Department of Human Genetics

Chair, Department of Human Genetics: Carole Ober
Human Genetics PhD Program Chair, François Spitz

Professors
- Habibul Ahsan, Public Health Sciences
- Graeme Bell, Medicine, Endocrinology
- Soma Das, Human Genetics
- Daniela Del Gaudio, Human Genetics
- Anna Di Rienzo, Human Genetics
- Yoav Gilad, Medicine, Genetic Medicine
- T. Conrad Gilliam, Human Genetics
- Lucy Godley, Medicine, Hematology/Oncology
- Bruce T. Lahn, Human Genetics
- Michelle Le Beau, Medicine
- Mary Sara McPeek, Statistics
- Ivan Moskowitz, Pediatrics, Cardiology
- Dan L. Nicolae, Statistics
- Marcelo Nobrega, Human Genetics
- John Novembre, Human Genetics
- Carole Ober, Human Genetics
- Olufunmilayo Olopade, Medicine
- Andrey Rzhetsky, Medicine, Computational Biomedicine and Biomedical Data Science
- François Spitz, Human Genetics
- Matthew Stephens, Human Genetics
- Joseph Thornton, Ecology and Evolution
- Olufunmilayo Olopade, Medicine, Hematology/Oncology

Clinical Professors
- Darrel J. Waggoner, Human Genetics

Associate Professors
- Luis Barreiro, Medicine, Genetic Medicine
- D. Allan Drummond, Biochemistry and Molecular Biology
- Brandon Pierce, Public Health Services

Assistant Professors
- Jeremy Berg, Human Genetics
- Mengjie Chen, Medicine, Genetic Medicine
- Andrew Dahl, Medicine, Genetic Medicine
- Xin He, Human Genetics
- Hae Kyung Im, Medicine, Genetics Medicine
- Yang Li, Medicine, Genetic Medicine
- Maanasa Raghavan, Human Genetics
- Matthias Steinrücken, Ecology & Evolution
- Lixing Yang, Ben May Department for Cancer Research
- Xiaochang Zhang, Human Genetics

Research Associate Professors
- Mark Abney, Human Genetics
- Sebastian Pott, Human Genetics
- Emma Thompson, Human Genetics
The graduate program in Human Genetics (https://hgen.uchicago.edu/) (HG) is the home within the Division of Biological Sciences for the study of basic principles of genetics and genomics as applied to human phenotypes. We provide broad training in experimental and computational genetics and genomics, statistical and population genetics, bioinformatics, and clinical genetics. A common theme throughout our research is the application of basic genetic principles and strategies to the study of disease mechanism, disease susceptibility, and the genetic architecture of complex traits. Within this framework, the goals of our program are to:

- Educate students broadly in the basic concepts and practices of human genetics and the disciplines of genomics, population genetics, bioinformatics and systems biology.
- Introduce students to the growing importance of evolutionary concepts and insights to the study of human disease and genetic variation.
- Prepare students for a new world of genetic medicine that will follow basic discoveries in our understanding of multifactorial inheritance.

Curriculum and Timeline - First Year (https://hgen.uchicago.edu/program/)

**Formal Coursework: Choice of Two Tracks:** Empirical Track (https://hgen.uchicago.edu/page/human-genetics-empirical-track-coursework/) or Computational Track (https://hgen.uchicago.edu/page/human-genetics-computational-track-coursework/)

To obtain a Ph.D. in the Division of Biological Sciences, nine graded courses are required as detailed below.


Training under the Empirical Track is focused on experimental techniques.

**Three [3] Required Courses:** HGEN 47400 Human Genetics I AND HGEN 46900 Human Variation and Disease

**Plus One [1] of the Following Core Elective Courses:** HGEN 47100 Intro Statistical Genetics OR HGEN 31100 Evolution of Biological Molecules OR HGEN 48600 Fundamentals of Computational Biology: Models and Inference OR ECEV 35600 Principles of Population Genetics I OR HGEN 47300 Genomics and Systems Biology OR MGCB 31300 Molecular Biology II OR MGCB 31400 Genetic Analysis of Model Organisms OR DVBI 36400 Developmental Mechanisms

**Plus Four [4] Elective Courses:** The electives can be selected according to the student's interests and the availability of courses


The Computational Track (https://hgen.uchicago.edu/page/human-genetics-computational-track-coursework/) curriculum trains students to address fundamental biological questions and to master the three skill sets that are essential to computational genomics research: probabilistic modeling, statistical inference, and computational algorithms & data structures. This curriculum is also unique in its focus on communication skills, both in terms of writing and speaking. This emphasis emerges from a perspective that computational biologists need to clearly explain complex algorithms and results in order both to effectively share their research products and to collaborate with diversely trained colleagues.

For additional information please click here to view the Doctoral Training in Computational Genomics (http://compbio.uchicago.edu/) website.

AND Three [3] Core Elective Courses Chosen from the Following List: Human Genetics I OR Genetic Analysis of Model Organisms OR Introductory Statistical Genetics OR Principles of Population Genetics I OR Evolution of Biological Molecules OR Biophysics of Biomolecules OR Human Variation and Disease OR Genomics and Systems Biology OR Quantitative Analysis of Biological Dynamics


Rotations

Students undertake short research projects in at least two different laboratories before beginning their dissertation research. The purpose of the rotation is to expose the student to different research environments, broaden his/her acquaintance with useful laboratory techniques, and introduce him/her to the conceptual framework of experimental design. The distribution of course offerings makes it difficult for students to undertake rotations in Autumn Quarter of the first academic year. Therefore, rotations are performed in the winter or spring and summer quarters. The winter and spring rotations last 10 weeks to coincide with the academic quarter. The summer rotation lasts 5 weeks, when the student is able to devote full-time to research. Students wishing to do a third rotation may do so during the second half of Summer Quarter.

Curriculum and Timeline - Second Year

Students spend the second year developing a research project and preparing a written proposal of dissertation research. This proposal is defended before a qualifying examination committee at the end of the academic year. Students satisfy any remaining course requirements and complete at least one of the two required Teaching Assistantships during this year.

Curriculum and Timeline - Advanced Years

After the qualifying exam, the student performs full-time thesis research while continuing to participate in program events such as seminars, journal clubs, etc. Students are welcome to audit courses in which they have an interest. Finally, in the final year of the program the student writes a dissertation describing his/her research, presents the work in a public seminar and defends it before his/her faculty examining committee. Please view the Human Genetics Handbook for a full description of the academic program and the courses available to our students.

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: 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.
Instructor(s): E. Ozkan, J. Piccirilli, 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): 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.  
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  
Equivalent Course(s): DVBI 31600, MGCB 31600, BCMB 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): MGCB 31900, DVBI 31900, BCMB 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  
she 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 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.  
Instructor(s): A DiRienzo

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-I. 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): BPHS 47300, IMMU 47300, BIOS 28407, 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