

KARNATAK UNIVERSITY, DHARWAD ACADEMIC (S&T) SECTION ಕರ್ನಾಟಕ ವಿಶ್ವವಿದ್ಯಾಲಯ, ಧಾರವಾಡ ವಿದ್ಯಾಮಂಡಳ (ಎಸ್&ಟಿ) ವಿಭಾಗ

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NAAC Accredited 'A' Grade 2014

No. KU/Aca(S&T)/SSL-394A/2022-23 1056

Date: 2 3 SEP 2022

#### ಅಧಿಸೂಚನೆ

ವಿಷಯ: 2022–23ನೇ ಶೈಕ್ಷಣಿಕ ಸಾಲಿನಿಂದ ಎಲ್ಲ ಸ್ನಾತಕ ಕೋರ್ಸಗಳಿಗೆ 3 ಮತ್ತು 4ನೇ ಸೆಮೆಸ್ಟರ್ NEP-2020 ಮಾದರಿಯ ಪಠ್ಯಕ್ರಮವನ್ನು ಅಳವಡಿಸಿರುವ ಕುರಿತು.

ಉಲ್ಲೇಖ: 1. ಸರ್ಕಾರದ ಅಧೀನ ಕಾರ್ಯದರ್ಶಿಗಳು(ವಿಶ್ವವಿದ್ಯಾಲಯ 1) ಉನ್ನತ ಶಿಕ್ಷಣ ಇಲಾಖೆ ಇವರ ಆದೇಶ ಸಂಖ್ಯೆ: ಇಡಿ 260 ಯುಎನ್ಇ 2019(ಭಾಗ–1), ದಿ:7.8.2021.

2. ವಿಜ್ಞಾನ & ತಂತ್ರಜ್ಞಾನ ನಿಖಾಯ ಸಭೆಯ ಠರಾವುಗಳ ದಿನಾಂಕ: 06.09.2022

3. ವಿಶೇಷ ವಿದ್ಯಾವಿಷಯಕ ಪರಿಷತ್ ಸಭೆಯ ನಿರ್ಣಯ ಸಂ. 01, ದಿನಾಂಕ: 17.09.2022

4. ಮಾನ್ಯ ಕುಲಪತಿಗಳ ಆದೇಶ ದಿನಾಂಕ: 22-09-2022

ಮೇಲ್ಕಾಣಿಸಿದ ವಿಷಯ ಹಾಗೂ ಉಲ್ಲೇಖಗಳನ್ವಯ ಮಾನ್ಯ ಕುಲಪತಿಗಳ ಆದೇಶದ ಮೇರೆಗೆ, 2022-23ನೇ ಶೈಕ್ಷಣಿಕ ಸಾಲಿನಿಂದ ಅನ್ವಯವಾಗುವಂತೆ, ವಿಜ್ಞಾನ & ತಂತ್ರಜ್ಞಾನ ನಿಖಾಯದ ಎಲ್ಲ ಸ್ನಾತಕ ಕೋರ್ಸಗಳ ರಾಷ್ಟ್ರೀಯ ಶಿಕ್ಷಣ ನೀತಿ (NEP)-2020 ರಂತೆ 3 ಮತ್ತು 4ನೇ ಸೆಮೆಸ್ಟರ್ಗಳಿಗಾಗಿ ವಿಶೇಷ ವಿದ್ಯಾವಿಷಯಕ ಪರಿಷತ್ ಸಭೆಯ ಅನುಮೋದಿತ ಪಠ್ಯಕ್ರಮಗಳನ್ನು ಪ್ರಕಟಪಡಿಸಿದ್ದು, ಸದರ ಪಠ್ಯಕ್ರಮಗಳನ್ನು ಕ.ವಿ.ವಿ. <u>www.kud.ac.in</u> ಅಂತರ್ಜಾಲದಿಂದ ಡೌನಲೋಡ ಮಾಡಿಕೊಳ್ಳಲು ಸೂಚಿಸುತ್ತಾ, ವಿದ್ಯಾರ್ಥಿಗಳು ಹಾಗೂ ಸಂಬಂಧಿಸಿದ ಎಲ್ಲ ಬೋಧಕರ ಗಮನಕ್ಕೆ ತಂದು ಅದರಂತೆ ಕಾರ್ಯಪ್ರವೃತ್ತರಾಗಲು ಕವಿವಿ ಅಧೀನದ / ಸಂಲಗ್ನ ಮಹಾವಿದ್ಯಾಲಯಗಳ ಪ್ರಾಚಾರ್ಯರುಗಳಿಗೆ ಸೂಚಿಸಲಾಗಿದೆ.

We Jahn

ಅಡಕ: ಮೇಲಿನಂತೆ

ಗೆ,

ಕರ್ನಾಟಕ ವಿಶ್ವವಿದ್ಯಾಲಯದ ವ್ಯಾಪ್ತಿಯಲ್ಲಿ ಬರುವ ಎಲ್ಲ ಅಧೀನ ಹಾಗೂ ಸಂಲಗ್ನ ಮಹಾವಿದ್ಯಾಲಯಗಳ ಪ್ರಾಚಾರ್ಯರುಗಳಿಗೆ. (ಕ.ವಿ.ವಿ. ಅಂರ್ತಜಾಲ ಹಾಗೂ ಮಿಂಚಂಚೆ ಮೂಲಕ ಬಿತ್ತರಿಸಲಾಗುವುದು)

ಪ್ರತಿ:

- 1. ಕುಲಪತಿಗಳ ಆಪ್ತ ಕಾರ್ಯದರ್ಶಿಗಳು, ಕ.ವಿ.ವಿ. ಧಾರವಾಡ.
- 2. ಕುಲಸಚಿವರ ಆಪ್ತ ಕಾರ್ಯದರ್ಶಿಗಳು, ಕ.ವಿ.ವಿ. ಧಾರವಾಡ.
- 3. ಕುಲಸಚಿವರು (ಮೌಲ್ಯಮಾಪನ) ಆಪ್ತ ಕಾರ್ಯದರ್ಶಿಗಳು, ಕ.ವಿ.ವಿ. ಧಾರವಾಡ.
- 4. ಅಧೀಕ್ಷಕರು, ಪ್ರಶ್ನೆ ಪತ್ರಿಕೆ / ಗೌಪ್ಯ / ಜಿ.ಎ.ಡಿ. / ವಿದ್ಯಾಂಡಳ (ಪಿ.ಜಿ.ಪಿಎಚ್.ಡಿ) ವಿಭಾಗ, ಸಂಬಂಧಿಸಿದ ಕೋರ್ಸುಗಳ ವಿಭಾಗಗಳು ಪರೀಕ್ಷಾ ವಿಭಾಗ, ಕ.ವಿ.ವಿ. ಧಾರವಾಡ.
- 5. ನಿರ್ದೇಶಕರು, ಕಾಲೇಜು ಅಭಿವೃದ್ಧಿ / ವಿದ್ಯಾರ್ಥಿ ಕಲ್ಯಾಣ ವಿಭಾಗ, ಕ.ವಿ.ವಿ. ಧಾರವಾಡ.



# KARNATAK UNIVERSITY, DHARWAD

# B.Sc. (Hons) Programme

DRAFT SYLLABUS FOR

# **GENETICS**

DISCIPLINE SPECIFIC COURSE (DSCC), OPEN ELECTIVE COURSE

(OEC) FOR SEM III & IV

UNDER

NATIONAL EDUCATION POLICY (NEP)

Effective from 2022-23

Karnatak University, Dharwad

# B.Sc. Semester-III

## Subject: Genetics Discipline Specific Course (DSCC) Course No.GEN103T Title of the Course: Biomolecules and Molecular Genetics(Theory)

The course Biomolecules and Molecular Genetics in III semester has two papers (Theory Paper-Ifor 04 credits & Practical Paper-II for 2 credits) for 06 credits: Both the papers are compulsory. Details of the courses are as under.

CourseN o./ Course code	Type of Course	Theory/Prac tical	Credits	Instruction hourperweek	Total No. of Lectures/Hours /Semester	Duration of Exam	FormativeA ssessment Marks	Summative Assessment Marks	Total Marks
GEN103T/ 033GEN011	DSCC	Theory	04	04	56hrs.	2 hrs	40	60	100

#### CourseOutcome(CO):

## After completion of course(Theory), students will be able to:

**CO1**: Describe the structure and function of biomolecules

- **CO 2:**Appreciate and illustrate the chemical composition of the genetic material and its multiplication
- **CO 3:**Describe the process of gene expression in prokaryotes and eukaryotes

CO 4: Explain the concept of transposition, mutation and DNA repair mechanism

GEN103T: Biomolecules and Molecular Genetics (Theory)	Total Hrs:56
Unit-I	14hrs
<b>Carbohydrates:</b> Structure and functions of carbohydrates (Monosaccharides, Disaccharides and polysaccharides). Metabolism of Carbohydrates: glycolysis. Citric acid cycle and gluconeogenesis. <b>Lipids:</b> Definition, Saturated and unsaturated fatty acids, Tri-acyl glycerol, phospholipids, glycolipids and steroids. <b>Amino acids and proteins:</b> Structure, classification and general properties of $\alpha$ -amino acids, organizations of protein-simple and conjugate protein- definition and examples. Peptide Linkages. Primary, secondary, tertiary and Quaternary structure of proteins and protein folding.	
Unit-II	14hrs
<ul> <li>Introduction: DNA (Hershey and Chase experiment) and RNA (Fraenkel and Singer experiment) as genetic material, Watson and Crick model.</li> <li>Structure and functions of DNA: Molecular structure of DNA, Chargaff's rule, forms of DNA - A, B and Z; Functions of DNA and RNA including ribozymes; denaturation and renaturation of DNA,</li> <li>DNA replication: Prokaryotes and Eukaryotes. Initiation, continuous and discontinuous synthesis. Enzymes and proteins involved in replication, Theta model and rolling circle model of replication.</li> </ul>	

Unit-III	14hrs
Transcription: Initiation elongation, termination in prokaryotes and eukaryotes, Post	
transcriptional modifications: Methylation, polyadenylation and RNA splicing. gene-	
silencing by RNA interference.	
Translation: Features of the genetic code; Types of RNA, structure of tRNAs, protein	
biosynthesis, aminoacyl-tRNA synthetases; Post, translational modification of Proteins.	
Principles of gene regulation: Negative and positive regulation; concept of operons,	
regulatory proteins, activators, repressors, DNA binding domains. <b>Regulation of gene expression:</b> Overview, regulation of gene expression in bacteria (Lac	
operon and Trp operon); regulation of gene expression in eukaryotes (regulation of	
galactose metabolism in yeast).	
Unit-IV	14hrs
Transposons: Bacterial Transposons - IS elements; P elements in Drosophila, AC-DS in	
Maize;	
Mutation: Mutagens- physical, chemical, Mutation- Definition of point mutation Types of	
mutations, Transition and transversion, base substitution- missense, non-sense, neutral	
and silent mutation. Frame shift mutation, Detection of mutation - Ames test;	
<b>DNA repair mechanism:</b> Mismatch repair, photo- reactivation, excision and SOS repair;	
Beneficial and harmful effects of mutation.	

## Books recommended :

- 1. Karp, G. (2009). Cell and molecular biology: concepts and experiments. John Wiley & Sons.
- 2. Lewin, B., Krebs, J., Kilpatrick, S. T., & Goldstein, E. S. (2011). Lewin's genes X. Jones & Bartlett Learning.
- Watson, J. D., Baker, T. A., Gann, A., Bell, S. P., Levine, M., &Losick, R. M. (2017). Molecular biology of the gene. Pearson Education; Seventh edition.
- 4. Lodish, H., Berk, A., Kaiser, C. A., Kaiser, C., Krieger, M., Scott, M. P., ... & Matsudaira, P. (2008). Molecular cell biology. Macmillan publisher.
- 5. Gupta, P. K. (2008). Molecular biology and genetic engineering. Deep and Deep Publications.
- 6. Rastogi, V. B. (2008). Fundamentals Of Molecular Biology. Ane Books Pvt Ltd.
- 7. Kumar A. & Shers S. S. (2020). Basic Concepts of Molecular Biology. Edubuds Publishing House, India.
- 8. Verma, P. S., & Agarwal, V. K. (2004). Cell Biology, Genetics, Molecular Biology, Evolution and Ecology. S. Chand Publishing.

# B.Sc.Semester-III

## Subject: Genetics Discipline Specific Course (DSCC)

#### Course No.-GEN1033P

#### Title of the Course: Biomolecules and Molecular Genetics(Practical)

CourseNo.	TypeofCo urse/ course code	Theory /Practical		hourperweek	Total No. of Lectures/ Hours/Semester			Summative Assessment Marks	TotalMar ks
GEN1033P / 033GEN012	DSCC	Practical	02	04	52 hrs	3 hrs	25	25	50

#### CourseOutcome(CO):

Aftercompletionofcourse(Practical), students will be able to:

**CO1**: Understand the working principle and handling of instruments.

biology

- **CO2:** Perform the isolation of DNA from various sources.
- **CO3:** Characterize the eye pigments in *Drosophila* using paper chromatography.
- **CO4:** Demonstrate the effects of mutation and appraise the applications of molecular markers.

#### ListoftheExperimentsfor52hrs/Semesters

- 1. Qualitative analysis of Carbohydrates
- 2. Qualitative analysis of Protein
- 3. Qualitative analysis of Lipid Estimation of reducing sugars
- 4. Estimation of proteins by FCR method
- 5. Estimation of total sugars by anthrone method
- 6. Estimation of reducing sugars by DNS method
- 7. Saponification value of oils and fats
- 8. Determination of molecular weight of proteins by SDS PAGE
- 9. Extraction of genomic DNA from by CTAB method
- 10. Extraction of RNA by lithium chloride method
- 11. Agarose gel electrophoresis for separation of genomic DNA
- 12. Quantification of nucleic acids using spectrophotometer
- 13. Separation of eye pigments in wild type and mutant Drosophila Using Thin Layer Chromatography.

\*Other practical may be added according to requirement and feasibility

#### Books recommended :

- 1. Satyanarayana, U. (2021). *Biochemistry, 6e-E-book*. Elsevier Health Sciences.
- 2. Walker, J. M. (2000). *Principles and techniques of practical biochemistry*. Cambridge University Press.
- 3. Mu, P., & Plummer, D. T. (2001). *Introduction to practical biochemistry*. Tata McGraw-Hill Education.
- 4. Chawla, R. (2014). *Practical clinical biochemistry: methods and interpretations*. JP Medical Ltd.
- 5. Wilson, K., Hofmann, A., Walker, J. M., &Clokie, S. (Eds.). (2018). *Wilson and Walker's principles and techniques of biochemistry and molecular biology*. Cambridge University Press.
- Jain, A., Jain, R., & Jain, S. (2020). Basic techniques in biochemistry, microbiology and molecular biology (pp. 9-10). New York, NY, USA:: Springer.
- 7. Green, M. R., & Sambrook, J. (2012). Molecular cloning. *A Laboratory Manual* 4<sup>th</sup> edition. CSH Press. USA.

#### **General instructions:**

Scheme of Practical Examination (distribution of marks): 25 marks for Semester endexamination

- 1. MajorQuestion ----- 6Marks
- 2. MinorQuestion------4Marks
- 3. Identification(A-E)-----8Marks
- 4. Viva-----2Marks
- 5. Journal-----5Marks

#### Total: 25marks

Note:SameSchememaybeusedforIA(FormativeAssessment)examination

# B.Sc.Semester-III

Subject: Genetics Open Elective Course (OEC) (OEC for other students) Course No.: GEN103E

# Title of the Course: Eugenics, Euphenics and Society (Elective

Theory)

CourseNo. / Course code	TypeofCo urse	Theory /Practica I	Credits	Instruction hourperwee k	Total No. ofLectures/Hours /Semester	Duration of Exam	Formative Assessment Marks	Summativ e Assessme nt Marks	Total Marks
GEN103E 003GEN051	OEC	Theory	03	03	42hrs	2 hrs	40	60	100

## Course Outcome(CO):

After completion of course, students will be able to:

CO1:explain the basic concepts of eugenics and assisted reproductive technology
 CO2:appraise the concept of preimplantation and prenatal diagnosis
 CO3:interpret the importance of genetic counselling
 CO4 :appraise the concept of gene therapy and its significance

GEN103E: Eugenics, Euphenics and Society	Total Hrs:42
Unit-I	14hrs
<b>Eugenics:</b> Concept, types-positive and negative Eugenics, Eugenics in United States, Nazism and decline of Eugenics.	
<b>Modern Eugenics:</b> Genetic engineering and modern reproductive technologies, invitro fertilization, female and male infertility, steps in IVF techniques, Sperm and Oocyte preservation.	
<b>Euphenics:</b> Concept, managing and treating genetic diseases–Type I diabetes, phenylketonuria and thyroid;	
<b>Euthenics:</b> Prevention of contagious diseases, environment pollution and parasitism; Ethical issues and evolutionary implications.	
Unit-II	14hrs
<b>Prenatal Diagnosis:</b> Indications for prenatal diagnosis; Methods- Noninvasive method- Ultrasonography and Fetal echocardiography, Invasive methods - Amniocentesis, Chorionic villus sampling and Fetoscopy; Pre-conceptional and pre-implantation genetic diagnosis- Teratogen exposure in early pregnancy, Genetic testing and screening.	
Unit-III	14hrs
<b>Gene Therapy:</b> Introduction, somatic and germ line gene therapy <i>Ex vivo</i> and <i>In vivo</i> gene therapy; Transgenic animal models, viral vectors, delivery methods; Gene Therapy and diseases- Cystic fibrosis, haemophilia; Cancer gene therapy, Gene therapy of non-heritable disorders; Cord blood banking and stem cell banking, Stem cell therapy	

#### Books recommended :

- 1. Gardner and Sutherland's chromosome abnormalities and genetic counselling, R.J. McKinlay Gardner and David J. Amor (2018).
- 2. Genetic counselling: ethical challenges and consequences, Dianne M. Bartels, Bonnie S. LeRoy, and Arthur L. Caplan (2011).
- 3. Ajay Paul (2000) Genetics- from genes to genomes,6th edition, Books and Allied (P), Ltd
- 4. Foundations of perinatal genetic counseling: a guide for counselors, Amber Mathiesen and Kali Roy (2018).
- 5. Rimon et al (2002) Principles and Practice of Medical Genetics, Vol I-III.
- 6. Martin H. Johnson & Barry Everitt. Essential reproduction.
- 7. Peter Snustad and Michael J Simmons (2009). Principles of Human Genetics. Fifth Edition. John Wiley & Sons, Inc.
- 8. Strachan T and Read A 2010 Human Molecular Genetics, Fourth Edition. Taylor and Francis
- 9. Ricki Lewis (2009) Human Genetics-Concepts and Application. Ninth Edition, McGraw-Hill College Publishers

# Details of Formative assessment (IA) for DSCC theory/OEC:40% weightage for total marks

Pedagogy: Lecture, Assignments, Interactive Sessions, ICT, Group Discussion

Formative Assessment 40 (Weightage in Marks includes: Written Tests, Activities/Assignment/Seminar/Presentation &Attendance)								
Assessment Occasion/type C1 C2 Total Marks								
Written Test (2)	10	10	20					
Seminar/Presentation/Activity	10		10					
Case work/Assignment/Field work/Project work etc		10	10					
Total	20	20	40					

# FacultyofScience

04-YearUGHonorsprogramme:2021-22

# GENERALPATTERNOFTHEORYQUESTIONPAPERFORDSCC/OEC (60marksforsemesterendExaminationwith2hrsduration)

Part-	A
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1. Questionnumber1-06carries2markseach.Answerany05questions :10marks

#### Part-B

2. Questionnumber07-11carries05Markseach.Answerany04questions :20marks

#### Part-C

- 3. Questionnumber12-15carries10Markseach.Answerany03questions :30marks
- 4. (Minimum1questionfromeachunitand10marksquestionmayhavesub questionsfor7+3or6+4or5+5ifnecessary)

## Total:60Marks

Note: Proportionate weightage shall be given to each unit based on number of hours prescribed.

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# **B.Sc.Semester-IV**

## Subject: Genetics Discipline Specific Course (DSCC) CourseNo.: GEN104T Title of the Course: Human Genetics and Genetic Counselling (Theory)

The course Human Genetics and Genetic Counselling in IV semester has two papers (Theory Paper –I for 04 credits &Practical paper-II for 2 credits) for 06 credits: Both the papers are compulsory. Details of the courses are as under.

CourseNo. / Course code	TypeofC ourse / course code	Theory /Practica I	Credits	Instruction hourperw eek	Total No. ofLectures/Hour s/Semester	Duration of Exam	FormativeA ssessment Marks	Summative Assessment Marks	Total Marks
GEN104T / 034GEN011	DSCC	Theory	04	04	56hrs	2hrs	40	60	100

## CourseOutcome(CO):

Aftercompletionofcourse(Theory), students will be able to:

**CO1:**Learn pattern on human disease inheritance

CO2: understand pedigree and risk calculation

**CO3**: assist in genetic counselling.

Syllabus-Course4 (Theory):Human Genetics and Genetic Counselling	Total Hrs:56
Unit-I	14hrs
Human Chromosomes: Normal Human Karyotype: Paris Nomenclature, Flow karyotyping	
(Quantification of DNA of individual chromosomes) Fluorescence Activated Cell Sorter (FACS).	
The genome project: History, organization and goals of human genome project; mapping	
strategies, current status of various maps;	
Genetic Diseases and Inheritance Pattern: Autosomal inheritance- Dominant (Ex. Adult polycystic kidney and Neurofibromatosis) Autosomal inheritance- Recessive (Ex. Albinism,	
Sickle cell anemia), X-linked – Recessive: (Ex. Duchenne muscular dystrophy-DMD) X-linked-	
Dominant : (Ex. XG blood group) Y-linked inheritance : Holandric gene (Ex. Testes	
determining factor - TDF) Multifactorial inheritance : (Ex. Congenital malformations: Cleft lip	
and palate, Rheumatoid arthritis and Diabetes) Mitochondrial diseases: (Ex. Leber's	
hereditary optic neuropathy)	

Unit-II	14hrs
<ul> <li>Unit 2: Introduction to immunology: antigens, antibodies, B and T Cells Immunity- Innate and acquired. Immune response - Humoral and Cell mediated Genetics of immune system – Antibody gene rearrangement and class switching. Inherited immunodeficiency- Ex. X-linked agammaglobulinemia. Major Histocompatibility Complex- Types, HLA disease associations. Transplantation, graft-rejection and immunosuppressors Concept of immunization</li> <li>Oncogenetics: A brief account of cancer-definition, types-Benign and Malignant; Sarcoma, Carcinoma, Lymphoma and Leukemia Properties of malignant cells, Types of genes - Proto oncogenes, Oncogenes, Difference between viral and cellular oncogenes, Tumor Suppressor genes-p53, pRB. Chromosomal abnormalities associated with the specific malignancies-Acute Promyelocytic Leukemia (APL), Chronic Myeloid Leukemia (CML) and Acute lymphoblastic leukemia (ALL)</li> </ul>	
	14hrs
<ul> <li>Prenatal diagnosis: Indications for prenatal diagnosis; Methods- Noninvasive method-Ultrasonography and Fetal echocardiography, Invasive methods - Amniocentesis, Chorionic villus sampling; pre-conceptional and pre-implantation genetic diagnosis- Teratogen exposure in early pregnancy, Genetic testing and screening.</li> <li>Gene therapy: Gene therapy with reference to SCID Stem cells- Properties, types and sources. A brief account on Cord blood banking and Stem cell therapy</li> </ul>	
Unit-IV	14hrs
<ul> <li>Genetic Counselling: Symbols used in pedigree studies, Pedigree analysis and construction, Pedigree analysis for the inheritance pattern of genetic diseases, Genetic Counselling. Introduction to Genetic counselling, historical over view, types and scope. Stages of counselling- History and pedigree construction, Examination, Diagnosis, Counselling &amp; Follow up.</li> <li>Counsellor: Definition, roles and responsibilities. Consultant - Definition, needs, rights; Individual and Group counselling; Process of Genetic Counselling - Information gathering, pedigree analysis, medical evaluation, physical examination and investigations ELSI in Genetic Counselling: Ethical, legal and social issues (ELSI), Acts and Amendments.</li> </ul>	

Booksrecommended.

- 1. Korf, B. R., & Irons, M. B. (2012). Human genetics and genomics. John Wiley & Sons.
- 2. Read, A. P. (2018). Human molecular genetics. Garland Science.
- 3. Pasternak, J. J. (2005). An introduction to human molecular genetics: mechanisms of inherited diseases. John Wiley & Sons.
- 4. Nussbaum, R., McInnes, R. R., & Willard, H. F. (2015). Thompson & Thompson genetics in medicine e-book. Elsevier Health Sciences.
- 5. Turnpenny, P. D., Ellard, S., & Cleaver, R. (2020). Emery's Elements of Medical Genetics E-Book. Elsevier Health Sciences.
- 6. Harper, P. (2010). Practical genetic counselling. CRC Press.

## B.Sc. Semester – IV Subject: Genetics Discipline Specific Course (DSCC) Course No: GEN104P Course Name: Human Genetics and Genetic Counselling (Practical)

CourseNo. / Course code	TypeofC ourse	Theory /Practica I	Credits	Instruction hourperw eek	Total No. ofLectures/Hour s/Semester	Duration of Exam	Formative Assessment Marks	Summative Assessment Marks	Total Marks
GEN104P /034GEN012	DSCC	Practical	02	04	52hrs.	3hrs	25	25	50

#### CourseOutcome(CO):

After completion of course (Practical), students will be able to:

- CO1: Collectpatient data
- CO 2 : construct and analyze pedigree
- CO 3 : calculate risk and probability of genetic diseases

#### List of the Experiments for 52hrs./Semesters

- 1. Study of Barr body in the Buccal epithelial cells
- 2. Study of drum sticks in Neutrophils of Blood smear
- 3. Blood Cell counting using Hemocytometer (RBC and WBC)
- 4. Leucocyte culture and karyotype analysis
- 5. Study of Normal Human Karyotype and identification of chromosomes
- Study of Abnormal KaryotypesDown's syndrome / Turner's syndrome / Klinefelter's syndrome.
- 7. Collecting data for genetic counselling and format preparation.
- 8. Pedigree construction techniques (manual and software aided)
- 9. Probability calculations prior, conditional, joint and posterior.
- 10. Pedigree analysis and Risk calculation in sex linked diseases
- 11. Pedigree analysis and Risk calculation in sex limited and sex influenced diseases
- 12. Pedigree analysis and Risk calculation in incomplete penetrance and expressivity
- 13. Mock genetic counselling activities

#### General instructions:

Scheme of Practical Examination (distribution of marks): 25 marks for Semester endexamination

- 1. MajorQuestion ----- 6Marks
- 2. MinorQuestion------4Marks
- 3. Identification(A-E)-----8Marks
- 4. Viva-----2Marks
- 5. Journal-----5Marks

**Total 25marks** 

#### Note:SameSchememaybeusedforIA(FormativeAssessment)examination

#### Booksrecommended.

- 1. Human Cytogenetics Vol.1& II, Rooney, Czepulkowshi
- 2. Inherited Eye Diseases, Diagnosis and clinical management, Saul Merin, Marcel Dekker Inc 1991
- 3. Human Molecular Genetics, 2nd edition, Tom Strachan and Andrew P. Read
- 4. The Biology Behind Genetic Counseling. Joanne Sutherland
- 5. Advanced Molecular Biology, 1st South Asian Edn 1998; Bios scientific publishers Ltd A concise reference by RM Twyman
- 6. PCR The Polymerase Chain Reaction, Birkhauser Boston Edn. 1994 Edited by Kary B Mullis, Francois Ferre, Richard A Gibbs
- 7. Genome Analysis, A laboratory manual, Vol 2, Detecting Genes, Bruce Birren et al; 1998 Cold Spring Harbour Laboratory Press
- 8. Genome Analysis, A laboratory manual, Vol 4, Mapping Genomes, Bruce Birren et al; 1999; Cold Spring Harbour Laboratory Press

# **B.Sc.Semester-IV**

**Subject: Genetics** 

**Open Elective Course (OEC for other students)** 

Course No.: GEN104E

#### Title of the Course: Human Genetic Disorders

CourseNo /Cours e code	TypeofCours e /Course code	Theory /Practica I	Credits	Instruction Hour per week	Total No. ofLectures/Hour s/Semester	Duration of Exam	FormativeA ssessment Marks	Summative Assessment Marks	Total Marks
GEN104E / 004GEN051	OEC	Theory	03	03	42hrs.	2hrs	40	60	100

## CourseOutcome(CO):

After completion of course, students will be having basic knowledge of:

- **CO1**: Mendelian genetics and inheritance of characteristics
- **CO 2** : Genotype phenotype correlation
- **CO 3 :** Extranuclear inheritance

Syllabus-OEC4:Title-Human Genetic Disorders		
Unit-I	14hrs	
<b>Human Chromosomes:</b> study of Normal Human Karyotype and abnormal karyotype. <b>Pattern of inheritance:</b> Autosomal dominant –PKU, Autosomal recessive – Sickle cell anemia, X-linked dominant – Fragile X syndrome, X linked recessive – DMD, Y linked inheritance and mitochondrial inheritance pattern.		
Unit-II	14hrs	
<b>Inborn errors of metabolism:</b> Disorders of carbohydrate metabolism–Galactosaemia, Disorders of amino acid metabolism – Alkaptonuria, Disorders of Lysosomal enzymes– Tay- Sachs disease, Disorders of Lipoprotein and lipid metabolism – Hyper Lipoproteinemia, Disorders of Purine metabolism-Lesch-Nyhan syndrome.		
Unit-III	14hrs	
Single gene diseases:Cystic fibrosis, hemophilia, oculocutaneous albinism. Multifactorial disorder:Diabetes, Parkinson's disease and schizophrenia. Gene Therapy: Cancer gene therapy, Gene therapy of non-heritable disorders; Cord blood banking and stem cell banking, Stem cell therapy.		

#### **Books recommended :**

- 1. Human Cytogenetics (vol. I &II) J.L. Hamerton
- 2. Human chromosomes: E.H. FORD
- 3. Human Genetics F. Vogel and A.G. Motulsky.
- 4. Genetics and Medicine M.W Thompson, R.R. Meinees and H.F Willard
- 5. Basic human genetics E.J. Mange and A.P. Mange.
- 6. Medical Genetics Jorde et al 7. New chromosomal syndromes: J.J. Yunis
- 7. Comprehensive clinical endocrinology: Besser and Thorner
- 8. Principles and practice of medicalgenetics:Rimoinetal

# Details of Formative assessment (IA) for DSCC theory / OEC: 40% weightage for total marks

Pedagogy: Lecture, Assignments, Interactive Sessions, ICT, Group Discussion

Formative Assessment 40 (Weightage in Marks includes: Written Tests, Activities/Assignment/Seminar/Presentation &Attendance)					
Assessment Occasion/type	C1	C2	Total Marks		
Written Test (2)	10	10	20		
Seminar/Presentation/	10		10		
Activity					
Case work/Assignment/Field work/Project work etc		10	10		
Total	20	20	40		

#### FacultyofScience 04-YearUGHonorsprogramme:2021-22

#### GENERALPATTERNOFTHEORYQUESTIONPAPERFORDSCC/ OEC (60marksforsemesterendExaminationwith2hrsduration)

#### Part-A

1. Questionnumber1-06carries2markseach.Answerany05questions	: 10mark	KS
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## Part-B

2.	$Question number 07 \hbox{-} 11 carries 05 Markseach. Answer any 04 questions$	: 20marks
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## Part-C

3. Questionnumber12-15carries10Markseach.Answerany03questions : 30 marks

(Minimum1 question from each unit and 10 mark squestion may have subque

stionsfor7+3or6+4or5+5ifnecessary)

## Total:60Marks

Note: Proportionate weightage shall be given to each unit based on number of hours prescribed.

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