LS 505A	Human Genetics	2 Credits
Name of the Faculty: Prof. R. Muthuswami*, Dr. Ekta Rai		

Sr.No.	Topic	Faculty Name/ Contact Hours
1.	**Organization of Human Genome** • Nuclear and mitochondrial genome • Mitochondrial genome organization, homoplasmy and heteroplasmy, • Karyotyping- G and R stain, C stain, FISH, and SKY • Protein coding genes- Alternative splicing, pseudogenes, gene families, Genes-within-genes, overlapping genes • Non-coding genes- tRNA, rRNA, small ncRNA, lncRNA, piRNA, endogenous siRNA • Repetitive elements- Satellite DNA, Mini satellites, microsatellites • Transposable elements- DNA transposons, LTR retroposons, non-LTR retroposons	RM/5
2.	**Mapping Techniques** • DNA markers in human genetics • Genetic mapping- Radiation hybrid mapping, Linkage analysis, LOD score • Physical mapping- Contig mapping, how the human genome was sequenced	ER/4
3.	**Mutations and Human Diseases** • Monogenic, oligogenic, and polygenic disorders • Mode of inheritance of monogenic disorders- dominant vs recessive; autosomal vs X-linked, pedigree analysis • Identifying disease genes- using genetic markers, position-dependent cloning, position-independent cloning • Allelic heterogeneity, Locus heterogeneity, Clinical heterogeneity, Compound heterozygosity • Penetrance and expressivity • Oligogenic disorders • Polygenic disorders- Linkage disequilibrium, GWAS studies to identify SNPs • Trinucleotide repeat disorders • Chromosomal aberrations • Genomic imprinting • Mitochondrial disorders	ER/12
4.	**Animal models for Human Diseases** • Different types of animal models • Creating animal models	RM/3
5.	**Gene Therapy and identification of mutations** • Virus based transfection strategies • Non-virus based transfection strategies • Gene therapy approaches for polygenic disorders	RM/4

Further Reading:

1. Human Molecular Genetics by Stratchan and Read