UG PROGRAM (4 Years Honors)
CBCS - 2020-21

Subject
Human Genetics

Syllabus and Model Question Papers

DETAILS OF COURSES AND CREDITS

Sem	Course No	Course Name	Course Type (T/P/L)	Hrs/Week	Credits	Max. Marks	Max. Marks
				Science: 4+2	Science: 4+2	Count/Internal/ Mid Assessment	Sem- End Exam
I	1	Genetics & Human Heredity	T	4	4	25	75
	2	Genetics and Human Heredity Lab	L	2	1	-	50
II	3	Human Genetics & Cytogenetic	T	4	4	25	75
	4	Human Genetics & Cytogenetic Lab	L	2	1	-	50
III	5	Human Molecular Genetics	T	4	4	25	75
	6	Human Molecular Genetics Lab	L	2	1	-	50
IV	7	Recombinant DNA and Stem Cell Technology	T	4	4	25	75
	8	Recombinant DNA and Stem Cell Technology Lab	L	2	1	-	50
	9	Statistics and Informatics In Human Genetics	T	4	4	25	75
	10	Statistics and Informatics in Human Genetics Lab	L	2	1	-	50

Note: * Course Type Code : T-Theory, L - Lab, P: Problem solving

4. Details of course-wise Syllabus

B.Sc.	Human Genetics	Credits: 4
Course-1	Genetics & Human Heredity	Hrs/Wk: 4

Aim and objectives of Course:

- To learn inheritance patterns of genes
- To get clear idea about sex determination
- To have knowledge about extra chromosomal inheritance
- To understand the genetic mapping
- To learn chromosomal abnormalities

Learning outcomes of Course:

- Comprehensive and detailed understanding of different types of inheritance patterns of genes and chromosomal abnormalities

UNIT I: 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 II: 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 III: 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 IV: 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 V: 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

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.

B.Sc.	Human Genetics	Credits: 1
Course-1(L)	Genetics & Human Heredity	Hrs/Wk: 2

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 & amp; 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 chromatin in buccal epithelial cells

1. **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 Courses) 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 Course-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.

2. Recommended Continuous Assessment methods:

Class tests
Seminars
Group discussions
Poster making
Power point presentation

5. Blue Print of Model Question Course(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)

Blue Print of Lab Model Question Course(Practical)

Time: 3hrs Max. Marks: 50M

1.	Major experiment	20M
2.	Minor experiment	15M
3.	Experiment principle	5M
4.	Record	5M
5.	Viva Voce	5M

MODEL QUESTION COURSE(Semester End)

B. Sc DEGREE EXAMINATIONS

SEMESTER -I
Course 1: GENETICS & HUMAN HEREDITY

Time: 3Hrs **Max.Marks:75**

Section - A

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

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

Section - B

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

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.

MODEL QUESTION COURSE
Semester End Examination - Practical
GENETICS & HUMAN HEREDITY LAB

Time: 3hrs **Max. Marks: 50M**

1.Major experiment **20M**

Prepare Mitotic slide with onion rootips by squash technique and report metaphase and anaphase stages

2.Minor experiment **15M**

Identify the ABO Blood group of the given subject and give report

3. Analyse the given pedigree and identify and identify the pattern of inheritance **5 M**

4. Record **5M**

5.Viva Voce **5M**

B.Sc.	Semester - II	Credits: 4
Course - 2	HUMAN GENETICS & CYTOGENETICS	Hrs/Wk: 4

Aim and objectives of Course

- To analyze complex traits inheritance
- To do mapping of Mendelian and complex traits
- To know the complications of pedigree analysis for monogenic and complex traits

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

UNIT I: 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 II: 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 III: Genetic Mapping of Mendelian and Complex characters

3.1 DNA Recombination and Crossing over
3.2 Identifying 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 IV: 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 V: 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

REFERENCE BOOKS:

1. Human Genetics: Concept and Application by Ricki Lewis 10th 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.

B.Sc.	Semester - II	Credits: 1
Course - 2(L)	Human Genetics & Cytogenetics Lab	Hrs/Wk: 2

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

HUMAN GENETICS & CYTOGENETICS LAB

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

2. **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 Courses) 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 Course-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.

5. Recommended Continuous Assessment methods:

Class tests
Seminars
Group discussions
Poster making
Power point presentation

MODEL QUESTION COURSE(Semester End)

B. Sc DEGREE EXAMINATIONS

SEMESTER -II

Course 2: HUMAN GENETICS & CYTOGENETICS

Time: 3Hrs **Max.Marks:75**

Section - A

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

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

Section - B

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

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.

MODEL QUESTION COURSE
Semester End Examination - Practical

Time: 3hrs **Max. Marks: 50M**

1.Major experiment **25M**
Analyse the dermatoglyphics of the given subject and give report.

2.Minor experiment **15M**
Prepare buccal smear and identify whether the given subject is barr body +ve or not.
3. Record **5M**
4.Viva Voce **5M**

B.Sc.	Semester-III	Credits: 4
Course-3	HUMAN MOLECULAR GENETICS	Hrs/Wk: 4

Aim and objectives of Course:

- To learn the structural organization of DNA, RNA & Proteins
- To get clear idea about the characteristics of DNA
- To have knowledge about the gene expression
- To understand the human chromosome and genome level organization

Learning outcomes of Course:

- Student will get clear idea of molecular level genome organization and Gene expression

UNIT I: DNA, RNA and Protein Structure

1.1.Building blocks and chemical bonds in DNA, – structure of DNA, A-B-Z and triplex DNA,
1.2 Building blocks and chemical bonds in RNA – Structure of RNA
1.3 Building blocks and chemical bonds in peptides- primary, secondary, tertiary and quaternary structure of proteins

UNIT II: DNA replication, recombination, Mutagenesis and DNA repair

2.1 Central dogma of molecular biology
2.2 DNA replication – semiconservative, semi-discontinuous, DNA replication machinery
2.3 DNA recombination
2.4 DNA mutagenesis
2.5 DNA repair

UNIT III: Human Chromosome Organization

3.1 Packaging of DNA – multiple hierarchies of DNA folding
3.2 Chromosomes as functional organelles –origins of replication, telomeres, centromeres
3.3 Heterochromatin and euchromatin

UNIT IV: Gene expression

4.1 DNA transcription
4.2 Post transcriptional modifications
4.3 Translation,
4.4 Post-translation processing

UNIT V:Human Genome organization

5.1 Mitochondrial genome – replication, genes, genetic code
5.2 Nuclear genome – protein coding genes, RNA genes
5.3 Nuclear genome – highly repetitive DNA, heterochromatin and transposon repeats

REFERENCE BOOKS:

1. Human Molecular Genetics by T. Strachan
2. Human Molecular Genetics by Gerard Meurant
3. Human Molecular Genetics by Christopher G Mathew.
4. Human Molecular Genetics by Sudbery
5. Human Genetics: From Molecules to Medicine by Christian Patrick Schaaf, Johannes Zschocke.

B.Sc.	Semester-III	Credits: 1
Course - 3(L)	Human Molecular Genetics Lab	Hrs/Wk: 2

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

HUMAN MOLECULAR GENETICS LAB

1. Extraction of DNA from human lymphocytes
2. Coursechromatography of amino acids
3. Electrophoresis: agarose gel electrophoresis, PAGE
4. Study of isozymes by PAGE
5. Comet assay to measure DNA damag
6. Problem based on homologous and site-specific recombination
7. Effects of mutagens on wt and repair deficient *E.coli* strains.
8. Preparation of Human chromosome spread and banding

4. **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: chromosome and genome organization
2.Student seminars (Individual presentation of Courses) on topics relating to: Gene expression
3.Quiz Programmes on: DNA, RNA and Protein structure
4.Individual Field Studies/projects: Barr body analysis
5.Group discussion on: DNA mutagenesis, recombination and Repair
6.Group/Team Projects on: Identification of mitochondrial inheritance patterns in mitochondrial diseases

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

3. Recommended Continuous Assessment methods:
Class tests
Seminars
Group discussions
Poster making
Power point presentation

MODEL QUESTION COURSE(Semester End)

B. Sc DEGREE EXAMINATIONS

SEMESTER -III

Course 3: HUMAN MOLECULAR GENETICS

Time: 3Hrs **Max.Marks:75**

Section - A

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

1. Post transcriptional modification.
2. Types of DNA.
3. Gene Expression
4. Structure of protein
5. Tautomerism
6. Barr body
7. Genetic code
8. Transposons

Section - B

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

9. (a) Write about the structure of DNA
   (or)
   (b) Write about different forms of RNA

10. (a) Explain the process of transcription
    (or)
    (b) Write about proteins synthesis

11. (a) Write about DNA replication
    (or)
    (b) Explain DNA recombination & repair

12. (a) Explain packaging of DNA in the chromosome
    (or)
    (b) Differentiate heterochromatin & euchromatin.

13. (a) Write about protein coding genes.
    (or)
    (b) Explain the differences between nuclear genome & Mitochondrial genome.

MODEL QUESTION COURSE
Semester End Examination - Practical

Time: 3hrs **Max. Marks: 50M**

HUMAN MOLECULAR GENETICS LAB

1.Major experiment	**25M**
Isolate DNA from the white blood cells and report	
2.Minor experiment	**15M**
Check the Quality of DNA by conducting Agarose gel electrophoresis	
3. Record	**5M**
4.Viva Voce	**5M**

B.Sc.	Semester-IV	Credits: 4
Course - 4	Recombinant DNA and Stem Cell Technology	Hrs/Wk: 4

Aim and objectives of Course:

- To learn about tools of recombinant DNA technology
- To have idea about applications of r DNA technology
- To have knowledge about cloning of Human diseased genes
- To understand biology of stem cells and applications

Learning outcomes of Course:

- Student will get clear idea about r DNA technology and stem cell technology and their applications in different fields.

UNIT I: Cell Based Cloning

1.1 Introduction to Cloning
1.2 Restriction endonucleases and other enzymes used in manipulating DNA molecules
1.3 Cloning vectors – plasmid vectors, lambda and cosmid vectors, P1 phage vectors, YAC, BAC, M13 or phagemid vectors, expression vectors
1.4 Introducing recombinant DNA into recipient cells
1.5 DNA libraries -generation of genomic and cDNA libraries; chromosomal DNA libraries

UNIT II: Screening Constructs

2.1 Nucleic Acid hybridization
2.2 Sequencing DNA- Sanger's method
2.3 PCR- Fundamentals, Designing of primers, Real-time PCR
2.4 Human-Rodent somatic cell hybrids

UNIT III: Cloning Human disease genes

3.1 Cloning human disease genes- functional candidate gene cloning, positional candidate gene cloning
3.2 Detection of mutations in human genes –SSCP analysis, DGGE, chemical mismatch cleavage
3.3 Detection of mutation in human gene – DNA sequencing, hetero-duplex analysis, protein truncation

UNIT IV:Applications of rDNA technology

4.1 DNA fingerprinting – use of mini-satellites for DNA fingerprinting, single locus probes, STRs
4.2 Genetic testing – prenatal testing, neonatal screening, diagnosis of genetic disease in children after birth, pre-symptomatic testing.
4.3 *In vivo, In vitro* gene therapy; vehicles for gene therapy; gene therapy for heritable and non-heritable genetic diseases.

UNIT V: Biology of stem cells and Applications

5.1 Historical perspectives, concept of stem cells
5.2 Cellular and molecular features of stem cells
5.3 Embryonic stem cells and germ stem cells
5.4 Foetal stem cells, adult stem cells and cancer stem cells
5.5 Medical need for stem cells and preservation of stem cells
5.6. Genetically engineered stem cells for gene therapy
5.7 Stem cell therapy – neurodegenerative disorders, cardiovascular disorders, metabolic disorders, hematopoietic disorders, organ disorders, autoimmune disorders, reproductive failures

RECOMMENDED TEXT BOOKS :

1. Gene Cloning and DNA Analysis (2010) 6th ed., Brown, T.A., Wiley-Blackwell publishing (Oxford, UK), ISBN: 978-1-4051-8173-0.
2. Principles of Gene Manipulation and Genomics (2006) 7th ed., Primrose, S.B., and Twyman, R. M., Blackwell publishing (Oxford, UK) ISBN:13: 978-1-4051-3544
3. Molecular Biotechnology: Principles and Applications of Recombinant DNA (2010) 4th ed., Glick B.R., Pasternak, J.J. and Patten, C.L., ASM Press (Washington DC), ISBN: 978-1-55581-498-4 (HC).
4. Human Molecular Genetics by Sudbery.

B.Sc.	Semester-IV	Credits: 1
Course - 4(L)	Recombinant DNA and Stem Cell Technology Lab	Hrs/Wk: 2

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

RECOMBINANT DNA AND STEM CELL TECHNOLOGY LAB

1. Isolation of plasmid DNA from *E. coli* cells.
2. Digestion of plasmid DNA with restriction enzymes.
3. Estimation of size of a DNA fragment after electrophoresis using DNA markers
4. Construction of restriction digestion maps from data provided
5. Recovery of DNA from low-melting temperature agarose gel
6. Preparation of competent cells of *E.coli*
7. Transformation of competent *E.coli* cells with plasmid DNA
8. Amplification of a DNA fragment by PCR.
9. Complementation of β-galactosidase for Blue and White selection.
10. Southern blotting
11. Western blotting.
12. Culturing cells – aseptic techniques, media
13. Sub-culturing and cell lines
14. Cryopreservation

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: PCR, nucleic acid hybridisation techniques, DNA sequencing
2. Student seminars (Individual presentation of Courses) on topics relating to: cloning of human diseased genes
3. Quiz Programmes on: Restriction enzymes, vectors of r DNA technology
4. Individual Field Studies/projects: Neonatal screening of genetic diseases
5. Group discussion on: Applications of r DNA technology and Stem cell technology
6. Group/Team Projects on: RFLP analysis of gene mutations.

B. General

1. Collection of news reports and maintaining a record of Course-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

MODEL QUESTION COURSE(Semester End)

B. Sc DEGREE EXAMINATIONS

SEMESTER -IV

Course 4: RECOMBINANT DNA AND STEM CELL TECHNOLOGY

Time: 3Hrs **Max.Marks:75**

Section - A

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

1. Plasmid vectors
2. Real time PCR
3. c-DNA Library
4. Neonatal screening
5. SSCP analysis
6. Embryonic stem cells
7. STR
8. Auto immune disorders

Section B

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

9. (a) Write an essay on cloning vectors.
   (or)
   (b) Explain the process of construction of DNA Library

10. (a) Write the process of DNA sequencing
    (or)
    (b) Explain the mechanism of PCR.

11. (a) Write the process of detection of mutations by
    1. SSCP analysis 2. DGGE 3. Chemical mismatch cleavage.
    (or)
    (b) Write any essay on human disease genes

12. (a) Write the methods of identifying genetic diseases in pre-natal condition.
    (or)
    (b) Write an essay on *In-vitro* gene therapy.

13. (a) Write about cellular and molecular features of stem cells
    (or)
    (b) Write an essay on stem cell therapy.

MODEL QUESTION COURSE
Semester End Examination - Practical

Time: 3hrs **Max. Marks: 50M**

RECOMBINANT DNA & STEM CELL TECHNOLOGY

1.Major experiment	**20M**
Isolate plasmid DNA from the given *E.coli* culture.	
2.Minor experiment	**15M**
Estimate the size of DNA fragments by using DNA markers	
3. Write the principle involved in polymerase chain reaction	**5 M**
4. Record	**5 M**
5.Viva Voce	**5 M**

B.Sc.	Semester-IV	Credits: 4
Course-5	Statistics and Informatics In Human Genetics	Hrs/Wk: 4

Aim and objectives of Course:

- To have clear idea about representation of data and analysis
- To learn about laws of probability and their applications
- To have knowledge about computer operations and internet
- To understand the usage of bio-informatics tools

Learning outcomes of Course:

- Student will get knowledge about statistical analysis of qualitative and quantitative characters, usage of computers and tools of bioinformatics

UNIT I: 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, coefficient of variation.

UNIT II: 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, Random Variables, Mathematical expectation and variances
2.3 Probability Distributions: Binomial, Poisson and normal distributions.
2.4 Bayes theorem

UNIT III: Correlation analysis, test of significance and ANOVA

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

UNIT IV: 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, mark up languages, application programs
4.3 Communication and Networks – network architecture, standards for exchange of information, internet services - email, WWW search engines

UNIT V: Bioinformatics

5.1 History of Bioinformatics
5.2 Databases and search tools – NCBI, EBI, Genome Net; Database mining tools – BLAST
5.3 Database archives – nucleic acid sequence databases, genome databases and genome browsers, protein sequence databases, databases of protein families, databases of structures, expression and proteomic databases, bibliographic databases
5.4 Gateways to archives –ENTREZ, PIR, ExPASy.

RECOMMENDED TEXT BOOKS:

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.

B.Sc.	Semester-IV	Credits: 1
Course-5(L)	Statistics and Informatics in Human Genetics Lab	Hrs/Wk: 2

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

Statistics and Informatics in Human Genetics Lab

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. Basics of Internet
12. Sequence retrieval (protein and gene) from NCBI, Structure download (protein and DNA) from PDB
13. Molecular file formats - FASTA, Gen Bank, Gen pept, GCG, CLUSTAL, Swiss-Prot, FIR

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: Mean, Median, Mode, Variance and Standard deviation
2. Student seminars (Individual presentation of Courses)
3. Quiz Programmes on: probability, Binomial expression, tests of association
4. Individual Field Studies/projects: Retrieving data by using Data mining tools like BLAST and FAST
5. Group discussion on: computer programming, Mark up languages.
6. Group/Team Projects on: Primer designing by BLAST

B. General

1. Collection of news reports and maintaining a record of Course-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

MODEL QUESTION COURSE(Semester End)

B. Sc DEGREE EXAMINATIONS

SEMESTER -IV

Course 5: STATISTICS AND INFORMATICS IN HUMAN GENETICS

Time: 3Hrs **Max.Marks:75**

Section - A

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

1. Histogram
2. Standard deviation
3. Probability law of multiplication
4. Student – t-test
5. Search engine
6. Poisson distribution
7. NCBI
8. BLAST

Section - B

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

9. (a) Write about graphical representation of data.

(or)

(b) Calculate measures of central tendency for the following data.

Class	0-10	10-20	20-30	30-40	40-50	50-60	60-70
f	2	5	10	20	15	12	8

10. (a) Write an essay on probability distribution.

(or)

(b) Explain laws of probability with suitable examples.

11. (a) Write an essay on correlation analysis.

(or)

(b) Write an essay on chi-square test for qualitative characters.

12. (a) Write about principles of computer operations.

(or)

(b) Write an essay on programming languages.

13. (a) Write about databases & search tools.

(or)

(b) Write an essay on nucleic acid sequence databases

MODEL QUESTION COURSE
Semester End Examination - Practical

Time: 3hrs **Max. Marks: 50M**

STATISTICS AND INFORMATICS IN HUMAN GENETICS

1.Major experiment **20M**

1. Calculate Mean and Standard deviation for the following data

Class	0-10	10-20	20-30	30-40	40-50	50-60	60-70
Frequency	2	5	10	20	15	12	8

2.Minor experiment **15M**

Calculate correlation coefficient for the following data

X	65	66	67	68	69	70	71
Y	67	68	66	69	72	71	69

3. What is WWW? Explain in-detail **5 M**
4. Record **5M**
5.Viva Voce **5M**