

## HUMAN GENETICS EMPIRICAL TRACK COURSES

### REQUIRED COURSES FIRST YEAR CURRICULUM

**TWO [2] REQUIRED COURSES AND TWO [2] CORE ELECTIVE COURSE AND FOUR [4] ELECTIVE COURSES PLUS TWO [2] ROTATIONS**

#### TWO [2] REQUIRED COURSES\*

**HGEN 47000 Human Genetics I. Ober, Nobrega, Waggoner.** This course covers classical and modern approaches to studying cytogenetic, Mendelian, and complex human diseases. Topics include chromosome biology, single gene and complex diseases, non-Mendelian inheritance, cancer genetics, human population genetics, and genomics. The format includes lectures and student presentations. *Autumn.*

**HGEN 46900 Human Variation and Disease. Berg, Raghavan.** This course focuses on principles of population and evolutionary genetics and complex trait mapping as they apply to humans. It will include the discussion of genetic variation and disease mapping data. *Spring.*

**\* A statistics course (e.g. HGEN 47400) of appropriate level given the student's background is also strongly recommended and will be chosen in consultation with the Curriculum/Student Affairs Committee.**

#### TWO [2] CORE ELECTIVE COURSES

**HGEN 47100 Intro Statistical Genetics. He, Im.** This course focuses on genetic models for complex human disorders and quantitative traits. Topics covered also include linkage and linkage disequilibrium mapping genetic models for complex traits, and the explicit and implicit assumptions of such models. *Winter.*

#### OR

**HGEN 31100 Evolution of Biological Molecules. Thornton, Drummond.** The course connects evolutionary changes imprinted in genes and genomes with the structure, function and behavior of the encoded protein and RNA molecules. Central themes are the mechanisms and dynamics by which molecular structure and function evolve, how protein/ RNA architecture shapes evolutionary trajectories, and how patterns in present-day sequence can be interpreted to reveal the interplay data of evolutionary history and molecular properties. Core concepts in macromolecule biochemistry (folding and stability of proteins and RNA, structure-function relationships, kinetics, catalysis) and molecular evolution (selection, mutation, drift, epistasis, effective population size, phylogenetics) will be taught, and the interplay between them explored. *Winter.*

#### OR

**HGEN 48600 Fundamentals of Computational Biology: Models and Inference. Novembre, Stephens.** Covers key principles in probability and statistics that are used to model and understand biological data. There will be a strong emphasis on stochastic processes and inference in complex hierarchical statistical models. Topics will vary but the typical content would include: Likelihood-based and Bayesian inference, Poisson processes, Markov models, Hidden Markov models, Gaussian Processes, Brownian motion, Birth-death processes, the Coalescent, Graphical models, Markov processes on trees and graphs, Markov Chain Monte Carlo. PQ: STAT 244 or equivalent. *Winter.*

#### OR

**ECEV 35600 Population Genetics I., Steinrücken.** Examines the basic theoretical principles of population genetics, and their application to the study of variation and evolution in natural populations. Topics include selection, mutation, random genetic drift, quantitative genetics, molecular evolution and variation, the evolution of selfish genetic systems, and human evolution. *Spring.*

#### OR

**HGEN 47300 Genomics and Systems Biology. Gilad.** This lecture course explores technologies for high-throughput collection of genomic-scale data, including sequencing, genotyping, gene expression profiling, and

assays of copy number variation, protein expression and protein-protein interaction. In addition, the course will cover study design and statistical analysis of large data sets, as well as how data from different sources can be used to understand regulatory networks, i.e., systems. Statistical tools that will be introduced include linear models, likelihood-based inference, supervised and unsupervised learning techniques, methods for assessing quality of data, hidden Markov models, and controlling for false discovery rates in large data sets. Readings will be drawn from the primary literature. Evaluation will be based primarily on problem sets. *Spring*.

**OR**

**MGCB 31300 Molecular Biology II. Ruthenburg, Staley, Lee.** The content of this course covers the mechanisms and regulation of eukaryotic gene expression at the transcriptional and post-transcriptional levels. Our goal is to explore research frontiers and evolving methodologies. Rather than focusing on the elemental aspects of a topic, the lectures and discussions highlight the most significant recent developments, their implications and future directions. *Spring*.

**OR**

**MGCB 31400 Genetic Analysis of Model Organisms. Bishop.** Fundamental principles of genetics discussed in the context of current approaches to mapping and functional characterization of genes. The relative strengths and weaknesses of leading model organisms are emphasized via problem-solving and critical reading of original literature. *Autumn*.

**OR**

**DVBI 36400 Developmental Mechanisms. Ferguson, Fehon.** This course provides an overview of the fundamental questions of developmental biology, with particular emphasis on the genetic, molecular and cell biological experiments that have been employed to reach mechanistic answers to these questions. Topics covered will include formation of the primary body axes, the role of local signaling interactions in regulating cell fate and proliferation, the cellular basis of morphogenesis, and stem cells. *Winter*.

## **ADDITIONAL REQUIRED COURSES**

**HGEN 31900 Introduction to Research.** Lectures on current research by departmental faculty and other invited speakers. A required course for all first-year graduate students in Human Genetics (“Allstars”). *Autumn*.

**HGEN 40300 Non-Thesis Research.** Laboratory rotations, and all research prior to passing the Qualifying Examination. *Autumn, Winter, Spring, Summer*.

**BSDG 55100 Responsible, rigorous, and reproducible conduct of research: R3CR.** Required of all BSD first-year doctoral students. The course is designed to stimulate thinking and facilitate discussion about the purpose and necessity of ethical conduct with respect to scientific and academic practices; to create personal awareness of the ethical dilemmas and choices that may be encountered in the course of a career in the sciences; to increase awareness and understanding of the importance of reproducible, rigorous, and transparent research; and to provide practical information regarding policies and procedures related to conduct in the Division of Biological Sciences at the University of Chicago. *Winter*.

## **PLUS FOUR [4] ELECTIVE COURSES**

### **ADDITIONAL ELECTIVE COURSES TO CHOOSE FROM TO FULFILL 4 COURSES**

#### **HUMAN GENETICS**

**HGEN 39900 Readings in Human Genetics. HG Faculty.** A course designed by students and faculty member. All reading courses must be approved by the Curriculum Committee prior to registration. *Autumn, Winter, Spring, Summer*.

**HGEN 47400 Introduction to Probability and Statistics for Geneticists. Abney.** This course is an introduction to basic probability theory and statistical methods useful for people who intend to do research in genetics or a similar scientific field. Topics include random variable and probability distributions, descriptive statistics, hypothesis testing and parameter estimation. Problem sets and tests will include both solving problems analytically and analysis of data using the R statistical computing environment. *Autumn*.

**HGEN 48800 Fundamentals of Computational Biology: Algorithms and Applications. He, Chen.** This course will cover principles of data structure and algorithms, with emphasis on algorithms that have broad applications in computational biology. The specific topics may include dynamic programming, algorithms for graphs, numerical optimization, finite-difference, schemes, matrix operations/factor analysis, and data management (e.g. SQL, HDF5). We will also discuss some applications of these algorithms (as well as commonly used statistical techniques) in genomics and systems biology, including genome assembly, variant calling, transcriptome inference, and so on. *Spring*.

**HGEN 36400 Molecular Phylogenetics. Thornton.** While evolution by natural selection is an elegantly simple phenomenon, modern research in evolutionary biology contains a variety of controversial, and sometimes confusing, topics. In this course, we will explore, as a group, a select list of controversial or confusing topics in evolutionary biology through a mix of student-led presentations and discussion of the primary literature. Each student will also write a review paper about his or her selected topic. *Autumn*.

### **BIOCHEMISTRY AND MOLECULAR BIOLOGY**

**BCMB 30400 Protein Fundamentals. Piccirilli, Arac-Ozkan, Ozkan,** The course covers the physical chemical phenomena that define protein structure and function. Topics include: three-dimensional structures of proteins; the principles of protein folding, molecular motion and molecular recognition; protein evolution, design and engineering; enzyme catalysis; regulation of protein function; proteomics and systems biology. Undergraduates are highly recommended to take BIOS 20200 (Introduction to Biochemistry) or equivalent before taking this course. *Autumn*.

### **DEVELOPMENTAL BIOLOGY**

**DVBI 35600 Vertebrate Developmental Genetics. Prince, Kratsios.** This advanced-level course combines lectures, student presentations, and discussion sections. It covers major topics in the developmental biology of vertebrate embryos (e.g., formation of the germ line, gastrulation, segmentation, nervous system development, limb patterning, organogenesis). The course makes extensive use of the current primary literature and emphasizes experimental approaches including embryology, genetics, and molecular genetics. *Winter*.

### **ECOLOGY AND EVOLUTION**

**ECEV 35901 Genomic Evolution I. Long.** Canalization, a unifying biological principle first enunciated by Conrad Waddington in 1942, is an idea that has had tremendous intellectual influence on developmental biology, evolutionary biology, and mathematics. In this course we will explore canalization in all three contexts through extensive reading and discussion of both the classic and modern primary literature. We intend this exploration to raise new research problems which can be evaluated for further understanding. We encourage participants to present new ideas in this area for comment and discussion. *Autumn*.

### **MOLECULAR GENETICS AND CELL BIOLOGY**

**MGCB 31200 Molecular Biology I. Rothman-Denes, Bishop.** Nucleic acid structure and DNA topology; methodology; nucleic-acid protein interactions; mechanisms and regulation of transcription in eubacteria, and of replication in eubacteria and eukaryotes; mechanisms of genome and plasmid segregation in eubacteria. *Winter*.

**MGCB 32000 Quantitative Analysis of Biological Dynamics, Munro, Rust.** T The basic focus of the course will be quantitative approaches to understanding organization and dynamics at the molecular, subcellular and cellular levels, and will rest on three pillars - modern imaging and image analysis, quantitative analysis and presentation of data, mathematical modeling and computer simulations. *Spring*.

**MGCB 31600 Cell Biology I. Glick, Turkewitz.** Eukaryotic protein traffic and related topics, including molecular motors and cytoskeletal dynamics, organelle architecture and biogenesis, protein translocation and sorting, compartmentalization in the secretory pathway, endocytosis and exocytosis, and mechanisms and regulation of membrane fusion. *Autumn*.

**MGCB 31700 Cell Biology II. Glotzer, Kovar.** This course covers the mechanisms with which cells execute

fundamental behaviors. Topics include signal transduction, cell cycle progression, cell growth, cell death, cancer biology, cytoskeletal polymers and motors, cell motility, cytoskeletal diseases, and cell polarity. Each lecture will conclude with a dissection of primary literature with input from the students. Students will write and present a short research proposal. *Winter*.

**MGCB 32000 Quantitative Analysis of Biological Dynamics, Munro, Rust.** The basic focus of the course will be quantitative approaches to understanding organization and dynamics at the molecular, subcellular and cellular levels, and will rest on three pillars - modern imaging and image analysis, quantitative analysis and presentation of data, mathematical modeling and computer simulations. *Spring*.

## **NEUROBIOLOGY**

**NURB 33400 Genetic Approaches in Neurobiology. Zhuang.** . This course is more technique oriented. The goal is to give a good coverage of different genetic approaches as well as different aspects of neurobiology. Topics are organized by genetic approaches as the following: 1) Transgenic. 2) Gene targeting. 3) Gene replacement. 4) Conditional knockout. 5) Genetic and optical control of neural activity. 6) Transgenic facilitated imaging. 7) Forward genetics and genetic screening. The selection of a variety of papers throughout the course aims to cover different neural pathways, neurotransmitters, receptor/channel types, signaling pathways, and functional implications (learning, memory, addiction, development etc). Specific emphasis will be on the integration of molecular, cellular and systems level approaches in understanding behavior. Lecture time will be devoted to the genetic approaches. Students will present and discuss papers. We will have 2-3 papers each lecture. *Spring*.

## **STATISTICS**

**STAT 24300 Numerical Linear Algebra.** This course is devoted to the basic theory of linear algebra and its significant applications in scientific computing. The objective is to provide a working knowledge and hands-on experience of the subject suitable for graduate level work in statistics, econometrics, quantum mechanics, and numerical methods in scientific computing. Topics include Gaussian elimination, vector spaces, linear transformations and associated fundamental subspaces, orthogonality and projections, eigenvectors and eigenvalues, diagonalization of real symmetric and complex Hermitian matrices, the spectral theorem, and matrix decompositions (QR, Cholesky and Singular Value Decompositions). Systematic methods applicable in high dimensions and techniques commonly used in scientific computing are emphasized. Students enrolled in the graduate level STAT 30750 will have additional work in assignments, exams, and projects including applications of matrix algebra in statistics and numerical computations implemented in Matlab or R. *Autumn*.

**STAT 24400 Statistical Theory and Methodology I.** This sequence is a systematic introduction to the principles and techniques of statistics, as well as to practical considerations in the analysis of data, with emphasis on the analysis of experimental data. This course is the first quarter of a two-quarter systematic introduction to the principles and techniques of statistics, as well as to practical considerations in the analysis of data, with emphasis on the analysis of experimental data. This course covers tools from probability and the elements of statistical theory. Topics include the definitions of probability and random variables, binomial and other discrete probability distributions, normal and other continuous probability distributions, joint probability distributions and the transformation of random variables, principles of inference (including Bayesian inference), maximum likelihood estimation, hypothesis testing and confidence intervals, likelihood ratio tests, multinomial distributions, and chi-square tests. Examples are drawn from the social, physical, and biological sciences. The coverage of topics in probability is limited and brief, so students who have taken a course in probability find reinforcement rather than redundancy. Students who have already taken STAT 25100 may choose to take STAT 24410 (if offered) instead of STAT 24400. Students taking either STAT 24400 or STAT 24410 will have appropriate preparation for STAT 24500. *Autumn, Winter*.

**STAT 24500 Statistical Theory/Method-2.** This course is the second quarter of a two-quarter systematic introduction to the principles and techniques of statistics, as well as to practical considerations in the analysis of data, with emphasis on the analysis of experimental data. This course continues from either STAT 24400 or STAT 24410 and covers statistical methodology, including the analysis of variance, regression, correlation, and some multivariate analysis. Some principles of data analysis are introduced, and an attempt is made to present the analysis of variance and regression in a unified framework. Statistical software is used. *Winter*.

**STAT 35500 Statistical Genetics.** This is an advanced course in statistical genetics. Prerequisites are Human Genetics 47100 and Statistics 24400 and 24500. Students who do not meet the prerequisites may enroll on a P/NP basis with consent of the instructor. Prerequisites are either Human Genetics 47100 or statistics preparation at the level of Statistics 24400 and 24500. This is a discussion course and student presentations will be required. Topics vary and may include, but are not limited to, statistical problems in linkage mapping, association mapping, map construction, and genetic models for complex traits. *Spring.*

**A complete list of courses is available on the [Human Genetics Graduate Program](#) website:**