ADIKAVI NANNAYA UNIVERSITY – RAJAMAHENDRAVARAM
HUMAN GENETICS
B.Sc. I YEAR – SEMESTER – I
(Revised Syllabus 2020-21)

4. Details of course-wise Syllabus

B.Sc.	Human Genetics (Semester-I)	Credits: 4+1
Paper-1	Genetics & Human Heredity	Hrs/Wk: 4+2

1. **Aim and objectives of Course:**

a) To learn inheritance patterns of genes
b) To get clear idea about sex determination
c) To have knowledge about extra chromosomal inheritance
d) To understand the genetic mapping
e) To learn chromosomal abnormalities

2. **Learning outcomes of Course**:
Comprehensive and detailed understanding of different types of inheritance patterns of genes and chromosomal abnormalities

3. **Detailed Syllabus: (Five units with each unit having 12 hours of class work):**

Unit-1: **Mendelian Genetics and Extensions**
1.1 Physical basis of Heredity – Structure and function of Cell, Nucleus and Chromosome
1.2 Mendelian Principles of inheritance – Law of segregation, Law of independent assortment –animal examples; Mendelian inheritance of human traits; Chromosome theory of inheritance.
1.3 Incomplete dominance and codominance; Multiple alleles, Lethal alleles, Pleiotropy, Penetrance and Expressivity – human examples
1.4 Two gene interactions – Epistatic, non-epistatic interactions; Polygenic inheritance in man and other animal organisms.
1.5 Genes and environment –norm of reaction, phenocopies, developmental noise

Unit-2 - **Sex Linked Inheritance and Sex Determination**
2.1 Sex Linked Inheritance - Sex linked inheritance in *Drosophila* and human; Sex limited and Sex influenced inheritance with examples.
2.2 Sex Determination - Sex determination in *Drosophila* – Genic balance theory
2.3 Sex determination in eukaryotes –heterogametic, homogametic, haplo-diploidy, role
of environmental factors, mosaics
2.4 Sex determination in mammals- and role of human Y chromosome, Dosage compensation

Unit-3 **Extra-chromosomal Inheritance**

3.1. Mitochondrial inheritance (petite mutations); Mitochondrial inheritance in man
3.2. Maternal inheritance-shell coiling in snail, *Ephestia* pigmentation
3.3. Infective heredity- symbionts in *Drosophila*, Kappa particles in *Paramecium*.
3.4. Epigenetics and genome imprinting in humans

Unit-4**: Linkage, crossing over and chromosome mapping**

4.1. Linkage, Crossing over and Recombination.
Linkage and chromosome mapping in eukaryotes – cytological basis of crossing over; recombination frequency, two factor and three factor crosses; interference and coincidence; Mitotic recombination
4.2 Linkage and chromosome mapping in prokaryotes – bacteria and bacteriophages – transformation, transduction, conjugation; gene mapping in bacteria.
4.3 Genetic definition of gene - Complementation test, intragenic complementation, rII locus of phage T4

Unit-5: **Variation in Chromosome number and structure**

5.1. Specialized chromosomes -Lampbrush chromosomes. Polytene chromosomes: Supernumerary chromosomes.
5.2.Variation in chromosome structure - Deletion, Duplication, Inversion, Translocation; Position effect
5.3. Variation in chromosome number - Euploidy and Aneuploidy in man

Recommended Text Books and Reference books:

1. Gardner, E.J., Simmons, M.J., Snustad, D.P. (1991). Principles of Genetics, John Wiley & sons, India.8th edition.
2. Snustad, D.P. and Simmons, M.J. (2010). Principles of Genetics, John Wiley & Sons Inc., India.5th edition.
3. Klug, W.S., Cummings, M.R., Spencer, C.A. (2012). Concepts of Genetics. Benjamin
Cummings, U.S.A. 10th edition.
4. Griffiths, A.J.F., Wessler, S.R., Carroll, S.B., Doebley, J. (2010). Introduction to Genetic
Analysis. W. H. Freeman and Co., U.S.A. 10th edition.

4. Details of Lab/Practical/Experiments/Tutorials syllabus:

Genetics and Human Heredity Lab

1. Mitosis & Meiosis through temporary squash preparation.
2.Mendel’s laws through seed ratios& Drosophila mutants.
3. Study of linkage, recombination, chromosome mapping using test cross data.
4. Study of human genetic traits: Sickle cell anaemia, Xeroderma Pigmentosum, Albinism. Tests for red-green Colour blindness, Widow’s peak, Rolling of tongue, Hitchhiker’s thumb and Attached ear lobe.
5. Incomplete dominance and gene interaction through seed ratios
6. Blood Typing: ABO groups & Rh factor.

7. Study of aneuploidy: Down's, Klinefelter's and Turner's syndromes.
8. Smear technique to demonstrate sex chromatinin buccal epithelialcells

5. **Recommended Co-curricular activities:** (Co-curricular activities should not promote copying from text book or from others' work and shall encourage self/independent and group learning)

A. Measurable:

1. Assignments on: Genetic mapping
   Problems on Mono-hybrid cross, Dihybrid cross and Gene interactions
2. Student seminars (Individual presentation of papers) on topics relating to:
   Inheritance patterns
   Extra chromosomal inheritance
   Sex determination
   Chromosome mapping
   Chromosomal abnormalities
3. Quiz Programmes on: Mendelian Genetics
4. Individual Field Studies/projects: Pedigree analysis
5. Group discussion on: Inheritance patterns
6. Group/Team Projects on: Identifying inheritance patterns of diseases from any hospital data.

B. General

1. Collection of news reports and maintaining a record of paper-cuttings relating to topics covered in syllabus
2. Group Discussions on:
3. Watching TV discussions and preparing summary points recording personal observations etc., under guidance from the Lecturers
4. Any similar activities with imaginative thinking.

6. **Recommended Continuous Assessment methods:**

Class tests
Seminars
Group discussions
Poster making
Power point presentation

5. Blue Print of Model Question Paper (Theory)

Semester End Examination

Time: 3hrs **Max. Marks: 75M**

Section A

Answer any FIVE questions. All questions carry equal marks. 5X5=25M
Total no. of questions =08.
Questions 1to 5 from each unit and 6 to 8 questions from any unit.

Section B

Answer ALL questions. All questions carry equal marks. 5X10=50M

TEN questions, TWO from each unit with internal choice (a or b)

6. MODEL QUESTION PAPER

Semester End Examination
HUMAN GENETICS
I SEMESTER

B.Sc.	BIOCHEMISTRY (Semester - I)	Max. Marks: 75M
Paper-I	GENETICS & HUMAN HEREDITY	3Hrs

Section A

I. Answer any FIVE questions. All questions carry equal marks. 5X5 = 25

1. Incomplete dominance & codominance
2. Epistasis
3. Haemophilia
4. Heterogametic female
5. Kappa particles
6. Crossing over
7. Translocation
8. Recombination

Section B

II. Answer ALL Questions. All questions carry equal marks 5X10 = 50

9. (a) Explain law of independent assortment with suitable examples.

(or)

(b) Write an essay on gene and environment.

10. (a) Explain the mechanism of sex-determination in *Drosophila*.

(or)

(b) Write about sex-linked inheritance in humans.

11. (a) Write an essay on mitochondrial inheritance in man.

(or)

(b) Explain in detail about infective heredity.

12. (a) Write an essay on linkage.

(or)

(b). Explain intragenic complementation by using rII locus of T4 bacteriophage.

13. (a) Write about aneuploidy.

(or)

(b) Explain in detail about lamp brush and polytene chromosomes.

B.Sc.	**HUMAN GENETICS (Semester - II)**	**Credits: 4+1**
Paper - 2	**HUMAN GENETICS & CYTOGENETICS**	**Hrs/Wk: 4+2 (T+L)**

1. Aim and objectives of Course

   a) To analyze complex traits inheritance
   b) To do mapping of mendelian and complex traits
   c) To know the complications of pedigree analysis for monogenic and complex traits

2. Learning outcomes of Course
   Help the student to understand the principles of linkage and chromosome mapping. Mapping provides clear idea about the diseased genes, their location and inheritance patterns

3. Detailed Syllabus: Five units (i.e., each unit having 12 hours of class work)

   Unit 1 Basic Human Genetics – Monogenic traits
   1.1 History of Human Genetics.
   1.2 Traits and their classification
   1.3 Pedigrees – family history, symbols, construction of pedigree
   1.4 Monogenic traits - autosomal inheritance, sex-linked inheritance, sex-limited and sex
   influenced inheritance, mitochondrial inheritance

1.5 Complications in pedigree patterns – non-penetrance, expressivity, pleiotropy, genetic heterogeneity, genomic imprinting, uniparental disomy, male lethality, X inactivation, consanguinity

Unit 2 Basic Human Genetics – Complex traits

2.1 Approaches to analysis of complex traits - Nature vs nurture, monozygotic and dizygotic twins
2.2 Polygenic inheritance of continuous traits – normal growth charts, dysmorphology
2.3 Polygenic inheritance of discontinuous traits – threshold model, liability and recurrence risk
2.4 Genetic susceptibility in multifactorial disorders – diabetes

Unit 3 Genetic Mapping of Mendelian and Complex characters

3.1 DNA Recombination and Crossing over
3.2Identifying recombinants and non-recombinants in pedigrees
3.3 Genetic and physical map distances – genetic markers, mapping of genetic traits
3.4 Two point mapping – LOD score analysis, multipoint mapping, Homozygosity mapping
3.5 Genetic mapping of complex traits – difficulties in mapping, allele sharing methods, sib pair analysis, allelic association, linkage disequilibrium mapping

Unit 4 Human Chromosomes

4.1 History of human cytogenetics
4.2 Cell cycle – mitosis and meiosis.
4.3 Human karyotype – banding, nomenclature of banding
4.4 Nomenclature of aberrant karyotypes

Unit 5 Chromosome anomalies

5.1 Common syndromes due to numerical chromosome changes
5.2 Common syndromes due to structural alterations (translocations, duplications, deletions, microdeletions, fragile sites)
5.3Common chromosome abnormalities in cancer

Recommended Text Books and Reference books:

1. Human Genetics: Concept and Application by Ricki Lewis 10 th Edition
2. Vogel and Motulsky's Human Genetics: Problems and Approaches
3. The Principles of Clinical Cytogenetics by Steven L. Gersen, Martha B. Keagle 3 rd edition.
4. Human Cytogenetics: Constitutional Analysis: a Practical Approach by Denise E. Rooney.

4. Details of Lab/Practical/Experiments/Tutorials syllabus:

1. Preparation of pedigree charts for blood group, tongue rolling, ear lobes and Color blindness.
2. Pedigree analysis for dominant and recessive autosomal and sex-linked traits.
3. Genetics of codominant genes – blood groups.

4. Barr Body analysis.
5. Dermatoglyphics
6. Polygenic inheritance – finger print ridge count
7. Preparation of metaphase chromosome spread using peripheral blood sample.
8. Sterilization techniques for leukocyte culture
9. Inoculation and Culture of human leucocytes
10. Preparation of metaphase plates and their staining and analysis
11. Human karyotyping – numericals on chromosome number.
12. Camera-lucida drawing of chromosomes.
13. Micrometric analysis of chromosomes.
14. Study of various abnormal karyotypes observed in humans.
15. G- banding of metaphase plates and their analysis
16. Sister Chromatid exchange analysis from peripheral blood

5. Recommended Co-curricular activities: (Co-curricular Activities should not promote copying from text book or from others' work and shall encourage self/independent and group learning)

   A. Measurable:
      1. Assignments on: Complications of pedigree analysis
         Genetic mapping
      2. Student seminars (Individual presentation of papers) on topics relating to:
         Monogenic traits
         Polygenic inheritance
      3. Quiz Programmes on: Pedigree analysis
         Monogenic traits
         Polygenic inheritance
      4. Individual Field Studies/projects: Syndromes
      5. Group discussion on: chromosomal aberrations
      6. Group/Team Projects on: Human karyotyping analysis of genetic disorders

   B. General
      1. Collection of news reports and maintaining a record of paper-cuttings relating to topics covered in syllabus
      2. Group Discussions on:
      3. Watching TV discussions and preparing summary points recording personal observations etc., under guidance from the Lecturers
      4. Any similar activities with imaginative thinking.

6. Recommended Continuous Assessment methods:

   Class tests
   Seminars
   Group discussions
   Poster making
   Power point presentation

SEMESTER – II
Semester End Examination - Theory
MODEL QUESTION PAPER

B.Sc.	HUMAN GENETICS (Semester: II)	Max. Marks: 75M
Paper - 2	HUMAN GENETICS & CYTOGENETICS	3Hrs

Section A

Answer any FIVE questions. All questions carry equal marks. 5X5 = 25

I.

1. Holandric inheritance
2. Pleiotropy
3. Concordance and Discordance
5. Polygenic inheritance
6. Sib pair analysis
7. Interphase
8. Turner's syndrome
9. Robertsonian translocation

Section B

II. Answer ALL Questions. All Questions carry equal marks 5X10 = 50

9. (a) Write about the inheritance of monogenic characters

(or)

(b) Write an essay on pedigree

10. (a) Explain the role of twin studies in understanding complex traits

(or)

(b) Write about genetic susceptibility of multifactorial disorders

11. (a) Write an essay on two-point mapping.

(or)

(b) Explain in detail about genetic mapping of complex traits.

12. (a) Write an essay on meiosis.

(or)

(b). Explain the different events in the history of human genetics

13. (a) Write about autosomal chromosomal abnormalities.

(or)

(b) Explain in detail about chromosomal abnormalities of cancer.