

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