THE TAMIL NADU Dr. M.G.R. MEDICAL UNIVERSITY,

69, ANNA SALAI, GUINDY, CHENNAI - 600 032.

Regulations, Syllabus & Guidelines

FOR

POST - DOCTORAL FELLOWSHIP PROGRAMME

IN

CYTOGENETICS

AS APPROVED IN 49TH SAB HELD ON 07.01.2015

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The Tamil Nadu Dr. M.G.R. Medical University, Chennai.

REGULATIONS FOR

POST - DOCTORAL FELLOWSHIP PROGRAMME IN CYTOGENETICS

1. PREAMBLE :

In exercise of the powers conferred by Section 44 of the Tamil Nadu Dr. M.G.R. Medical University, Chennai Act 1987 (Tamil Nadu Act 37 of 1987) the Standing Academic Board of the Tamil Nadu Dr. M.G.R. Medical University, Chennai hereby makes the following Regulations for Post - Doctoral Fellowship Programme in Cytogenetics.

2. SHORT TITLE AND COMMENCEMENT :

The course shall be called as "POST-DOCTORAL FELLOWSHIP PROGRAMME IN CYTOGENETICS" of the Tamil Nadu Dr. MGR Medical University, Chennai.

This shall come into force from the academic year 2015-2016.

The regulations framed are subject to modification from time to time by the Standing Academic Board.

3. AIM AND OBJECTIVES :

Keeping in view the explosion of knowledge in modern medicine, the University introduces a series of Post - Doctoral Fellowship Programmes in different disciplines (speciality or sub - speciality), wherein suitable candidates will be imparted training in the concerned area.

These Post - Doctoral Fellowship Programmes aims that the candidate gets exposure in the concerned disciplines with particular emphasis on their clinical skills. The course is meant to give intensive hands - on clinical training with periodic evaluation by experienced teaching staff.

The Post -Doctoral Fellowship programmes offered by this University cannot be equated with DM / M.Ch.

4. DURATION OF THE COURSE :

The Duration of the Courses is 2 years. The **First One Year** can be considered as training period and **Second Year** be the period of study. At the end of **Second Year** the candidates be permitted to appear for the examination.

5. ELIGIBILITY FOR ADMISSION :

a. Candidates who have passed Medical Post Graduate degree Course recognized by the Medical Council of India are eligible to join in **POST-DOCTORAL FELLOWSHIP PROGRAMME IN CYTOGENETICS.** This includes M.D. (Anatomy) / M.D. (Path.) / DNB (Anatomy or Path.). One year Post - PG experience is not required for admission to this course.

b. The Ph.D. in the concerned speciality is not considered to be eligible to apply for the Post - Doctoral Fellowship Programme in Cytogenetics.

c. Candidates who have obtained P.G. degree from any recognized University, within India, but outside the state of Tamil Nadu will have to produce Migration Certificate from their qualifying University. No Objection Certificate issued by the National Board of Examinations, New Delhi, is equivalent to Migration Certificate.

d. The candidates shall obtain the Eligibility Certificate before starting of One year pre-training and Eligibility Certificate will not be issued after completion of pre-training in other Institutions.

e. The candidates shall obtain Eligibility Certificate before the cut off date for admission in that particular Academic year.

f. The candidates has to complete his / her One year pre - training in the same Institution(s) in which the candidate desires to undergo the course.

6. ELIGIBILITY CRITERIA :

M.D. (Anatomy) / M.D. (Path.) / DNB (Anatomy or Path.) candidates are eligible for the Post - Doctoral Fellowship Programme in Cytogenetics.

7. AGE :

No upper Age limit fixed for admission to Post - Doctoral Fellowship

Programme in Cytogenetics.

8. ELIGIBILITY CERTIFICATE :

Candidates who have passed the qualifying examination as stated in Regulation No.6 above other than the Tamil Nadu Dr. M.G.R. Medical University, Chennai shall obtain an "Eligibility Certificate" from this University by remitting the prescribed fees along with the application form and required documents before seeking admission in any one of the affiliated Medical Institutions. The application form is available in the University website: www.tnmgrmu.ac.in.

9. MIGRATION CERTIFICATE :

On completion of Post Doctoral Fellowship Programme in Cytogenetics, Migration Certificate will not be issued.

10. CONDONATION OF BREAK OF STUDY :

The Break of Study for a period of less than 90 days can be condoned by the Course Director and the Break of Study for the period of more than 90 days and less than one year is to be condoned by the University authorities. The application form is available in the University website: www.tnmgrmu.ac.in.

11. RE-ADMISSION OF THE BREAK OF STUDY :

1. The course shall be completed within the period of double the duration from the date of admission.

2. The Regulation for Re-admission are as per the University's common Regulation for Re-admission.

3. The candidates having break period in the One year pre-training shall complete the balance period of training before starting the 2nd year of study in the Post Doctoral Fellowship Programme in Cytogenetics.

12. NO. OF ATTEMPTS ALLOWED FOR EXAMINATION:

The candidates of 2 year Post Doctoral Fellowship courses in Cytogenetics

shall be allowed for a maximum of five attempts with a period of 4 years including the first appearance.

13. ADMISSION :

The admission for the Post - Doctoral Fellowship Programme in Cytogenetics is twice in a year (i.e.,) 1st January and 1st July.

(a) Admission upto 31st January - 28th February is the last date for Registration.

(b) Admission upto 31st July - 31st August is the last date for Registration.

An Institution can admit max. of Two (2) students per year for Post-Doctoral Fellowship Programme in Cytogenetics i.e, either in January/July sessions or 1 candidate for each session (January or July). However, the total number of candidates per institution per course shall not exceed Two (2) per year.

14. SELECTION OF CANDIDATES :

The candidates for the Post-Doctoral Fellowship Programme in Cytogenetics can be selected by the centers on their own and the selection to be transparent.

a. The candidates selected by the Institutions will become bonafide student of this University only after registration.

b. The Post-Doctoral Fellowship Programmes of the Affiliated Institutions will be published in the University website.

15. COMMENCEMENT OF THE COURSE :

The one / two years Post - Doctoral Fellowship Programmes will commence on 1st January & 1st July of every year and the candidates are expected to get registered with this University within 30 days of their selection by the Affiliated Institutions. (i.e. 28th February & 31st August).

16. CURRICULUM :

The Regulation, Guidelines, Curriculum and the Syllabus for the Post

Doctoral Fellowship Programmes is prescribed in these regulations are subject to modification by the Standing Academic Board from time to time.

17. REGISTRATION :

A Candidate admitted into 'POST-DOCTORAL FELLOWSHIP PROGRAMME IN CYTOGENETICS' under any one of the affiliated institutions of this University shall register his / her name with this University by submitting the prescribed application form for registration duly filled in, along with the prescribed fee and a declaration in the format to the Controller of Examination of this University through the affiliated institution within 30 days from the cut-off date prescribed for admission. The application should have the date of admission of the course.

18. SCHEME OF EXAMINATION :

Commencement of examination for the Post Doctoral Fellowship Programme in Cytogenetics is on any day within the calendar month of January / July. The examination will be conducted with one internal Examiner i.e. the Course Director who is the Convener of the examination and Two External Examiner of which One from Government & One from Private Institution. The maximum age limit for the examiner is 70.

19. EXAMINATION PATTERN :

There is no theory examination for the Post Doctoral Fellowship Programme in Cytogenetics. The Institution must have periodical assessment on the performance of the students by maintaining a log book.

20. ATTENDANCE :

90% attendance is mandatory to become eligible for the examination and will be certified by the Course Director.

21. MINIMUM / MAXIMUM MARKS FOR PRACTICAL / CLINICAL / ORAL & INTERNAL ASSESSMENT :

The Examination pattern is as follows :

Exam	Maximum	Minimum
Practical	100	50
Orals / Viva	100	50
Internal Assessment	100	50
Log Book	50	25

The log book will be assessed by Examiners during the Clinical Examination. Projects presentation is mandatory and 25% of IA is for the project.

22. EXAMINATION :

There is No Theory Examination. Only Clinical Examination & Viva will be conducted.

A candidate who undergoes the Post-Doctoral Fellowship Programme in Cytogenetics shall satisfy the required eligibility criteria to appear for the Examination.

23. CLINICAL EXAMINATION HOURS :

The Clinical Examination will be conducted between 9 a.m. to 5.00 p.m.

24. PRACTICAL EXAMINATION :

Minimum 10 and Maximum 20 OSCE Stations will be given for examination (Objective Structured Clinical Examination).

Internal Assessment marks and attendance are to be submitted to the University one month before the Examination.

25. EXAMINATION CENTRE :

In case of more than one Centre conducting the same Course, the Examination Centre will be decided by the Controller of Examinations of this University and the Course Director of the Centre will be the Convener of the examination.

26. STIPEND :

The University will not give any stipend to the candidates admitted for Post-Doctoral Fellowship Programme in Cytogenetics, and the University will not interfere, if the private institutions are willing to give a stipend to the candidates admitted under them.

27. LOG BOOK :

The Log Book shall be verified by the Course Director periodically and should be submitted to the examiners at the time of examination for evaluation and only the marks to be sent to the University for result processing.

Submission of Log Book is compulsory to appear for University examination.

If the examiners suggest minor corrections and resubmission of the Log Book, the results of the candidates shall be with held and the Log Book shall be resubmitted by the candidate within three months. The Log Book shall be revalued by the same set of examiners and after getting approval, the results of the candidate shall be published.

28. Project :

Project should be submitted at the end of the year and it carries 25% of marks in the internal Assessment.

29. PROGRAMME DIRECTOR :

Must have 15 years of work experience with a teaching experience of 10 years and must possess Diploma / Fellowship / Ph.D. in the concerned field.

Programme Director should be responsible for

1. Journal Club
2. C.M.E. Programmes
3. Internal Assessment
4. Hands on Training
5. Knowledge about complications.

30. VACATION :

There is no vacation.

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Annexure 5

Course Name: ***Post Doctoral Fellowship in Cytogenetics***

Course content: Cytogenetics is an evolving field. Obtaining chromosomes for study requires cell culture and is labour-intensive. Training in morphology and pattern recognition is necessary to be able to identify chromosomes correctly. Abnormalities have to be described using the international system for human cytogenetic nomenclature (ISCN).

The course will focus on the following skills that are necessary to enable the trainee to be a competent diagnostician and to function in a supervisory capacity in a clinical cytogenetics laboratory.

- Cell culture techniques to obtain chromosomes for study.
- Methods to identify and characterize chromosomal abnormalities.
- The morphology of normal and abnormal chromosomes.
- Performance and interpretation of fluorescence in situ hybridization tests.
- Use of cytogenetic nomenclature (ISCN) to write reports for the above tests.
- Performance and interpretation of related molecular diagnostic techniques.

Teaching Methods:

- Seminars/ Lectures
- Case Discussions
- Laboratory Posting for hands on experience with cytogenetic techniques

Course content – Theory

- Structure and organisation of the human genome
- DNA structure, variation, replication and repair
- Transcription, translation and post translational processing
- Structure of genes and functional DNA elements
- Regulation of gene expression
- Various types of repeats in human genome
- Types of polymorphisms in the human genome
- Mutation – types and effects
- Epigenetics, imprinting and DNA methylation
- Cell cycle, mitosis, and meiosis, recombination
- Gametogenesis and fertilization
- Uniparental disomy
- Chromosome structure, morphology and variation
- Lyon hypothesis and X inactivation
- Autosomal and sex chromosome aneuploidy syndromes
- Structural abnormalities of chromosomes

- Microdeletion syndromes
- Chromosome breakage syndromes
- Cytogenetics of spontaneous miscarriage and infertility, polysomies
- Sexual differentiation and intersex
- Prenatal diagnosis – amniocentesis, chorionic villus sampling, cordocentesis
- Pseudomosaicism, mosaicism and chimerism
- Role of cytogenetics in cancer
- Chromosomal abnormalities in haematological malignancies
- Chromosomal abnormalities in solid tumours
- The genetic and molecular basis of cancer
- Inherited cancer predisposition
- Indications for chromosome analysis
- Cytogenetic nomenclature (ISCN)
- Mendelian inheritance
- Atypical patterns of inheritance
- Single gene disorders
- Common disorders with multifactorial inheritance
- Molecular basis of genetic disease
- Role of genes and cellular and molecular mechanisms in development
- Genotypes and phenotypes in populations
- Hardy Weinberg equilibrium
- Genetic drift and founder effect
- Calculation of conditional probability and genetic risk
- Bayesian probabilities
- Methods of mutation analysis
- Mapping and disease gene identification
- Molecular cytogenetics – fluorescence in situ hybridisation, multiplex ligation dependent probe amplification, DNA microarrays,

<u>Course content for fellowship in Cytogenetics - Practical skills</u>

Cytogenetic techniques and microscopy to analyse chromosomes from

- blood
- bone marrow

- amniotic fluid
- chorionic villus samples
- solid tissues including products of conception, tumours and skin

Specifically, the candidate must be able to

- Initiate, maintain and harvest in-vitro suspension and adherent cultures of diagnostic human specimens from a variety of tissues as listed above
- Prepare metaphase spreads from human chromosomes.
- Perform appropriate staining techniques (G-,R-, Q-,C-, NOR, distamycin-DAPI) know their uses and limitations and be able to interpret the findings .
- Perform other cytogenetic tests such as Mitomycin C testing for chromosome breakage and sister chromatid exchange.
- Perform fluorescence in-situ hybridization **(FISH) analysis** for
  - aneuploidy
  - microdeletion
  - translocation
  - amplification
  - in constitutional and acquired abnormalities and interpret the result.
- Prepare reagents required for these tests
- Use light, phase contrast, fluorescence and inverted microscopes and be able to maintain them.
- Use automated karyotyping systems for karyotyping and FISH.
- Detect and correct errors in the above techniques.
- Maintain quality control systems in the laboratory.

Interpreting and communicating the findings of cytogenetic analysis

The candidate must be

- able to develop a professional opinion as to the causation, severity, and likely outcome of the abnormality.
- able to write a written report which clearly conveys diagnostic information and recommendations to the requesting physician.
- able to communicate with clinicians to seek or provide appropriate information and inferences about a patient by oral (face-to-face or telephone) communication and follow up patient outcomes by consultation with clinicians.
- aware of when to seek further expert opinion.

Other skills:

To be able to

- **advise referring physicians regarding** the
  - most appropriate test for a particular clinical question
  - relative diagnostic strengths and the limitations of any proposed investigation.
  - relevant samples and anticoagulants or transport media used for specific samples, eg., use of sodium heparin versus EDTA for blood, bone marrow aspirates, buccal smears, chorionic villus and amniotic fluid samples, products of conception, skin biopsy, muscle biopsy, lymph node biopsy, and cerebrospinal or any other body fluid.

- requirement for duplicate samples, and positive and negative control samples
- specimen transport conditions (time after collection, temperature and container) for the dispatch of samples.

- **Maintain patient** confidentiality and privacy.

- **Contribute** appropriately to teaching, clinico-pathological conferences, quality control and other relevant meetings.

Laboratory management

To be able to run a laboratory with regard to current standards and to

- **handle specimens appropriately with respect to**
  - transport time and conditions to ensure specimen integrity and reduce the likelihood of a suboptimal /failed culture.
  - receipt, identification and allotting a laboratory accession number.
  - data entry including the recording of all relevant information on both patient and sample .
  - safe handling, storage, retention and disposal of samples.
- **organise and monitor**
  - workflow to ensure that routine samples are processed and analysed as per the stated turn-around times and priorities urgent or out-of-hours work.
  - staff for best use of time
- **use** and maintain appropriate instrumentation & automation systems.
- **maintain** a standard operating procedure manual.
- **ensure** that specimens are selected, indexed and stored appropriately.
- **keep** accurate records.
- **retrieve** specimens or records promptly when required for examination and review.
- **be** familiar with, and maintain the requirements for
  - quality control
  - calibration of equipment
  - standardisation of tests
- **be aware of**
  - costs of reagents, their preparation and usage
  - sources of patient and laboratory variation
- **maintain** a safe environment including methods of waste disposal as per prescribed standards.
- **trouble-shoot** and validate in-house laboratory tests.
- **train** personnel as required.

Related molecular cytogenetics / genetics skills

Prepare nucleic acids as required for testing programs, including cDNA and modified / substituted DNA, to an appropriate purity standard.

Perform standard polymerase chain reactions.

Be able to advise on the appropriate use of

- PCR / hybridisation based testing (Southern analysis)

- DNA sequencing
- linkage studies
- microsatellite based studies
- M-FISH/SKY/ microarrays
- analytical methods for known mutations /common mutation screen

Suggested reading - Books

- Principles of Clinical Cytogenetics by Steven L Gersen and Martha B Keagle
- Thompson and Thompson Genetics in Medicine by Robert L Nussbaum, Roderick R McInnes, Huntington F Willard
- Chromosome abnormalities and genetic counseling by R J McKinlay Gardner and Grant R Sutherland
- The AGT cytogenetics laboratory manual by Margaret J Barch, Turid Knutsen, and Jack L Spurbeck
- International System for Human Cytogenetic Nomenclature (ISCN) 2013, L G Shaffer, ML Slovak and N Tommerup

Suggested reading – Journals

- American Journal Of Human Genetics
- BMC genetics
- BMC medical genetics
- Human Molecular Genetics
- Clinical Dysmorphology
- Journal of Genetics
- Molecular Cytogenetics
- Cancer Genetics and Genome Research
- Genes, Chromosomes and Cancer
- Nature genetics

Departments involved in the training programme:

- **Cytogenetics** – Ten months to gain adequate exposure to constitutional as well as cancer cytogenetics* with an option of spending one month in a laboratory in a recognised institution.
- **Haematology** – six weeks in the molecular genetics and the flow cytometry laboratories for familiarity with the related laboratory tests.
- **Pathology and Clinical Pathology** – two weeks for familiarity with bone marrow morphology and

with molecular pathology.

Other faculty in related specialties who will be involved in the training (not counted for seats)

1. Head / Professor, Department of Haematology
2. Head / Professor, Department of Pathology
3. Head / Professor, Department of Clinical Pathology, Immunohematology and Blood Transfusion

Exit examination:
Written, practical tests and viva voce as for other courses.
1. Theory
Two papers of three hours each, consisting of essays and short notes and will cover basic cytogenetics including the structure and organisation of chromosomes and DNA, constitutional chromosomal abnormalities, cancer cytogenetics, principles of laboratory management and quality control.
2. Practical examination
Duration: Three hours – dry practical to test analysis, interpretation and problem solving, interpretation of findings and correct use of ISCN.

3. Oral examination (viva voce)

In addition, the candidate must do either of the following:

1. Submit an article (cases series, not case report) for publication in an indexed journal.

2. Submit a case-book of 10 cases as follows: at least one prenatal, one postnatal, one haematology /oncology and one FISH study from cases sent for diagnosis during the training period. One patient may appear only once. Each case should be given a unique identity number so that details may be checked. Word limit: 25,000, excluding references.
Patient names/ identification must not be present, but the cases should be numbered so that they can be identified.

Authority who evaluates/accepts the above: Professor/Head of the Research committee.

Last date for submission: One month before course completion.

This will need to be accepted by the external examiners at the time of the final examination (as per MCI norm).

Detailed syllabus
Course Content – Post Doctoral Fellowship in Cytogenetics
Practical Skills

I. Basic Practical Skills

1. Cytogenetics techniques, microscopy and related skills

- Set up, maintain and harvest in vitro culture for diagnostic human specimens.
- Prepare metaphase spreads according to laboratory guidelines.
- Select, perform and interpret routine and special stains, and detect and correct errors in these processes.
- Maintain laboratory microscopes and use light, phase contrast, fluorescence and inverted microscopy appropriately.
- Be able to use automated karyotyping systems.
- Specifically should to be able to perform accurate identification of normal and abnormal chromosomes and common aberrations.

2. **Perform cytogenetic analysis of**

- blood
- amniotic fluid cells
- chorionic villus samples
- products of conception
- normal tissues (skin or other) for fibroblast cytogenetics
- bone marrow and other malignant tissues

3. **Perform FISH analysis for**

- aneuploidy
- microdeletion
- translocation
- chromosome painting

4. **Provide a professional opinion on each case, (**based on the available information**)**

- As to the nature, causation, severity, and likely outcome of the condition.
- Should be able to access appropriate information to assist in the interpretation of data.

5. **Communicate an opinion**

- By preparing a written report which contains all appropriate diagnostic information recommendations to the requesting clinician in a timely fashion,
- Where appropriate, be able to discuss a case to the referring clinician
- Be aware of when to seek further expert opinion.

II. Other related skills

1. Be able to advise clinicians regarding the

- appropriate choice and selection of tests, given the clinical question to be answered,
- Relevant samples and preservatives required for specific tests: eg blood, bone marrow aspirates, chorionic villus, and amniotic fluid, products of conception, skin, lymph nodes, body fluids and buccal swabs.
- Relative diagnostic strengths and the limitations of any proposed investigation.
- Requirement for duplicate samples, and positive and negative control samples.

- Specimen transport conditions for the dispatch of samples, including timeliness and temperature.

2. Request provision of pedigree information for familial conditions if required.
3. Maintain patient confidentiality and privacy.
4. Contribute appropriately to teaching, clinico-pathological conferences, morbidity and mortality reviews, quality and audit committees and other similar meetings.

III. Handling of Specimens

- Ensure specimen transport time and conditions have been appropriate to guarantee specimen integrity and timeliness with reference to the relevant laboratory procedure
- Apply the principles of appropriate specimen receipt, identification and laboratory accession.
- nsure procedures for data entry include the recording of adequate information on both patient and sample for repeat future family testing.
- Apply principles of safe sample handling, storage, retention and subsequent disposal.
- Apply laboratory-specified workflow procedures to prioritize routine, urgent and out of hours work.

Iç. Production, Validation, Analysis, Reporting and Storage of Laboratory Data

i) Be familiar with the requirements for
   - specimens
   - quality control
   - calibration of equipment
   - standardisation of tests
   - sources of patient and lab variation
   - reagent preparation and usage
   - waste disposal as per existing standards
   - costs
   - record keeping
   - training of personnel as required

ii) Design, trouble-shoot and validate in-house laboratory tests.
iii) Analyse, interpret, record, report, compile and check morphological, qualitative and quantitative test results in the context of the clinical question.
iv) Maintain a standard operating procedure manual.
v) Be able to use and maintain appropriate instrumentation & automation systems**.**
vi) Ensure that specimens are selected, indexed and stored appropriately.
vii) Be able to retrieve specimens, records and showing examples of specific diseases or processes for examination and review.

ç. Laboratory environment

- Be able to run a laboratory with regard to current standards
- Maintain a safe environment
- Organize staff
- Participate in continuing education

çI. Related molecular genetics skills

- Prepare nucleic acids as required for testing programs, including cDNA and modified / substituted DNA, to an appropriate purity standard.
- Design oligonucleotide primers to maximize PCR sensitivity and specificity.
- Establish PCR reagent mixes for different PCR applications.
- Perform standard PCRs.
- Be familiar with, and be able to advise on the appropriate use of
  - standard and allele restricted PCR
  - modified PCR for methylation studies
  - modified PCR for mutation screening
  - quantitative PCR methodologies
  - hybridisation based testing (Southern analysis)
  - DNA sequencing
  - linkage studies
  - microsatellite based studies
  - M-FISH/SKY
  - analytical methods for known mutations and common mutation screens

Detailed syllabus

Course Content – Post Doctoral Fellowship in Cytogenetics

Theory

Genome and Genetic structures

- DNA structure and variation
- Epigenetics and DNA methylation
- Chromosome structure, morphology and variation
- Structure and organisation of the human genomes, & comparisons with other phyla
- Structure, variation, function and identification of genes and functional DNA elements
- Mapping, linkage disequilibrium and haplotypes in the human genome
- Various types of repeats in human genome

Molecular and cellular physiology

- DNA replication and repair
- RNA transcription and processing
- Translation and post translational processing
- Mitosis and the cell division cycle
- Meiosis, recombination and chromosomal segregation
- Fertilisation, early embryogenesis and malformation
- Mosaicism and chimerism

- Twins and twinning

Mutation / polymorphisms

- Mutation and polymorphisms
- Microdeletions & microduplications
- Biological basis of non-disjunction & aneuploidy
- Segregation of chromosomal structural anomalies
- Types of polymorphisms in the human genome
- Mutation scanning techniques for known and unknown variants

Inheritance

- Standard patterns and modifiers of inheritance
- Complex, multifactorial and quantitative traits
- Population genetics & Hardy Weinberg equilibrium Methods of mutation analysis

Cancer genetics

- The biological basis of cancer
- Inherited cancer predisposition
- Molecular aberration in cancer

Principles of genetic testing

- Direct and indirect laboratory testing
- Clinical: molecular correlates. Phenotype/genotype relationships
- Calculation of conditional probability and genetic risk
- Bayesian probabilities
- Relevant aspects of epidemiology and statistics
- Population screening

Cytogenetics

- Uses of karyotyping
- Chromosomal preparations and banding techniques
- Cytogenetic nomenclature
- FISH techniques
- Molecular techniques with direct relevance to cytogenetic analysis

- Assessment of chromosome breakage
- Karyotyping systems in clinical cytogenetics

Clinical relevance of constitutional and acquired chromosome abnormalities

- Constitutional chromosomal disorders
- Autosomal and sex chromosome aneuploidy
- Polysomies
- Structural abnormalities of chromosomes
- Cryptic chromosomal translocations
- Microdeletion syndromes
- Uniparental disomy
- Abnormal segregation of structural anomalies

Pregnancy

- Fertility and infertility
- First trimester screening
- Prenatal testing by amniocentesis
- Prenatal testing by CVS
- Mosaicism and pseudomosaicisim
- Cytogenetics of spontaneous miscarriage

Development

- Embryogenesis and malformations
- Sex determination & differentiation
- Chromosome causes of intersex
- Chromosome breakage syndromes

Cancer Cytogenetics

- Molecular basis of cancer
- Common karyotypic abnormalities in leukemia
- Significance of karyotype in leukemia
- Significance of karyotype in solid tumours

Laboratory safety

Suggested reading - Books

Principles of Clinical Cytogenetics

Steven L Gersen and Martha B Keagle

Thompson and Thompson Genetics in Medicine

Robert L Nussbaum, Roderick R McInnes, Huntington F Willard

Chromosome abnormalities and genetic counseling

R J McKinlay Gardner and Grant R Sutherland

The AGT cytogenetics laboratory manual

Margaret J Barch, Turid Knutsen, and Jack L Spurbeck

International System for Human Cytogenetic Nomenclature (ISCN) 2013

L G Shaffer , ML Slovak and N Tommerup

Suggested reading – Journals

American Journal Of Human Genetics

BMC genetics

BMC medical genetics

Human Molecular Genetics

Clinical Dysmorphology

Journal of Genetics

Molecular Cytogenetics

Cancer Genetics and Genome Research

Genes, Chromosomes and Cancer

Nature genetics