Department of Human Genetics

Chair: Carole Ober

Professors

• Habibul Ahsan, Public Health Sciences
• Graeme Bell, Biochemistry and Molecular Biology
• Soma Das
• Anna Di Rienzo
• Elliot Gershon, Psychiatry and Behavioral Neuroscience
• Yoav Gilad
• T. Conrad Gilliam
• Lucy Godley, Medicine
• Bruce T. Lahn
• Michelle M. Le Beau, Medicine
• Natalia Maltsev
• Mary Sara McPeek, Statistics
• Dan L. Nicolae, Statistics
• Marcelo Nobrega
• John Novembre
• Carole Ober
• Andrey Rzhetsky, Medicine
• Matthew Stephens
• Joseph Thornton, Ecology and Evolution
• Olufunmilayo Olopade, Medicine
• Darrel J. Waggoner
• Kevin White

Associate Professors

• Mark Abney
• Ivan Moskowitz, Pediatrics

Assistant Professors

• Mengjie Chen, Medicine
• D. Allan Drummond, Biochemistry and Molecular Biology
• Daniela Del Gaudio
• Xin He
• Hae Kyung Im, Medicine
• Yang Li, Medicine
• Zejuan Li, Medicine
• Vincent J. Lynch
• Brandon Pierce, Public Health Sciences
• Lixing Yang, Medicine
• Xiaochang Zhang

The Department of Human Genetics offers training in a number of fields of human genetics such as human disease, classical genetics, complex trait genetics, population and evolutionary genetics, cytogenetics, neurogenetics, systems biology, pharmacogenetics and developmental human genetics. This coursework is intended for graduate students who plan to pursue research careers and teaching in the emerging areas of modern biology, and is intended for medical students, advanced undergraduate and graduate students in other programs. The Ph.D. program places great emphasis on sound preparation in human genetics, statistical genetics, and molecular biology.

The Degree of Doctor of Philosophy

A Ph.D. candidate must fulfill certain formal coursework requirements, pass one preliminary and one qualifying examination, and present a satisfactory dissertation describing the results of original research.
The department expects a knowledge of and proficiency in human genetics. This requirement will normally be met by fulfilling the formal coursework described here, but degree programs are flexible. Courses taken at other institutions, in other programs, or as part of the Pritzker School of Medicine curriculum may substitute for HG courses with approval of the Curriculum Committee. To fulfill the requirements for a Ph.D., nine graded courses are required. In the Department of Human Genetics, a student must take the following three required courses:

<table>
<thead>
<tr>
<th>Course Code</th>
<th>Course Title</th>
<th>Units</th>
</tr>
</thead>
<tbody>
<tr>
<td>MGCB 31400</td>
<td>Genetic Analysis of Model Organisms</td>
<td>100</td>
</tr>
<tr>
<td>HGEN 47000</td>
<td>Human Genetics-1</td>
<td>100</td>
</tr>
<tr>
<td>HGEN 46900</td>
<td>Human Variation and Disease</td>
<td>100</td>
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One of the following:

- HGEN 47100 Intro Statistical Genetics 100
- DVBI 35600 Vertebrate Development 100
- MGCB 31300 Molecular Biology-II 100
- ECEV 35600 Principles of Population Genetics-1 100

The remaining 4 courses are electives chosen from a host of courses in the Biological Sciences Division and Statistics Department. All courses are to be approved by an assigned academic advisor. These courses and many more are designed to develop greater proficiency in your particular sub discipline.

A student is also required to do two laboratory rotations before selecting an advisor and laboratory in which to pursue a Ph.D. dissertation. These rotations will be graded and together will be equivalent to one elective. All students are required to serve as a teaching assistant for two quarters.

During the second year, students select a thesis advisor and begin laboratory research. To complete the Ph.D. degree, they must prepare, under the general direction of an appointed doctoral committee, a dissertation based upon their original research. A public seminar describing the results of the dissertation research must be presented and the dissertation must be successfully defended before the doctoral committee.

Application

For information about applying to our graduate program, please visit:https://apply-bsd.uchicago.edu/apply/.

Human Genetics Courses

**HGEN 30100. Appl of Scientific Advncmt to Disease Detection & Management. 75 Units.**

**HGEN 30400. Protein Fundamentals. 100 Units.**

The course covers the physical/chemical phenomena that define protein structure and function. Topics include: the principles of protein folding, molecular motion and molecular recognition; protein evolution, design and engineering; enzyme catalysis; regulation of protein function and molecular machines; proteomics and systems biology. Workshop on X-ray Crystallography: The workshop is an addendum to Protein Fundamentals and is required for all BCMB students. This one week workshop will provide students with an intensive introduction to protein structure determination by x-ray crystallography. In addition to lectures, an extensive laboratory component will give students the opportunity to carry out protein crystallization, data collection (at Argonne), structure determination, refinement, model building and validation.

**Instructor(s):** E. Ozkan, D. Arac

**Terms Offered:** Autumn

**Equivalent Course(s):** MGCB 30400, BCMB 30400

**HGEN 31100. Evolution of Biological Molecules. 100 Units.**

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.

**Instructor(s):** A. Drummond, J. Thornton

**Terms Offered:** Winter

**Prerequisite(s):** Comfort with basic computer programming (course will use Python and R); undergraduate biology, chemistry, calculus, and introductory statistics.

**Equivalent Course(s):** BCMB 31100, ECEV 31100

**HGEN 31400. Genetic Analysis of Model Organisms. 100 Units.**

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.

**Equivalent Course(s):** MGCB 31400, DVBI 31400, BCMB 31400
HGEN 31600. Cell Biology I. 100 Units.
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.
Instructor(s): A. Turkewitz, B. Glick
Terms Offered: Autumn
Equivalent Course(s): BCMB 31600, DVBI 31600, MGCB 31600

HGEN 31800. Current Topics in Human Genetics. 50 Units.
This course will expose students to current research topics in Human Genetics through the Seminar Series. This is a required ½ credit course for all Human Genetics students and will be graded Pass/Fail. (Autumn, Winter, Spring)
Instructor(s): Ann Di Rienzo
Terms Offered: Autumn Spring Winter

HGEN 31900. Introduction to Research. 100 Units.
Lectures on current research by departmental faculty and other invited speakers. A required course for all first-year graduate students.
Instructor(s): Staff
Terms Offered: Autumn Winter
Equivalent Course(s): DVBI 31900, BCMB 31900, MGCB 31900, GENE 31900

HGEN 36400. Molecular Phylogenetics. 100 Units.
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.
Instructor(s): J. Thornton, A. Drummond
Terms Offered: Spring, offered in alternate (even) years
Note(s): not offered in 2018-19
Equivalent Course(s): ORGB 36400, ECEV 36400

HGEN 39500. Historical and Conceptual Foundations of DevoEvo. 100 Units.
The goal of this course is to explore the historical and conceptual foundations of Developmental Evolution (DevoEvo) through readings and group discussions of historical and philosophical literature on evolutionary and developmental biology, in particular the role developmental biology played in the formulation of evolutionary theory and its subsequent banishment from the Modern Synthesis. The course begins with a review of nineteenth-century scientific and evolutionary thought, including an examination of competing theories of evolution (Theistic Evolutionism, Lamarckism, Orthogenesis, and Mutation Theory) and their contribution (or lack thereof) to modern evolutionary biology. We then explore how (and why) developmental biology was excluded from the formulation of the Synthesis and Neo-Darwinian thought, and examine the source of continued conflicts between Neo-Darwinism and DevoEvo. The course concludes with a discussion of what (if anything) DevoEvo can contribute to evolutionary theory that other research programs cannot (for example, what kinds of phenomena do developmental mechanisms contribute more to the explanation of evolutionary processes than population genetic mechanisms?).
Instructor(s): V. Lynch
Terms Offered: Winter
Prerequisite(s): For Biology Majors: Three quarters of a Biological Sciences Fundamentals sequence including BIOS 20187 or BIOS 20235.
Equivalent Course(s): ORGB 39500, BIOS 21418

HGEN 39900. Topics: Human Genetics. 300.00 Units.

HGEN 40300. Non-Thesis Rsch: Human Genetics. 300.00 Units.
Research conducted by graduate students prior to the qualifying exam.

HGEN 40400. Thesis Research. 300.00 Units.
Dissertation Research conducted by graduate students.
Instructor(s): A DiRienzo
Terms Offered: Autumn Spring Summer Winter

HGEN 46900. Human Variation and Disease. 100 Units.
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.

HGEN 47000. Human Genetics-1. 100 Units.
This course covers classical and modern approaches to studying cytogenic, Mendelian, and complex diseases. Topics include chromosome biology, single gene and complex disease, non-Mendelian inheritance, cancer genetics, human population genetics, and genomics. The format includes lectures and student presentations.
Instructor(s): C. Ober, M. Nobrega, D. Waggoner
HGEN 47300. Genomics and Systems Biology. 100 Units.
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 statistic 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.
Instructor(s): Y. Gilad Terms Offered: Spring
Prerequisite(s): Three quarters of a Biological Sciences Fundamentals sequence including BIOS 20187 or BIOS 20235 and STAT 23400 or BIOS 26210 and BIOS 26211
Equivalent Course(s): BIOS 28407, IMMU 47300, BPHS 47300, CABI 47300

HGEN 47400. Introduction to Probability and Statistics for Geneticists. 100 Units.
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.
Instructor(s): M. Abney Terms Offered: Autumn

HGEN 48600. Fundamentals of Computational Biology: Models and Inference. 100 Units.
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.
Instructor(s): J. Novembre, M. Stephens Terms Offered: Winter
Prerequisite(s): STAT 244
Equivalent Course(s): STAT 35450

HGEN 48800. Fundamentals of Computational Biology: Algorithms and Applications. 100 Units.
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.
Instructor(s): Xin He, Mengjie Chen Terms Offered: Spring
Equivalent Course(s): STAT 35460

HGEN 70000. Advanced Study: Human Genetics. 300.00 Units.
Advanced Study: Human Genetics
Font Notice

This document should contain certain fonts with restrictive licenses. For this draft, substitutions were made using less legally
restrictive fonts. Specifically:

Times was used instead of Trajan.
Times was used instead of Palatino.

The editor may contact Leepfrog for a draft with the correct fonts in place.