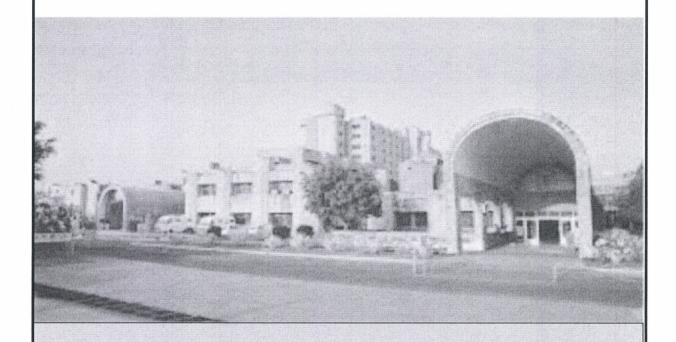
Global GREEN
Regional BLUE
National ORANGE
Local (State)





# Curriculum M.Sc. Genetic Counseling

Department of Medical Genetics
Sanjay Gandhi Postgraduate Institute of Medical Sciences,
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Lt Col Val
Execution

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#### **PREAMBLE**

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The last two decades have seen enormous advances in our understanding of human health and diseases and the contribution of genetic factors in determining normal homeostasis. Not only the technology, but the emergence of medical genetics as a clinical specialty has created the need for good support from laboratory and genetic counseling services. There is a need to create a large trained and competent manpower in India to translate the advances in genetics to healthcare and patient services. Specialty of Medical Genetics is unique in its way as compared to other clinical specialties as counseling is an important part of patient care. Genetic counseling is a psychoeducational process involving communication of complex scientific information as applicable to the patient/ family specific situation. This involves time; interpretation of family and medical histories and genetic test results; communication skills to educate about the implications of test results, genetic concepts, management and preventive measures, etc. The process also involves many psychosocial issues including informed choices and adaptation to the risk and/or conditions. Limited number of training centers and trained clinical geneticists make it difficult to optimally address all these aspects. Hence a well-trained paramedical person can be of great utility in the clinic for the clinical geneticist and patients and families by devoting more time for counseling. United States of America and Europe have regular programs in genetic counseling for over 2 decades now. Medical genetics is a well-established specialty in India though the number of trained medical geneticists is still limited. Genetic counselors are essential part of clinical genetics services and for a large country like India we need more service providers in medical genetics. India, with its huge population, only has very few programs in genetic counseling. This calls for an initiative to develop a training program in genetic counseling at our centre, the first in India to start the DM Medical Genetics course.

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#### INFORMATION ABOUT THE COURSE

# 1. Title

Master of Science (M.Sc.) in Genetic Counseling

# 2. Need and Scope: Objectives

The program aims to:

- Provide academic and vocational training to become professional genetic counselors
- Teach the students the knowledge of basic genetics, genetic diagnostics as applicable in clinical situations and genetic counseling
- Provide training in principles and skills of genetic counseling
- Equip students with the necessary skills to enable them to contribute to the research and development of the profession
- Promote awareness on the importance of the psychosocial impact of human genetic technology among patients, health and social care professionals and the wider society
- To provide necessary training to pursue research (PhD) in genetics-related area after obtaining the degree

#### 3. Definitions

#### a. Genetic Counseling

Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates:

i. Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.

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- ii. Education about inheritance, testing, management, prevention, resources and research.
- iii. Counseling to promote informed choices and adaptation to the risk or condition.

(A new definition of Genetic Counseling: National Society of Genetic Counselors' Task Force report. National Society of Genetic Counselors' Definition Task Force, Resta R, Biesecker BB, Bennett RL, Blum S, Hahn SE, Strecker MN, Williams JL.J Genet Couns. 2006 Apr;15(2):77-83).

#### b. Genetic Counselor

Genetic counselors are health professionals with specialized graduate degrees and experience in the areas of medical genetics and counseling. Most enter the field from a variety of disciplines, including biology, genetics, nursing, psychology, public health, and social work. They work in close association and supervision of clinical geneticists and / or clinicians to enhance the genetic counseling process. Genetic counselors work as members of a health care team, providing information and support to families who have members with birth defects or genetic disorders

and support to families who have members with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions. They identify families at-risk, investigate the problem present in the family, interpret information about the disorder, analyze inheritance patterns and risks of recurrence and review available options with the family.

Genetic counselors also provide supportive counseling to families, serve as patient advocates and refer individuals and families to community or state support services. They serve as educators and resource people for other health care professionals and for the general public. Some counselors also work in administrative capacities. Many engage in research activities related to the field of medical genetics and genetic counseling. (Adopted by the National Society of Genetic Counselors, Inc. 1983)

# 4. Expected outcome of the program: Scope of practice

a. Clinical Genetics: The trained genetic counselor will work under supervision and in close collaboration with Medical Geneticist / Clinical Geneticist or a

clinician taking care of patients and families with genetic disorders. He/she will not examine the patient, order laboratory investigations or counsel the patient independently without the advice, order or supervision of the medical practitioner as mentioned above. The genetic counselor's scope of practice will include the following:

- i. Explain the nature of genetics evaluation to clients, obtain and review medical and family histories, based on the referral indication, and document the family history using standard pedigree symbols.
- ii. Identify additional client and family medical information relevant to risk assessment and consideration of differential diagnoses, and assist in obtaining such information.
- iii. Research and summarize pertinent data from the published literature, databases, and other professional resources, as necessary for each client.
- iv. Synthesize client and family medical information and data obtained from additional research as the basis for risk assessment, differential diagnosis, genetic testing options, reproductive options, follow-up recommendations, and case management.
- v. Assess the risk of occurrence or recurrence of a genetic condition or birth defect, using a variety of techniques, including knowledge of inheritance patterns, epidemiologic data, quantitative genetics principles, statistical models, and evaluation of clinical information, as applicable.
- vi. Explain to clients, verbally and/or in writing, medical information regarding the diagnosis or potential occurrence of a genetic condition or birth defect, including etiology, natural history, inheritance, disease management and potential treatment options.
- vii. Discuss available options and delineate the risks, benefits and limitations of appropriate tests and clinical assessments.
- viii. Document case information clearly and concisely in the medical record and in correspondence to referring physicians, and discuss case information with other members of the healthcare team, as necessary.

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ix. Assist clients in evaluating the risks, benefits and limitations of participation in research, and facilitate the informed consent process.

x. Identify and access local, regional, and national resources such as support groups and ancillary services; discuss the availability of such resources with clients; and provide referrals, as necessary.

xi. Plan, organize and conduct public and professional education programs on medical genetics, patient care and genetic counseling issues.

# b. Counseling and Communication

 Develop a genetic counseling agenda with the client(s) that includes identification and negotiation of client/counselor priorities and expectations.

 Identify individual client and family experiences, behaviors, emotions, perceptions, values, and cultural and religious beliefs in order to facilitate individualized decision making and coping.

iii. Assess client understanding and response to medical information and its implications, and educate client appropriately.

iv. Interpret reports and explain to the patient / consultand.

v. Utilize appropriate interviewing techniques and empathic listening to establish rapport, identify major concerns and engage clients in an exploration of their responses to the implications of the findings, genetic risks, and available options/interventions.

vi. Identify the client's psychological needs, stressors and sources of emotional and psychological support in order to determine appropriate interventions and/or referrals.

vii. Promote client-specific decision making in an unbiased non-coercive manner that respects the client's culture, language, traditions, lifestyle, religious beliefs and values.

viii. Use knowledge of psychological structure to apply client-centered techniques and family systems theory to facilitate adjustment to the occurrence or risk of occurrence of a congenital or genetic disorder.

#### c. Professional Ethics and Values

- Recognize and respond to ethical and moral dilemmas arising in practice, identify factors that promote or hinder client autonomy, and understand issues surrounding privacy, informed consent, confidentiality, real or potential discrimination and potential conflicts of interest.
- ii. Advocate for clients, which includes understanding client needs and perceptions, representing their interests in accessing services, and eliciting responses from the medical and social service systems as well as the community at large.
- iii. Recognize personal limitations in knowledge and/or capabilities and seek consultation or appropriately refer clients to other providers.
- iv. Maintain professional growth, which includes acquiring relevant information required for a given situation, keeping abreast of current standards of practice as well as societal developments, and seeking out or establishing mechanisms for peer support.
- v. Respect a client's right to confidentiality, being mindful of local, state and federal regulations governing release of personal health information.

#### 5. Outcome: Prospects and placement

India with its huge population and burden of genetic disorders does not have any formal university approved genetic counseling program. With the advances in the field of medical genetics, a lot of hospitals are expected to provide genetic counseling services to the patients and the society. The program will also train the candidates in research and they will have the option of pursuing higher qualifications like PhD in Medical/Human Genetics. The trained candidate/will be

able to provide genetic counseling for common genetic conditions under the guidance of a medical geneticist: Down syndrome, Duchenne muscular dystrophy, thalassemia, spinal muscular atrophy, amniocentesis, chorionic villus sampling and genetic testing. The other areas include (not limited to) registries, research grants, clinical trials, public health programs like population based screening for common disorders, newborn screening and industrial placements. The genetic counselors will also be able to work in departments of obstetrics, neurology, cardiology, oncology etc. as genetic disorders account for a significant number of disorders in any medical specialty and genetic counseling is an important part of the management of patients and families with genetic disorders.

The trained candidates will also be tutors / trainers for M Sc (genetic counseling) program.

#### 6. Duration

The MSc courses have 4 semesters with 1.5 years didactic and practical followed by 6 months of Research project/Dissertation. The course curriculum and duration of the courses in hours is provided with respective courses in the 'Courses' section of the website <a href="https://www.cmtsgpgi.org">www.cmtsgpgi.org</a>.

The entire curriculum is split into semesters of 6 months duration each; and semester examinations will be performed on a 6 monthly basis.

The maximum permissible duration of courses is - 4 years

# 7. Eligibility for Admission

- i) The maximum age shall be 30 years as prescribed in the advertisement for admission to the course. Any age relaxation shall be as per State Government rules.
- Bachelor's degree in Nursing or Human Genetics or Human Biology or Biochemistry or Medical Biotechnology or Molecular Biology or Life sciences, Botany, Zoology, Biomedical science or Radiography & Imaging Technology or Medical Laboratory Technology or Public Health or M B B S / BDS.
- iii) Candidates passing B.Sc. through correspondence course shall not be eligible.

iv) Candidates with degree in alternative / traditional medicine like Ayurveda, Homeopathy, Unani, BAMS, BHMS will not be eligible.

# 8. Method of Selection (As per common CMT rules)

Selection of the candidates shall be strictly on the basis of merit obtained in the entrance examination of the Institute. The entrance examination will be conducted in English language. The scheme of entrance examination will be as under:

- (i) The general standards of entrance examination will be that of B.Sc. or an equivalent examination of an Indian University. The pattern of the examination will be based on the qualifying degree standard and comprises of Biology, chemistry, Physics/ Mathematics and general English.
- (ii) Candidate should secure a minimum of 50% marks (percentile) in entrance examination.
- (iii) In case of two or more candidates belonging to the same category obtaining equal marks in the entrance examination, their merit will be determined in order of preference as under:
  - a. Candidate older in age shall be preferred
  - b. Candidate obtaining higher marks in qualifying examination shall be preferred.
- (iv) Selection of the students by the Entrance examination conducted by the Institute, will be followed by or counselling/ Interview as per Institute norms. Candidates attending the interview cum seat counselling need to bring all original documents for verification and come prepared for fee payment and admission which will be done immediately as described below
- (v) The Interview Board/Counselling Committee will comprise of the Principal/ Nodal Officer/ Head / Course co-ordinator/nominee of the Department as approved by the Head of the Institute, whose recommendations shall be final for the selection of the students.
- (vi)During subsequent counseling (s) the seat will be allotted as per the merit of the candidate depending on the availability of seats on that particular day.

Candidate who fails to complete the admission formalities, on the notified date(s) will forfeit the claim for admission and placement in the waiting list except if permitted by the competent authority under special circumstances.

# 9. Course Fee

Annual Fees for the course will be as per CMT norms and is subject to change as per decision of the Institute from time to time:

Sl.No.	Items	1st year Ins	tallment	2 <sup>n</sup> year instalment Rs.		
		Rs.				
		1st	2nd	3rd	4th	
1.	Admission fees	5000				
2.	Course fees	35000	35000	35000	35000	
3.	Examination fees	+2500	2500	+2500	2500	
4.	Enrollment fees	500				
5.	Degree/Certificate/fees		300		300	
6.	Migration Certificate fee				300	
7.	Caution Money (Refundable)	10000				
8.	Library Fees	250	250	250	250	
9.	Hostel fees (excluding mess charges) *	3000	3000	3000	3000	
	Total (excluding Hostel	53,250	38050	37,750	38,350	
	fees)					

# 10. Total Number of Seats

4 candidates will be admitted in each academic year.

# 11. Hostel Facility

Hostel will be provided to the students in the campus, if available.

# 12. Internship

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Lt Col Varun Bajpai vSM Executive Registrar SGPGIMS,Lucknow All candidates after successful completion of the course will undergo 180 days of compulsory internship in the department before the award of the degree with at least 90% attendance (excluding the holidays).

Interns will be paid a stipend of Rs.8000/- per month (fixed) during the internship (subject to approval by competent authority).

#### 13. Academic Year

Academic session will start from August / September month of every year in accordance to the rules and regulation setup by the CMT, SGPGIMS, Lucknow.

Academic Year is defined as two consecutive (one odd + one even) semesters.

Sessions / Exams	Tentative schedule**
Start of session	August-September
Classes of odd semester papers (1st & 3rd) will be held during	Sep, Oct, Nov, Dec
Internal assessment of 1 <sup>st</sup> & 3 <sup>rd</sup> semester	Nov-Dec
Timing for conducting odd semester (1st & 3rd) final exams	Jan
Classes of even semester papers (2 <sup>nd</sup> & 4 <sup>th</sup> ) will be held during	Feb, Mar, Apr, May,
	June
Internal assessment of 2 <sup>nd</sup> & 4 <sup>th</sup> semester	May-June
Timing for conducting even semester (2 <sup>nd</sup> & 4 <sup>th</sup> ) final exams	July-August

<sup>\*\*</sup> will be dictated by the timing of NEET examination/year

#### 14. Medium Of Instruction

English shall be the medium of instruction for the subjects of study as well as for the examination including entrance examination.

# 15. Teaching Faculty

- i. Faculty, Concerned Departments of SGPGIMS, Lucknow
- ii. Faculty, College of Medical Technology, SGPGIMS, Lucknow
- iii. Faculty working in other departments of SGPGIMS, Lucknow
- iv. Guest faculty as and when required for specialized teaching.
- v. Adjunct faculty as and when required for specialized teaching
- vi. Structured Practical classes/ demonstration may also be taken by Senior residents/ laboratory

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vii. Technicians (minimum BSc with 1 year working experience/Diploma with 3 years' experience) in respective departments of SGPGIMS, only after approval by competent authority.

#### 16. Course Structure

a) The student will attend Out Patient Department and work with clinical geneticists to learn pedigree drawing, evaluation of genetic aspects of the case and learn / participate in genetic counseling under supervision.

The student will be posted in laboratories to observe and understand the principles of genetic tests, report preparation, data entry. On non-OPD days the student will review the literature for case evaluation, prepare case summaries and work up for genetic counseling sessions. The students will also attend the morning academic sessions of the Department.

**Table 1: Semester wise Credit distribution:** 

#### Semester I

Paper	Subject	Hours/v	veek			Maximum	marks	
		Lectur	Tutoria	Clinical/l	Cred	Internal	Extern	Tot
		es (1	ls	ab work	it	assessm	al	al
		hour	(1hour	(2		ent	exam	
		=1	=1	hours=1				
		credit)	credit)	credit)				
MGC1	Introduction	2	1	2	4	30	70	100
01	to Medical							
	Genetics				:			
MGC1	Cytogenetics	2	1	2	4	30	70	100
02	and							
	Chromosom							
	al Disorders							
MGC1	Principles of	2	1	18	12	30	70	100
03	Genetic							
	Counseling							
	and							
	Communicat							
	ion Skills							
Total		6	3	22	20	90	210	300

#### Semester II

n	G 1	77 / 1	34 ' 1
Paper	Subject	Hours/week	Maximum marks

		Lectur	Tutoria	Clinical/l	Cred	Internal	Extern	Tot
		es	ls	ab work	it	assessme	al	al
						nt	exam	
MGC1 04	Common Presentatio ns in Clinical Genetics	2	1	10	8	30	70	100
MGC1 05	Basic Genetics & Genetic Counselin	2	1.	6	6	30	70	100
MGC1 06	Molecular Genetics	2	1	6	6	30	70	100
Total		6	3	22	20	90	210	300

# Semester III

Paper	Subject	Hours/w	reek			Maximum	marks	
		Lectur es	Tutoria ls	Clinical/l ab work	Cred it	Internal assessme nt	Extern al exam	Tota l
MGC20	Genetic disorder s – Part I	2	1	10	8	30	70	100
MGC20 2	Prenatal Genetic s & Fetal Medicin e	2	1	6	6	30	70	100
MGC20 3	Genetic Disorde rs Part II	2	1	6	6	30	70	100
Total		6	3	22	20	90	210	300

# Semester IV

Paper	Subject	Hours/week				Maximum marks		
		Lectur es	Tutori als	Clinical/ lab work	Cred it	Internal assessm ent	Extern al exam	Tot al
MGC2 04	Genetic disorders – Part III	2	1	10	8	30	70	100

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MGC2	Genomic	2	1	6	4	30	70	100
05	medicine and							
	Research							
MGC2	Preventive	2	1	6	4	30	70	100
06	Genetics							
	Dissertation/T	-	•	-	4	-	-	-
	hesis							
Total		6	3	22	20	90	210	300

• Dissertation/Project should be done concurrent with academic, lab and patientrelated work and should be completed by early part of second session of second year

• <u>Course</u>—wise objectives and outcomes are given in Annexure I and Detailed curriculum in Annexure II

# b) Organization and scheduling of the course:

**Postings in laboratories:** The candidates will be made to undergo posting in laboratories to observe cytogenetics, molecular techniques and biochemistry as necessary.

**Patient care, teaching and research:** The candidate is expected to participate in evaluation, testing and counseling for genetic diseases in OPDs; make ethics committee submissions, interact/liaison with ethics committees and any agency involved in research projects undertaken in the department as and when needed, fill case record forms, and participate in audits and inspections.

*Case summaries*: Each candidate is expected to write 50 case summaries illustrating all the issues involved in genetic counseling covering the common genetic conditions.

Suggested cases/families and conditions for training: Down syndrome (5), Duchenne muscular dystrophy (5), spinal muscular atrophy (5), thalassemia (5), amniocentesis (5), chorionic villus sampling (5), familial cancers (5), recurrent pregnancy loss (5), neural tube defects (5) and fetal anomalies detected by antenatal ultrasound (5). Exposure to counseling for teratogen exposure, fetal autopsy, diagnostic testing (pre and post-test counseling),

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research testing, consanguinity, preconception counseling, newborn screening and presymptomatic screening are encouraged.

**Development of patient counseling/education material:** The candidates must prepare patient education or counseling aids (brochures or pamphlets) for at least two common genetic conditions under the close supervision of the guide or teacher.

#### Seminars and Case Discussion

Students enrolled in M.Sc. Genetic Counseling course shall have to deliver Seminars on a recent innovative topics of Medical Genetics. Seminar will be of 45-minutes duration followed by questions session of 15 minute by the audience comprising of faculty and students. The speaker has to write an abstract to be distributed during Seminar in addition to two copies of write-up giving relevant details of the background of the subject, methods used and references/sources of collection of presentation material.

Clinical case presentations with stress on genetic counseling will also be mandatory for the students.

#### 17. Syllabus

Course curriculum and syllabi for the course shall be as prescribed by the Academic Board of the Institute from time to time. However, to start with, a detailed and comprehensive syllabus in this regard is being annexed herewith (Annexure-1).

# 18. Equipments for Training

Students will be provided exposure/ demonstration of laboratory techniques used in genetic diagnosis and research. They will be encouraged to observe preparation and interpretation of reports.

# 19. Method of Examination & Evaluation (As per CMT rules)

The examination of all the courses required for the M.Sc. Degree shall be consisting of Continuous Internal Assessment and End-Semester Examination. The Continuous Internal Assessment and the End-Semester Examination shall have the weightage of 30% and 70%, respectively

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Examinations will be conducted as Internal assessments and External Assessment/ Semester examination, as defined below

# (i) Internal Assessment (IA)

One internal assessment will be conducted in each semester after completion of about 50-75% of syllabus and at least a month or more before the semester examination. Weightage of the Internal assessment will be 30% of the total marks of the subject in the final paper. The Institute will conduct internal assessment exams based on timeline set by the College of medical technology, SGPGIMS. Internal Assessment will be conducted at the level of course coordinator. A student who fails to appear in an internal assessment examination due to valid reason will be allowed one more chance to take the same examination.

Table-2: Internal Assessment marks distribution

Paper	Subject Title		Maximum Marks
		Theory	Practical + Log book
	Semester	I	· .
MGC-101	Introduction to Medical Genetics	20	Case evaluation I- 10
MGC -102	Cytogenetics and Chromosomal	20	Laboratory analysis, Report
	Disorders		interpretation I - 10
MGC -103	Principles of Genetic Counseling	20	Genetic counseling case I-10
	and Communication Skills		
***************************************	Semester	II	
MGC -104	Common Presentations in Clinical	20	Case evaluation II - 10
	Genetics		
MGC-105	Basic Genetics & Genetic	20	Genetic counseling case II-10
	Counseling		
MGC-106	Molecular Genetics	20	Laboratory analysis, Report
			interpretation II - 10

	Semester III							
MGC-201	Genetic disorders – Part I	20	Case evaluation I- 10					

MGC -202	Prenatal Genetics & Fetal Medicine	20	Laboratory analysis, Report
		interpretation I - 10	
MGC -203	Genetic Disorders Part II	20	Genetic counseling case I-10
	Semester	IV	\$1.00 Text \$1.00 text \$1.00 text
MGC -204	Genetic disorders – Part III	20	Case evaluation II - 10
MGC -205	Genomic medicine and Research	20	Genetic counseling case II-10
MGC -206	Preventive Genetics	20	Laboratory analysis, Report
			interpretation II – 10

Note: The practical examination will include:

Clinical case evaluation -2 cases, cases for genetic counseling -2 cases and laboratory experiments- 2.

A student must secure at least 50% of total marks fixed for internal assessment for a particular subject in order to be eligible to appear in examination in that subject. The internal assessment marks obtained by the students will be added to the total marks of annual exam of the course.

# ii) Work Diary/Log Book

Every candidate shall attend symposia, seminars, conferences, journal review meetings & lectures as prescribed by the department. Every candidate shall maintain a work diary and record of his/her participation in the training program. The work diary and record shall be scrutinized and certified by the concerned faculty members. Internal assessment based on log book will be done each year carrying 30 marks. The log book shall be scrutinized and certified by the concerned faculty and course coordinator and will be submitted atleast 01 month before the final examination.

# iii) Annual / semester Examination (Theory & Practical) (As per CMT rules) External Assessment/ Semester Examination

There shall be a 6 monthly semester exam/external examination at the end of each semester in the form of theory and practical exam. The candidate shall be required to appear in every paper of the semester as specified in the course.

A standard model format of the End-Semester Examination paper consisting of 70 marks shall be as following,

Long Answers (05), 10 marks each – to attempt any 4

Short Answers (08), 5 marks each - to attempt any 6

Table -3: Subjects and Distribution of Marks for Semester Exams

Paper	Subject	Maximum Marks					
		Theory	Practical +Viva	Internal Assessment	Total		
	S	emester ]					
MGC-101	Introduction to Medical Genetics	50	Case evaluation I-20	30	100		
MGC -102	Cytogenetics and Chromosomal Disorders	50	Laboratory analysis, Report interpretation I - 20	30	100		
MGC -103	Principles of Genetic Counseling and Communication Skills	50	Genetic counseling cases I-20	30	100		
	Se	emester I	I				
MGC -104	Common Presentations in Clinical Genetics	50	Case evaluation II-20	30	100		
MGC-105	Basic Genetics & Genetic Counseling	50	Genetic counseling cases II-20	30	100		
MGC-106	Molecular Genetics	50	Laboratory analysis, Report interpretation II - 20	30	100		

Semester III					
MGC-201	Genetic disorders – Part I	50	Case	30	100
			evaluation I-20		
MGC -202	Prenatal Genetics & Fetal	50	Laboratory	30	100
	Medicine		analysis,		
		,	Report		
			interpretation I		
			- 20		
MGC -203	Genetic Disorders Part II	50	Genetic	30	100
			counseling		
			cases I-20		
	Se	mester IV	7		
MGC -204	Genetic disorders - Part	50	Case	30	100
	III		evaluation II-		
			20		

MGC -205	Genomic medicine and	50	Laboratory	30	100
	Research		analysis,		
		ė	Report	ej.	
			interpretation	a a	
			II - 20		
MGC -206	Preventive Genetics	50	Genetic	30	100
			counseling		
			cases II-20		

Note: The practical examination will include:

Clinical case evaluation -2 cases, cases for genetic counseling -2 cases and Interpretation of laboratory tests -2.

#### iv) Thesis / Dissertation

- a) Each candidate pursuing M.Sc. Genetic counseling Course is required to carry out a dissertation work on a selected topic under the guidance of a recognized post graduate teacher. The completed thesis/dissertation should be submitted in the first session of second year. The dissertation is aimed to train the candidate in research methods and techniques. It includes identification of problem, formulation of hypothesis, search and review of literature, getting acquainted with recent advances, collection of data, analysis of data, interpretation of results and drawing conclusions.
- b) Every candidate shall submit two hard copies of synopsis containing particulars of proposed dissertation work in the second session of first year. The synopsis in prescribed proforma shall be sent through proper channel to the Head, Department of Medical Genetics.
- c) The Departmental Committee shall review the synopsis and if found suitable, shall register the dissertation topic. No change in the dissertation topic or guide shall be made without prior approval of the Departmental Committee.
- d) The dissertation shall be written under the following headings:
  - Introduction
  - Aims or objectives
  - Review of literature

- Materials and methods
- Results
- Discussion
- Conclusion
- Summary
- References
- Tables
- Annexure
- e) The dissertation text shall not be less than 50 pages and shall not exceed 100 pages excluding references, tables, questionnaires and other annexure. It should be neatly typed in double spacing on one side of A4 size paper and hard bound properly (spiral binding not acceptable). A declaration by the candidate that the work was done by him/her shall be included. The guide and head of the department shall certify the bonafide of the dissertation. f) Four copies of dissertation shall be submitted to the Institute through proper channel along with a soft copy on CD not later than beginning of second session of second year. It shall be assessed by two examiners appointed by the Institute (one internal and one external). No marks shall be awarded for dissertation but the Degree will only be awarded when the submitted dissertation is found to be satisfactory by the institute.

# g) Guide for Dissertation

Guide of a student shall be a faculty in the department of Medical Genetics of SGPGIMS. The student- guide ratio will be 2:1 i.e. a guide shall supervise dissertation work of not more than 02 students per academic year.

- h) Viva Voice Examination after submission of dissertation
- Those candidates who complete the dissertation will be called for a viva voce examination.
- The viva voce examination will be conducted by the Board of Examiners. The Board will consist of the Course co-ordinators and or other Faculty members of the department, and one external examiner.
- If a candidate fails to satisfy the examiners at the viva voce examination, the Board of examiners may recommend that the candidate be permitted to present the same dissertation again and submit to a further viva voce examination within a period of 1 month as intimated by the Institute.

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 He/she will be awarded degree only after passing all semesters, as defined, and satisfactory completion of the dissertation.

# 20. Attendance (As per CMT rules)

i)To be eligible to appear at an examination, a student must have satisfied the following conditions. A candidate must secure minimum 80% physical attendance in overall, with at least -

a.75% attendance in theoretical

b.80% in Skills training (practical) for qualifying to appear for the final examination.

ii)Special classes conducted for any purpose shall not be considered for the calculation of percentage of attendance.

- iii)Condonation for Attendance
- a. Condonation may be granted by the Principal/Nodal Officer to the extent of 10% in exceptional cases i.e. serious illness & hospitalization, accident, mishap in the family or deputation by the college for any specific work for which the period of his/her absence shall not be counted towards the calculation of attendance on the condition that students concerned submit a certificate to that effect from the appropriate authority.
- b. There is no clause of Distance Education/Correspondence in any of the MSc courses.

#### 21. Appointment of Examiners (As per CMT rules)

- 1. Convenor Head, Concerned Department, SGPGIMS, Lucknow
- Internal examiner Course coordinator of the Concerned Department, SGPGIMS and or Faculty, Concerned Department or Faculty, College of Medical Technology, SGPGIMS, Lucknow.
- 3. External Examiner (01) External examiner Tutor / Lecturer / faculty working in Concerned/ Related Department of a teaching hospital, or paramedical college belonging to the specialty concerned. An external examiner will be from another institute with minimum of 5 years post graduate teaching experience.

#### 22. Promotion and Pass Criteria (as per CMT rules)

A credit-based-grading- system for the purpose of assessment of the students will be performed.

Results will be generated as the as per the UGC guidelines on adoption of choice-based credit system as well as in percentage.

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# Computation of SGPA and CGPA

**Credit:** A unit by which the course work is measured. It determines the number of hours of instructions required per week. One credit is equivalent to one hour of teaching (lecture or tutorial) or two hours of practical work/field work per week.

- a. 1 credit = 1 hour of instruction (lecture/tutorial) per week (1 credit course = 15 contact hours of instruction per semester)
- b. 4 credits = 4 hours of instruction per week (4 credit course = 60 contact hours of instruction per Semester)
- c. 1 credit = 2 hours of laboratory work/field work per week (1 credit course = 30 hours of laboratory work/semester)

Credit Point: It is the product of the grade point and the number of credits for a course. Grade Point: It is a numerical weight allotted to each letter grade on a 10-point scale. Letter Grade: It is an index of the performance of students in a said course. Grades are denoted by letters O, A+, A, B+, B, C, P and F. A letter grade is assigned to a student on the basis of evaluation of her/his performance in a course on a ten point scale as below.

# Table for conversion of percentage into grade points

Letter Grade	Numerical Grade Point	Class Interval (in %)
O (Outstanding)	10	Above 90 and ≤ 100
A+ (Excellent)	9	Above 80 and ≤ 90
A (Very Good)	8	Above 70 and $\leq$ 80
B+ (Good)	7	Above 60 and $\leq$ 70
B (Above Average) **	6	Above 50 and $\leq$ 60
C (Average)	5	Above 45 and $\leq$ 50
P (Pass)	4.5	40 to 45
F (Fail)	0	< 40
Ab (Absent)	0	Absent

<sup>\*\*</sup>as per the recommendations of Ministry of family and health welfare, National Commission for Allied Health Sciences, 50% marks individually in theory and practical in each paper is considered Pass.

Credit Point/score: It is the product of grade point and number of credits for a course (= Grade point x Credits)

(b) SGPA (Semester grade point average) is the ratio of sum of the product of the number of credits with the grade points scored by student in all the paper, the sum of the number of credits of all the courses undergone by a student.

Sum of credit scores of all papers of a semester (grade  $SGPA = \frac{point \ scored \ x \ credits)}{total \ credit \ points \ for \ that \ semester}$ 

- (c) The CGPA (Cumulative Grade Point Average) is also calculated in the same manner taking into account all the courses undergone by a student over all the semesters of the course.
- (d) The SGPA shall be calculated at end of each semester and CGPA shall be calculated after clearing all papers of 1<sup>st</sup> to 4<sup>th</sup> semesters and shall be rounded off to 2 decimal places.
- (e) The following reference for calculating maximum marks and obtained marks for a Programme/Course will be followed -

For Maximum Marks -

- 1 Credit Course = 25 marks course
- 2 Credit Course= 50 marks course
- 3 Credit Course= 75 marks course 4 Credit Course= 100 marks course

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#### Results

Pass criteria – Student must attain at least 50% marks in each Theory, and Practical independently / separately for each individual paper conducted at the end of that semester. The marks obtained in the internal assessment of the paper and semester examination will be added to compute the percentage.

**Promoted Provisionally**— The candidate may be promoted from one academic year to the next if they pass in more than 50% papers of the odd and even semesters taken together in those academic years. However, he/she will have to re-appear for the backlog/failed papers as and when they are conducted by the Institute in the subsequent odd/even semesters.

**Detained** - A candidate will be detained if he/she fails in more than 50% papers of Semester I and Semester II taken together in 1<sup>st</sup> academic year and will have to re-join classes with the junior batch and become eligible/complete attendance for re-appearing in the examinations. He/she will have to pay the tuition fees and examination fees again as per norms.

If a student is not eligible for appearing in any semester exam, either due to attendance shortage, break, non-payment of college fees or exam fees, or inability to fill exam form, then that student will also be treated as detained and will take re-admission in the same academic years with junior batch.

A candidate will be allowed to take these backlog papers in not more than 4 attempts (1<sup>st</sup> + 3). Failing to appear in an examination due to reasons mentioned above will be considered as 1 attempt.

No extra time beyond the stipulated time as mentioned below.

Maximum permissible duration of courses (inclusive of internship) is - 4 years

If he/she fails to complete the course during this period/or fails to clear a paper in the aforementioned 4 attempts, his registration for the course will be terminated.

Carry over benefit / Promoted Provisionally: A candidate shall be promoted provisionally from odd to the even semester ( $1\rightarrow 2$ ,  $3^{rd}\rightarrow 4^{th}$ ) in the first and second academic year, irrespective of number of papers of failure in the respective odd semester.

The candidate will be promoted provisionally to 3<sup>rd</sup> semester, if he/she has cleared more than 50% papers Semester I + II; he/she will be detained in the respective academic year if he/she does not clear more than 50% papers Semester I + II.

- The candidate failing in more than 50% papers of semester III+IV, will be detained and will re-join course with the junior batch and pay the required tuition fees and examination fees as indicated.
- o The candidate failing in semester 3 or 4 need not take classes again, but will have to clear the backlog papers as and when conducted, after paying due examination fees. o The candidate will appear in the failed/backlog odd and even semester papers as and when they are being conducted by the Institute after submitting the examination fees.
- The candidate will be awarded degree only after having passed all semesters and completed dissertation/research project satisfactorily as defined below.

#### **Grace Marks**

A student shall be eligible for grace marks, provided he / she appeared in all the papers of a semester and failed in only one paper in either theory or practical. A maximum of 5 grace marks will be granted in either theory or practical and not both, before declaration of result of that semester.

#### Re-Evaluation and Re-totaling

Re-evaluation and re-totaling shall be allowed *in only theory papers any 02 papers*. The candidate will have to submit an application to the Principal/Nodal Officer for the same within 8 days of the date of the declaration of the results.

Fees as applicable must be submitted as per Institute norms.

In case, on re-evaluation, there is any answer left unchecked, the same will be placed before the Board of Examiners of the respective subjects for evaluation. The information regarding re-addition of marks will be intimated to the candidate within 45 days.

# 23. Award of Degree:

The degree of M.Sc. Genetic counseling will be awarded to the candidates only after he/she completed the following:

- (i) Has passed all the examinations of 1st and 2nd year.
- (ii) Has satisfactorily completed the full period of training and dissertation.
- (iii) His/her work and conduct during the period of training have been satisfactory.
- (iv) Degree will be awarded at the convocation held at the Institute.

#### **24.** Vacation (As per the institute norms)

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Students will observe vacation as under:

(i) Winter break

10 days in December

(ii) Summer break

10 days in June

(iii) Sick leave

10 days

#### 25. Ordinance:

Notwithstanding the integrated nature of this course which is spread over two academic years, the ordinance in force at the time a student joins the course shall hold good only for the examination held during or at the end of the academic year and nothing in this ordinance shall be deemed to debar the Institute from amending the ordinance and the amended ordinance, if any, shall apply to all the students, whether old or new.

#### Annexure-I

#### SYLLABUS OF THE COURSE

# MGC101: Introduction to Medical Genetics

- 1. History of Medical Genetics & Pedigree drawing
- 2. Genetics in clinical practice
- 3. Mendelian Inheritance
- 4. Non Mendelian Inheritance
- 5. Multifactorial Inheritance
- 6. Population Genetics
- 7. Consanguinity and Inbreeding
- 8. Clinical case Preparation
- 9. Genotype and Phenotype
- 10. Human Anatomy and birth defects

# MGC102: Cytogenetics and chromosomal disorders

1. Chromosomal basis of heredity

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- 2. Cell culture, karyotyping and cytogenetic nomenclature
- 3. Chromosomal abnormalities Numerical & structural
- 4. Indications of karyotyping
- 5. Prenatal cytogenetics
- 6. Down syndrome & common aneuploidies
- 7. Sex chromosomal abnormalities
- 8. Phenotypes Structural chromosomal abnormalities [Balanced & Unbalanced]
- 9. Contiguous gene syndromes
- 10. Family with structural chromosomal abnormality: Risk of recurrence

# MGC103: Principles of Genetic Counseling and Communication Skills

- 1. Definition of genetic counseling & indications
- 2. Principles of genetic counseling
- 3. Steps of genetic counseling
- 4. Risk calculations for monogenic and multifactorial disorders
- 5. Counseling for adult onset disorders
- 6. Basics of communication
- 7. Communication of risks
- 8. Giving bad news
- 9. Psychosocial issues
- 10. Evaluation of genetic counseling

# **MGC104: Common Presentations in Clinical Genetics**

- 1. Intellectual disability
- 2. Autism & other developmental disabilities
- 3. Recurrent pregnancy losses & bad obstetric history
- 4. Congenital malformations
- 5. Short stature
- 6. Multiple malformation syndromes

- 7. Regression of milestones
- 8. Disorders of sexual development and differentiation
- 9. Genetics of common disorders
- 10. Common neonatal problems

# MGC105: Basic genetics and Genetic Counseling

- 1. Genetic testing in Children Issues
- 2. Educational aids for patients and family
- 3. Genetic counseling for autosomal dominant disorders
- 4. Genetic counseling for autosomal recessive disorders
- 5. Genetic counseling for X-linked recessive disorders
- 6. Genetic counseling for X-linked dominant and semi-dominant disorders
- 7. Counseling issues in chromosomal disorders
- 8. Counseling in disorders with non- mendelian inheritance
- 9. Legal and social issues in Genetic disorders
- 10. Ethical issues in Genetic disorders

#### **MGC 106: Molecular Genetics**

- 1. Structure of gene and genome
- 2. PCR and sequencing
- 3. Types of Mutations and Effects
- 4. Methods to detect copy number variations
- 5. Nomenclature of DNA variations
- 6. Recombinant DNA technology in diagnostics
- 7. Next generation sequencing: technique and applications
- 8. Guidelines for prediction of pathogenicity
- 9. Bioinformatics and databases
- 10. DNA fingerprinting

# MGC201: Genetic disorders - Part I

- 1. Dysmorphism and related syndromes
- 2. Phenotype nomenclature (HPO)

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- 3. Thalassemia and other hemoglobinopathies
- 4. Genetic disorders of coagulation & bleeding
- 5. Hearing impairment
- 6. Disorders of neuromuscular system
- 7. Inborn errors of metabolism acute presentation
- 8. Inborn errors of metabolism chronic presentation
- 9. Disorders of skeletal system
- 10. Connective tissue disorders

# MGC202: Prenatal Genetics & Fetal Medicine

- 1. Prenatal diagnosis: Techniques (Fetal sampling & Preimplantation diagnosis)
- 2. Pre test & post test counseling, PCPNDT
- 3. Fetal imaging
- 4. Fetal therapy
- 5. Next Generation Sequencing in prenatal diagnosis
- 6. Teratogens
- 7. Preconceptional screening
- 8. Neural tube defects and major malformations
- 9. Soft markers on fetal ultrasound
- 10. Genetic counseling in special situations

# MGC203: Genetic disorders - Part II

- 1. Imprinting defects
- 2. Triplet repeat disorders
- 3. Mitochondrial disorders
- 4. Oligogenic disorders
- 5. Multifactorial disorders
- 6. Mosaicism and related disorders
- 7. Cancer biology
- 8. Familial cancer syndromes
- 9. Genetics of common cancers
- 10. Issues in counseling for cancer

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#### MGC204: Genetic disorders – Part III

- 1. Disorders of Endocrinology
- 2. Genetic disorders related to skin
- 3. Genetic disorders related to eye
- 4. Cardiomyopathies and other cardiac disorders
- 5. Immunodeficiency disorders
- 6. Genetic disorders related to Digestive tract and Hepatobiliary disorders
- 7. Genetic disorders of renal system
- 8. Overgrowth disorders
- 9. Structural malformations of brain

#### MGC205: Genomic medicine and Research

- 1. Pharmacogenomics and personalized medicine
- 2. Treatment of genetic disorders
- 3. Gene therapy
- 4. Novel Strategies for treatment
- 5. Counseling for genetic research participation and consent taking
- 6. Electronic health records & Disease based registry
- 7. Patient advocacy
- 8. Secondary findings in genetic counseling
- 9. Education to other health care providers
- 10. Laboratory diagnostic errors: causes and Issues

#### **MGC206: Preventive Genetics**

- 1. Concepts of population based screening
- 2. New born screening
- 3. Prevention of thalassemia and other hemoglobinopathies
- 4. Screening for Down syndrome
- 5. Prevention of Neural tube defects
- 6. Population based registry of birth defects
- 7. High risk pregnancies for genetic disorders
- 8. Organizing genetic services at community level
- 9. MTP and PCPNDT act
- 10. Extended family screening: Reaching relatives

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#### A. Recommended books and other resource material:

# a) Books recommended for regular study

- Genetic Counseling: Practice and Principles by Angus Clarke, Taylor & Francis
- 2. A guide to Genetic Counseling by Uhlmann WR, Schuette JL and Yashar B, John Wiley & Sons
- 3. Thompson and Thompson Genetics in Medicine by Nussbaum RL, McInnes RR, Willard HF, WB Saunders Company
- 4. Guide to Genetic Counseling by <u>Uhlmann</u> W R, <u>J. Schuette</u> J L, <u>Yashar</u> B, Wiley Blackwell
- 5. Chromosomal abnormalities and genetic counseling by Gardner RM, Sutherland GR, Oxford University Press
- 6. Genetic Counseling Practice: Advanced Concepts and Skills by Veach PM, LeRoy BS and Bartels DM, Willey -Blackwell

# b) Reference books

	Authors/Editors	Title	Publisher
1	Phadke SR	Genetics for Clinicians	Prism Books
2	Read A, Donnai	New Clinical Genetics	Scion
	D		Publishing
			Ltd
3	Scriver CR, Beaudet	The metabolic and molecular basis	McGraw Hill
	AL, Sly WS, Valle D	of inherited disease	
5	Strachan T, Read AP	Human molecular genetics	BIOS Scientific
			Publishers Ltd,
			Oxford
6	Nussbaum, McInnes,	Thompson & Thompson Genetics in	Elsevier
	Willard	Medicine	

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7	Turnpenny, Ellard	Emery's Elements of Medical	Elsevier
		Genetics	
8	Stevenson RE, Hall	Human malformations and related	Oxford
	JG	anomalies	University Press
9	Gorlin RJ, Cohen	Syndromes of the head and neck	Oxford
	MM Jr, Henneckem	·	University Press
	RCM		
10	Graham JM Jr	Smith's recognizable patterns of	WB Saunders
		human deformation	
11	Taybi H, Lachman	Radiology of syndromes, metabolic	Mosby Year
	RS	disorders and skeletal dysplasias	Book Publishers
12	Gersen SL, Keagle	Principles of clinical cytogenetics	Humana Press
	МВ		
13	Milunsky A	Genetic disorders and the fetus:	The Johns
		Diagnosis, prevention and treatment	Hopkins
			University Press,
			London
14	Romero R, Pilu G,	Prenatal diagnosis of congenital	Appleton &
	Ghindini A, Hobbins	anomalies	Lange
	JC, Jeanty P		
15	Keeling JW	Fetal and neonatal pathology	Springer-
			Verlang, London
16	Wigglesworth JS,	Textbook of fetal and perinatal	Blackwell
	Singer DB	pathology	Scientific
			Publications,
			Boston
17	Friedman JM,	Teratogenic effects of drugs: A	The Johns
	Polifka JE	resource for clinicians (TERIS)	Hopkins
			University Press
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18	Hoffee PA	Medical molecular genetics	Fence Creek
			Publishing,
			Madison,
			Connecticut
			Publishers
19	Rimoin DL, Connor	Emery and Rimoin's Principles and	Churchill
	JM, Pyeritz RE	Practice of Medical Genetics	Livingstone
20	Harper PS	Practical Genetic Counseling	Butterworth-
			Heinemann
21	Aase JM	Diagnostic Dysmorphology	Plenum Medical
			Book Co.
22	Jones KL, Smith	Smith's Recognizable Pattern of	Saunders
	DW	Human malformations	
23	Callen PW	Ultrasonography in Obstetrics and	Saunders
		Gynecology	Elsevier
24	Rodeck CH, Whittle	Fetal Medicine: Basic Science and	Wiley-Blackwell
	MJ	Clinical Practice	
25	Jorde LB, Carey JC,	Medical Genetics	Mosby
	Bamshad MJ		
L			

# c) Journals

	Name of the journal	Publisher	
1	Nature Genetics	Nature Publishing Group	
2	Nature Reviews Genetics	Nature Publishing Group	
3	American Journal of Human	University of Chicago Press (American	
	Genetics	Society of Human Genetics)	
4	Clinical Genetics	Blackwell Publishing	
5	Journal of Medical Genetics	BMJ Publishing Group	
6	American Journal of Medical	Wiley InterScience	
	Genetics A and C		
7	Journal of Genetic Counseling	Springer	

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8	Clinical Dysmorphology	Lippin Williams & Wilkins
9	Prenatal Diagnosis	John Wiley & Sons, Ltd.

# d) Databases

Online Mendelian Inheritance in Man (OMIM), London Medical Data base and POSSUM and TERIS, Phenomizer, gnomAD, ClinVar, ClinGEN

# **Annexure II**

# **Objectives and Outcomes of the courses**

Cours e	Title	Objective of course	Outcome
1st Year	•		
Semeste	r I		
MGC- 101	Introduction to Medical Genetics	<ul> <li>Pedigree drawing</li> <li>Principles of Mendelian inheritance</li> <li>Non-mendelian and multifactorial inheritance</li> <li>Understanding basic concepts of population genetics</li> <li>Consanguinity and inbreeding</li> <li>Clinical case preparation</li> </ul>	<ul> <li>Draw pedigree and compile clinical case records – 10 cases/week</li> <li>Determine possible modes of inheritance from family history/pedigree</li> <li>Understand degrees of consanguinity and relatedness</li> <li>Recognize common birth defects</li> </ul>

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		Recognizing     common birth     defects	
MGC - 102	Cytogenetics and Chromosomal Disorders	defects  Chromosomal basis of heredity  Cell culture, karyotyping and cytogenetic nomenclature  Chromosomal abnormalities — Numerical & structural	<ul> <li>Perform karyotype and reporting of 10 samples</li> <li>Know clinical indications for chromosomal testing</li> <li>Know the tests for chromosomal abnormalities with advantages and limitations</li> <li>Understand different chromosomal</li> </ul>
		<ul> <li>Indications of karyotyping</li> <li>Prenatal cytogenetics</li> <li>Down syndrome &amp; common aneuploidies</li> <li>Sex chromosomal abnormalities</li> </ul>	<ul> <li>abnormalities</li> <li>Know phenotype and chromosomal abnormalities of associated with Down syndrome, trisomy 13, 18, Turner syndrome, Klinefelter syndrome</li> <li>Understand different structural chromosomal</li> </ul>
		<ul> <li>Phenotypes         Structural         chromosomal         abnormalities         [Balanced &amp;             Unbalanced]</li> <li>Contiguous gene         syndromes</li> <li>Family with         structural         chromosomal         abnormality: Risk         of recurrence</li> </ul>	aberrations and clinical implications.

		<ul> <li>Short stature</li> <li>Multiple         malformation         syndromes</li> <li>Regression of         milestones</li> <li>Disorders of sexual         development and         differentiation</li> <li>Genetics of         common disorders</li> <li>Common neonatal         problems</li> </ul>	Able to do genetic counseling for common conditions like Down syndrome, DMD, Turner syndrome, couples with recurrent pregnancy loss and bad obstetric history. etc
105 &	asic Genetics c Genetic counseling	<ul> <li>Genetic testing in Children         <ul> <li>Issues</li> </ul> </li> <li>Educational aids for patients and family</li> <li>Genetic counseling for autosomal dominant disorders</li> <li>Genetic counseling for autosomal recessive disorders</li> <li>Genetic counseling for X-linked recessive disorders</li> <li>Genetic counseling for X-linked dominant and semidominant disorders</li> </ul>	<ul> <li>Able to do genetic counseling for any monogenic condition</li> <li>communicate concepts of asymptomatic carrier in recessive disorders, penetrance and variable expressivity, germline mosaicism</li> <li>Appropriate and effective use of educational aids</li> <li>Pretest and post-test counseling in minors</li> <li>Identify ethical issues in various case scenarios</li> <li>Understand legal implications in certain clinical settings e.g PCPDNT act, insurance/employmen t discrimination etc.</li> </ul>

		Counseling issues     in chromosomal     disorders	Prepare patient information sheets for chosen conditions- 2
		Counseling in     disorders with non-	
		mendelian	
		inheritance	
		Legal and social	
		issues in Genetic	
		disorders	
		• Ethical issues in	
		Genetic disorders	
MGC-	Molecular	Structure of gene	Read and analyze
106	Genetics	and genome	sequencing reports  • Design primers for
		PCR and	PCR
		sequencing	Perform Sanger
		Types of Mutations	sequencing -10
		and Effects	samples  • Use standard
	·	Methods to detect	nomenclature for
		copy number	denoting genetic
		variations	variations  • Application of
		Nomenclature of	standard guidelines
		DNA variations	for pathogenicity
		Recombinant DNA	classification of
		technology in	variants • Prepare reports for
		diagnostics	Sanger/ next
		Next generation	generation
		sequencing:	sequencing – 10 samples
		technique and	• Correlation of
		applications	prioritized variants in
		Guidelines for	NGS analysis
		prediction of	
		pathogenicity	1
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2 <sup>nd</sup> Yea	r	<ul> <li>Bioinformatics and databases</li> <li>DNA fingerprinting</li> </ul>	
MGC- 201	Genetic disorders – Part I	<ul> <li>Dysmorphism and related syndromes</li> <li>Phenotype nomenclature (HPO)</li> <li>Thalassemia and other hemoglobinopathie s</li> <li>Genetic disorders of coagulation &amp; bleeding</li> <li>Hearing</li> </ul>	<ul> <li>Annotation of phenotype with HPO terms and documentation for all cases being processed for NGS.</li> <li>Perform syndrome search using databases</li> <li>Genetic work-up and counseling for Hemoglobinopathies and Hemophilia</li> <li>Understanding genetics and genetic counseling of common neuromuscular</li> </ul>
		<ul> <li>impairment</li> <li>Disorders of neuromuscular system</li> <li>Inborn errors of metabolism – acute presentation</li> <li>Inborn errors of metabolism – chronic presentation</li> <li>Disorders of skeletal system</li> </ul>	disorders like SMA, DMD  Genetic evaluation and counseling for Achondroplasia, hypochondroplasia  Genetic evaluation and counseling for non-syndromic deafness

		Connective tissue     disorders	
MGC - 202	Prenatal Genetics & Fetal Medicine	<ul> <li>Prenatal diagnosis:         Techniques (Fetal sampling &amp; Preimplantation diagnosis)</li> <li>Pre test &amp; post test counseling,         PCPNDT</li> <li>Fetal imaging</li> <li>Fetal therapy</li> <li>Next Generation Sequencing in prenatal diagnosis</li> <li>Teratogens</li> <li>Preconceptional screening</li> <li>Neural tube defects and major malformations</li> <li>Soft markers on fetal ultrasound</li> <li>Genetic counseling</li> </ul>	<ul> <li>Pretest counseling for couples undergoing invasive prenatal testing for high-risk pregnancies</li> <li>Post -test counseling following prenatal diagnosis</li> <li>Genetic counseling for common prenatally detected conditions like increased NT, short long bones, neural tube defects etc.</li> <li>Genetic counseling for USG detected soft markers</li> <li>Determination of teratogenicity and genetic counseling</li> <li>Genetic counseling for preimplantation genetic testing and other artificial reproductive technologies.</li> </ul>
MGC -	Genetic	<ul><li>in special situations</li><li>Imprinting defects</li></ul>	Genetic evaluation
203	Disorders Part II	<ul> <li>Triplet repeat     disorders</li> <li>Mitochondrial     disorders</li> <li>Oligogenic     disorders</li> </ul>	and counseling for common imprinting disorders like Prader Willi, Angelman etc.  Genetic counseling for triplet repeat disorders and communicate

Semester IV	<ul> <li>Multifactorial disorders</li> <li>Mosaicism and related disorders</li> <li>Cancer biology</li> <li>Familial cancer syndromes</li> <li>Genetics of common cancers</li> <li>Issues in counseling for cancer</li> </ul>	concept of anticipation  • Understanding and communication of polygenic risk scores  • Genetic evaluation and counseling for hereditary cancer syndromes including pre-symptomatic testing
MGC - Genetic 204 disorders – Part III	<ul> <li>Disorders of         Endocrinology</li> <li>Genetic disorders         related to skin</li> <li>Genetic disorders         related to eye</li> <li>Cardiomyopathies         and other cardiac         disorders</li> <li>Immunodeficiency         disorders</li> <li>Genetic disorders         related to Digestive         tract and         Hepatobiliary         disorders</li> <li>Genetic disorders         of renal system</li> </ul>	<ul> <li>Genetic evaluation and counseling of conditions like epidermolysis bullosa, retinitis pigmentosa, cardiomyopathies</li> <li>Determine possible genetic etiologies of any disorder through clinical case study, pedigree analysis and database enquiries.</li> </ul>

		Overgrowth	
		disorders	
		Structural	
		malformations of	
		brain	
MGC - 205	Genomic medicine and	• Pharmacogenomics	Database search for
203	medicine and Research	and personalized	pharmacogenomic variants
		medicine	Basic knowledge of
		• Treatment of	available treatment for
		genetic disorders	common genetic disorders
		Gene therapy	<ul><li>Glsorders</li><li>Counseling and</li></ul>
		Novel Strategies	consent taking for
		for treatment	genetic research
		Counseling for	participation • Preparation of ethics
		genetic research	committee submission
		participation and	documents for
		consent taking	research projects
		Electronic health	Maintenance of     database like OPD
		records & Disease	patient data, DNA
		based registry	banking data etc.
		Patient advocacy	Knowledge about  notions support and
		<ul> <li>Secondary findings</li> </ul>	patient support and advocacy groups
		in genetic	
		counseling	
		<ul> <li>Education to other</li> </ul>	
		health care	
		providers	
		Laboratory	·
		diagnostic errors:	
		causes and Issues	
		THE TOTAL	
MGC -	Preventive	• Concepts of	Genetic counseling for
206	Genetics	population based	newborn screening,
		screening	1

•	New born
	screening

- Prevention of thalassemia and other hemoglobinopathie
   s
- Screening for Down syndrome
- Prevention of Neural tube defects
- Population based registry of birth defects
- High risk
   pregnancies for
   genetic disorders
- Organizing genetic services at community level
- MTP and PCPNDT act
- Extended family screening:
   Reaching relatives

- genetic carrier screening
- Application of cascade/extended familial screening
- Counseling for Screening and prevention of Neural tube defects, Down syndrome, thalassemia
- Identification and counseling of high risk pregnancies.
- Preparation of a poster or audiovisual aid for public awareness regarding an area of community genetics.