ZOE456: HUMAN GENETICS

Teaching 10 hours/Unit

COURSE OUTCOME

1. The process of cell division and sex determination in human
2. Genesis and effect of Chromosomal abnormalities in human
3. Structure of DNA and Gene, Mutation, DNA fingerprinting
4. Mendelian and Non Mendelian inheritance in human
5. Basis of autosomal dominant and recessive traits and sex linked inheritance
6. Principle of hardy-Weinberg's law and important of twin study
7. Genetic counselling and it's important for the genetic disorder

UNIT-I

Cytogenetics: Cell cycle, Mitosis, Meiosis, Gametogenesis, Fertalization.Human Chromosomes- Chromosome Morphology, Karyotyping and its application. Sex Chromatin-Barr Body, Lyon's Hypothesis, Sex determination, Genetic significance of X Inactivation. Milestones in the development of genetics

UNIT-II

Chromosomal Aberrations: Structural aberrations, abnormalities: Structural abnormalities: Deletions, translocation, Insertion, Inversion, Isochromosomes, Ring chromosomes. Factors Playing role in Chromosomal Aberrations. Antosomal Abnormalities-Down's syndrome (Trisomy-21), Edward's syndrome (Trisomy-18), Patau's syndrome (Trisomy-13). Sex chromosome abnormalities- Klinefelter's syndrome and Turner's syndrome

UNIT-III

Molecular Genetics: Structure of Nucliec acid, Types of DNA-Unique sequences, Satellite DNA, Interspersed repetitive DNA sequences, Single nucleotide polymorphism (SNPs) Short Tandem repeats(STRs), Variable number of tandem repeats (VNTRs), Restriction fragment length polymorphism (RFLP), Mitochondrial DNA, Triplet code. Brief introductions to Genes and its structure, Mutation, Gene bank, Recombinant DNA, DNA fingerprinting technology, DNA markers used for tracing human ancestry.

UNIT-IV

Modes of inheritance: Mendel and Mendelism, Mendel's laws,Brief Introduction to Alleles, Phenotype, Genotype, Dominant and Recessive alleles, Wild type and mutant alleles, Codominant Alleles, Lethal Alleles, Multiple Alleles, Heterozygotes, Homozygotes, Penetrance and Expressivity. Pedigree analysis, Mode of Mendelian inheritance in human

(single gene disorders): Autosomal dominant, autosomal recessive, X-linked dominant,X-linked recessive. Non mendelian Inheritance -Polygenic/complex inheritance and extra chromosomal inheritance – Erythroblastosis Fetalis in Humans

UNIT-V

Population Genetics: Definition of population genetics, Calculation of allele frequencies (MN and ABO blood groups); Random mating, Hardy-Weinberg's Law, factors influencing Hardy-Weinberg equilibrium, Endogamy, consanguineous marriage. Eugenics, Twins and type of twins Prenatal diagnosis: Invasive and non-invasive techniques, Amniocentesis, Chorionic villus sampling, Ultrasound, Foetoscopy, Foetal blood sampling (FBS), Maternal serum screening, Carrier screening for autosomal recessive and X-linked disorders, Genetic Counselling.

REFERENCES

1. Elrod, S., & Stansfield, W. (2010) *Schaum's Outline of Genetics*. McGraw Hill Professional.
2. Falconer, D. S., and Mackay, T.F. (1996) *Introduction to quantitative genetics*, Essex, England: Longman.
3. Gangane, S.D. (2017) *Human Genetics*, 5th ed. New Delhi, Elsevier.
4. Gersen, S., & Keagle, M. (2013). The principles of clinical cytogenetics. S. L. Gersen, & M. B. Keagle (Eds.). New York: Springer
5. Hartl, Daniel L., and Elizabeth W. Jones (1998) *Genetics: principles and analysis*, Sudbury, Mass: Jones and Bartlett Publishers.
6. Jorde LB, Carey JC and Bamshad, M.J.(2009) Medical Genetics, Elsevier Publication
7. Orlando J. Miller (2000) Human Chromosomes Springer-Verlag New York
8. Snustad, D P., and Simmons M.J. (2012) *Principles of genetics*. Hoboken, NJ: Wiley.
9. Strachan T and Read, A.P. (2011), Human Molecular Genetics, Garland Science/Taylor and Francis Group Publication, 4th Edition.
10. Steven L. Gersen and Martha B. Keagle (1999) The Principles of Clinical Cytogenetics Humana Press