diet, food, nutrients affects our genome.
- It describes chages in epigenome due to availability of metabolites.
- The chapter describes how human body is susceptible to disease due to changes in nutrition.

Unit 1: **10hrs**

Nutrition and common diseases – An introduction **5Hrs**

Introduction to nutrition and its importance, Impact of exercise, Nutrition and obesity, Nutrition and diabetes, Nutrition and cancer, Nutrition and cardiovascular diseases

Molecular genetic basis **5Hrs**

Nutrient sensing mechanisms via nuclorecoptors, Function and action of PPARs, Circadian control of metabolic process. Adaptation of human genome to dietary change, Vitamin D and skin color association, Integrative personal omics profiling.

Unit 2: **10Hrs**

Nutritional signaling and epigenetics

Metabolism and epigenetic signaling, One carbon metabolism and DNA methylation Nutritional triggered epigenomic changes in mice and humans. Function of sirtuins in Aging, Age associated Nutrient sensing pathways, AMPK signalin and calorie restriction in drosophila and mice

Unit 3 **10Hrs**

The link between diseases and Nutrition

Haromonal regulation in Obesity, Genetics of obesity, Diabetes – Insulin signaling pathway, glucose homeostasis, genetics of Diabetes, central role of FOXO transcription factors, thrifty gene hypothesis. Metabolic syndromes- genetic and epigenetic basis of metabolic syndromes. Mechanisms of hypertension and atherosclerosis.

Recommended books

1. Nutrigenomics by Carsten Carlberg, Stine Marie Ulven, Ferdinand Molnar, Springer publications

SEMESTER 3

AEC-5 Infertility & Assisted Reproductive Technology

(30 hours, LTP – 2+0+0)

Learning Objectives

- The student will be able to identify the causes of female and male infertility and describe appropriate workup, testing and treatment.
- The student should be able to discuss assisted reproductive technologies, including IVF and embryo transfer, intracytoplasmic sperm injection, sperm preparation techniques, and embryo cryopreservation, as well as indications and protocols for these procedures.
- Identify patients at increased risk of genetic disorders, and common indications and techniques for genetic screening, testing and counseling.

Unit 1

Oogenesis and Folliculogeneis: Oocyte retrieval and selection. Preparation and evaluation of oocytes for ICSI. Hyaluronic acid binding-mediated sperm selection for ICSI.
Spermatogenesis and Andrology: Evaluation of sperm. Sperm preparation techniques. Sperm chromatin assessment.
Fertilization and Embryos in assisted reproduction technology (ART): Embryology, In Vitro Fertilization (IVF), Analysis of Fertilization, Morphological Assessment of Embryos and Oocytes, Embryo Transfer Techniques, Cryopreservation and Vitrification, Time Lapse Videos.

Unit 2

Endometrial Receptivity and Female Factor Sterility: Window of implantation, endometrial cycle, Assessment of Receptivity, Impact of Ovarian Stimulation.
Male Infertility: Etiology and Pathophysiology, Clinical and Endocrinal Evaluation.
Genetics of Infertility: Cytogenetic Abnormalities, Genetics of y chromosome-Derived Infertility, Molecular Genetic Testing.
Female Factor Infertility: Uterine, Cervical, Tubal and Fallopian tube factors, Infertility and Molecular Genetics in Females.

Unit 3

Advances and Dilemmas in Assisted Reproductive Technologies, Preimplantation Genetic Screening of Embryos, Preimplantation Genetic Diagnosis, Preimplantation Genetic Diagnosis and HLA typing, OMICS in Infertility.

Recommended books

1. Dhastagir Sultan Sheriff (2018) Infertility, Assisted Methods of Reproduction and Hormonal Assays, IntechOpen publisher
2. National Research Council. (2002). Scientific and Medical Aspects of Human Reproductive Cloning. Washington, DC: The National Academies
3. Gey Becker (2000) The Elusive Embryo: How Women and Men Approach New Reproductive Technologies. University of California Press. Berkeley, CA. Publication

SEMESTER 3
AEC-6 Scientific Writing
(30 hours, LTP – 2+0+0)

Learning objectives:
This course is aimed at teaching the fundamentals of effective scientific writing. The primary focus is to introduce the students to the format of scientific writing, the peer review process, and ethical issues in publication. Additionally, fundamental professional skills in writing CV/ Resume and presentation will also be addressed. The course will be presented in two segments: Part (1) lectures- 20 hours; and Part (2) presentations by the students- 10 hours.

Unit I - Basics of effective writing **(3 Hrs)**
Introduction to the characteristics of good writing, crafting the ideas and themes, the right choice of words, voice, grammar and style, formation of better sentences and paragraphs, organization and streamlining the writing process.

Unit II – Professional Skills **(5 Hrs)**
Effective oral presentation, professional email and what's app etiquette, use of social media as a professional tool, writing an effective CV/ resume, applying for internship or approaching for research opportunities.

Unit III– Structure of scientific articles **(8 Hrs):**
Different types of scientific writing, differences in structure between reviews and research articles; literature review, composition of an abstract, title, introduction, methods, results and discussion, preparing figures and figure legends, bibliography, process involved in publishing scientific articles, issues in scientific writing (plagiarism, authorship, ghost writing), peer review process.

Unit IV – Science Careers **(4 Hrs)**
Different career options in science, communicating science to layman, Pursuing PhD- international and national opportunities.

Recommended books

1. How to write and publish a scientific paper by Robert A. Day and Barbara Gastel.
2. The craft of scientific writing by Michael Alley.
3. The scientists guide to writing by Stephen B. Heard.
4. The elements of style by William Struck and White.
5. An utterly correct guide to clarity and style by Benjamin Dreyer.

Semester 4
Masters Research Project
(18 credits)
VBA-1 Industry internship
(6 credits)