

# SECTION – I (Course Content)

## GENETICS

(Handouts/ monographs – to be prepared and distributed to the students)

Lecture: 8

hrs

Practicals: 6

hrs

## TOPICS

### 1. Introduction :

1 hr

#### A. Definition:

- a. Medical genetics
- b. Cytogenetics
- c. Clinical genetics

#### B. History:

- a. Gregor Mendel
- b. Mendelian laws of inheritance
- c. Mitosis
- d. Meiosis

### 2. Chromosomes:

1 hr

- a. Structure
- b. Classification of human chromosomes
- c. Karyotyping- methodology
- d. Sex chromatin
- e. Lyon hypothesis

### 3. Chromosomal disorders:

- Importance of non-disjunction
- Numerical abnormalities
  - Polyploidy
  - Aneuploidy
  - Trisomy and monosomy
  - Down's syndrome
  - Patau's syndrome
  - Edwards's syndrome
  - Klienfelter's syndrome
  - Turner's syndrome
  - Mosaicism
  - Causes of numerical abnormalities
- Structural abnormalities
  - Deletion, inversion, translocation and ring chromosomes
  - Isochromosomes, chromosomal fragile sites, fragile X chromosome

### 4. Chromosome at molecular level:

1 hr

- Structure of DNA- RNA
- Genetic code
- Genetic mutation
- Mutagens

**5. Clinical genetics**

1 hr

- Pedigree chart
- Inheritance

**6. Diagnosis of Genetic disease**

1 hr

- Prenatal diagnosis
  - Indications
  - Chorionic villi biopsy
  - Maternal sera
  - Amniocentesis

**7. Genetic Counselling :**

1 hr

- Definition
- Indication
- Basis of gene therapy

**8. Revision :**

1 hr

**SECTION – II**  
**(Course Content under Level – I, II, III)**  
**GENETICS**

(Handouts/ Monographs to be prepared and distributed to the students)

S.NO	TOPIC	LEVEL 1	LEVEL 2	LEVEL 3	PRACTICALS	DEMONSTRATION
1	INTRODUCTION	<i>Definition:</i> <ul style="list-style-type: none"> <li>• Medical genetics</li> <li>• Clinical genetics</li> </ul> <i>History</i> <ul style="list-style-type: none"> <li>• Gregor Mendel</li> <li>• Mendelian laws of inheritance</li> </ul>			<ul style="list-style-type: none"> <li>• Draw the different stages of mitosis</li> <li>• Draw the X-chromatin (Barr body)</li> </ul>	<ul style="list-style-type: none"> <li>• Demonstration of mitosis and meiosis under the microscope</li> <li>• Demonstration of Barr body under the microscope</li> </ul>

		<i>Cell division:</i> <ul style="list-style-type: none"> <li>• Mitosis</li> <li>• Meiosis</li> </ul>				
2	<b>CHROMOSOME - I</b>	<ul style="list-style-type: none"> <li>• Structure of chromosome</li> <li>• Classification of human chromosomes</li> <li>• Sex chromatin</li> <li>• Lyon hypothesis</li> <li>• Karyotyping:</li> </ul> <p>Methodology</p>			<ul style="list-style-type: none"> <li>• Prepare a karyotype from the given photocopy of numbered G-banded chromosomal spread</li> </ul>	Demonstration of normal male and female chromosome spreads focused under the microscope
3	<b>CHROMOSOME - II</b>	<ul style="list-style-type: none"> <li>• Structure of DNA, RNA</li> <li>• Genetic code</li> <li>• Gene mutation</li> <li>• Mutagens</li> </ul>				<ul style="list-style-type: none"> <li>• Demonstration of DNA using a model</li> </ul>

4	<b>GENETIC DISEASES- I</b>	<ol style="list-style-type: none"> <li><b>1. SINGLE GENE DISORDERS:</b> <ul style="list-style-type: none"> <li>• Autosomal dominant</li> <li>• Autosomal recessive</li> <li>• Sex linked dominant</li> <li>• Sex linked recessive</li> </ul> </li> <li><b>2. CHROMOSOMAL DISORDERS:</b> <ol style="list-style-type: none"> <li><b>a. Numerical abnormalities:</b> <ul style="list-style-type: none"> <li>➤ Polyploidy</li> <li>➤ Aneuploidy (Trisomy &amp; monosomy)</li> <li>➤ Down's syndrome</li> <li>➤ Patau's syndrome</li> <li>➤ Edward's syndrome</li> <li>➤ Klienfelter's syndrome</li> <li>➤ Turner's syndrome</li> </ul> </li> <li><b>b. Mosaicism</b></li> <li><b>c. Causes of numerical abnormalities</b></li> <li><b>d. Non-disjunction</b></li> <li><b>e. Anaphase lag</b></li> <li><b>f. Structural abnormalities:</b> <ul style="list-style-type: none"> <li>➤ Deletion, inversion, translocation, ring</li> </ul> </li> </ol> </li> </ol>	<ul style="list-style-type: none"> <li>➤ Cri-du chat syndrome</li> <li>➤ Fragile X-chromosome</li> </ul>			<ul style="list-style-type: none"> <li>• Clinical cases or pictures of the clinical cases</li> </ul>
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		chromosome ➤ Isochromosome, fragile sites <b>3. MULTIFACTORIAL          DISORDERS</b>				
6	<b>CLINICAL GENETICS</b>	<ul style="list-style-type: none"> <li>• Pedigree chart/ symbols</li> </ul>				
7	<b>DIAGNOSIS OF GENETIC DISEASES</b>	<b>Prenatal diagnosis:</b> <ul style="list-style-type: none"> <li>• Indications</li> <li>• Amniocentesis</li> <li>• Chorionic villus biopsy</li> <li>• Maternal serum screening</li> </ul>				<ul style="list-style-type: none"> <li>• Demonstration (live or video) of the techniques</li> </ul>
8	<b>GENETIC COUNSELLING AND NEW DEVELOPMENTS</b>	<ul style="list-style-type: none"> <li>• Definition</li> <li>• Indications</li> <li>• Gene therapy</li> <li>• Human genome</li> <li>• Human cloning</li> </ul>				

**GENETICS : PRACTICALS**  
**(Each of two hours duration\*)**

1. Mitosis, meiosis, Barr body :
  - Stages of mitosis, meiosis are focused under the microscope
  - To draw the different stages of mitosis and meiosis
  - To draw the Barr body which is focused under the microscope
  
2. Preparing a pedigree chart :
  - Symbols used for preparing a pedigree chart are provided
  - To make a pedigree considering the student as a proband
  
3. Karyotyping and clinical features of common genetic syndromes :
  - To prepare a karyotype from the photocopy of the numbered G banded chromosomes
  - Normal male and female chromosomal spreads are focused under the microscope for demonstration
  - Observation and writing of the clinical features of Down's syndrome. Patau's syndrome. Edward's syndrome. Klinefelter's syndrome and Turner's syndrome shown in photographs.

\* **Workbook also to be completed**