GENERIC ELECTIVE (BIOMED-GE-) GENETIC BASIS OF DISEASES

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)
		Lecture	Tutorial	Practical/ Practice		
Genetic Basis of Diseases	**4**	**3**	**-**	**1**	**XII Passed**	**Basic knowledge of Biology**

Learning Objectives

- The course is designed to provide insight about the importance of our genetic material.
- Students will be taught different types of changes that can take place in our genetic material and their repercussions.
- Students will be briefed as to how even minor changes in such a complex genetic system can lead to serious defects and disorders.

Learning outcomes

Having successfully completed this course, students will understand:

- The basic structural arrangement of our genetic material, its location within the cells and how it contributes to the unique features of each individual organism.
- Possible changes that can occur in the chromosomes at the macro level and what serious consequences this might have to the bearing individuals will be taught to the students.
- Not only the structural features but also the correct dose of the chromosomes present in our cells plays an important role in regulating normal body functioning. The same will be taught by citing examples of disorders associated with both extra as well as deficient chromosome numbers.
- The basic Mendelian pattern of inheritance. Students will also learn about different changes that can occur within a single gene, the diseases associated with them and how these changes can be inherited from one generation to the next.

SYLLABUS

Unit I: Organization of human genome (09 Hrs)

Basic structure of DNA and chromosomes, euchromatin, heterochromatin. A brief overview of the human nuclear and mitochondrial genome, Concept of allele, haploid and diploid. Genetic Variations-Polymorphism vs mutations. Types of mutations: Somatic vs germline.

Unit II: Structural chromosomal abnormalities (06 Hrs)

Different types of structural chromosomal abnormalities (deletions, duplications, inversions and translocations) and their associated disorders (Cri-du-chat, Wolf-Hirschhorn, Charcot-Marie-Tooth disease Type 1, Pallister Killian, Hunter syndrome, Walker-Warburg, CML).

Unit III: Numerical Chromosomal abnormalities (06 Hrs)

Concept of non-disjunction anaphase lagging, genomic imprinting, uniparental disomy, euploidy, aneuploidy and associated disorders (Down Syndrome, Edward Syndrome, Patau Syndrome, Turner Syndrome, Klinefelter Syndrome, Prader-Willi Syndrome, Angelman Syndrome).

Unit IV: Monogenic Disorders (12 Hrs)

Mendelian inheritance (autosomal and sex-linked). Types of gene mutations (substitution, indels, dynamic) and associated disorders: (Achondroplasia, Huntington's disease, sickle cell anaemia, cystic fibrosis, thalassemia, Rett Syndrome, haemophilia, colour blindness, phenylketonuria, albinism, maple syrup urine disease, alkaptonuria).

Unit V: Other genetic disorders (07 Hrs)

Multifactorial disorders like Cancer, Alzheimer's disease, Arthritis, Diabetes

Unit VI: Genetic counselling (05 Hours)

Invasive and non-invasive methods of prenatal diagnosis and screening (Down syndrome, Thalassemia). Genetic counselling for risk assessment and possible treatment and management strategies.

Practical component (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. PCR for polymorphism detection
2. Study of chromosomal abnormalities through karyotypes
3. Pedigree charts for disorders likeHuntington's disease, colour blindness, sickle cell anaemia
4. Pedigree analysis for determining inheritance and risk assessment
5. Case studies for disorders like cancer, diabetes
6. Case studies for genetic counselling
7. Determination of linkage and cross-over analysis (through two point test cross and three point test cross data).
8. Analysis of Tetrads from *Saccharomyces cerevisiae.*

Essential readings:

- Klug, W. S., Cummings, M., Spencer, C. A., Palladino, M. A., Darrell K. (2019). 12th Edition. Concepts of genetics. San Francisco, NY:Pearson ISBN-13: 9780134604718.
- Snustad, D.P. and Simmons, M.J. (2019). 7th Asia Edition. Principles of genetics. New York, USA: John Wiley and Sons. ISBN-13: 9781119657552.
- Strachan, T. and Read, A. (2018). 5th Edition. *Human molecular genetics*. Florida, USA: CRC Press, Garland Science. ISBN: 978-0815345893.
- Gardner E. J., Simmons M. J. and Snustad D. P. (2006). 8th edition Principles of genetics. USA. Wiley. ISBN-13: 978-8126510436.

Suggestive readings:

- 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: 978-0471298069.