

ADIKAVINANNAYAUNIVERSITY::RAJAHMAHENDRAVARAM B.Sc Human Genetics Syllabus (w.e.f:2020-21A.B)

#### Skill Enhancement Courses (SECs) for Semester -V, From 2022-23(Syllabus-Curriculum)

#### Structure of SECs for Semester-V

#### (To choose One pair from the Four alternate pairs of SECs)

Year	Semester	Paper Code	Paper Title	Course Type (T/L/P)	Hrs./Week (Science:4+2)	Credits (Science:4+1)	Max. Marks Cont/Internal /Mid Assessment	Max. Marks Sem-EndExam	
				1	A - PAIR		•		
	v	HGT 6A	Clinical Genetics & Genetic Counseling	Т	4	4	25	75	
3 <sup>rd</sup>		HGP 6A	Clinical Genetics & Genetic Counseling Lab	L	2	1		50	
	Elective	HGT 7A	Laboratory Diagnosis In Genetics	Т	4	4	25	75	
	Papers *Any OnePair	HGP 7A	Laboratory Diagnosis in Genetics Lab	L	2	1		50	
	Offer an				B - PAIR				
	Elective Paper A Or B Or C	HGT-6B	Human Genome Project And Genomes	Т	4	4	25	75	
		HGP-6B	Human Genome Project and Genomes Lab	L	2	1		50	
		HGT-7B	Molecular Techniques In Genetic Engineering	Т	4	4	25	75	
			HGP-7B	Molecular Techniques in Genetic Engineering Lab	L	2	1		50
				·	C - PAIR				
		HGT-6C	Developmental & Behavioral Genetics	Т	4	4	25	75	
		HGP-6C	Developmental and Behavioral Genetics Lab	L	2	1		50	
		HGT-7C	Molecular Pathology In Human Diseases	Т	4	4	25	75	
		HGP-7C	Molecular Pathology in Human Diseases Lab	L	2	1		50	



#### Note: \*Course type code: T: Theory, L: Lab, P: Problem solving

\*Note: FIRST and SECOND PHASES (2 spells) of APPRENTICESHIP between 1st and

2nd year and between 2nd and 3rd year (two summer vacations)

\*Note: THIRD PHASE of APPRENTICESHIP Entire 5th / 6th Semester

**Note:-1:** For Semester-V for the domain subject Biochemistry, any one of the three pairs of SECs shall be chosen as courses 6 and 7, i.e., 6A & 7A or 6C & 7C. The pair shall not be broken (ABCD allotment is random, not on any priority basis).

**Note:-2:** One of the main objectives of Skill Enhancement Courses (SEC) is related to the domain subject in students. The syllabus of SEC will be partially skill oriented. Hence, teachers shall also impart practical training to students on the field skills embedded in the syllabus citing related real field situations.



B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:4
Course: 6A	Clinical Genetics & Genetic Counseling	Hrs/Wk:4

#### **UNIT-1 GENETIC DISORDERS I**

- 1.1 Monogenic diseases Cystic fibrosis, Tay-Sachs syndrome, Marfan syndrome
  - 1.2 Inborn errors of metabolism Phenylketonuria, Maple syrup urine syndrome, Galactosemia
  - 1.3 Genome imprinting syndromes Prader Willi and Angelman syndrome

# **UNIT-2 GENETIC DISORDERS II**

- 2.1 Genomic syndromes Neurofibromatosis I
- 2.2 Neurogenetic disorders Charcot Marie Tooth syndrome, spinal muscular atrophy,
  - Alzhemiers diseases, syndromes due to triplet nucleotide expansion
- 2.3 Muscle genetic disorders dystrophies, myotonias, myopathies

# **UNIT-3 GENETIC DISORDERS III**

- 3.1 Genetic Disorders of Haemopoetic systems- sickle cell anaemia, thalassemias, hemophilia
  - 3.2 Genetic disorders of eye coloblindness, retinitis pigmentosa, glaucoma
- 3.3 Complex polygenic syndromes artherosclerosis, Diabetes mellitus
- 3.4 Mitochondrial syndromes

# **UNIT-4 GENETIC COUNSELLING**

- 4.1 Role of genetic counseling
- 4.2 Causes and factors for seeking counselling
- 4.3 Dysmorphology
- 4.4 Prenatal, preimplantation and postnatal diagnosis

# **UNIT-5 PRACTICAL GENETIC COUNSELING**

5.1 Process of genetic counselling - Constructing a family tree, diagnostic information, risks and odds, estimation of risks

- 5.2 Genetic counselling in Mendelian disorders
- 5.3 Genetic counselling in Non-Mendelian disorders
- 5.4 Ethical and legal issues in genetic counselling



B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:1
Course: 6A	Clinical Genetics & Genetic Counseling Lab	Hrs/Wk:2

- 1. Metaphase chromosome preparations from bone marrow of mouse, rat, human
- 2. Chromosome preparation from lymphocyte culture
- 3. G-banding, C-banding, R-banding
- 4. Karyotyping
- 5. Meiosis in mouse testis
- 6. Sex chromatin (buccal mucosa, hair bud)
- 7. Micronuclei assay
- 8. Chromosome preparation from chorionic villi, stem cells, cell line
- 9. Sister Chromatid Exchange (SCE)
- 10. Molecular markers for tumor detection
- 11. Genetic counseling (pedigree analysis in disease conditions, risk calculation)
- 12. Y-chromosome microdeletion
- 13. Biochemical tests for sugar, albumin, Creatine phosphokinase-CPK, glucose 6 phosphate dehydrogenase-G6PD

# **Suggested Readings**

- 1. Chen, Harold Atlas of Genetic Diagnosis and CounselingSpringer 2012.
- 2. Thompson and Thompson & Thompson Genetics in Medicine, Robert L. Nussbaum, Roderick R. McInnes, Huntington F. Willard (eds)



B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:4
Course: 7A	Laboratory Diagnosis in Genetics	Hrs/Wk:4

#### **UNIT-1 BASICS OF CELL CULTURE & INSTRUMENTATION**

1.1 Techniques of cell cultures (short term lymphocyte, primary and secondary cell cultures, maintenance of cell lines)

- 1.2 Spectrophotometer, centrifugation
- 1.3 ELISA, radioactivity detection
- 1.4 Mass spectrometry
- 1.5 High performance liquid chromatography

#### **UNIT-2 TECHNIQUES IN CHROMOSOME ANALYSIS**

- 2.1 Techniques of chromosome analysis (a) Chromosome preparation from cultured lymphocytes, cell lines and solid tumors (b) Karyotyping, C-,G-banding and fluorescence banding, nomenclatures of bandings (c) *In-situ* hybridization techniques (d) Meiotic chromosomes in mouse testis
- 2.2 Chromosomal anomalies and disorders Numerical (polyploidy, aneuploidy, autosomal, sex- chromosomal) Structural (deletion, duplication, translocation, inversion, isochromosome, ring chromosome) Chromosomal abnormalities in cancer

2.3 Microscopy -Metaphase chromosome preparations from bone marrow of mouse, rat, human, Sex chromatin (buccal mucosa, hair bud), Comet assay, Micronuclei assay, Chromosome preparation from chorionic villi, Sister Chromatid Exchange (SCE)

# UNIT-3 GENETIC DISORDERS & MOLECULAR TECHNIQUES FOR DISEASE IDENTIFICATION

- 3.1 Genetic Disorders Classification of genetic disorders, Single gene Disorders (Cystic Fibrosis, Marfan's syndrome), Multifactorial disorders (Diabetes, Atherosclerosis, Schizophrenia)
- 3.2 Molecular Techniques PCR-RFLP, Multiplex-PCR, SSCP, MALDI-TOF

3.3 Disease identification and Genetic tests for following disorders: (a) Thalassemia, Fanconi, Sickle Cell anaemia, Fragile-X syndrome, Alzheimer's disease (b) Duchenne Muscular Dystrophy/Becker's Muscular Dystrophy, Huntington's disease (c) Allelic susceptibility test for multifactorial disorders (Neural Tube Defect, Cleft Lip and Palate, Cardio Vascular Disorder, Male infertility)



# **UNIT-4 BIOCHEMICAL GENETIC DIAGNOSIS**

4.1 Biochemical tests: sugar, albumin, urea, protein, globulin, vitamin ;

4.2 Biochemistry and diagnostic tests of following diseases -Duchenne Muscular Dystrophy (DMD) (Creatine phosphokinase-CPK),Phenylketonuria-PKU (phenylketone) ,G6PD deficiency syndrome (G6PD) , Endocrine disorders related to thyroid and reproduction (TSH, T3, T4, Estradiol, Testosterone, LH, FSH)

#### **UNIT-5 GENETIC COUNSELLING AND PRENATAL DIAGNOSIS**

- 5.1 Causes and factors for seeking counselling
- 5.2 Dysmorphology
- 5.3 Ethical and legal issues in genetic counselling
- 5.4 Prenatal and preimplantation diagnosis





B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:1
Course: 7A	Laboratory Diagnosis in Genetics Lab	Hrs/Wk:2

- 1. Metaphase chromosome preparations from bone marrow of mouse, rat, human
- 2. Chromosome preparation from lymphocyte culture
- 3. G-banding, C-banding, R-banding
- 4. Karyotyping
- 5. Fluorescence in-situ Hybridization (FISH)
- 6. Meiosis in mouse testis
- 7. Sex chromatin (buccal mucosa, hair bud)
- 8. Comet assay
- 9. Micronuclei assay
- 10. Chromosome preparation from chorionic villi, stem cells, cell line
- 11. Sister Chromatid Exchange (SCE)
- 12. Molecular markers for tumor detection
- 13. Bcr-abl (RT-PCR)
- 14. Genetic counseling (pedigree analysis in disease conditions, risk calculation)
- 15. Prenatal diagnosis of Thalassemia
- 16. Y-chromosome microdeletion

17. Biochemical tests for sugar, albumin, Creatine phosphokinase-CPK, glucose 6 phosphate dehydrogenase-G6PD

#### **Suggested Readings**

1. Primrose, SB and Twyan RM. *Principles of gene manipulation and genomics*. 7th edition.Blackwell Science, 2006.

2. Watson, Myers and Caudy. *Recombinant DNA: Genes and Genomes-A short course*. 3rd edition. 2006. Freeman W.H. and Company.

3. Fundamentals of Molecular Diagnositics by David E. Bruns, Edward R. Ashwood, Carl A Burti4. Human Genetics: From Molecules to Medicine by Christian Patrick Schaaf, Johannes Zschocke, Lorraine Potocki



B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:4
Course: 6B	Human Genome Project and Genomes	Hrs/Wk:4

#### UNIT 1 Genome Organization and Study

1.1 Genome – general features, features of eukaryotic nuclear genomes

1.2 Genomes, transcriptomes and proteomes

1.3 Genome diversity – significance of genomes – bacteria, yeast, *Caenorhabditis*, *Homo sapiens*, *Arabidopsis*.

#### **UNIT 2 Mapping Genomes**

2.1 Genetic mapping – pedigree analysis, DNA markers – RFLPs, SSLPs, SNPs

2.2 Physical mapping – restriction mapping, FISH, Radiation hybrid mapping, STS mapping

2.3 Sequencing genome- assembly of contiguous DNA sequence, shotgun method, clonecontig method, whole-genome shotgun sequencing

#### **UNIT 3 Genome Projects**

3.1 Human genome project, HapMap Project, 1000 genome project, ENCODE project

- 3.2 Other genome projects.
- 3.3 Applications and proposed benefits of HGP –ELSI.

#### **UNIT 4 Understanding Genome sequence**

- 4.1 Locating the genes in a genome sequence
- 4.2 Determining the functions of individual genes
- 4.3 Transcriptome microarrays
- 4.4 Proteome protein profiling

#### **UNIT 5 Molecular phylogenetics**

- 5.1 Phenetics and cladistics
- 5.2 Reconstruction of DNA based phylogenetic tree

5.3 Applications of molecular phylogenetics – evolutionary relationship between humans and primates; origin of AIDS; human prehistory.



B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:1
Course: 6B	Human Genome Project and Genomes Lab	Hrs/Wk:2

- 1. Isolation and purification of genomic DNA.
- 2. Detection of SNPs using SNP specific primers and PCR.
- 3. Study of VNTR's in human genome as the polymorphic loci.
- 4. Design primers for PCR based detection of the gene and mapping primers on the genome
- 5. Introduction to NCBI websites
- 6. Introduction to database:protein data bank, nucleic acid database, Genbank .
- 7. Web based analysis to retrieve a nucleotide sequence from NCBI,
- 8. Sequence alignment using BLASTn, BLASTp, CLUSTALW.
- 9. Gene finding tools GenScan, GLIMMER
- 10. Introduction to proteomics Protparam, GOR, unPredict, SWISSMODEL .
- 11. Visualization software Rasmol
- 12. Generating phylogenetic tree using PHYLIP

#### **Suggested Readings**

- 1. Human Genome Project by James Toriello .
- 2. Understanding the Human Genome Project by Michael A Palladino.

3. Human Genes and Genomes: Science, Health, Society by Leon E Rosenberge, Diane Drobnis Rosenberg.

4. From Genes to Genomes: Concepts and Applications of DNA Jeremy W Dale, Malcolm von Schantz, Nick Plant .

5.Genomes 3 by Terence A Brown.

6. Principles of Gene Manipulation and Genomics by Sandy B Primrose and Richard Twyman.



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B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:4
Course: 7B	Molecular Techniques in Genetic Engineering	Hrs/Wk:4

# UNIT-1 NUCLEIC ACID ISOLATION AND AGAROSE GEL ELECTROPHORESIS. (9hours)

Conventional and kit method for isolation of nucleic acids-Plasmid DNA-Genomic DNA from Bacterial cells, Plant cells, animal cells-RNA isolation and m-RNA purification –Agarose purification-Agarose gel electrophoresis-Staining techniques –Pulse feild gel electrophoresis

# **UNIT-2 PCR TECHNIQUES**

Principle of Polymerase Chain Reaction (PCR)-Components of PCR reaction and optimization of PCR –Gene specific primer- Inverse PCR, Hot-start PCR, Loop mediated PCR – Reverse transcription PCR and Real time PCR. Chemistry of primer synthesis

# **UNIT-3 HYBRIDIZATION METHODS**

Probes –Labelling of probes-Radio active and non-radio active probes-Detection techniques, Southern hybridization, Northern hybridization, Western blotting

# UNIT-4 DNA SEQUENCING AND GENE SYNTHESIS (9 hours)

Sangers's method of DNA sequencing – Manual and automated methods. Pyrosequencing-massive parallel 454-sequencing, Illumina sequencing, SOLID sequencing, single molecule sequencing

# **UNIT-5 PROTEIN TECHNIQUES**

Electrophoresis of protein –native and denaturing conditions, capillary and gel electrophoresis, 3D gel electrophoresis, ELISA ,Yeast hybrid system-one hybrid system, phage display



(0.1

(9 hours)

(9hours)

(9 hours)

B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:1
Course: 7B	Molecular Techniques in Genetic Engineering Lab	Hrs/Wk:2

- 1. Primer designing
- 2. Insertion deletion polymorphism
- 3. DNA Finger printing RFLPs and VNTRs
- 4. Amplification and purification of DNA fragments
- 5. ARMS-PCR
- 6. Multiplex PCR
- 7. Nested PCR
- 8. DNA sequencing methods
- 9. SDS-Gel electrophoresis
- 10. Southern blotting
- 11. Northern blotting
- 12. Western blotting

# **Suggested Reading**

1.Fredrick M.Ausubel.RogerBrent,Robert E Kingstone,David D. Moore,Seidman J. G,JohnA.Smith and Kevin Struhl, "Current Protocols in Molecular Biology",John Wiley & Son,Inc.2003.

2. DanielC. Liebler "Introduction to Proteomics", Human Press, 2002.



B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:4
Course: 6C	Developmental and Behavioral Genetics	Hrs/Wk:4

#### **UNIT-1: Germ Cells and Fertilization**

Germ Cells

Spermatogensis

Oogenesis

Fertilization and Gastrulation

#### **UNIT-2: Molecular Aspects of Development**

Maternal effector gene

Gap genes

Pair rule gene

Segment polarity genes

Homeotic genes

#### UNIT-3: Genetics of Embryonic Development in Drosphila

Overview of Drosphila development

Zygotic genes and segment formation

#### **UNIT- 4: Molecular aspects of flower development**

Flower Development in Arabidopsis

Development, Role of Homeotic Selector Gene

#### **UNIT-5: GENETIC CONTROL OF BEHAVIOUR**

Introduction, Behaviour in Invertebrates, Honeybee, *Drosophila* – Genetic basis of alcoholism, genetic basis for sexual orientation. Courtship behaviour in various animals.



B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:1
Course: 6C	Developmental and Behavioral Genetics Lab	Hrs/Wk:2

- 1. Study of development in chick embryo
- 2. Dissection of imaginal disc in Drosophila larvae
- 3. Life cycle of Drosophila, husbandary and handling.
- 4. Role of shh signaling in chick development
- 5. Observation of live and plastic embedded chick embryos
- 6. The maternal effect gene in Drosophila

#### **Suggested Reading**

The cell – Bruce Alberts

Emery's Elements of Medical Genetics- Robert. F. Mueller, Ian. D. Young.

Principles of Development - Wolpert; Developmental Bidgy by Scoff F. Gilbert

Principles of Genetics – Snustad, Simmons, Jenkins.



B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:4
Course: 7C	Molecular Pathology in Human Diseases	Hrs/Wk:4

#### UNIT 1 Human diseases I

1.1 Etiology, pathology and symptoms of genetically inherited diseases – PKU, Alkaptonuria, Galactosemia, Von Gierke disease, LeschNyhan syndrome, Gout, sickle cell anaemia, beta thalassemia, diabetes

1.2 Mode of infection, symptoms and epidemiology of disease caused by viruses (HIV, Hepatitis B, Rabies, HSV-1)

1.3 Mode of infection, symptoms and epidemiology of disease caused by bacteria – typhoid, syphilis, TB

# UNIT 2 Human diseases II

2.1 Mode of infection, symptoms and epidemiology of disease caused by fungi – aspergillosis, histoplasmosis.

- 2.2 Mode of infection, symptoms and epidemiology caused by protozoa –malaria, amoebiasis.
- 2.3 Cancer genetics tumor suppressor genes, oncogenes, Molecular basis of oncogenesis

#### **UNIT 3 Basic Instrumentation principles and techniques**

- 3.1 Principles of electrophoresis and immunoblotting
- 3.2 Principles of DNA sequencing and methods of genotyping and mutation analysis
- 3.3 Principles and applications of PCR
- 3.4 In situ hybridization techniques ISH, FISH

#### **UNIT 4 Genetic testing for hereditary disorders**

- 4.1 Genetic testing for thalassemia
- 4.2 Genetic testing for familial colorectal cancer
- 4.3 Genetic testing for familial breast and ovarian cancer

4.4 EGFR mutation in lung cancer, HER2 amplification in breast cancer, FISH test for early bladder cancer detection, KRAS mutation detection for colorectal cancer

#### **UNIT 5 Molecular diagnosis of infectious diseases**

5.1 Principles of HPV testing and methods of genotyping

5.2 Hepatitis B virus infection – testing for viral load and HBV DNA mutants detection

5.3 Molecular techniques -NestedPCR, Real Time PCR for different clinical applications

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B.Sc Human Genetics Syllabus (w.e.f:2020-21A.B)

B.Sc	Semester – V (Skill Enhancement Course- Elective)	Credits:1
Course: 7C	Molecular Pathology in Human Diseases Lab	Hrs/Wk:2

- 1. Preventing contamination.
- 2. Extract and assess the purity of DNA.
- 3. Agarose gel electrophoresis
- 4. Set up PCR.
- 5. Evaluate Southern blot data
- 6. Analyze PCR product using agarose gel electrophoresis and interpret results
- 7. Demonstration of karyotyping
- 8. Isolate cellular RNA, purify mRNA
- 9. Set up RT-PCR using commercial kit
- 10. Analyze RT-PCR results by agarose gel.

#### **Suggested Reading:**

- Basic Concepts of Molecular Pathology Series: Molecular Pathology Library, Vol. 2Cagle, Philip T. Allen, Timothy C. (Eds.)Springer 2009
- **2.** Molecular Pathology: The Molecular Basis of Human Disease; William B. Coleman, Gregory J. Tsongalis (Eds.);Academic Press;
- **3.** Genomics and Personalized Medicine Huntington F. Willard, Geoffry S. Ginsburg; Elsevier 2009
- **4.** Medical Genetics, 4th Edition;Lynn B. Jorde, John C. Carey, and Michael J. Bamshad,Mosby
- DNA from A to Z & Back Again; Carol A. Holland and Daniel H. Farkas; AACC Press 2008
- 6. Molecular Genetic Pathology, 1st ed.; Liang Cheng and David Zhang; Humana Press 2008

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B.Sc Human Genetics Syllabus (w.e.f:2020-21A.B)

# Model Question Paper Semester and Examination

# **6A: CLINICAL GENETICS AND GENETICS COUNSELLING**

# Time : 3 Hrs

I.

MaxMarks:75M 5×5=25M

- 1. Galactosaemia
- 2. Marfan Syndrome

Answer any 5

- 3. Alzhemiers disease
- 4. Duchenne muscular dystrophy
- 5. Sickle cell anemia
- 6. Pre-implantation diagnosis
- 7. Mendelian disorders
- 8. Construction of family tree
- II. Answer all of the following
- 9a. Write an essay on Inborn errors of metabolism (**Or**)
- 9b. What is Genome Imprinting
- 10a. Explain in detail about Muscle genetic disorders (Or)
- 10b. Write an essay on Genomic syndromes
- 11a. What are the Haemotopoitic genetic disorders (Or)
- 11b. Write about Mitochondrial syndromes
- 12a. What is Genetic counseling (**Or**)
- 12b. Explain Invasive techniques of pre-notal diagonasis
- 13a. Write in detail about the process of genetic counseling(**Or**)
- 13b.What are the Ethical and legal issues in the genetic counselling



5×10=50M

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# MODEL QUESTION PAPER Semester End Examination- Practical 6A: CLINICAL GENETICS AND GENETICS COUNSELLING

Time : 3 Hrs	MaxMarks:50M	
1. Major experiment	<b>20M</b>	
1. Observe whether the given subject is sex chromatin positive or not by Blood		
smear preparation.		
2. Minor experiment	15M	
Prepare Idiogram for the given karyotype		
<b>3.</b> Analyse the given Pedigree and calculate risk for the next	generation5M	
4. Record	5M	
5. Viva voce	5M	

# Model Question Paper Semester and Examination

# 7A: LABORATORY DIAGNOSIS IN GENETICS

# Time : 3 Hrs

Part-A

5×5=25M

MaxMarks:75M

5×10=50M

# Answer any Five of the following

- 1. HPLC
- 2. Aneuploidy
- 3. C-Banding
- 4. MALDI-TOF
- 5. Fragile X Syndrome
- 6. Pre implantation diagnosis
- 7. Neural tube defects
- 8. FISH

# Part-B

# Answer all of the following

9. a) Write about different techniques used in cell culture

Or

- b) Discuss the principles involved in spectrophotometer and centrifugation
- 10. a) Write about numerical chromosomal abnormalities

Or

- b) Explain insitu hybridisation techniques
- 11. a) Mention any two molecular techniques which are used in the detection of genetic Disorders
  - b) What is allelic susceptibility test for multifactorial disorders
- 12. a) Write about biochemistry and diagnostic tests of Duschene muscular dystrophy and G-6 P.D. deficiency.

Or

b) What are the thyroid related diseases and techniques used in their diagnosis

13. a) write about pre natal diagnosis of genetic defects

# Or

b) What are the ethical and legal issues in genetic counselling





# MODEL QUESTION PAPER Semester End Examination- Practical 7A: LABORATORY DIAGNOSTICS IN GENETICS

Time : 3 Hrs	MaxMarks:50M	
1. Major experiment	<b>20M</b>	
1. Observe whether the given subject is sex chron smear preparation.	natin positive or not by Buccal	
2. Minor experiment	15M	
Write about Karyotyping		
<b>3.</b> Analyse the given Pedigree and calculate risk for t	e e	
4. Record	5M	
5. Viva voce	5M	

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B.Sc Human Genetics Syllabus (w.e.f:2020-21A.B)

# Model Question Paper Semester and Examination

#### **6B: HUMAN GENOME PROJECT AND GENOMES**

# Time : 3 Hrs

# MaxMarks:75M

# I. Answer any FIVE of the following

5×5=25M

- 1. Transcriptomes.
- 2. RFLP
- 3. Hap Map Project
- 4. Phenetics
- 5. Phylogenetic tree
- 6. Shot gun method.
- 7. Pedigree analysis
- 8. HGP.

# II. Answer ALL the following question 5×10=50M

9. (a) Write an essay on eukaryotic nuclear genome.

(or)

- (b) Write about transcriptomes & proteomes.
- 10.(a) Write an essay on physical mapping.

(or)

- (b) Write about different genome sequencing methods.
- 11.(a) Write about human genome project.

(or)

(b) What are the applications and proposed benefits of HGP.

12.(a) Write an essay on protein profiling.

(or)

- (b) Write about process of locating genes in a genome sequence.
- 13.(a) Write the process of reconstructing phylogenetic tree.

# (or)

(b) What are the applications of molecular phylogenetics

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# MODEL QUESTION PAPER Semester End Examination- Practical 6B HUMAN GENOME PROJECT AND GENOMES

Time : 3 Hrs	MaxMarks:50M
1. Major experiment	<b>20M</b>
Isolation of DNA from WBC.	
2. Minor experiment	<b>15M</b>
Sequence alignment using BLAST	
<b>3.</b> PCR Primer designing	5M
4. Record	5M
5. Viva voce	<b>5</b> M

# Model Question Paper Semester and Examination

# 7B: MOLECULAR TECHNIQUES IN GENETIC ENGINEERING

# Time : 3 Hrs

# 5×5=25M

MaxMarks:75M

- I. Answer any 5
- 1. Isolation of RNA
- 2. Hot-Star PCR
- 3. Real time PCR
- 4. Pulse- field gel electrophorets
- 5. Radi<mark>o</mark> active probes
- 6. Capillary electrophoreses
- 7. Illumina sequency
- 8. Western blotting
- II Answer ALL the following question

9a. Write about Isolation of genomic DNA

# (Or)

- b. Write an essay on Agarose gel electrophoresis
- 10a. Explain the Mechanism involved in Polymerate chain reacation

# (Or)

b. Write about RTPCR

11a. Explain in detail about labeling of probes

# (Or)

b. Write about Blotting techniqus

12a. Write Sayer's Method of DNA sequencing

# (Or)

b. What is pyroseqnencing

13a. Explain in detail about ELISA

# (Or)

b. Write about Electrohoresis of Proteins







# MODEL QUESTION PAPER Semester End Examination- Practical 7B: GENETIC ENGINEERING LAB

Time : 3 Hrs	MaxMarks:50M	
Major experiment	<b>20M</b>	
1. Amplify the given DNA sample by using Invitro gene closing technique		
2. Minor experiment Explain in detail about primer designing	15M	
<b>3.</b> Write the principle involved in ARMS PCR	5M	
4. Record	<b>5</b> M	
5. Viva voce	<b>5</b> M	



# Model Question Paper Semester and Examination

# **6C: DEVELOPMENT & BEHAVIOURAL GENETICS**

# Time : 3 Hrs

I. Answer any 5

- 1. Germ cells
- 2. Spermatogensis
- 3. Pair rule gene
- 4. Gap gene
- 5. Drosophila development
- 6. Courtship behaviour
- 7. Homeotic selector gene
- 8. Honeybee behavioural genetics

# II Answer ALL the following question

9 a. Explain the process of oogenesis

# (Or)

- b .Write about fertilization ad gastrulation
- 10a. What are Maternal effect genes

# (Or)

Write about segment Polarity genes

11a. Write about genetic background of embryo development in Drosophila

# (Or)

- b. Write about Zygotic genes and segment formation
- 12a. Explain the process of flower development in Arabidopsis

# (Or)

b. Explain the role of Hoemotic selector gene

13a. Write about Genetic control of Behaviour

# (Or)

b. Explain in detail about genetic basis of alcoholism

MaxMarks:75M 5×5=25M



# MODEL QUESTION PAPER Semester End Examination- Practical DEVELOPMENT AND BEHAVIOURAL GENETICS LAB

Time : 3 Hrs Ma	xMarks:50M
Major experiment	<b>20M</b>
<b>1.</b> Dissect drosophila larra and report about imaginal disc	
2. Minor experiment	<b>15M</b>
Report life cycle of drosophila	
<b>3.</b> Write about role of SHH signaling in chick developmen	t <b>5M</b>
4. Record	5M
5. Viva voce	<b>5</b> M

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B.Sc Human Genetics Syllabus (w.e.f:2020-21A.B)

# Model Question Paper Semester and Examination

# 7C: MOLECULAR PATHOLOGY IN HUMAN DISEASES

# Time : 3 Hrs

# MaxMarks:75M

 $5 \times 5 = 25M$ 

# I. Answer any FIVE of the following

- 1. Phenylketoneuria.
- 2. Epidemiology of rabies
- 3. Mode of infection of aspergillosis
- 4. Tumor suppressor genes
- 5. Immunoblotting
- 6. FISH
- 7. Thalessaemia
- 8. Nested PCR.

# II. Answer ALL of the following

9. (a) Write a note on infection, symptoms and epidemiology of HIV.

(or)

(b) Write an essay on mode of infection, symptoms and epidemiology of typhoid.

10.(a) Write in detail about molecular basis of oncogenesis.

(or)

- (b) Write a note on aspergillosis and histoplasmosis.
- 11.(a) Write an essay on electrophoresis.

(or)

- (b) Write a note on principles & applications of PCR.
- 12.(a) Write an essay on genetic testing of colorectal cancer.

(or)

- (b) Write about familial breast cancer & ovarian cancer.
- 13.(a) Write about principles of HPV testing.

(or)

(b) Write in detail about real time PCR for different clinical applications.

# 5×10=50M



# MODEL QUESTION PAPER Semester End Examination- Practical 7C: MOLECULAR PATHOLOGY IN HUMAN DISEASES

Time : 3 Hrs	MaxMarks:50M	
1. Major experiment	<b>20M</b>	
1. Test the quality of given DNA by conducting Agroose gel electrophores		
2. Minor experiment	15M	
Write about the components required for PCR and describe its setup		
<b>3.</b> Write about the principle involved in RT-PCR	5M	
4. Record	5M	
5. Viva voce	5M	