BTH402 **MOLECULAR GENETICS** **Hours: 52**

Course outcome

This course explains and discusses the following:

CO 1. Mendelian laws of inheritance, deviations and exceptions to these laws.
CO 2. various types of recombination in Bacteria including transformation, transduction and conjugation
CO 3. various types of mutations at the molecular level and types of DNA repair to fix the mutations upon DNA damage.
CO 4. mobile genetic elements-transposable elements, mechanism of translocation and their distribution from prokaryotes to higher organism.
CO 5. population genetics, and about genotype and allelotype frequency calculation.
CO 6. forward and reverse genetics along with gene silencing techniques and gene knockout

UNIT I (13 hrs)

Mendelian genetics, symbols and terminology, principle of segregation, principle of independent assortment, multiple alleles, interaction of genes, pleiotropy; Deviations and exceptions to Mendelian ratios – variation of dominance, multiple alleles, sex-linkage, linkage and crossing over and chromosome mapping. Sex determination, dosage compensation and extrachromosomal inheritance (e.g. *Chlamydomonas*, snail, *Neurospora* and yeast).

UNIT II (13 hrs)

Identification of DNA as genetic material, experiments of Griffith, Avery MacLeod and McCarthy. Molecular mutation (mechanisms of missense, nonsense, transition, transversion and frame-shift mutation, lethal mutation, origin of spontaneous mutation and control)
Recombination in bacteria: Transformation, transduction and conjugation. DNA damage – mechanical and chemical; types of DNA repair, photo-reactivation, base excision, recombination, mismatch, SOS.

UNIT III (13 hrs)

C-value paradox, co-linearity of genes, split genes, gene families. Study of model systems: *Drosophila, Arabidopsis* and human beings. Chromosome analysis, karyotyping, cytogenetic mapping, Fluorescent In-situ Hybridization (FISH) Technique, Comparative genomic hybridization. Human Cytogenetics: Human karyotype construction. Mendelian and chromosome based heritable diseases and syndromes (colour blindness, retinoblastoma, haemophilia, cystic fibrosis, sickle cell anaemia, Down's syndrome, Klinefelters's syndrome, Turner's syndrome, Edward's syndrome and Cri-du-chat syndrome), Prenatal diagnosis (amniocentesis and chorionic villus sampling). Genetic counseling.

UNIT IV (13 hrs)

Transposable elements, Discovery, types and their significance in bacteria and Eukaryotes.
Population and evolutionary genetics: Genetic variation, Hardy-Weinberg equilibrium, inbreeding, outbreeding and changes in allelic frequency. Epigenetics, functional perturbation, knockdown (interference RNA, small interference RNA), knockout technology, micro RNA.Genetics and evolution.

References

1) Basic Genetics. Hartl D.L. & Jones E.W. Jones & Bartlett Pub., 1998
2) Genes. Lewin B., Oxford Univ. Press, 2000