Syllabus

M.Sc. Genetic Counselling

2023-2025 batch

(CBCS)

(Semester I-IV)

Sri Guru Ram Das University of Health Sciences, Amritsar

ABOUT THE COURSE

Genetic counselling is a recognised professional career for delivering the genetic and genomic healthcare. The new M.Sc. Genetic Counselling course is a vocational training programme for career of genetic counsellors who are equipped with the knowledge, skills and competencies in various areas of genetic counselling. It will provide the trainee theoretical knowledge and developing skills for application and translation of the scientific, clinical and psychosocial aspects of genetic counselling in dealing with diverse range of common and rare genetic diseases. The role of genetic counsellor is rapidly expanding with the availability and relatively easy access for genetic and genomic testing.

Currently, there is unprecedented access to information about genetic conditions. Families and communities are anxious on disease risks and likelihood of health issues or complications related to complexities of genomics. This requires the need for professionals, like genetic counsellors, who are qualified and capable of extending help to patients, families, clinicians, policy makers and society at large to understand and act on information and explanation offered. The profession of Genetic Counselling offers a multitude of options for health science students to work in Hospitals, Oncology and other specialist clinics, Diagnostic laboratories, Advocacy groups, Fertility (assisted reproduction/ IVF) & Foetal medicine centres, Public Health Institutes, Research & Teaching Departments, and various NGO's.

MISSION

The mission is to provide students a comprehensive training programme of genetic counselling with a focus on basic and applied knowledge of genetics & genomics, clinical experience and counselling skills in an environment that values and supports equity and inclusion. Through rigorous coursework, field experience and research, the programme aims to train highly competent genetic counsellors who are adequately prepared to serve in both clinical and non-clinical roles in the community and to lead future generations of genetic counsellors and genetic healthcare providers. The programme values compassion, honesty, dedication, ethics and personal integrity, and encourages professional collaboration, pursuit of excellence and commitment to continued professional development.

LEARNING OBJECTIVES

The MSc Genetic Counselling Programme offers the unique opportunity for learning and vocational training in the following areas-

i. Basic Courses in Genetics, Molecular Biology, Cell Biology.
ii. Basic elements of Human Genetics, specifically patterns of inheritance, clinical cytogenetics, clinical molecular/ DNA diagnostics, epigenetics and non-traditional inheritance, mitochondrial genetics, and genome polymorphisms including copy number changes.
iii. Genetic clinics assisting clinicians in diagnosis and management through the multi-disciplinary team working with different clinical disciplines. This aspect of the course would require clinical portfolio.
iv. vi) Organisation and conduct of genetic counselling with particular emphasis on psychological and social aspects. This aspect of the course would require the genetic counselling portfolio.
v. Close working links with genetic or genomic diagnostic laboratory including practical opportunity for generation and interpretation of results with the help of laboratory scientists and bio-informatician.
vi. Opportunity to learn and explore issues related to ethics, law and society, collectively referred to as ELSI.

vii. Opportunity for independent research project for developing research skills and ability to explore inter-related deep insights into the subject. The research project would be disseminated in the form of thesis, peer- reviewed publications and/or presentations in academic and clinical conferences.

viii. Finally, a trained Genetic Counsellor will have the knowledge and skill to help patients and families make the best decisions for themselves based on sound knowledge and available testing and treatment or management options applicable for both common and rare genetic disorders.

DURATION OF COURSE:

Duration of Master of Science in Genetic Counselling shall be of two years (4 semesters).

ELIGIBILITY FOR ADMISSION

a) The M.Sc. Programme is open to a candidate who has passed Undergraduate degree in Biological Sciences (B.Sc. Human Genetics, Medical Stream, Biotechnology)/ MBBS or equivalent examination from a Statutory Institution/University.

OR

b) Any other examination recognized by the Board of Management of this University as an equivalent course/examination thereto, from time to time.

EVALUATION OF STUDENTS

Continued Internal Assessment has to be followed by giving at least four examinations, quiz, assignment, field work etc.

Postgraduate Programme: Purely internal evaluation for courses and external evaluation for thesis/dissertation/project work.

EXAMINATION SCHEDULE

1. At least four examinations shall be held–First hourly examination (weightage 10%), Mid-term examination (weightage 25%), Second hourly examination (weightage 10%) and end semester examination (weightage not less than 50%) and 5% attendance weightage. The weightage for Continued Internal Assessment and the End Semester Examination will be equal
2. If practical is a part of theory course it will have weightage up to 30%.
3. The teacher may decide to give an assignment or a class seminar in lieu of the first or second hourly examination with intimation to Head of the Department.
4. All postgraduate examinations will be held in the regular class and conducted by the class teacher under supervision of Head of the Department.
5. All the teachers offering courses shall submit a midterm report to COE in a prescribed format within one week of holding the midterm examination.
6. All the examinations will be held during the week earmarked by the Registrar in the academic calendar.

ELIGIBILITY FOR APPEARING IN END SEMESTER EXAMINATION

1. 50% aggregate marks in First Hourly, Second Hourly and Mid Term Examination.
2. 75% attendance in class lectures and practical classes.

3. Up to 10% lecture shortage may be condoned by paying fine.
4. List of students detained in different courses to be displayed by the concerned Head of the Department well in advance of filling examination forms.

The 10-point scale and letter grades

Percent marks obtained	Corresponding Credit point	Letter Grade
100	10.00	O (Outstanding)
90	9.00	A+ (Excellent)
80	8.00	A (Very good)
70	7.00	B+ (Good)
60	6.00	B (Above average)
50	5.00	**C (Average) For PG programme 5.00 is PASS**
Less than 50	FAIL	FAIL

EXAMINATIONS AND EVALUATION OF STUDENTS

1.1. In case a student could not appear in any component of the Continuous Internal Assessment of a course due to medical reasons or under other exceptional circumstances (supported by documentary evidence), a separate examination in that component will be arranged by the concerned teacher with the approval of respective HOD before the beginning of End Semester Examination.

1.2. A student will be permitted to appear in the End-Semester Examination as per the Conduct of Examination Rules after filling up the prescribed examination form, payment of the prescribed examination fee, satisfying the attendance requirement and fulfilling other eligibility criteria.

1.3. The question paper pattern of the End Semester Examination will be prescribed by the concerned Board of Studies and at least two sets of question papers for these examinations will be submitted to COE in the beginning of the semester, one of these sets will be passed on to the concerned HOD one hour before the Examination. It will be compulsory to pass the End Semester Examination for successful completion of the course.

1.4. Unless prescribed in the Regulations and the Scheme of Examination of a particular programme, a candidate will be deemed to have completed his/her course successfully if he/she obtains minimum 50% marks/Grade point of 5.00/ as per the 10-point scale.

1.5. The Distribution of marks in the final semester of course will be of 30% weightage for written examination (VSAQ's/MCQs), 30% weightage will be research dissertation and 40% of marks

weightage will be given to clinical portfolio with internal and external assessment related to specialist clinical field chosen by the student.

1.6. If a student fails in the End Semester Examination, a supplementary examination within six weeks of declaration of result will be arranged for such students by the respective department with the help of COE. The marks for all other components as applicable will however, be carried forward in such cases. The students will be required to deposit the examination form along with prescribed fee for all such examinations.

1.7. The marks obtained in the Continued Internal Assessment and all the examinations will be shown to the students. The evaluation scheme will also be explained to students.

1.8. At the end of semester, the result for each course is compiled by the concerned teacher, discussed with the head of department and compiled result submitted by HOD to COE.

Transcript:

Based on the above recommendations on Letter grades, grade points and SGPA and CGPA, the transcript for each semester and a consolidated transcript indicating the performance in all semesters may be issued.

Re-Entry after Break of Study:

I. The University regulations for readmission are applicable for a candidate seeking re-entry to a program.
II. Students admitted the program and absenting for more than 3 months must seek readmission into the appropriate semester as per university norms.
III. The student shall follow the syllabus in vogue (currently approved/is being followed) for the program.
IV. All re-admissions of students are Course to the approval of the Vice-Chancellor.
V. All dropped students with CGPA of less than 5.0 in PG programmes, have right to petition for readmission to first year class against vacant seats.
VI. The student shall apply to the Registrar within seven days of his/her dropping from the university.
VII. The case will be reviewed by a petition committee.
VIII. Readmission shall be permitted only once.
IX.

A Ranking

i. The first two ranks of the programme will be decided on the basis of grades of CGPA in the courses.
ii. In case of a tie, marks % of core courses only will be taken into account.

B Normal, Maximum and Minimum Credit Load in a Semester

i. In a programme, the normal full-time programme of work in a semester shall be 24 credit hours.
ii. A maximum of 30 and a minimum of 18 credit hours may be taken by a student.
iii. The PG students on 'Good Standing' may be allowed to register a maximum of 34 credit hours (4 extra credit hours) during their semesters by the Dean, provided these courses registered have been offered on regular basis.

PROGRAMME CONTINUATION / DISCONTINUATION:

The continuation/discontinuation and Exit with Degree shall be governed as follows:

I. A candidate shall be allowed to continue the programme provided he/she maintains a CGPA of 5.0 both in all theory and lab courses at the end of the even semesters (e.g., 2nd, 4th for the academic programmes). Otherwise, the candidate shall remain in the same year till he/she maintains the required CGPA as 5.0.

II. Further, if a student who has been detained due to shortage of attendance in more than 50% course in a semester and due to that he/she is not able to maintain minimum criteria of required CGPA 5.0, then such student will be detained in entire semester and shall have to attend regular classes of all courses in next academic year of said semester.

III. A candidate shall have to re-appear in semester examination of the courses with Fail/Absent grade (as per Grade Assignment Table), as and when the same course is offered during regular course of study. Such students shall retain their internal/sessional marks.

IV. Attendance requirement for appearing in End Semester Examination of each of the semesters shall be 75%.

V. A student who has been detained due to shortage of attendance shall not be allowed to be promoted to next semester and he/she will be required to repeat only those courses where the student could not make up required 75% attendance in a semester. That is, attendance of a student shall be considered course-wise and if any student fails to maintain minimum criteria of 75% attendance in a course, then he/she will be detained in that particular course only and all such students will have to reappear for that course(s) and also attend the class of that particular course with the next batch of students of said semester.

VI. Maximum three attempts including main exam (main exam + two backlogs) shall be provided to all students to clear his/her backlogs/arrear in order to get promoted in next year/semester. That is, maximum two academic years are permissible for a student for the completion of the academic programme/course. In no situation a student will be allowed to take more than two academic years, for any reason whatsoever, including for the reasons of detention for shortage of attendance or deficiency of CGPA during the whole term of completion of the course/programme.

VII. A student who has already availed two additional academic years to clear his/her backlogs/arrears in order to be eligible for the Degree in stipulated time period, will not get any further chance and therefore admission of such student(s) would be automatically stand cancelled.

CORRECTION OF ERRORS

In case of any error is detected in the marks recorded on the award list, the examiner(s) concerned shall make a request to correct the mistake to the Principal through the Head of the department, and shall attach relevant documentary evidence. A committee consisting of the following members shall take suitable remedial measures depending upon the merit of the case.

a) Dean/Principal (Chairman)
b) Head of the department.
c) Two Faculty Member nominated by the Dean

Sri Guru Ram Das University of Health Sciences, Amritsar
Course Structure for the M.Sc. Genetic Counselling

Course code	Course Title	Course Type	L+T+P	Total Credits
Semester-I				
MGC.501	Principles and Practice in Medical Genetics-I	Core Course	3+0+0	3
MGC.502	Fundamental of Genetic Counselling Skills	Core Course	4+0+0	4
MGC.503	Clinical applications of Cytogenetics & Molecular Techniques	Core course	4+0+0	4
MGC.504	Molecular Basis of Human Diseases	Core course	3+0+0	3
RMB.505	Research Methodology & Biostatistics	Interdisciplinary course	3+1+0	4
MGC.599	Seminar I & Journal Club	Core course	2+0+0	2
MGC.506P	Medical Genetics Clinic Practical	Core course	0+0+2	2
		Total Credits	**19+1+2**	**22**
Semester-II				
MGC.507	Principle and Practice in Medical Genetics-II	Core Course	3+0+0	3
MGC.508	Fundamentals of Personalized Medicine	Core Course	3+0+0	3
MGC.509	Dysmorphology	Core Course	3+0+0	3
MGC.510	Reproductive Genetics	Core Course	3+0+0	3
MGC.511	Foundation of Genomic Medicine	Core Course	3+0+0	3
MGC.512P	Prenatal Genetic Counselling Clinical Practical	Core Course	1+0+1	2
MGC.513P	Advanced Clinical Genetics Practical	Core Course	1+0+1	2
MGC.514P	Neurogenetics Clinical Practical	Core course	1+0+1	2
MGC.600	Synopsis writing	Core course	1+0+0	1
		Total Credits	**19+0+3**	**22**
Semester-III				
MGC.515	Clinical Genetics & Management of Metabolic Disease	Core course	3+0+0	3
MGC.516	Cancer Genetic Counselling	Core course	3+0+0	3
MGC.517	Translational Genomics	Core course	3+0+0	3
MGC.518	Ethical, Legal and Social Issues in Genetic Counselling	Core elective course	3+0+0	3
MGC.599	Seminar II	Core course	1+0+0	1
MGC.519	Regulations in Human Genome Research	Core Course	2+0+0	2
MGC.520	Bioethics & Intellectual Property Rights	Elective Course	2+0+0	2
MGC.521P	Cancer Genetic Counselling Clinical	Core Course	0+0+2	2

	Practical			
MGC.522 P	Metabolic Genetics Clinical Practical	Core Course	0+0+2	2
MGC.523 P	Cardiovascular Genetics Clinical Practical	Core Course	0+0+2	2
		Total Credits	**17+0+6**	**23**
Semester-IV				
MGC-600	Project /Dissertation Work	Core course	0+0+20	20
MGC.550	Clinical Rotation	Core course	0+0+20	20
		Total Credits	**0+0+40**	**40**
		Grand Total Credits	**56+0+51**	**107**
Project/Dissertation work: Project/Dissertation work will begin in third semester. The proposal for Project/Dissertation work shall be finalized in second semester.				

DISRIBUTION OF MARKS

Semester –I

Course code	Title	Total Marks
MGC.501	Principles and Practice in Medical Genetics –I	100
MGC.502	Fundamental of Genetic Counselling Skills	100
MGC.503	Clinical Applications of Cytogenetics & Molecular Techniques	100
MGC.504	Molecular Basis of Human Diseases	100
RMB-505	Research Methodology & Biostatistics	100
MGC.506 P	Medical Genetics Clinic Practical	50
MGC.599	Seminar I & Journal Club	50
	Total Marks	**600**

Semester –II

Course code	Title	Total Marks
MGC.507	Principles and Practice in Medical Genetics -II	100
MGC.508	Fundamentals of Personalized Medicine	100
MGC.509	Dysmorphology	100
MGC.510	Reproductive Genetics	100
MGC.511	Foundation of Genomic Medicine	100
MGC.512 P	Prenatal Genetic Counselling Clinical Practical	50
MGC.513 P	Advanced Clinical Genetics Practical	50
MGC.514 P	Neurogenetics Clinical Practical	50
MGC.600	Synopsis Writing	-
	Total Marks	**650**

Semester –III

Course code	Title	Total Marks
MGC.515	Clinical Genetics & Management of Metabolic Disease	100
MGC.516	Cancer Genetic Counselling	100
MGC.517	Translational Genomics	100
MGC.518	Ethical, Legal and Social Issues in Genetic Counselling	100
MGC.519	Regulations in Human Genome Research	50
MGC.520	Bioethics & Intellectual Property Rights	50
MGC.521 P	Cancer Genetic Counselling Clinical Practical	50
MGC.522 P	Metabolic Genetics Clinical Practical	50
MGC.523 P	Cardiovascular Genetics Clinical Practical	50
MGC.599	Seminar II	50
	Total Marks	**700**

Semester –IV

MGC.600	Dissertation–3 Months	100
MGC.550	Clinical Rotation-3 Months	50
	Total Marks	**150**
	GRAND TOTAL	**2100**

SKILLS TO BE ACQUIRED DURING CLINICAL TRAINING IN GENETIC COUNSELLING

A. Communication skills

1. Ability to communicate information regarding genetic concepts, genetic diseases and the personal and medical consequences of genetic disease to individual clients (most of whom will have no scientific training), the general public and other health professionals in English and another language (e.g., Hindi, Punjabi, Haryanvi, etc.).
2. Ability to obtain information from the patient/family both with respect to the medical situation that brought them to genetic counselling and with respect to their own primary concerns.
3. Ability to respond to those concerns with medical or genetic information, with psychological support, and where appropriate, with referral to social service agencies or other health professionals.
4. Ability to help the family understand, where appropriate, the normal psychological reactions to death of a child, loss of a pregnancy or coping with a family member with genetic disease and to provide the requisite support.
5. Ability to explain the alternatives available to each family, both with respect to medical management, life expectations and social services, and with respect to psychological adjustments.
6. Ability to convey information without infringing upon individual social, cultural or religious beliefs. This will in many cases require some discussion of the ethical ambiguities inherent in the provision of genetic services.

B. Assessment skills

1. Ability to identify the concerns of the patient/family (or community group) with respect to a specific genetic disease or genetic disorders in general (i.e., why is a genetic counsellor being consulted?).
2. Ability to assess the clients' level of background knowledge, so that counselling and information may be focused appropriately.
3. Ability to identify families and clients whose levels of stress or distress require a professional psychological referral.
4. Ability to identify the most appropriate referrals for particular patients and families, both for medical and non-medical support.
5. Ability to identify situations in which particular ethical/legal issues are likely to arise, and to counsel/inform accordingly.

C. Technical skills/knowledge

During the course of clinical training, it is expected that the student will consolidate his/her knowledge of basic medical genetics so that it can be applied to individual patient problems. The student's ability to do this will be demonstrated by: displaying basic knowledge of commonly occurring genetic disorders, conducting appropriate literature searches to learn about genetic disorders, and conveying this information to colleagues and patients in a concise and accurate fashion. The student should become knowledgeable about each of the following areas.

1. Medical terminology and the literature of human and medical genetics
2. Cytogenetics

3. Biochemical Genetics
4. Molecular Diagnosis and Genomics
5. Dysmorphology, teratology, inherited congenital and adult-onset diseases, and other pertinent aspects of medical genetics.
6. Social and family support services available within the particular geographic area of practice, Ethical, Legal and Social Issues concerning the Genetic Counselling
7. Use of databases pertinent to the practice of medical genetics, dysmorphology, and teratology and variants of uncertain significance.
8. Population genetics, Epidemiology and Screening.

D. Administrative skills

1. Ability to obtain medical records and/or information as required.
2. Ability to communicate appropriately in writing with patients and colleagues.
3. Ability to organize and schedule clinic activities, including patient visits, diagnostic tests and follow-up.
4. Ability to refer families to the appropriate social service and/or support agencies.

PRACTICALS FOR THE PROGRAMME

MGC.506P: Medical Genetics Clinic Practical

SKILLS TO BE ACQUIRED DURING CLINICAL TRAINING IN GENETIC COUNSELLING

Communication skills

1. Ability to communicate information regarding genetic concepts, genetic diseases and the personal and medical consequences of genetic disease to individual clients (most of whom will have no scientific training), the general public and other health professionals.
2. Ability to obtain information from the patient/family both with respect to the medical situation that brought them to genetic counselling and with respect to their own primary concerns.
3. Ability to respond to those concerns with medical or genetic information, with psychological support, and where appropriate, with referral to social service agencies or other health professionals.
4. Ability to help the family understand, where appropriate, the normal psychological reactions to death of a child, loss of a pregnancy or coping with a family member with genetic disease and to provide the requisite support.
5. Ability to explain the alternatives available to each family, both with respect to medical management, life expectations and social services, and with respect to psychological adjustments.
6. Ability to convey information without infringing upon individual social, cultural or religious beliefs. This will in many cases require some discussion of the ethical ambiguities inherent in the provision of genetic services. Methods for obtaining medical and family histories, approaches to evaluation of individuals and families with genetic disorders, and techniques for providing genetic counselling.

MGC.512P: Prenatal Genetic Counselling Clinical Practical

Hands-on training in prenatal genetic counselling for cell-free foetal DNA and maternal serum screenings, abnormal screening results, ultrasound anomalies, teratogens, family history of genetic conditions, recurrent miscarriage and advanced maternal age. Students provide genetic counselling, case management and follow up in a supervised setting.

MGC.513P: Advanced Clinical Genetics Practical

Hands-on training in clinical genetics diagnostic and counselling clinics. Students participate in providing genetic evaluation, risk assessment and counselling to patients with indications including dysmorphic features; abnormal growth; difficulties with neurological, psychological and /or intellectual functioning; family history of genetic disorders; and abnormal genetic test results.

MGC.522P: Metabolic Genetics Clinical Practical

Hands-on training in metabolic genetics outpatient clinics and inpatient settings with introduction to the roles of the multidisciplinary biochemical genetics team. Students gain experience with the

new-born screening programme and conditions including PKU, galactosemia, organic acid disorders, and lysosomal storage diseases.

MGC.523P: Cardiovascular Genetics Clinical Practical

Covers presentation, long-term management and screening recommendations, risk assessment, genetic testing options and psychosocial issues associated with inherited cardiovascular conditions including cardiomyopathy, familial hypercholesterolemia, aortopathies and congenital heart defects. Students provide genetic counselling and follow-up in a supervised setting.

MGC.521P: Cancer Genetic Counselling Clinical Practical

Practicum includes presentation, long-term comprehensive management, risk assessment, genetic testing options and psychosocial issues associated with inherited oncology conditions. Students provide genetic counselling and follow-up in a supervised setting and become familiar with the genetic counsellor's role in multidisciplinary tumour board meetings.

MGC.514P: Neurogenetics Clinical Practical

Hands-on training in clinical neurogenetics diagnostic and counselling clinics. Students will participate in providing genetic evaluation, risk assessment and counselling to patients with indications including inherited structural brain anomalies, epilepsy syndromes, muscular dystrophies, developmental delays, autism, mitochondrial disorders, and adult-onset neurodegenerative conditions.

Clinical Rotations:

In the departments of Obstetrics & Gynaecology, Paediatrics, Medicine, Cancer hospital to maintain logbooks of approximately 50 cases (combined) of prenatal, paediatric, adult and oncology.

Laboratory Rotations:

Lab rotations in Biochemistry, Pathology and Microbiology laboratories to understand relevant tests and for knowing the normal values for haematology, NBS /Antenatal screening, lipid profile, sugars, hemograms, HPLC etc and autoimmune panels for infertility and BOH and SLE etc.

MGC.501: Principles and Practice in Medical Genetics-I **Credits: 3**

Unit No.	Content	Lectures
1.	Impact of Genetics in Medicine, Clinical Genetic services, Organisation of Genetic services	08
2.	General concepts on Enzyme deficiencies and diseases, various spectrums of genetic diseases and their patterns of inheritance, Detection of Carriers	15
3.	Mutations, Types of Mutations, Clinical consequences of mutations	10
4.	Disorders of sexual development, Chromosomal instability syndromes, Chromosomal microdeletion syndromes, Prion diseases.	12
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Gardner, A. and Davies, T. (2017). *Human Genetics*. Viva Books, New Delhi, published by arrangements with Scion Publishing Limited, 2nd ed.
2.	Gibson, G. (2015). *A Primer of Human Genetics*. Sinauer.
3.	Rimoin, D.L., Pyeritz and Korf, P.R. (2013). *Emery and Rimoin's Principles and Practice of Medical Genetics*, Academic Press, New York, 6th ed.
4.	Turnpenny, P.D. and Ellard, S. (2015). *Emery's Elements of Medical Genetics*. Elsevier, 15th ed.

MGC.502: Fundamentals of Genetic Counselling Skill **Credits: 4**

Unit No.	Content	Lectures
1.	Pedigree taking and analysis-taking information, drawing pedigree, pedigree symbols, identifying mode of inheritance, calculating recurrent risk	09
2.	Communication and Counselling	10
3.	Patterns and mechanism of inheritance-Autosomal dominant, Autosomal recessive, X and Y linked inheritance, mitochondrial inheritance, patterns of multifactorial disease inheritance, non-traditional types of inheritance-imprinting, dynamic mutations, epigenetics, methylation, parental imprinting	15
4.	Genetic Counselling Skills, practice guidelines for telehealth genetic counselling	10
5.	Philosophy and theories of genetic counselling, preparing a Brochure and poster on various issues of counselling and translating them in another language	10
6.	Psychological aspects of genetic counselling-basic Psychology-Grieving process, differences in perception and psychosocial issues like patriarchal society and blame for male child on female	10
	Total	**64**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Gardner, A. and Davies, T. (2017). *Human Genetics*. Viva Books, New Delhi, published by arrangements with Scion Publishing Limited, 2nd ed.
2.	Gibson, G. (2015). *A Primer of Human Genetics*. Sinauer.
3.	Rimoin, D.L., Pyeritz and Korf, P.R. (2013). *Emery and Rimoin's Principles and Practice of Medical Genetics*, Academic Press, New York, 6th ed.
4.	Green S *et al*., (2023). An Evidence Based Practice Guideline f The National Society of Genetic Counselors for Telehealth Genetic Counseling. *Journal for Genetic Counselling* 32: 4-17.
5.	Clarke, A. (2020). *Peter Harper's Practical Genetic Counselling*, Edward Arnold Publishers, 8th ed.

MGC.503: Clinical Applications of Cytogenetics & Molecular Techniques **Credits: 4**

Unit No.	Content	Lectures
1.	Human chromosomes- nomenclature, identification, Autosomes and Sex Chromosomes, ISCN nomenclature	15
2.	Types of chromosomal anomalies, Numerical anomalies, Structural anomalies	10
3.	Chromosomal and genic sex determination, X-inactivation/Dosage Compensation, X-linked diseases in males and females, Correlation of chromosomal abnormalities to disorders	10
4.	Tests for assessing chromosomal anomalies, Methods of chromosome analysis- Karyotyping and chromosomal banding, Fluorescent In-Situ Hybridization, Comparative Genomic Hybridization	09
5.	Basics of Molecular Genetics	05
6.	Molecular genetic testing techniques- PCR, RT PCR, PCR-RFLP, NGS, TaqMan Assay,	15
	Total	**64**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Li, M. and Pinkel, D. (2006). Clinical Cytogenetics and Molecular Cytogenetics. *J Zhejiang Univ Sci B* 7(2):162-3. PMID: 16421976; PMCID: PMC1363764.
2.	Nowakowska, B. and Bocian, E. (2004). Molecular Cytogenetic Techniques and their Application in Clinical Diagnosis. *Med Wieku Rozwoj* 8(1):7-24. Polish. PMID: 15557693.
3.	Gersen, S.L. and Keagle, M.B. (2005). *The Principles of Clinical Cytogenetics*. Humana Press, USA, 2nd Edition.
4.	Czepulkowski, B. (2004). *Analysing Chromosomes*. Bios Scientific Publishers Ltd., Oxford, 2nd Edition.

MGC-504: Molecular Basis of Human Disease **Credits:3**

Unit No.	Content	Lectures
1.	Molecular basis of human diseases- perspectives of genetic counselling in specific organ system, Molecular Genetics and Genetic counselling in organ systems	08
2.	Neuromuscular disorders, Disorders of Central Nervous system, Disorders of Mental Function, Disorders of Bone and Connective Tissue	10
3.	Oral and Craniofacial disorders, Cardiovascular and Respiratory disorders, Disorders of Gastrointestinal tract, Renal and Urinary tract disorders	08
4.	Skin Disorders, Disorders of Eye, Deafness	10
5.	Endocrine and Reproductive Disorders	09
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Jameson, L.J. (ed.) (1998). *Principles of Molecular Medicine*. New Jersey: Humana Publishers.
2.	Strachan, T., Read, A.P. (2010). *Human Molecular Genetics*, London: Garland Publishers.4th edition.
3.	Karpati, G., Hilton-Jones, D. and Griggs, R.C. (eds) (2001). *Disorders of Voluntary Muscle*, Cambridge: Cambridge University Press.7th edition.
4.	Rosenburg, R.N., Di Mauro, S., Paulson, H.L., Ptacek, L. and Nestler, E.J. (2007). *Molecular and Genetic Basis of Neurologic and Psychiatric Disease*. New York: Lippincott, Williams & Wilkins.
5.	Gorlin, R.J., Cohen, M.M. and Hennekam, R.J.M. (2001). *Syndromes of the Head and Neck*, 4th edition. New York: Oxford University Press.
6.	Kumar, D. (2020). *Clinical Molecular Medicine,* Elsevier Academic Press, 2nd edition.

RMB-505: Research Methodology & Biostatistics **Credits: 4**

Unit No.	Content	Lectures
1.	Planning of Research: planning process, selection of a problem for research, formulation of the selected problem, hypothesis formation, measurement, research design/plan. Sampling: sampling techniques or methods, choice of sampling techniques, sample size, sampling and non-sampling errors.	08
2.	Clinical epidemiology and types of medical research: descriptive studies, analytic studies, interventional studies, clinical trials.	08
3.	Sample variability and significance: testing statistical hypothesis, tests of significance, Z-test, one-tailed and two tailed tests.	08
4.	Variability and its measures: Types, biological, real, experimental, measures of variability, range, semi-interquartile, range (Q), mean deviation, standard deviation (SD), coefficient of variation (CV), standard error of mean, applications and uses, standard error of difference between two means of large sample, small sample, t-test unpaired, paired, variance ratio test, analysis of variance. Normal distribution and normal curve: Demonstration of a normal distribution, normal curve, standard normal deviate(z), asymmetrical distributions. Probability (chance): Addition law of probability, multiplication law, binomial probability distribution, probability chance from shape of normal distribution or normal curve.	08
5.	The Chi-square Test: Alternate test to find significances of difference in two or more than two proportions, as a test of association between two events in binomial or multinomial samples, as a test goodness of fit, calculation of x^2 value, restrictions in application of x^2 test, Yates corrections.	05
6.	Correlation and regression: Calculation of correlation coefficient from ungrouped series, regression, calculation of regression coefficient(b), regression line, standard deviation of the Y measurement for the regression line.	08
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	John, V. B. and John, V. K. (2016). *Research in Education*, Pearson publishers. 10th edition
2.	Finney, D.J. (1980). *Statistics for Biologists*. Chapman and Hall Ltd.
3.	Bland, M. (2006). *An Introduction to Medical Statistics*. Oxford University Press, 3rd edition.
3.	Chawla, D. and Sondhi,N. (2016). *Research Methodology Concepts and Cases*, Vikas Books Publishers, 2nd edition.
4.	Donald, H. M. and Theresa, L.W. (2006). *Research Methods*, Cengage Learning India Pvt. Ltd, 5th edition.
5.	Gurumani, N. (2007). *Research Methodology for Biological Sciences*. M.J.P. Publishers, India.

MGC-507: Principles and Practice in Medical Genetics-II **Credits: 3**

Unit No.	Content	Lectures
1.	Single gene disorders, Genetics of Common disorders	10
2.	Molecular Genetics of Haemophilia A and B, Molecular Genetics of thalassemia, Sickle Cell disease	10
3.	Genetic Lipodystrophies, Gene mapping and molecular pathology, Molecular analysis of Mendelian disorders, Treatment of genetic disorders, CRISPR-Cas9, Gene Therapy	06
4.	National Rare Disease Policy 2021, OECD guidelines on Human Biobanks and Genetic Research databases	09
5.	Genetics of overgrowth syndromes, An approach to diagnosis of skeletal dysplasias, Duchenne Muscular Dystrophy: Symptoms, Diagnosis and Therapy, Neural Tube defects- Cause and prevention in India	10
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Rimoin, D. L., Pyeritz, R. E. and Korf, B. (Eds.). (2013). *Emery and Rimoin's Essential Medical Genetics*. Elsevier.
2.	Jorde, L. B., Carey, J. C. and Bamshad, M. J. (2019). *Medical genetics e-Book.* Elsevier Health Sciences.
3.	Passarge, E. (2021). Origins of Human Genetics. A Personal Perspective. *European Journal of Human Genetics*, *29*(7):1038-1044.
4.	Pyeritz, R. E., Korf, B. R. and Grody, W. W. (Eds.). (2020). *Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics: Metabolic Disorders*. Academic Press.

MGC.508: Fundamentals of Personalized Medicine **Credits: 3**

Unit No.	Content	Lectures
1.	Pharmacogenomics- Genomic perspectives of Precision Medicine and healthcare	15
2.	Nutrigenomics	15
3.	Types and applications of Stem cell and regenerative medicine, HLA Typing and its applications	15
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Weinshilboum, R. M. and Wang, L. (2017). Pharmacogenomics: precision medicine and drug response. *In Mayo Clinic Proceeding Elsevier* 92(11):1711-1722.
2.	Müller, M. and Kersten, S. (2003). Nutrigenomics: goals and strategies. *Nature Reviews Genetics* 4(4): 315-322.
3.	Mahla, R. S. (2016). Stem cells applications in regenerative medicine and disease therapeutics. *International journal of cell biology* 76(9):11-60.

MGC.509: Dysmorphology **Credits: 3**

Unit No.	Content	Lectures
1.	Purpose of genetic evaluation in a Dysmorphic child	10
2.	Evaluation of a dysmorphic child, Description of Common terminologies depicting facial anomalies, diagnostic approach to a dysmorphic child	15
3.	Syndrome diagnosis and clinical management, Aetiological basis of malformation syndromes, molecular basis of congenital malformations, Genetic recurrence risks in malformation syndromes	08
4.	Various dysmorphology databases and their uses	02
5.	Classification of birth defects, Minor and major congenital abnormalities, Teratogenic effect on development, CHARGE and VACTERAL association, Single gene defects, Multifactorial inheritance, Role of genetic counselling in dysmorphology.	10
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Jones, K.L. (2006). *Smith's Recognizable Patterns of Human Malformation*. New York: Elsevier
2.	Stevenson, R.E, Hall, J.G. and Goodman, R.M. (2006). *Human Malformations and Related Anomalies*. New York: Oxford University Press.
3.	Winter, R. and Baraitser, M. (2002). *The London Dysmorphology Database*. Oxford: Oxford University Press.
4.	Jones, R.E. and Lopez, K.H. (2014). *Human Reproductive Biology*. Academic Press (Elsevier), 4th edition.
5.	Fundukian, L.J. (2010). *Pearls of Dysmorphology. The GALE Encyclopaedia of Genetic disorders*. China Translation and Printing Services Limited, China, Vol. I and II, 3rd edition.

MGC.510: Reproductive Genetics **Credits: 3**

Unit No.	Content	Lectures
1.	Reproductive Biology and Embryology, Male/ Female reproductive anatomy, Pregnancy and Child birth	10
2.	Prenatal Procedures and Screening tests, Amniocentesis, CVS, Ultrasound Screening, Maternal serum Screening, NIPT, new reproductive technologies	10
3.	Bad Obstetric history	5
4.	Congenital anomalies and teratogenicity, termination of pregnancy-medical and psychological implications, Suitable testing of POC	10
5.	Primary/Secondary Infertility–causes, Assisted reproductive techniques, PGT	10
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Gardner, R.J.M. and Sutherland, G.R. (2003). *Chromosome Abnormalities and Genetic Counselling*. Oxford: Oxford University Press.
2.	Scriver, C.K., Beaudet, A.L., Sly, W.S. and Valle, D. (eds) (2001). *Metabolic and Molecular Basis of Inherited Disease*. New York

MGC.511: Foundation of Genomic Medicine **Credits: 3**

Unit No.	Content	Lectures
1.	Introduction to Genomic Medicine, Genomic Counselling and Genomic Medicine	10
2.	Genomics and Society, Genomic Medicine principles and practice, Genomics and Clinical Medicine	13
3.	Genomics and Health in developing world, Genetic disorders of the Indian Subcontinent	12
4.	Population Aspects of Genetic Counselling and Genetic Screening	05
5.	Interpretation of genomic data, application of genomics to clinical practice, Communicating genomic information to patients	05
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Ginsburg, G. S. and Willard, H. F. (2009). Genomic and personalized medicine: foundations and applications. *Translational Research* 154(6): 277-287.
2.	Zlotogora, J. (2009). Population programs for the detection of couples at risk for severe monogenetic diseases. *Hum Genet* 126: 247-53.

MGC.515: Clinical Genetics & Management of Metabolic Disease **Credits:3**

Unit No.	Content	Lectures
1	Inborn Errors of Metabolism-New Born Screening programme	15
2.	Focus on biochemistry of genetic disorders resulting in metabolic problems with the processing and storage of molecules, Lysosomal storage disorders, amino acid metabolism, disorders of carbohydrate metabolism, disorders affecting glycosylation, purine and pyrimidine metabolism, lipoprotein and lipid metabolism	15
3.	Common relevant syndromes	15
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Pourfarzam, M. and Zadhoush, F. (2013). Newborn Screening for inherited metabolic disorders; news and views. *J Res Med Sci* 18(9):801–8. PMCID: PMC3872591.
2.	Mak, C.M., Lee, H.C., Chan, A.Y. and Lam, C.W. (2013). Inborn errors of metabolism and expanded newborn screening: review and update. *Crit Rev Clin Lab Sci* 50(6):142-62. doi: 10.3109/10408363.2013.847896. PMID: 24295058.

MGC.516: Cancer Genetic Counselling **Credits:3**

Unit No.	Content	Lectures
1.	Basics and types of cancer, Origin and classification of cancer, Genetic basis of cancer, Role of tumour suppressors and oncogenes in cancer, Role of microRNAs in cancer.	12
2.	Genetics and inheritance of hereditary cancers, neoplasia syndromes, evaluation of personal and family cancer history, Cancer risk assessment process- review medical/pathology records	10
3.	Assess eligibility for genetic testing, provide cancer risk assessment counselling for patients and their family members, Relevant genetic testing for a variety of hereditary cancer syndromes	10
4.	Psychosocial aspects of counselling patients with or at risk for cancer, in adult and paediatric setting	08
5.	Somatic testing for Cancer therapy	05
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Peng,Y. and Croce, C. M. (2016). The role of MicroRNAs in human cancer. *Signal transduction and targeted therapy* 1(1):1-9.
2.	Zeng, C., Bastarache, L. A., Tao, R., Venner, E., Hebbring, S., Andujar, J. D. and Denny, J. C. (2022). Association of pathogenic variants in hereditary cancer genes with multiple diseases. *JAMA oncology* 8(6):835-844.
3.	Riley, B. D., Culver, J. O., Skrzynia, C., Senter, L. A., Peters, J. A., Costalas, J. W. and Trepanier, A. M. (2012). Essential elements of genetic cancer risk assessment, counselling, and testing: updated recommendations of the National Society of Genetic Counsellors. *Journal of Genetic Counselling* 21:151-161.
4.	American Society of Clinical Oncology. (2003). American Society of Clinical Oncology policy statement update: genetic testing for cancer susceptibility. *Journal of clinical oncology: official journal of the American Society of Clinical Oncology* 21(12):2397-2406.

MGC.517: Translational Genomics **Credits:3**

Unit No.	Content	Lectures
1.	Genomic variant analysis and Clinical interpretation	15
2.	ACMG-ACP Guidelines for annotation 2021, Database familiarization for help in diagnosis, genotype/phenotype correlation	09
3.	Protein sequence analyses, protein microarray, protein modifications and protein- protein interaction analyses, Computational analyses for proteins identification and their function, Protein biomarkers for disease diagnosis, Proteomics in drug discovery.	10
4.	Clinical Application of Genome Editing	05
5.	CAR-T Therapy (Chimeric Antigen Receptor therapy)	04
6.	Nutrigenetics to Precision Nutrition	02
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Li, Q. and Wang, K. (2017). InterVar: clinical interpretation of genetic variants by the 2015 ACMG-AMP guidelines. *The American Journal of Human Genetics* 100(2):267-280.
2.	Yu, X., Schneiderhan-Marra, N. and Joos, T.O. (2010). Protein microarrays for personalized medicine. *Clinical Chemistry* 56(3):376-387.
3.	Schubert, M. L., Schmitt, M., Wang, L., Ramos, C. A., Jordan, K., Müller-Tidow, C. and Dreger, P. (2021). Side-effect management of chimeric antigen receptor (CAR) T-cell therapy. *Annals of Oncology* 32(1):34-48.
4.	Bordoni, L. and Gabbianelli, R. (2019). Primers on nutrigenetics and nutri (epi) genomics: Origins and development of precision nutrition. *Biochimie* 160:156-171.

MGC.518: Ethical, Legal and Social Issues in Genetic Counselling **Credits:3**

Unit No	Content	Lectures
1.	Ethical principles for healthcare practice, Human rights (including foetus), Genetic Laws and Guidelines, Genome Policy	8
2.	Cultural Competence, Impact of illness/ or disability on the individual, family and society, Insurance, employment and discrimination issues related to genetic conditions	8
3.	Quality control in Clinical Practice, Quality control in Genetic Testing, Regulation of Genetic Testing, Direct-to-Consumer testing	8
4.	Biological Data storage, access and sharing policy of India (2019)	8
	Total	**45**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Taylor, A. L. (1999). Globalization and biotechnology: UNESCO and an international strategy to advance human rights and public health. *American Journal of Law & Medicine* 25(4):479-541.
2.	Badzek, L., Henaghan, M., Turner, M. and Monsen, R. (2013). Ethical, legal, and social issues in the translation of genomics into health care. *Journal of Nursing Scholarship* 45(1):15-24.

MGC.519: Regulations in Human Genome Research **Credits:2**

Unit No.	Content	Lectures
1.	Overview of Human Genome Editing and Overarching Principles for Governance	06
2.	Rules Governing Laboratory Research on Human Gametes and Embryos	05
3.	Universal Declaration on the Human Genome and Human Rights	06
4.	Science and Bioethics of CRISPR-Cas9 Gene Editing, Outlining the Ethical Questions on CRISPR-Cas9 for Gene Editing, WHO issues new recommendations on human genome editing for the advancement of public health	05
5.	National policy for genome research, Regulation of Emerging gene technologies in India, National Guidelines for gene therapy product development and Clinical trials	10
	Total	**32**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Charo, R.A., Hynes, R.O., Beier, D.W. *et al*. (2017). *Human genome editing: science, ethics, and governance*. National Academies of Sciences, Engineering, and Medicine. National Academies Press. 2nd edition.
2.	Cribbs, A.P. and Perera, S.M.W. (2017). Science and Bioethics of CRISPR-Cas9 Gene Editing: An Analysis Towards Separating Facts and Fiction. *Yale J Biol Med*. 90(4):625-634. PMID: 29259526; PMCID: PMC5733851.
3.	Sudbery, P. and Sudbery I (2009). *Human Molecular Genetics*. Pearson Education, UK, 3rd Edition.
4.	Watson, J.D., Myers, R.M., Caudy, A.A. and Witkowski, J.A. (2007). *Recombinant DNA Genes and Genomes – A Short Course*. Cold Spring Harbor Laboratory Press, 3rd Edition.

MGC-520: Bioethics and Intellectual Property Rights **Credits: 2**

Unit No.	Content	Lectures
1.	Introduction to Bioethics, Bioethical issues related to Healthcare &Medicine	02
2.	Bioethics- Definition, historical aspects, types and scope, rights-based ethics theories, Duty based ethics theories and utilitarian ethics, basic concepts & principles of autonomy, non-maleficence, beneficence and justice	05
3.	Animal ethics, Health policy privacy, Ethical dilemmas: Contextual vignettes in medical genetic advances- Genetic screening, gene editing, genome ownership, genetic discrimination, genetic insurance, genetic privacy	05
4.	Prudence of investigation confidentiality, Patients' bill of rights, Disposal of investigative material, Integrity, Blood transfusion	05
5.	Rational drug prescribing, Clinical trials, Risk minimization, Animal ethics, Drugs & Cosmetics Act, The Patients Act, The MTP Act, The prenatal diagnostic techniques Act, The transplantation of Human Organs Act, Brain death & organ retrieval	05
6.	Medicolegal aspects of medical records	02
7.	Introduction to Intellectual Property: Concept of Intellectual Property, Kinds of Intellectual Property Patents, Copyrights Designs, Trademarks, Geographical Indication, Infringement of IPR, Its protection and Remedies Licensing and its types	08
	Total	**32**

Suggested Books/Additional reading material:

Sr. No.	Authors/ Name of Books/Publisher
1.	Beauchamp, T.L., Walters, L., Kahn, J.P. and Mastrianni, A.C. (2013). *Contemporary Issues in Bioethics*, Wadsworth Publishing Co. 8th edition.
2.	Classic Philosophical Questions by Gloud (2008) *Case book series and booklets by UNESCO Bioethics Core Curriculum. Program and meeting document.* 8th edition. Encyclopaedia of Bioethics 5 vol set, (2003) ISBN-10: 0028657748
3.	Ganguli, P. (2001). *Intellectual property rights* Ganguli-Tata McGraw Hill. ISBN-10: 0074638602.

Teaching Activities

- **Departmental Seminar**

  A monthly educational seminar is sponsored by the Department of Genetics. Presenters include invited guest speakers, departmental faculty, fellows and students. Genetic counselling students are required to present a seminar related to their research project during the second year of training.

- **Community Outreach**

  Students have the opportunity to participate in invited talks to local schools and community groups.

- **Journal Club**

  Participants discuss genetic counselling and clinical genetics topics of interest. Genetic counselling students are required to select, present and lead discussion about one current journal article at a journal club during their second year.

- **Syndrome Review**

  Once or twice each month, students attend review lectures on various genetic syndromes. During each meeting, two students provide slide presentations on assigned syndromes.