FACULTY OF LIFE SCIENCES

SYLLABUS

For

Bridge Course in HUMAN GENETICS

(Under Credit Based Continuous Evaluation Grading System)

(Semester: III–IV)

Session: 2015-16

GURU NANAK DEV UNIVERSITY AMRITSAR

Bridge Course in Human Genetics (Semester System)
(Under Credit Based Continuous Evaluation Grading System)

SCHEME OF COURSES
(2015-2016)

Semester-III

Course No.	C/E/I	Course Title	L	T	P	Total Credits
Core Courses (19 Credits)						
HGL-422	C	Medical Genetics	3	1	0	4
HGL-501	C	Quantitative Genetics	3	0	0	3
HGL-502	C	Functional Genomics	3	0	0	3
HGL-505	C	Human Embryology and Developmental Genetics	3	0	0	3
HGS-506	C	Seminar	0	0	1	1
HGP-422	C	Medical Genetics Practical	0	0	2	2
HGP-502	C	Functional Genomics Practical	0	0	3	3
Elective Course (3 Credits)						
HGL-511	E	Immunogenetics in Health and Disease	3	0	0	3
HGL-514	E	Genetics of Human Pathogens and Infectious Diseases	3	0	0	3
Interdisciplinary/Optional Course (3 or 4 Credits) —						
–	I	To be taken from outside the department	3	0	0	3
Total Credits			**18**	**1**	**6**	**25**

1. The students are required to opt for one 'Elective' paper.
2. The students are also required to take one paper (Interdisciplinary/Optional) of 3 or 4 credits of their choice from any other department of Guru Nanak Dev University Campus, Amritsar.

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Semester-IV

Course No.	C/E/I	Course Title	L	T	P	Total Credits
Core Courses (18 Credits)						
HGL-472	C	Clinical Genetics and Genetic Counselling	3	1	0	4
HGL-551	C	Perspectives of Human Genome and Bioethics	3	0	0	3
HGL-552	C	Applied Human Molecular Genetics	3	0	0	3
HGL-554	C	Structural Biology and Pharmacogenomics	3	0	0	3
HGP-472	C	Clinical Genetics Practical	0	0	2	2
HGP-552	C	Applied Human Molecular Genetics Practical	0	0	3	3
Elective Course (3 Credits)						
HGL-411	E	Regulatory Approaches in Human Genetics	3	0	0	3
HGL-412	E	Biological Demography	3	0	0	3
Interdisciplinary/Optional Course (3 or 4 Credits) —						
–	I	To be taken from outside the department	3	0	0	3
Total Credits			**18**	**1**	**5**	**24**

1. The students are required to opt for one 'Elective' paper.
2. The students are also required to take one paper (Interdisciplinary/Optional) of 3 or 4 credits of their choice from any other department of Guru Nanak Dev University Campus, Amritsar.

Bridge Course in Human Genetics (Semester-III)
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HGL-422 MEDICAL GENETICS

Credits: 3-1-0

Unit-I

Growth and development of medical genetics (1956-to-present), Role of genetics in medicine, Types of genetic disorders — single gene, chromosomal, multifactorial. Genetic disorders with classical Mendelian inheritance with examples, Medical relevance of meiosis and mitosis, Dominance and recessiveness, Mechanism of dominance, Concept of phenotype and relationship between genotype and phenotype in genetic disease.

Unit-II

Concepts of genetic heterogeneity, Reduced penetrance, Variable expressivity, Pseudoautosomal inheritance, Genomic imprinting, Mosaicism, Uniparental disomy and Pleiotropy with suitable examples, Consequences of consanguineous marriages, Mitochondrial diseases.

Unit-III

Pharmacogenetic diseases (Hereditary disorders with altered drug response, Malignant hyperthermia, G-6-PD deficiency), Chromosomal instability syndromes, Sex chromosomal and differentiation anomalies, Chromosomal microdeletion syndromes, Prion diseases, Gene mapping in DMD, CF, HD.

Books Recommended

1. Hudgins, L., Toriello, H.V. Enns, G.M. and Hoyme, H.E. (Eds.) (2014). Signs and Symptons of Genetic Conditions: A Handbook. Oxford University Press, London, 1st ed., p. 560.
2. Jorde, L.B., Carey, J.C. and Bamshad, M.J. (2010). Medical Genetics. Mosby, 4th ed.
3. Korf, B.R. and Irons, M.B. (2013). Human Genetics and Genomics. John Wiley and Sons, Hoboken, 4th ed.
4. Nussbaum, R.L., McInnes, R.R. and Willard, H.F. (2007). Thompson & Thompson Genetics in Medicine. Saunders, 7th ed.
5. Rimoin, D.L., Pyeritz, R.E. and Korf, B.R. (2013). Emery and Rimoin's Essential Medical Genetics. Academic Press, New York, 1st ed.
6. Rimoin, D.L., Pyeritz, R.E. and Korf, B.R. (2013). Emery and Rimoin's Principles and Practice of Medical Genetics. Academic Press, New York, 6th ed.
7. Rosenberg, L.E. and Rosenberg, D.D. (2012). Human Genes and Genomes: Science, Health, Society. Academic Press, 1st ed.
8. Speicher, M., Antonarakis, S.E. and Motulsky, A.G. (Eds.) (2010). Vogel and Motulsky's Human Genetics: Problems and Approaches. Springer Verlag, 4th ed.
9. Strachan, T., Goodship, J. and Chinnery, P. (2014). Genetics and Genomics in Medicine. Garland Science, New York, 1st ed.
10. Tobias, E.S., Connor, M. and Ferguson-Smith, M. (2011). Essential Medical Genetics. Wiley-Blackwell, 6th ed.
11. Turnpenny, PD. (2012). Emery's Elements of Medical Genetics. Churchill Livingstone, 14th ed.

Bridge Course in Human Genetics (Semester-III)
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HGL-501 QUANTITATIVE GENETICS

Credits: 3-0-0

Unit-I

Calculation of population mean and variation, Average effects of genes, Measurements of total genetic variances, Genetic and environmental components of variance, Heritability estimation through twin data, Measures of heterozygosity and gene diversity (through Nei and others), Selection favouring heterozygotes, Test for genetic heterogeneity for two alleles, Descriptive epidemiology, incidence and prevalence.

Unit-II

The linkage disequilibrium and measures of D. Polygenic model for quantitative traits, Kinship and genetic sharing. Notation of genetic distances with different index, Measurements of standard genetic distances of Nei, Natural selection and sex ratio, Measurements of genetic similarities and distance between the populations, Analysis of racial differences on the basis of polymorphic genes.

Unit-III

Estimation of genetic correlation, Resemblance between relatives and concept of heritability, Gene fixation probability and fixation time, Effective population size and population bottlenecks, Concept and significance of genetic admixture, Estimating admixture proportions from frequencies of a single allele, Quantitative trait loci (QTL), Introduction to molecular phylogenetics, Unweighted pair group method (UPGMA), Neighbour-joining (NJ) method, Genetic diversity within and among population, Phenotypic plasticity, Cultural transmission, Wright's F-statistics and G-statistics.

Books Recommended

1. Balding, D.J., Bishop, M. and Cannings, C.C. (Eds.) (2007). Handbook of Statistical Genetics. John Wiley and Sons, England, Vol. 1 and 2, 3rd ed.
2. Brandon, R.N. (1996). Concepts and Methods in Evolutionary Biology. Cambridge University Press, USA.
3. Cavalli-Sforza, L.L. and Bodmer, W.F. (1971). The Genetics of Human Population. W.H. Freeman and Co., San Francisco, CA.
4. Falconer, F.S. and MacKay, T.F.C. (1996). Introduction to Quantitative Genetics. ELBS/Longman, England, 4th ed.
5. Hamilton, M.B. (2009). Population Genetics. Wiley-Blackwell, USA.
6. Hartl, D.L. and Clark, A.G. (2007). Principles of Population Genetics. Sinaur Associates, Inc., Massachusetts, 4th ed.
7. Hedrick, P.W. (2011). Genetics of Population. Jones and Bartlett Publishers, Massachusetts, 4th ed.
8. Neale, B., Ferreira, M.A.R., Medland, S.E. and Posthuma, D. (Eds.) (2008). Statistical Genetics: Gene Mapping through Linkage and Association. Taylor and Francis Group, USA.

Bridge Course in Human Genetics (Semester-III)
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HGL-502 FUNCTIONAL GENOMICS

Credits: 3-0-0

Unit-I

Split genes and overlapping genes, DNA hybridization/denaturation, DNA cot curves, Multigene families and repetitive DNA, Genetic mechanisms resulting in sequence exchanges, pathogenic mutations and repeated sequences, Structure and function of microRNAs, snRNA, RNA interference, Role of microRNAs in human disease diagnostics.

Unit-II

Genetic approaches in treating human diseases, Principles and applications of gene therapy, Methods of transferring transgenes into target cells, Gene therapy for cystic fibrosis, Duchenne muscular dystrophy, bleeding disorders and severe combined immunodeficiency syndrome.

Unit-III

DNA sequencing and its applications, DNA profiling, DNA fingerprinting and its applications, DNA microarray and its applications, Transcriptomics, Evolution of mitochondrial genome, eukaryotic nuclear genome, human sex chromosome, human DNA sequence families and DNA organization, gene structure and modern humans.

Books Recommended

1. Brown, T.A. (2005). Genetics: A Molecular Approach. Bios Scientific Publishers Ltd., Oxford, 3rd ed.
2. Brown, T.A. (2006). Genomes 3. Bios Scientific Publishers Ltd., Oxford, 3rd ed.
3. Brown, T.A. (2010). Gene Cloning and DNA Analysis: An Introduction. Blackwell Publishing Co., Oxford, 6th ed.
4. Strachan, T. and Read, A. (2010). Human Molecular Genetics. Garland Publishers, London, 4th ed.
5. Sudbery, P. and Sudbery I (2009). Human Molecular Genetics. Pearson Education, UK, 3rd ed.
6. Primrose, S.B. and Twyman, R.M. (2003). Principles of Genome Analysis and Genomics. Blackwell Publications, London.
7. Watson, J.D., Myers, R.M., Caudy, A.A. and Witkowski, J.A. (2007). Recombinant DNA Genes and Genomes – A Short Course. Cold Spring Harbor Laboratory Press, 3rd ed.

Bridge Course in Human Genetics (Semester-III)
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HGL-505 HUMAN EMBRYOLOGY AND DEVELOPMENTAL GENETICS

Credits: 3-0-0

Unit-I

Fertilization and molecular events during fertilization, and prenatal development of human embryo up to three germinal layers. differential gene activity and cell differentiation, Gastrulation, cleavage patterns, fate map during gastrulation, Notochord formation, Neurulation, Formation of somites, Structure of somites, Formation of blood vessels.

Unit-II

Implantation, Formation and types of placenta, Genetic and molecular control of development of head and neck region, development of nervous system.

Unit-III

Genetic and molecular control of development of limbs, gastrointestinal system and cardiovascular system, Genetics of sex determination in humans and development of urogenital system.

Books Recommended

1. Gilbert, S.F. (2011). Developmental Biology. Sinnauer Association, Inc., Sunderland, MA.
2. Mueller, R.F. and Young, I.D. (2010). Emery's Elements of Medical Genetics. Churchill Livingstone, New York, 11th ed.
3. Nussbaum, R.L., McInnes, R.R. and Willard, H.F. (2001). Thompson and Thompson Genetics in Medicines. W.B. Saunders and Co., New York, 6th ed.
4. Sadler, T.W. (2010). Langman's Medical Embryology. Williams and Wilkins, Philadelphia, USA, 11th ed.
5. Snustad, P. and Simmons, M.J. (2010) Principle of Genetics. John Wiley and Sons, Inc., N.J., USA.
6. Singh, I. and Paul, G.P. (2009). Human Embryology. McMillan Co., New Delhi.

Bridge Course in Human Genetics (Semester-III)
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HGS-506 SEMINAR

Credits: 0-0-1

The paper is based on the seminars delivered by the students on current topics related to various disciplines of Human Genetics.

Bridge Course in Human Genetics (Semester-III)
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HGP-422 MEDICAL GENETICS PRACTICAL

Credits: 0-0-2

Analysis of different modes of inheritance, Numericals and case studies on medical genetics, Karyotyping from abnormal chromosomal preparations, Genetic databases — OMIM, London dysmorphology database, Possum, Repertox, Human cytogenetics database, Online medical genetics resources – OMIM, Gene clinics, Gene tests, ESHG; Sites for patients — Genetic Alliance, Family village.

Bridge Course in Human Genetics (Semester-III)
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HGP-502 FUNCTIONAL GENOMICS PRACTICAL

Credits: 0-0-3

Preparation of different stock solutions for DNA extraction and electrophoresis, Preparation of working solutions from stock solutions, Extraction of DNA from human blood by organic method, DNA quantification, Amplification of genomic DNA by PCR, PCR-RFLP, Qualitative and quantitative estimation of carbohydrates, Lipids and proteins.

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Bridge Course in Human Genetics (Semester-III)
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HGL-511 IMMUNOGENETICS IN HEALTH AND DISEASE

Credits: 3-0-0

Unit-I

Immunoassays to assess primary and secondary antigen-antibody interaction, Evaluation of lymphocyte function, Monoclonal and genetically engineered antibodies, Cell culture systems, Experimental animal models in immunogenetics.

Unit-II

Autograft, isograft, allograft, xenograft, Immunological basis of graft rejection, Clinical characteristics of allograft rejection, HLA typing, Circumvention of alloimmune response, Maternal-fetal incompatibility, Role of MHC in resistance and susceptibility to diseases.

Unit-III

Tumor antigens, Limitations of effectiveness of immune response against tumors, Immunodiagnosis of tumor antigens, Types and properties of immunotolerance, T-cell tolerance to self-antigens, B-cell tolerance to self-antigens, Central tolerance, Peripheral tolerance, Acquired allogeneic tolerance.

Books Recommended

1. Abbas, A.K., Lichtman, A.H. and Pillai, S. (2010). Cellular and Molecular Immunology. WB Saunders Co., USA.
2. Benjamini, E., Coico, R. and Sunshine, G. (2009). Immunology: A Short Course. John Wiley and Sons, Inc., New York.
3. Delves, P.J., Martin, S.J., Burton, D.R. and Roit, I.M. (2006). Essential Immunology. Blackwell Scientific Publications, Oxford.
4. Kindt, T.J., Goldsby, R.A. and Osborne, B.A. (2007). Kuby Immunology. W.H. Freeman and Co., New York.
5. Parham, P. (2009). The Immune System. Taylor and Francis Group, New York.

Bridge Course in Human Genetics (Semester-III)
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HGL-514 GENETICS OF HUMAN PATHOGENS AND INFECTIOUS DISEASES

Credits: 3-0-0

Unit-I

Introduction to pathogens, common bacterial, viral and protozoal pathogens and their molecular interaction with the host, Common strategies to colonize host, Co-evolution between host and pathogen, Evolution of virulence and pathogenicity, Pathogenicity islands, Evolution of antibiotic resistance (MRSA strains), Molecular mechanisms for origin of new pathogens.

Unit-II

Role of host genetic factors in infectious diseases, Genetic theory of infectious diseases, Genetic epidemiology of infectious diseases, Identifying infectious disease susceptibility genes, Use of mouse models for genetic susceptibility to infectious diseases, Genetic diversity in MHC and immune responses to infectious diseases.

Unit-III

Genetic determinants and major genes affecting common infectious diseases, Mycobacterial diseases; Tuberculosis and Leprosy, Viral Diseases, Hepatitis-B and Hepatitis-C, HIV/AIDS, Common protozoal diseases, Malaria, RBC genes and Malaria, Leishmaniasis, Genetic variation in response to vaccination.

Books Recommended

1. Alberts, B., Johnson, A. and Lewis, J. (2002). Molecular Biology of the Cell. Garland Science, New York, 4th ed.
2. Kimman, T.G. (2001). Genetics of Infectious Disease Susceptibility. Springer Science and Business Media.
3. Richard, A., Kaslow, M.D., Janet, M., McNicholl, M.D. and Hill, A.V.S. (2008). Genetic Susceptibility to Infectious Diseases. Oxford University Press, USA.
4. Willey, J., Sherwood, L. and Woolverton, C. (2010). Prescott's Microbiology. McGraw-Hill Higher Education, 8th ed.

Bridge Course in Human Genetics (Semester-IV)
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HGL-472 CLINICAL GENETICS AND GENETIC COUNSELLING

Credits: 3-1-0

Unit-I

Clinical Applications of Medical Genetics — Prenatal diagnosis: purpose, indications and techniques — invasive and non-invasive, Amniocentesis, Chorionic villus sampling, Ultrasound, Fetoscopy, Maternal serum screening, Fetal cells in maternal blood, Preimplantation diagnosis, Effect of mutagenic and teratogenic exposure in early pregnancy.

Unit-II

Carrier screening — criteria for heterozygous screening programmes, Carrier testing for autosomal recessive and X-linked disorders, Risk calculations, Population screening for genetic disease — adult, newborn, Clinical utilization of presymptomatic and predispositional testing, Presymptomatic testing for genetic diseases and malignancy.

Unit-III

Genetic Counselling — Process, Role of genetic counsellors, Diagnostic problems in genetic counselling, Psychosocial aspects of genetic counselling, Genetic care pathway and preventive management guidelines. Role of social workers, nutritional, occupational, physical, speech therapist, psychologists and school professionals in genetics, Gene therapy and stem cell research in clinical genetics.

Books Recommended

1. Cassidy, S.B. and Allanson, J.E. (2010). Management of Genetic Syndromes. Wiley-Blackwell, 3rd ed.
2. Gardner, R.J.M., Sutherland, G.R. and Shaffer, L.G. (2011). Chromosome Abnormalities and Genetic Counselling. Oxford University Press, 4th ed.
3. Gunder, L.M.and Sudbury, S.A.M. (2011). Essentials of Medical Genetics for Health Professionals. Jones and Bartlett Learning.
4. Hudgins, L., Toriello, H.V. Enns, G.M. and Hoyme, H.E. (Eds.) (2014). Signs and Symptons of Genetic Conditions: A Handbook. Oxford University Press, London, 1st ed., p. 560.
5. Jorde, L.B., Carey, J.C. and Bamshad, M.J. (2010). Medical Genetics. Mosby, 4th ed.
6. Korf, B.R. and Irons, H.B. (2013). Human Genetics and Genomics. John Wiley and Sons, Hoboken, 4th ed.
7. Nussbaum, R.L., McInnes, R.R. and Willard, H.F. (2007). Thompson & Thompson Genetics in Medicine. Saunders, 7th ed.
8. Mummery, C., Wilut, I., Van de Stolpe, A. and Roelen, B.A.J. (Eds.). (2011). Stem Cells: Scientific Facts and Fiction. Academic Press, London.
9. Pritchard, D.J. and Korf, B.R. (2013). Medical Genetics at a Glance. Wiley-Blackwell, 3rd ed.
10. Read, A. and Donnai, D. (2010). New Clinical Genetics. Scion, U.K., 2nd ed.
11. Rimoin, D.L., Pyeritz, R.E. and Korf, B.R. (2013). Emery and Rimoin's Essential Medical Genetics. Academic Press, New York, 1st ed.
12. Rosenberg, L.E. and Rosenberg, D.D. (2012). Human Genes and Genomes: Science, Health, Society. Academic Press, 1st ed.
13. Strachan, T., Goodship, J. and Chinnery, P. (2014). Genetics and Genomics in Medicine. Garland Science, New York, 1st ed.
14. Strachan, T. and Read, A. (2011). Human Molecular Genetics. 4th ed., Garland Publishers, New York.
15. Tobias, E.S., Connor, M. and Ferguson-Smith, M. (2011). Essential Medical Genetics. Wiley-Blackwell, 6th ed.

Bridge Course in Human Genetics (Semester-IV)
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HGL-551 PERSPECTIVES OF HUMAN GENOME AND BIOETHICS

Credits: 3-0-0

Unit-I

The Human Genome Project, Goals and organization, Benefits and ELSI, Draft and complete sequence of the Human Genome, Overview of Human Genome Diversity Project, Environmental Human Genome Project, Cancer genome anatomy project, The SNP/Hap Map project, The Phenome project, The Metabolome project..

Unit-II

Bioethics — Its historical aspects, Types and scope, Rights'-based ethics theories, Duty-based ethics theories and utilitarian ethics, Basic aspects of autonomy, Non-maleficience, beneficience and justice, Informed consent and confidentiality, Universality of bioethics, Bioethical maturity, Environmental ethics.

Unit-III

Ethical dilemmas — Contexual vignettes in medical genetic advances, Genetic selection, Germline gene therapy, Genetic screening, Health and disease, Genetic determinism, Genome ownership, Genetic discrimination, Genetic insurance, Genetic privacy, Genome patenting, Assisted reproduction.

Books Recommended

1. Almond, B. and Parker, M. (Eds.) (2003). Ethical Issues in New Genetics: Are Genes Us? Ashgate Publishing Ltd., England.
2. Beauchamp, T.L. and Childress, J.F. (1994). Principles of Biomedical Ethics. Oxford University Press, New York.
3. Bryant, A., Baggott la Velle, L. and Searle, J. (Eds.) (2002). Bioethics for Scientists. John Wiley and Sons Ltd., England.
4. Jecker, N.S., Jonsen, A.R. and Pearlman, R.A. (2010). Bioethics: An Introduction to History, Methods and Practice. Jones and Bartlett, New Delhi, 2nd ed.
5. Macer, D.R.J. (1998). Bioethics is Love of Life: Alternative Textbook. Eubios Ethics Institute Publications, Japan.
6. Macer, D.R.J. (Ed.) (2000). Ethical Challenges As We Approach the End of the Human Genome Project. Eubios Ethics Institute Publications, Japan.
7. Quackenbush, J. (2011). The Human Genome: The Book of Essential Knowledge. Imagine, New York.
8. Rosenberg, L.E. and Rosenberg, D.D. (2012). Human Genes and Genomes: Science, Health, Society. Academic Press, 1st ed.
9. Yashon, R.K. and Cummings, M.R. (2012). Human Genetics and Society. Oxford University Press.

Websites and Electronic References

http://www.nhgri.nih.gov/
http://www.ncbi.nlm.nih.gov/
http://www.hgmp.mrc.ac.uk./GenomeWeb/
http://www.ncbi.nlm.nih.gov/genemap99/
http://www.ncbi.nlm.nih.gov/CGAP
http://www.ncbi.nlm.nih.gov/HUGO
http://www.ebi.ac.uk/~sterk/genome-MOT/
http://www-fp.mcs.anl.gov/~gaasterland/genomes.html
http://www.jax.org/
http://www.biol.tsukuba.ac.jp/
http://www.nuffieldbioethics.org

Bridge Course in Human Genetics (Semester-IV)
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HGL-552 APPLIED HUMAN MOLECULAR GENETICS

Credits: 3-0-0

Unit-I

Principles and strategies in identifying disease gene, Genetic and physical mapping of human genome, Mapping and cloning of human disease genes, Direct and indirect DNA testing and its applications, DNA-based diagnosis of genetic diseases, Population screening.

Unit-II

Comparative genomic hybridization (CGH), DNA chips, DNA dragnets, Principles and application of denaturing high pressure liquid chromatography (dHPLC), Multiplex ligation-dependent probe amplification (MLPA), Mass spectrometry (MS), Tandem mass spectrometry (TMS), Next generation sequencing (NGS) and its applications.

Unit-III

Molecular genetics of retinoblastoma, Glaucoma, Marfan syndrome, DMD, Cystic fibrosis, Huntington's disease, Complex human diseases like NIDDM, Hypertension, Cardiovascular disorders, Obesity, Molecular genetics of mitochondrial disorders.

Books Recommended

1. Brown, T.A. (2002). Genomes. Oxford Scientific Publications, London.
2. Brown, T.A. (2005). Genetics: A Molecular Approach. Bios Scientific Publishers Ltd., Oxford, 3rd ed.
3. Brown, T.A. (2006). Genomes 3. Bios Scientific Publishers Ltd., Oxford, 3rd ed.
4. Brown, T.A. (2010). Gene Cloning and DNA Analysis: An Introduction. Blackwell Publishing Co., Oxford, 6th ed.
5. Gleck, B.R. and Pasternack, J.R. (2003). Molecular Biotechnology: Principles and Applications of Recombinant DNA. ASM Press, Washington.
6. Korf, B.R. (2007). Human Genetics and Genomics. Blackwell Scientific Publication, USA, 3rd ed.
7. Lewin, B. (2008). Genes-IX. Jones and Barlett Publishers, Inc., USA.
8. Schena, M. (2003). Microarray Analysis. Wiley-Liss, New York.
9. Strachan, T. and Read, A. (2010). Human Molecular Genetics. Garland Publishers, London, 4th ed.
10. Sudbery, P. and Sudbery, I. (2009). Human Molecular Genetics. Pearson Education, UK, 3rd ed.

Bridge Course in Human Genetics (Semester-IV)
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HGL-554 STRUCTURAL BIOLOGY AND PHARMACOGENOMICS

Credits: 3-0-0

Unit-I

Tertiary structures of DNA, Hoogstein base pairing, Triple helices, DNA toplogy, Supercoiling, Common structural motifs in RNA, General principles of protein structure, Primary structure, Peptide unit, Basic dipeptide unit, Introduction to Ramachandran plot, Secondary and tertiary structures, α-helix, β-sheet, Common three dimensional folds/motifs found in proteins and their mode of interaction with DNA.

Unit-II

Prediction of secondary structure from primary structure, Protein folding, Levinthal paradox, Physical nature of non-covalent interactions, Experimental methods for protein purification, Overview of methods used for determination of protein structure, Introduction to structural databases for protein studies.

Unit-III

Pharmacogenomics of obesity-related hypertension, Diabetes and other cardiovascular diseases, Basics of drug discovery, Role of functional genomics in new drug discovery and drugable genome, Genome-guided identification and validation of drug targets, Role of SNPs in drug discovery and development.

Books Recommended

1. Brandon, C. and Tooze, J. (1999) Introduction to Protein Structure. Garland Publishers, London.
2. Hartl, D.L. and Jones, E.W. (2005). Genetics: Analysis of Genes and Genomes. Jones and Barlett Publishers, Massachusetts.
3. Jain, K.K. (2001). Drug Discovery: Current Trends and Future Prospects. Urch Publishers, London.
4. Leach, A.R. (2001). Molecular Modelling. Principles and Applications. Addison Wesley Longman, Essex.
5. Nelson, D.L. and Cox, M.M. (2008). Lehninger's Principles of Biochemistry. W.H. Freeman and Co., New York.

HGP-472 CLINICAL GENETICS PRACTICAL

Credits: 0-0-2

Case studies, Designing proformas (Thalassemia, Primary ammenorhea, Mental retardation, Recurrent abortions, Prenatal screening questionnaire, Taking history, Preconceptional screening and counselling, Case management of selected genetic diseases (Congenital anomalies, Down syndrome, Achondroplasia, Marfan syndrome, Dysmorphology, and Clinical teratology). Genetic registers, importance and maintenance.

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HGP-552 APPLIED HUMAN MOLECULAR GENETICS PRACTICAL

Credits: 0-0-3

DNA isolation, Amplification of genomic DNA by PCR, Agarose gel electrophoresis of amplified products, Polyacrylamide gel electrophoresis (PAGE), PCR-SSCP analysis, Demonstration of RT-PCR, Biological database searches (MEDLINE, NCBI, ENSEMBL, UCSC, GDB, PDB, HGMD, RetNet).

Bridge Course in Human Genetics (Semester-IV)
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HGL-411 REGULATORY APPROACHES IN HUMAN GENETICS

Credits: 3-0-0

Unit-I

Historical overview for regulatory approaches for research in Human Genetics and protection of human subjects (Eugenics, Experiments during and after World War-II, The Tuskegee Trial, Body Doe Case); Oaths, Codes and Declarations — *Charak samitha*, The Hippocratic oath (old and modern versions), The Nuremberg code, Declaration of Geneva, Declaration of Helsinki.

Unit-II

Overview of reprogenetics, Consumer genetics, Personal genomics, Public health genetics, Community genetics, Regenerative medicine, Organ transplantation, Euthanasia, Intellectual property rights, International guidelines and regulations for conducting Human Genetics research — Universal Declaration on the Human Genome and Human Rights, International Ethical Guidelines for Biomedical Research involving Human Subjects (CIOMS-WHO).

Unit-III

The Indian guidelines for genetically-modified organisms (GMOs), Stem cell research, ART and for conducting biomedical research on human subjects. Relevant Indian Legislation — Drugs and Cosmetics Act, The Patents Act, The Medical Termination of Pregnancy Act, The Preconception and Prenatal Diagnostic Techniques Act, The Transplantation of Human Organs
Act.

Useful Links

1. Department of Biotechnology, Ministry of Science and Technology, Government of India, Ethical Policies on the Human Genome, Genetic Research and Services. (www.dbtindia.nic).
2. Indian Council of Medical Research, Consultative Document on Ethical Guidelines for Biomedical Research on Human Subjects. (www.icmr.nic.in).
3. Indian Council of Medical Research, Guidelines for preparing Standard Operating Procedures (SOP) for Institutional Ethics Committee for Human Research. (www.icmr.nic.in).
4. Indian Council of Medical Research, National Guidelines for Accreditation, Supervision and Regulation of ART Clinics in India. (www.icmr.nic.in).
5. Indian Council of Medical Research, Statement on Specific Principles on Human Genetics Research. (www.icmr.nic.in).
6. Indian Council of Medical Research and the Department of Biotechnology, Draft Guidelines for Stem Cell Research and Therapy. (www.icmr.nic.in; www.dbtindia.nic).
7. Indian Council of Medical Research and Ministry of Health and Family Welfare, Good Clinical Laboratory Practices. (www.icmr.nic.in).
8. Statement on Gene Therapy Research (adopted by the Human Genome Organization, HUGO). (www.hugo-international.org).
9. WIPO (World Intellectual Property Organization) (www.wipo.int).
10. World Medical Association Declaration of Helsinki (Ethical Principles for Medical Research Involving Human Subjects) (www.cioms.ch; www.who.int).
11. WTO. Agreement on Trade-Related Aspects of Intellectual Property Rights (TRIPS) is part of the agreements of the World Trade Organization (www.wto.org).

Bridge Course in Human Genetics (Semester-IV)
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HGL-412 BIOLOGICAL DEMOGRAPHY

Credits: 3-0-0

Unit-I

Some basic concepts of demography — Family, Marriage, Reproductive span, Fertility, Fecundity, Sterility, Morbidity, Mortality, Stillbirth, Live-birth, Foetal death, Abortion, Child death, Birth order, Parity, Cohort, Growth rate, and Migration.

Unit-II

Sources of demographic data, Factors affecting fertility, mortality, migration, sex ratio, Population structure, Density of population, Determinants of population growth.

Unit-III

Principles of epidemiology, Epidemiology of communicable and non-communicable diseases, Concept of health and diseases, Family planning programme in India, Urbanization, Factors responsible for urban-rural population distribution.

Books Recommended

1. Hinde, A. (1998). Demographic Methods. Oxford University Press, Inc., New York.
2. Krishna Reddy, M.M. (1996). An Introduction to Demographic Behaviour in India. Kanishka Publishers, Distributors, New Delhi.
3. Krishnan, P., Tuan, C.-H. and Mahadevan, K. (1992). Readings in Population Research. B.R. Publishing Corpn., Delhi.
4. Majumdar, P.K. (2010). Fundamentals of Demography. Rawant Publications, Jaipur.
5. Raj, H. (2004). Fundamentals of Demography. Surjeet Publications, Delhi.
6. Shrivastava, O.S. (1994). Demography and Population Studies. Vikas Publishing House Pvt. Ltd., New Delhi.
7. Srinivasan, K. (1998). Basic Demographic Techniques and Applications. Sage Publications, New Delhi.