DISCIPLINE SPECIFIC CORE COURSE- 17 (BIOMED-DSC-17) HUMAN GENETICS

CREDIT DISTRIBUTION, ELIGIBILITY AND PRE-REQUISITES OF THE COURSE

Course title & Code	Credits	Credit distribution of the course			Eligibility criteria	Pre-requisite of the course (if any)	Department offering the course
		Lecture	Tutorial	Practical/ Practice			
Human Genetics **BIOMED-DSC-17**	**4**	**3**	**-**	**1**	**Class XII Passed**	**Basic Knowledge of Genetics**	**Biomedical Science**

Learning objectives

The Learning objectives of this course are as follows:

- This course is designed to develop an appreciation for the groundwork carried out so far in areas that contributed to our understanding of human genetics and diseases, relates to how it has been built on the numerous genetic studies carried out over decades to contribute to the understanding of the relationship between genotype and phenotype.
- The course will also introduce the sequencing of the Human Genome and new methods of investigating biological function, research into the genetic and molecular basis of human disease.

Learning outcomes

Having successfully completed this course, students shall be able to:

- Students will understand the patterns of inheritance of monogenic traits from pedigree data for both Mendelian and non-Mendelian traits.
- They will comprehend the techniques and advances in the analysis of DNA, identification of genes involved in diseases, and gene/sequence mapping strategies.
- Students will be able to describe objectives, tools, approaches and outcomes of the Human Genome Project (HGP). They will be aware of the ethical and societal issues raised by the new knowledge derived by using new technologies.
- Students will be able to apply principles of genetics at population level.
- They will understand the genetic basis of common diseases and methods of prenatal diagnosis.
- Students will be able to proficiently explore relevant literature, web sites and databases for research into human genetics.

SYLLABUS OF BIOMED-DSC-17:

Unit- I: Inheritance for Monogenic Traits (08 hrs)

History of Human Genetics: Early Greek concepts about inheritance, Cytogenetics history (the works of Winiwater, Painter and Tjio and Levan), Landmark achievements of Galton, Garrod etc. Patterns of Inheritance: Recapitulation of principles of human inheritance pattern through pedigree analysis: Autosomal inheritance- dominant, recessive, sex-linked inheritance, sex- limited and sex- influenced traits and mitochondrial inheritance. Deviations from the basic pedigree patterns- non-penetrance, variable expressivity, pleiotropy, late onset, anticipation, consanguinity and its effects, mosaicism and chimerism, genetic heterogeneity, uniparental disomy, and genomic imprinting.

Unit- II: Genetic and Physical Maps (06 hrs)

Genetic markers and their applications. Overview of genetic maps. Physical maps (different types- restriction, cytogenetic maps, use of FISH in physical mapping, radiation hybrids and clone libraries in STS mapping)

Unit- III: Identification of Human Disease Genes (08 hrs)

Principles and strategies, positional and candidate gene approaches, (examples- HD, CFTR), concept of twin and adoption studies.DNA sequencing (Principles of Maxam-Gilbert and Sanger Method, introduction to NGS with an example of illumina based sequencing), DNA fingerprinting, polymorphism screening (genotyping of SNPs and microsatellite markers)

Unit- IV: Human Genome Project (04 hrs)

History, organization and goals of human genome project, Tools (Vectors- BAC, PAC, YAC)) and approaches (Hierarchical and whole genome shotgun sequencing), outcomes ethical issues and applications in human diseases

Unit- V: Population Genetics (05 hrs)

Genotypic and allelic frequencies, Hardy-Weinberg Equilibrium, linkage disequilibrium, haplotype construction (two loci using SNPs and/or microsatellites).

Unit- VI: Clinical Genetics **(08 hrs)**

Inborn errors of metabolism and their genetic basis (example- phenylketonuria), genetic disorders of hematopoietic systems (examples- sickle cell anemia and thalassemia), genetic basis of color blindness, familial cancers (example- retinoblastoma) and mental retardation.

Prenatal Diagnosis: Brief introduction, methods of prenatal diagnosis (invasive and non-invasive such as Amniocentesis, Chorionic villus sampling, Ultrasonography, Fetoscopy, Maternal serum screening, Fetal cells in maternal blood) and its application with examples of Aneuploidy and Thalassemia.

Pharmacogenetics and Pharmacogenomics(genetic polymorphism in drug metabolism genes e.g. cytP450 and GST and their effect on drug metabolism and drug response), genetic counseling.

Unit- VII: Guided short project **(06 hrs)**

Short project involving, data analysis/*in silico* analysis of genomes/ literature-based project; guiding the students through identification of the project, discussions on approach and methodology, and strategies for data analysis.

Practical **(30 hrs)**

(Wherever wet lab experiments are not possible the principles and concepts can be demonstrated through any other material or medium including videos/virtual labs etc.)

1. Pedigree construction of some common phenotypic characteristics of humans.
2. Pedigree analysis and risk assessment.
3. Restriction mapping/ STS mapping from the given data.
4. Demonstration of DNA fingerprinting.
5. Polymorphism analysis using PCR.
6. Analysis of the given DNA sequencing data based on Maxam-Gilbert and Sanger sequencing methods.
7. Study of Hardy-Weinberg equilibrium by PTC tasting and ABO blood grouping.
8. Video based demonstration of tools for prenatal diagnosis.
9. Exploring DNA, RNA, and Protein Sequence Databases for retrieval of a desired human sequence and sequence alignment using BLAST.
10. Preparation of human metaphase chromosomes and Giemsa staining.

Essential readings:

- Strachan, T. and Read, A. (2018). 5th Edition. *Human molecular genetics*. Florida, USA: CRC Press, Garland Science. ISBN: 978-0815345893.

- Pasternak, J.N. (2005). 2nd Edition. *An introduction to human molecular genetics*. New York, USA: Wiley-Liss. ISBN: 978-0-471-47426-5.
- Cantor, C.R. and Smith, C.L. (1999). 1st Edition. *Genomics: The science and technology behind the human genome project*. New York, USA: Wiley-Interscience. ISBN: 9780471599081.

Suggestive readings:

- Brown, T.A. (2023). 5th Edition. *Genomes 4*. New York, USA: Garland Science. ISBN-13: 978-0815345084.
- Speicher, M.R., Antonarakis, S.E. and Motulsky, A.G. (2010). 4th Edition. *Vogel and Motulsky's Human genetics: Problems and approaches*. Berlin, Germany: Springer Verlag. ISBN: 978-3540376538.
- Wilson, G.N. (2000). 1st Edition. *Clinical genetics: A short course*. New York, USA: Wiley-Liss, ISBN: 978-047129806.