Optional course

LS 506 Human Genetics (2 credits)

R Muthuswami/New faculty

S No	Topic	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	5
2.	Mapping Techniques • DNA markers-RFLP, AFLP, SSR, RAPD • Genetic mapping- Radiation hybrid mapping, Linkage analysis, LOD score • Physical mapping- Contig mapping, how the human genome was sequenced • Introduction to NGS and its applications	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	12
4.	Animal models for Human Diseases Different types of animal models Creating animal models	3

5.	Gene Therapy and identification of mutations Virus based transfection strategies Non-virus based transfection strategies Gene therapy approaches for polygenic disorders	4

Recommended Reading:
Human Molecular Genetics by Stratchan and Read\