

Medical Genetics	
Course No.	Gen.1303
Credit hour	2 (Theoretical)
Prerequisite	None

Aims & objectives

This course provides the student with the principles of Human cytogenetics and molecular biology techniques. It demonstrates the practical applications of this technology in a diagnostic laboratory. Topics include, but are not limited to, DNA/RNA isolation, hybridization, Polymerase Chain Reaction, and restriction enzyme analysis.

Syllabus

General subject areas include:

1. The scientific basis of Human Genetics.
2. Chromosomes and its relation to heredity.
3. Cell reproduction and divisions (cell growth cycle; Mitosis & Meiosis).
4. Chromosome Structure and nomenclature.
5. Numerical Chromosomal Anomalies (Trisomy; Monosomy; Mosaic aneuploidy; polyploidy).
6. Structural chromosomal Anomalies (Deletion; Duplication; Translocation; Inversion).
7. Karyotype and its techniques.
8. Barr body and performance of Barr test.
9. Patterns of inheritance (Pedigree symbols and construction) and interpretation of karyotype.
 - a. Autosomal dominant inheritance.
 - b. Autosomal recessive inheritance.
 - c. X- Linked inheritance.
 - d. Mitochondria inheritance.
10. The nature of genes.
11. The replication of DNA.
12. The genetic code and its transcription and translation.
13. Regulation of gene expression.
14. The organization of eukaryotic and prokaryotic genomes.
15. Genetic manipulation of DNA sequences etc.

Instructional Methods

- Lecture.
- Discussion.
- Demonstration.
- Case studies.
- Tutorials.

Suggested Method of Evaluation

In-term examinations	50 Marks.
Attendance and participations	10 Marks.
Final examinations	40 Marks.
Total Marks	100 Marks.

Instructional and Resource Materials

1. Handouts.
2. Internet resources.
3. Supplies used in hospitals.
4. Case studies.
5. Text books.

Textbooks & References

- Essential Medical Genetics By: J.M.Connor & M.A.Ferguson Smith.
- Essential of Genetics By: W.S.Klug & M.R. Cummings.