LSC 525 - Human Genetics

1. Genes in pedigree - Behaviour of monogenic, oligogenic and polygenic traits/diseases, behaviour and characteristics of the patterns of inheritance of the DNA sequence, allelic, locus and clinical heterogeneity; complementation study through pedigrees.

2. Complications to the basic pedigree patterns - pseudo-dominant pedigree patterns, non-penetrance, imprinting, anticipation, germ line and somatic mosaicisms.

3. General organization of human genome, mitochondrial genome organization, nuclear genome organization; size and banding of human chromosomes; distribution of tandems and interspersed repetitive DNA, gene distribution and density in human nuclear genome; organization of genes coding for rRNA, mRNA, small nuclear RNA.

4. Overlapping genes, genes within genes, gene families, pseudo genes, truncated genes and gene fragments

5. Gene mapping; Human Genome project –techniques and technology involved in genome mapping, low and high resolution mapping; approaches to physical and genetic mapping; principles and strategies for identifying unknown disease or susceptibility genes; beyond genomics – the physical and genetic mapping the post genomic era.

6. Pathogenic mutations, polymorphism versus mutations, types of mutations; DNA repair associated syndromes, Molecular pathology – evaluation and database of pathogenic mutations; mitochondrial mutations.

7. Animal models for human diseases, types of animal models, transgenic animals modelling complex diseases, procedures of production, detection and use in the study of different diseases.

8. Principles of molecular genetic based therapies, classical gene therapy, gene therapy for inherited disorders; gene therapy for neoplastic disorders and infectious diseases; genetic testing of diseases; ethics of genetic testing and gene therapy.

Suggested Readings

1. Human Molecular Genetics – by Strachen and Read

2. Principles of Human Genetics – by Curt Stern