FACULTY OF LIFE SCIENCES

SYLLABUS

FOR

Pre-Ph.D. Course in Human Genetics

(Credit Based Evaluation & Grading System)

Examinations: 2019-20

GURU NANAK DEV UNIVERSITY AMRITSAR

Pre Ph.D. Course in Human Genetics
(Credit Based Evaluation & Grading System)

Programme Code: HGZ

Scheme of Course

Course No.	C/E/I	Course Title	L	T	P	Total Credit	Total Marks		
Core Courses (5 Credits)							**Max Marks**	**Mid Sem**	**End Sem**
HGS-921	**C**	**Seminar**	**1**	**0**	**0**	**1**	**100**	**20**	**80**
LSL-901	**C**	**Research Methodology**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
Elective Courses (9 Credits)									
HGL-901	**E**	**Advances in Genomics and Human Genetics**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGL-903	**E**	**Genetics in Medicine**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGL-905	**E**	**Applied Human Population Genetics**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGL-906	**E**	**Genomics to Proteomics**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGL-907	**E**	**Prevention and Management of Genetic Diseases**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
Interdisciplinary/Optional Course (4 Credits)									
–	**I**	**To be offered from outside the department**	**4**	**0**	**0**	**4**	**100**	**20**	**80**

1. The students are required to take one compulsory paper ‘LSL-901 Research Methodology’ (Core) and one paper ‘HGS-921 Seminar’ (Core) of the department, and one paper Interdisciplinary/Optional) of their choice from any other department of Guru Nanak Dev University Campus, Amritsar.
2. These courses may be opted from the same department or in an allied field/s in other departments of the University (depending on their area of specialization/research interest) and at least one of these courses should be from outside the department.
3. The courses no. HGL-901, HGL-903, HGL-905, HGL-906 and HGL-907 will also be offered as interdisciplinary courses for the students of other departments of Guru Nanak Dev University Campus, Amritsar.

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LSL-901 - Research Methodology

Credits 3-1-0
Time: 3 Hours **Max. Marks: 100**
Mid Semester Marks : 20
End Semester Marks : 80

Mid Semester Examination: 20% weightage
End Semester Examination: 80% weightage

Instructions for the Paper Setters:
Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Note: The course will be numerical oriented to train the students for the analysis of research data. Use of calculators will be allowed in the examination.

SECTION-A

1. **Descriptive statistics:** Statistical expressions, central tendency, dispersion of data (arithmetic and geometric), moments, skewness, kurtosis, sample size estimation.
2. **Probability**: Concept of probability, conditional probability, distributions: Normal, Poisson, binomial, 't', χ^2, F-distributions.

SECTION-B

3. **Testing of hypothesis**: Central limit theorem, null hypothesis and alternative hypotheses, Z-test, Student's t-test, χ-square, F-test, sample size, confidence intervals.odds ratio, index numbers, Probit analysis.
4. **Correlation and regression analysis**: Linear correlation and regression, exponential regression, logarithmic regression, reciprocal regression, Michael-Menten's regression, logistic regression, Gompertz regression, monomolecular regression.

SECTION-C

5. **Multiple correlation and regression**: MLR with 2 and 3 independent variables, quadratic and cubic polynomial regressions, Beta regression, sine curve, multiple correlation, partial correlation, path analysis, time series analysis.
6. **Experimental designs:** Experimental designs, central composite designs with 2 and 3 factors.

SECTION-D

7. **Analysis of Variance**: Assessing normality, one way and 2-way ANOVA, Tukey's multiple comparison test, HSD.
8. **Multivariate analysis**: Cluster analysis and dendrogram, principal component analysis, factor analysis, artificial neural networks.
9. **Non-parametric tests:** Wilcoxon's, Mann-Whitney's tests, Spearman's rank correlation, Kendall's Tau.
10. **Basic Greek and Latin words:** The students will learn Greek alphabet and more than 100 basic roots and words used in science.

Note: The students will be asked to submit an assignment of computer softwares designed by them on the basis of the Research methodology syllabus.

References:

1. Bailey, N.T.J. (1995). Statistical Methods in Biology. Cambridge University Press, Cambridge.
2. Kothari, C.R. (2004). Research Methodology: Methods and Techniques, New Age International Publishers, New Delhi.

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HGL-901 ADVANCES IN GENOMICS AND HUMAN GENETICS
(Elective Paper)

Credits: 3-0-0
Time: 3 Hours **Max. Marks: 100**
Mid Semester Marks : 20
End Semester Marks : 80

Mid Semester Examination: 20% weightage
End Semester Examination: 80% weightage

Instructions for the Paper Setters:
Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A
Molecular markers and disease, Mitochondrial genetics, Genetics of simple and complex diseases, Copy nuclear variation (CNV) and human health, Genetics of autoimmune disease and cancer, Studying human gene function through animal models, Genetic testing

Section-B
Microarray and its applications in genomic studies, Real Time PCR - Principle and applications in human genetics and human health. Next generation DNA sequencing and its applications in the diagnosis of human disease.

Section-C
Transcritomics, Metagenomics. Genotyping technologies for genetic research, Study design for monogenic and complex disorders: Linkage analysis, Genome-wide linkage studies, Candidate gene studies, GWAS and its applications

Section-D
Pharmacogenomics- Principles and practices, Human genome and public health, Gene therapy vs gene doping, Epigenomics: CpG islands, DNA methylation; miRNA and its role in studying complex disorders, 'Flip-Flop' phenomena for gene reversal; Missing inheritance.

Recommended Books:
1. Annual Review of Genomics and Human Genetics (2009). Academic Press, New York, Vol. 10.
2. Brown, T.A. (2006). Gene Cloning and DNA Analysis. Blackwell Publishing Co., Oxford, 6th ed.
3. Strachan, T. and Read, A.P. (2010). Human Molecular Genetics. Garland Publishers, New York, 4thed.
4. http://arjournals.annualreviews.org/toc/genom/10/1.

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HGL-903: GENETICS IN MEDICINE
(Elective Paper)

Credits: 3-0-0

Time: 3 Hours **Max. Marks: 100**

Mid Semester Marks : 20
End Semester Marks : 80

Mid Semester Examination: 20% weightage
End Semester Examination: 80% weightage

Instructions for the Paper Setters:
Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Developmental Origin of Health and Disease and Transgenerational inheritance, Stem cells in Regenerative Medicine, Cord Blood banking and applications.

Section-B

Reprogenetics, Pre-conceptional diagnosis, Cell- free fetal Nucleic acids testing. Public Health Genomics

Section-C

Whole Genome and Exome Sequencing in Medicine, Secondary and Incidental findings in genomic sequencing, Concept of exosomes

Section-D

Domains of Personal Genomics, Personalized Medicine, Precision Medicine Initiative, Systems Medicine.

Recommended Books:

1. Gibson, G. (2015). A Primer of Human Genetics. Sinauer.
2. Korf, B.R. and Irons, H.B. (2013). Human Genetics and Genomics. John Wiley and Sons, Hoboken, 4th ed.
3. Nussbaum, R.L., McInnes, R.R. and Willard, H.F. (2015). Thompson & Thompson's Genetics in Medicine. Saunders, 8th ed.
4. Read, A. and Donnai, D. (2015). New Clinical Genetics. Scion, UK, 3rd ed.
5. Rimoin, D.L., Pyeritz and Korf, P.R. (2013). Emery and Rimoin's Principles and Practice of Medical Genetics, Academic Press, New York, 6th ed.
6. Skirton, H. and Patch, C. (2017). Genetics for the Health Sciences. Viva Books, New Delhi, published by arrangements with Scion Publishing Limited.
7. Strachan, T. Goodship, J. and Chinnery P. (2015). Genetics and Genomics in Medicine. Garland Publishers, New York.
8. Turnpenny, P.D. and Ellard, S. (2015). Emery's Elements of Medical Genetics. Elsevier, 15th ed.

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HGL-905 APPLIED HUMAN POPULATION GENETICS
(Elective Paper)

Credits: 3-0-0

Time: 3 Hours **Max. Marks: 100**

Mid Semester Marks : 20
End Semester Marks : 80

Mid Semester Examination: 20% weightage
End Semester Examination: 80% weightage

Instructions for the Paper Setters:
Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section- A

Application of human population genetics. Probability in population genetics, addition and multiplication rule. Means, variance and confidence intervals, null hypothesis, type I and II errors, statistical power of the test, multiple comparisons and standard Bonferroni or Dunn-Sidate correction. Procedure and principle to obtain p-value.

Section- B

Relation between inbreeding coefficient and F-statistics. Wahlund's principle and fixation index. Population stratification. Effective population size, factors such as inbreeding, founder effect and bottleneck influences on effective population size.

Section-C

Foundation of genetic epidemiology. Descriptive epidemiology, incidence, point and period prevalence. Phenotypic aggregation within families. Basic types of study design, descriptive studies, analytical studies. Varieties of biased study design and steps for minimizing bias.

Section- D

Models for the ethics of population genetic research. Biobank's ethics and governance frame work. Recruitment of participants. Consent, race, ethnicity and genetics.

Books Recommended

1. Balding, D.J., Bishop, M and Cannings (eds) (2007). Handbook of Statistical Genetics. John Wiley and Sons Ltd., England, Vol. I,II.
2. Hedrick, P.W. (2005). Genetics of Population. Jones and Bartlett Publishers, Massachusetts.
3. Indrayan, A (2008). Medical Biostatics. Chapman and Hall, New York.
4. Kothari, C.R. (2009). Research Methodology: Methods and Techniques. New Age International Publishers, New Delhi.

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HGL-906 GENOMICS TO PROTEOMICS
(Elective Paper)

Credits: 3-0-0
Time: 3 Hours **Max. Marks: 100**
Mid Semester Marks : 20
End Semester Marks : 80

Mid Semester Examination: 20% weightage
End Semester Examination: 80% weightage

Instructions for the Paper Setters:
Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A
Molecular genetics of eye diseases, Molecular basis of bones and musculo-skeletal diseases.

Section-B
Human genomic sequence and annotations, Comparative homologies of human genomic sequence, evolutionary changes and SNPs, Genetic variations and molecular evolution, functional characterizations of mutant alleles, Genotype-phenotype correlations, Human phenotype ontologies.

Section-C
Gene to protein, Characterization of proteins, HPLC, FPLC, 2-DE analyses, Mass spectrometry and their applications.

Section- D
Protein sequence analyses, protein microarray, protein modifications and protein- protein interaction analyses, Computational analyses for proteins identification and their function, Protein biomarkers for disease diagnosis, Proteomics in drug discovery.

Recommended Books:

1. Azuaje, F. and Dopazo, J. (2006). Data Analysis and Visualization in Genomics and Proteomics. John Wiley and Sons, Inc., USA.
2. Baxevanis, A.D. and Ouellette, B.F.F. (2001). Bioinformatics: A Practical Guide to the Analysis and Proteins. John Wiley and Sons, Inc., USA.
3. Baxevanis, A.D. and Ouellette, B.I.I. (Eds.) (2005). Bioinformatics: A Practical Guide to the Analysis of Genes and Proteins. John Wiley and Sons, Inc., USA, 3rd ed.
4. Brown, J.R. (2008). Comparative Genomics: Basic and Applied Research. CRC (Taylor and Francis). 5. Brownstein, M.J., Khodursky and Arkady, B. (Eds.) (2003). Functional Genomics. Humana Press, New Jersey.
5. Copeland, R.A. (1994). Methods for Protein Analysis. A Practical Guide to Laboratory Protocols, Chapman and Hall, USA.
6. Dale, J.W. and Schantz, M.V. (2002). From Genes to Genomes: Concepts and Applications of DNA Technology. John Wiley and Sons, Inc., USA.
7. Dunham, I. (Ed.) (2003). Genome Mapping and Sequencing. Horizon Scientific Press, UK.
8. Figeys, D. (Ed.) (2005). Industrial Proteomics Applications for Biotechnology and Pharmaceuticals. John Wiley and Sons, Inc., USA.
9. Hart, W.M. (1992). Adler's Physiology of the Eye. Mosby Year Book, Inc., USA, 9th ed.
10. Jaffe, N.S. and Horwitz, J. (1992). Lens and Cataract. Gower Medical Publishing Co., Vol. 3.
11. Mount, D.W. (2004). Bioinformatics: Sequence and Genome Analysis. Cold Spring Harbor Laboratory, New York, 2nd ed.
12. O'Connor, C.D. and Hames, B.D. (2008). Proteomics. Scion Publishing Ltd.
13. Taylor, G.R. and Day, I.N.M. (Eds.) (2005). Guide to Mutation Detection. John Wiley and Sons, Inc., USA.
14. Twyman, R.M. (2004). Principles of Proteomics. Taylor and Francis Publishers.

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HGL-907 PREVENTION AND MANAGEMENT OF GENETIC DISEASES
(Elective Paper)

Credits: 3-0-0

Time: 3 Hours **Max. Marks: 100**
Mid Semester Marks : 20
End Semester Marks : 80

Mid Semester Examination: 20% weightage
End Semester Examination: 80% weightage

Instructions for the Paper Setters:
Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A
Genetic evaluation and counselling in common genetic conditions, Interviewing techniques, Psychosocial and emotional issues, Case preparation, Pedigree construction, Communication, Consent for all investigations.

Section-B
Dysmorphology examination, Importance of facial features and physical measurements, interpretation of research findings, Impact of genetic disorders on patient's family, Treatment options and limitations.

Section-C
Risk assessment for congenital diseases, Sex anomalies, Mental retardation, Recurrent abortions, and Cancer for families and particular ethnic groups,

Section-D
Searching online resources and support groups, Issues in health-care systems, Legal issues in counselling, Methods of outreach progress, Importance of genetic database, management and confidentiality.

Books Recommended

1. Harper, P.S. (1998) Practical Genetic Counselling. Butterworth Heineman, 5th Ed.
2. Mendelow, B., Ramsay, M., Chetty, N. and Stevens, W. (2009). Molecular Medicine for Clinicians. Wits University Press, Johnnesburg.
3. Pritchard, D. and Korf, B. (2008). Medical Genetics at a Glance. Blackwell Publishers, Oxford, 2nd Ed.
4. Thompson, J.S. (2007). Thompson and Thompson Genetics in Medicine, Saunders/ Elsevier, Philadelphia, 7th Ed.
5. Weil, J. (2000). Psychosocial Genetic Counselling. Oxford University Press Inc., New York.