

**Adikavi Nannaya University**  
**CBCS Semester System**  
**B.Sc. Human Genetics**  
**Semester - V**

**PAPER-V : STATISTICS AND INFORMATICS IN HUMAN GENETICS**

**Unit 1 Descriptive Statistics**

- 1.1 Methods of presentation and interpretation of data – frequency distribution, graphical representation of data, histogram, frequency polygon, frequency curve.
- 1.2 Measures of Central tendency – mean, median, mode
- 1.3 Measures of Dispersion - standard deviation, variance.

**Unit 2 Elementary Probability**

- 2.1 Mathematical definition of probability of an event, Use of permutations and combinations in calculations of Probability
- 2.2 Conditional probability, Additive and Multiplication law of Probability.
- 2.3 Probability Distributions: Binomial, Poisson and normal distributions.

**Unit 3 Correlation analysis, test of significance and ANOVA**

- 3.1 Correlation and regression analysis— Relationship between variables
- 3.2 Test of significance – statistical and scientific hypothesis, null and alternative hypothesis, procedure of hypothesis testing,
- 3.3 Test of significance – student's t test, chi-square test.
- 3.4 ANOVA – general idea of one way and two way analysis

**Unit 4 Computers, operating systems and Internet**

- 4.1 Principles of computer operations –basic computer architecture, hardware architecture
- 4.2 Principles of computer operations – software architecture, operating systems, Programming languages –traditional and scripting languages, Java, markup languages, application programs
- 4.3 Internet Services, email, WWW search engines

**Unit 5 Bioinformatics**

- 5.1 History of Bioinformatics
- 5.2 Databases and search tools – NCBI, EBI, GenomeNet; Database mining tools – BLAST
- 5.3 Database archives – nucleic acid sequence databases, genome databases and genome browsers, protein sequence databases, databases of protein families,.

## **Practicals**

1. Frequency distribution
2. Various types of graphs
3. Mean, Median, Mode
4. Standard deviation, variance and coefficient of variation
5. Testing of hypotheses regarding population mean
6. Testing of hypotheses about the difference between population means
7. Chi-square test
8. Testing of Correlation Coefficient
9. Fitting of simple linear regression
10. One-way ANOVA & Two-way ANOVA
11. Internet basics
12. Sequence retrieval (protein and gene) from NCBI, Structure download (protein and DNA) from PDB
13. Molecular file formats - FASTA, GenBank, Genpept, GCG, CLUSTAL, Swiss-Prot, FIR.

## **Suggested Readings**

1. Fowler, J., Cohen, L. and Jarvis, P. (1998). Practical Statistics for Field Biology. John Wiley and Sons, 2nd ed. .
2. Bland, M. (2006). An Introduction to Medical Statistics. Oxford University Press, 3rd ed.
3. Finney, D.J. (1980). Statistics for Biologists. Chapman and Hall Ltd.
4. Wayne, W, Daniel (1999). Biostatistics: A Foundation for Analysis in Health Sciences. John Wiley and Sons, 7th ed.

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**Paper - VI: Human Biochemical and Cytogenetics**

**UNIT-1**

The concept of Genetic Polymorphism

Blood Groups: ABO, MN and Rh systems, ABH saliva secretion

**UNIT-2**

Hemoglobin – Structural Diversity & Hemoglobinopathies

Serum Proteins Haptoglobin

Heterochromatin and genetic inactivation : Lyon's hypothesis

**UNIT-3**

Inborn errors of metabolism : Albinism, Phenylketoneuria and Alkaptonuria.

Pharmacogenetics: glucose-6-phosphatedehydrogenase deficiency, Pseudocholinesterase deficiency

**UNIT-4**

Human somatic chromosomes - Nomenclature : Standardization in Human Cytogenetics - Denver Report (1960), London Report (1953), Chicago Report (1966), Paris Report (1971)

Morphological variability of Human chromosomes.

**UNIT-5**

Chromosomal abnormalities in Man - Numerical aberrations –Classical syndromes (Down syndrome, Edward syndrome, Patau syndrome, Turner syndrome, Klienfelter syndrome)

Structural aberrations- Cri-du- chat syndrome, Wolf-Hirsch horn syndrome

**III Year Practical VI: Practicals in Human Biochemical and Cytogenetics.**

Slide test for sickle cell Haemoglobin, Hb typing on Paper electrophoresis. BCB dye test for G6PD enzyme deficiency, Hb estimation by Haemoglobinometer. Starch - agarose gel electrophoresis for Hb and G6pD systems, single chemical screening tests and paper chromatography for screening amino acidurias.

Analysis of Interphase Nuclei - Buccal smear and blood smear Preparation of Karyotypes of normal male and female Metaphase drawing