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
- Richard Hudson, Ecology and Evolution
- Bruce T. Lahn
- Michelle M. Le Beau, Medicine
- Natalia Maltsev
- Mary Sara McPeek, Statistics
- Dan L. Nicolae, Statistics
- Carole Ober
- Andrey Rzhetsky, Medicine
- Matthew Stephens
- Joseph Thornton, Ecology and Evolution
- Olufunmilayo Olopade, Medicine
- Darrel J. Waggoner
- Kevin White
- Huntington F. Willard

Associate Professors
- Mark Abney
- Ivan Moskowitz, Pediatrics
- Marcelo Nobrega
- John Novembre
- Abraham Palmer

Assistant Professors
- D. Allan Drummond, Biochemistry and Molecular Biology
- Daniela Del Gaudio
- Xin He
- Zejuan Li, Medicine
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>
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<tbody>
<tr>
<td>MGCB 31400</td>
<td>Genetic Analysis of Model Organisms</td>
<td>100</td>
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<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:

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<tr>
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<tbody>
<tr>
<td>HGEN 47100</td>
<td>Intro Statistical Genetics</td>
<td>100</td>
</tr>
<tr>
<td>MGCB 31500</td>
<td>Genetic Mechanisms</td>
<td>100</td>
</tr>
<tr>
<td>DVBI 35600</td>
<td>Vertebrate Development</td>
<td>100</td>
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<tr>
<td>MGCB 31300</td>
<td>Molecular Biology-II</td>
<td>100</td>
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<tr>
<td>ECEV 35600</td>
<td>Principles of Population Genetics-1</td>
<td>100</td>
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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 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): R. Keenan, S. Koide, Kossiakoff 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): ECEV 31100, BCMB 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.
Instructor(s): A. Palmer, D. Bishop, E. Ferguson, J. Malamy Terms Offered: Autumn
Equivalent Course(s): DVBI 31400, BCMB 31400, MGCB 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. Quarter

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): MGCB 31900, BCMB 31900, DVBI 31900, GENE 31900

HGEN 40400. Thesis Research. Units.
No description available.
Instructor(s): A DiRienzo Terms Offered: Autumn, Winter, Spring, Summer

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. Spring

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. Autumn
Instructor(s): C. Ober, M. Nobrega, D. Waggoner

HGEN 47100. Intro Statistical Genetics. 100 Units.
This course focuses on genetic models for complex human disorders and quantitative traits. Topics covered also include linkage and linkage disequilibrium mapping and genetic models for complex traits, and the explicit and implicit assumptions of such models.
Instructor(s): J. Novembre, B. Pierce, L. Chen Terms Offered: Winter
Equivalent Course(s): BIOS 21216
HGEN 47300. Genomics and Systems Biology. 100 Units.
This lecture course explores the technologies that enable high-throughput collection of genomic-scale data, including sequencing, genotyping, gene expression profiling, assays of copy number variation, protein expression and protein-protein interaction. We also 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 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 are drawn from the primary literature.
Instructor(s): Y. Gilad Terms Offered: Spring
Prerequisite(s): STAT 23400 or Statistics in the Biomath Sequence
Equivalent Course(s): IMMU 47300, BIOS 28407

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, A. Skol Terms Offered: Autumn