The guidelines in this handbook are official policies of the Department of Human Genetics. Students and faculty of the department are expected to follow these policies. Students with questions not answered by this handbook are encouraged to contact the Graduate Affairs Administrator or the Chair of the Curriculum/Student Affairs Committee.
ACADEMIC AND HUMAN GENETICS CALENDAR

Autumn Quarter 2021

September 6    Labor Day
September 13-17 Prelim Examinations Week
September 20-24 Orientation week
September 27   Autumn Quarter classes begin
October 20     Dissertation Office Draft Deadline for Autumn 2021 graduation
November 5     Winter 2022 Quarter rotation decisions due
November 5     Final Dissertation Submission Deadline Autumn 2021 graduation
November 22-26 Thanksgiving Break
December 11    Autumn 2021 Quarter ends

Winter Quarter 2022

January 3      Winter Quarter classes begin
January 17     Martin Luther King, Jr. Day observance
January 26     Dissertation Office Draft Deadline for Winter 2022 graduation
February 4     Spring Quarter rotation decision due
February 11    Final Dissertation Submission Deadline Winter 2022 graduation
March 7        Thesis Advisory Committee members due (Second year students)
March 12       Winter Quarter ends

Spring Quarter 2022

March 25       Spring Quarter classes begin
April 4        Thesis Advisory Committee members due (Second year students)
April 3        Dissertation Office Draft Deadline Spring 2022 graduation
May 9*         Date for Qualifying Exams due (Second year students)
May 16         Summer Quarter rotation decision due (First year students)
April 29       Final Dissertation Submission Deadline Spring 2022 graduation
May 30         Memorial Day Holiday
June 3         Divisional Academic Ceremony - Spring 2022
June 4         Spring 2019 Convocation
June 4         Spring Quarter ends
May - June     Qualifying Exams held

Summer Quarter 2022

June 13 or 21 (TBD)  Summer Quarter begins
July 4              Independence Day observance
August 26           Summer Quarter ends

*The oral Qualifying Exam does not need to be taken on May 9th must be scheduled by that date. The exam must be completed by the end of Spring Quarter 2022. Extension for Qualifying Exam may be granted with the approval of the Curriculum/Student Affairs Committee.

Useful Links
Academic Calendar; BSD & University Policies; Department of Human Genetics; Dissertation Office; Human Genetics Graduate Program; Office of Graduate and Postdoctoral Affairs; Office of International Affairs Resources for Current Students; UChicago Forward
HUMAN GENETICS ADMINISTRATION

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OTHER UNIVERSITY OFFICES

Main Number - University  773/702-1234
Main Number - Hospitals  773/702-1000
Campus Police  (Call 123 from any University phone)  773/702-8181
University Voice Directory  773/702-1610
Student Health  773/702-4156
Student Counseling  773/702-9800
Student Disability Services  773/702-6000
Student Insurance (USHIP)  773/834-4543 (option #2)

HUMAN GENETICS GRADUATE PROGRAM WEBSITE
First Year

The first year of graduate study is spent taking classes, exploring research opportunities, and performing laboratory rotations.

Graduate students in the BSD are required to take nine courses for the PhD. Most classes are completed within the first year. In addition to the course requirements, students are required to attend HGEN 31900, Introduction to Research (often referred to as “AllStars”), to learn about faculty research programs within the Molecular Biosciences Cluster. Students are also required to complete two lab rotations, which are short research projects in at least two different laboratories. These rotations are done during the first academic year and must be completed before beginning dissertation research in the autumn quarter of the second year. A third rotation in the summer quarter is optional.

First-year students are also required to attend the Human Genetics Seminar Series, Work-in-Progress Seminars, and Human Genetics Journal Club when their class schedule permits.

Second Year

Just prior to the start of the second year, students take the Preliminary Examination as a first step towards candidacy for the PhD.

By the beginning of the second year, students choose a research advisor.

A minimum of coursework may continue through the second year. Most of the second year is spent developing a research project and preparing the student to submit a written proposal for dissertation research. This proposal must be defended before an oral Qualifying Examination (QE) committee before the end of spring quarter. Extension for Qualifying Exam may be granted with the approval of the Curriculum/Student Affairs Committee (CSAC). Passing the QE permits the student to enter into candidacy for the PhD.

The members of the QE committee are chosen by the student in consultation with their advisor. Committee members must be approved by the CSAC. The QE committee is comprised of the student’s research advisor and three or four other faculty members. The QE committee selects a chairperson from the committee members who is not the research advisor. At least two of the QE committee members must have an appointment in the Department of Human Genetics. If a student chooses an advisor who does not have a primary or secondary appointment in the Department, the student and prospective faculty advisor must submit a petition for approval to the CSAC. When choosing an advisor outside of the department, it is important to design a project that is appropriate for a PhD in human genetics. In addition, the thesis committee must include at least two members with a primary appointment in the Department of Human Genetics.

Second year students are required to attend the Human Genetics Seminar Series, Work-in-Progress Seminars, and Human Genetics Journal Club. In addition, second year students are expected to present at the Human Genetics Journal Club.

Advanced Years (Third years and beyond)

After the QE, the student performs full-time thesis research. The dissertation research period should take approximately 16-20 quarters, with the total duration of coursework and research not to exceed 24 quarters.

Advanced year students are required to attend the Human Genetics Seminar Series, Work-in-Progress Seminars, and Human Genetics Journal Club.

Finally, each graduating student writes a dissertation describing their research, presents the work in a public seminar and defends it before their faculty examining committee.
Evaluation

The Department of Human Genetics expects every student, throughout their term in the program, to have numerous informal conversations with the members of the CSAC, professors in their courses, their research advisor and doctoral committee members. This allows students to obtain frequent appraisals of and constructive advice on their progress. At least one formal evaluation of each student's progress must take place each academic year. In the first and second years, the evaluation is based on the student's performance in courses, laboratory rotations and the preliminary and qualifying examinations. In advanced years, the research advisor and doctoral committee report to the CSAC on the student's dissertation research progress after annual committee meetings. If the CSAC is apprised of deficiencies in performance, the student will receive a letter describing those deficiencies with suggestions as to how they might be remedied.

Graduate Education Administrator

The Graduate Education Administrator, Sue Levison, provides assistance to students on a variety of questions and problems as they arise. The office is located in CLSC 1111. The office phone number is 773/702-2464; Cell phone is 773/307-2237 and e-mail address is slevison@bsd.uchicago.edu.

Requirements for the PhD Degree

A PhD candidate must fulfill certain formal coursework requirements, pass preliminary and qualifying examinations and present a satisfactory dissertation describing the results of original research.

All graduate students are required to serve as a Teaching Assistant in two courses for academic credit before the PhD degree is awarded. Courses can be undergraduate, graduate, or medical, but must be in the Biological Sciences Division. For those students who feel unprepared for teaching, a TA Training Course is offered every academic year (Spring quarter). This course may count as one of the two TA credits.

The department expects a knowledge of and proficiency in human genetics. This requirement will normally be met by fulfilling the formal coursework listed below, but the program is flexible. Courses taken at other institutions, in other departments, or as part of the Medical School curriculum may substitute for genetics courses with approval of the CSAC.

Formal Coursework (Empirical and Computational Course Tracks)

Eight courses and at least two lab rotations are required to fulfill the requirements for a PhD. In order to maintain full-time status, students are expected to take three courses each quarter or two courses and a lab rotation during the Autumn, Winter or Spring quarters and one (or two) rotations during the summer quarter. One elective course may be taken in the second year. Anything less than full-time student status requires the approval of the CSAC. Students must complete eight graded courses and two rotations prior to taking the QE at the end of their second year. It is permissible for students to take the QE during the quarter they satisfy their final course requirement and no later than the ninth quarter from their matriculation quarter.

Human Genetics Empirical Track Coursework
(Three required courses AND two core electives AND three elective courses PLUS two rotations)

Three Required Courses
HGEN 47000 Human Genetics I (Autumn) AND HGEN 47500 Genetic Mechanisms from Variation to Evolution. AND GEN 46900 Human Variation and Disease (Spring)
Two Core Electives Courses (Chose from the following)
HGEN 47100 Intro Statistical Genetics (Winter) OR HGEN 31100 Evolution of Biological Molecules (Winter) OR HGEN 48600 Fundamentals of Computational Biology: Models and Inference (Winter) OR HGEN 47300 Genomics and Systems Biology (Spring) OR HGEN TBD Quantitative Genetics for the 21st Century (Spring) OR MGCB 31400 Genetic Analysis of Model Organisms (Autumn) OR DVBI 36400 Developmental Mechanisms (Winter) OR ECEV 35600 Population Genetics (Spring) I OR ECEV 35600 Population Genetics I (Spring) OR MGCB 31300 Molecular Biology II (Spring)

Plus Two Lab Rotations
BSDG 40100 Section 11 Non-Thesis Research / BSDG 40200 Section 11 Non-Thesis Research. Laboratory rotations, and all research prior to passing the Qualifying Examination. (Autumn, Winter, Spring, Summer)

Plus Three Electives:
Students must take three electives (selected by the student from a list approved by the CSAC), including one statistics course at a level appropriate to the student’s background and research interests.

ADDITIONAL ELECTIVE COURSES TO CHOOSE FROM TO FULLFILL 3 COURSES
HGEN 39900 Readings in Human Genetics (Autumn, Winter, Spring, Summer); HGEN 47400 Introduction to Probability and Statistics for Geneticists (Autumn); HGEN 48800 Fundamentals of Computational Biology: Algorithms and Applications (Spring); HGEN 36400 Molecular Phylogenetics (Autumn); BCMB 30400 Protein Fundamentals (Autumn); DVBI 35600 Vertebrate Developmental Genetics (Winter); ECEV 35901 Genomic Evolution I (Autumn) MGCB 31200 Molecular Biology I (Winter); MGCB 32000 Quantitative Analysis of Biological Dynamics (Spring); MGCB 31600 Cell Biology I (Autumn); MGCB 31700 Cell Biology II (Winter); MGCB 32000 Quantitative Analysis of Biological Dynamics (Spring); NURB 33400 Genetic Approaches in Neurobiology (Spring); STAT 24300 Numerical Linear Algebra (Autumn); STAT 24400 Statistical Theory and Methodology I (Autumn, Winter); STAT 24500 Statistical Theory/Method-2 (Winter); STAT 35500 Statistical Genetics (Spring)

ADDITIONAL REQUIRED COURSES
HGEN 31900 Introduction to Research. "Allstars" (Autumn)
BSDG 55100 Responsible, rigorous, and reproducible conduct of research: R3CR. Winter

Human Genetics Computational Track Coursework
(Three required courses AND three core electives AND two additional elective courses PLUS two rotations)

Three required courses in Computational Biology and Statistics

Plus Three Core Elective Courses Chosen from the Following List
HGEN 47000 Human Genetics I (Autumn) OR MGCB 31400 Genetic Analysis of Model Organisms (Autumn) OR HGEN Genetic Mechanisms from Variation to Evolution (Autumn) 47500 HGEN 47100 Introductory Statistical Genetics (Winter) OR ECEV 35600 Principles of Population Genetics I (Winter) OR ECEV 31100 Evolution of Biological Molecules (Winter) OR BCMB 32200 Biophysics of Biomolecules (Spring) OR HGEN 46900 Human Variation and Disease (Spring) OR HGEN xxxxx Quantitative Genetics for the 21st Century (Spring) HGEN 47300 Genomics and Systems Biology (Spring) OR MGCB 32000 Quantitative Analysis of Biological Dynamics (Spring)

Plus Two Lab Rotations
BSDG 40100 Section 11 Non-Thesis Research / BSDG 40200 Section 11 Non-Thesis Research. Laboratory rotations, and all research prior to passing the Qualifying Examination. (Autumn, Winter, Spring, Summer)
Plus Two [2] Additional Elective Courses Chosen from the Following List


ADDITIONAL REQUIRED COURSES

HGEN 31900 Introduction to Research. “Allstars” (Autumn)
BSDG 55100 Responsible, rigorous, and reproducible conduct of research: R3CR. Winter

A complete list of courses is available on the Academic Information System and Human Genetics Graduate Program website

Additional Information for Formal Coursework (Empirical and Computational Course Tracks)

- Students must petition the CSAC for approval of courses not listed in this handbook as “approved”.
- At least three of the four electives and all of the requirements must be completed before attempting the Preliminary Exam.
- All four electives must be taken before the Qualifying Exam.
- One elective course may be taken pass/fail. Additional courses may be taken pass/fail subject to the approval of the instructor and the Curriculum Committee.
- If a student wishes to take an undergraduate course to fulfill an elective requirement, they must petition the CSAC for approval.
- Students should note that several courses have prerequisites for enrollment or require the consent of the instructor. Students entering the department with advanced coursework at the graduate level should inquire whether this coursework can substitute for required load of elective credits.

Students are expected to have a grade average of "B" or higher in their required courses and a “B” average overall. Students who fail to maintain a “B” average after the second quarter will be placed on academic probation. Students concluding their first year with less than a “B” average or less than a “B” in a required course will be terminated from the program after spring quarter unless otherwise recommended by the CSAC. Students who receive a D or F in any course, during any quarter, will be put on immediate academic probation, regardless of the grade average.

Introduction to Research

All first-year students are required to attend the HGEN 31900 Introduction to Research course (“AllStars”) during the Autumn quarter. This course is designed to provide incoming students with information on the variety of faculty research opportunities available and experience with oral presentations. This course is offered pass/fail. Strict compliance with the attendance policy is required for a passing grade.

Scientific Ethics Courses (Responsible, rigorous, and reproducible conduct of research: R3CR)

All first-year students are required to attend a scientific ethics class organized by the Dean of Students Office. This course is offered during the spring quarter and features sessions on scientific ethics that often involve examining case studies. The course organizer distributes announcements with the title of each talk and the name of the faculty members who will present.

A second training in the ethical conduct of research is required for students still registered four years after their initial training. Senior ethics training content is more closely aligned with research areas and so this training is coordinated by the individual graduate programs.
Prescribed Courses

In some instances, a student's undergraduate training may not have prepared him/her for a required course. In such cases, the CSAC will prescribe an appropriate graduate or undergraduate course if necessary. In some such cases, the prescribed course can be counted as a graduate elective.

Reading Courses

Reading courses taken for a grade must be approved by the CSAC prior to registration. Every reading course must conform to the following requirements: 1) it must meet weekly, 2) the instructor must provide a syllabus for the course and an evaluation of the student's performance, both of which will become part of the student's file, and 3) the student must submit a written paper.

Laboratory Rotations

Students are required to perform at least two laboratory rotations before selecting an advisor and laboratory to pursue a PhD dissertation.

The purpose of lab rotations is to expose the student to different research environments, broaden their acquaintance with useful laboratory techniques and introduce them to the conceptual framework of experimental design. Students undertake short research projects in at least two different laboratories before beginning their dissertation research. At the end of the rotation, the student should write a two- to three page report on their rotation project(s) and/or prepare an oral presentation, at the discretion of the mentor. The mentor will grade the report or presentation and will provide the CSAC with an evaluation of the student’s performance.

Rotations are typically performed in the Winter, Spring and Summer quarters during the first year of study. In exceptional circumstances a student can begin a rotation the summer prior to the start of the first academic year. During the Autumn, Winter and Spring quarter the rotation lasts ten weeks, coinciding with the academic quarter. One ten week or two five week rotations is done during the Summer quarter when the student can devote full time to research.

Students arrange their own rotations by contacting potential mentors directly. All Human Genetics faculty members are potential rotation mentors. After the student and mentor have agreed on the time period for the rotation, the student and mentor will complete the Divisional Rotation Form which is then given to the Graduation Education Administrator for additional processing. The Program Chair and the CSAC will review the completed Divisional Rotation Form let the student know if the rotation has been approved. Students who would like to rotate with a faculty member who does not have a primary or secondary appointment in the Department of Human Genetics must petition the CSAC for approval.

Teaching Assistantships

All graduate students are required to serve as a Teaching Assistant in two courses for academic credit before the PhD degree is awarded. Courses can be undergraduate, graduate, or medical, but must be in the Biological Sciences Division.

The ability to communicate verbally and to teach are important skills for a successful research career. As such, all students are required to serve as teaching assistants (TAs) for two quarters, with responsibilities that may include leading discussion groups, writing problem sets, and running laboratories. Students normally undertake their teaching assistantships during the second and third years. A course designed to train graduate students to be an effective TA may be taken in lieu of one of the two assistantships. The student must receive approval from the CSAC prior to accepting a TAship. The two required TAships must be completed prior to the end of their fourth year of study. Student MAY NOT fulfill a TAship requirement during the last quarter of their graduate studies.

Students with extensive teaching experience at the graduate level (e.g. while studying for a Master's degree) are permitted to petition for waiver of the teaching requirement. The petitioner must provide documented evidence of
prior teaching experience. For additional information, please contact the Graduate Education Administrator.

Students may not TA for pay before completing the requirement; they may only TA the same course twice if there are significant changes in responsibilities and opportunities to learn new skills in teaching.

**Preliminary and Qualifying Examinations**

**The Preliminary Examination**

The Biological Sciences Division requires that "a general oral or written qualifying examination, separate from course examinations, must be passed by the student upon the major subject offered and such subordinate subjects as may be required by the Department concerned." In the Department of Human Genetics, this examination will be given in the September after completion of the first year. Students will be given a set of questions covering broad areas in classical and human genetics.

The exam lasts for approximately two hours. To prepare for the exam students are allowed to use books, reference materials and lecture/seminar notes to answer the questions. Students are also free to discuss the questions among themselves and with faculty.

Students will be given the full list of Preliminary Examination questions at the beginning of the Summer quarter. There are two sections to the Preliminary Examination. The first section is required; students need to prepare to answer all the questions in the required section. The remaining questions are divided into four topics. Students will choose two of the four topics. In the second part of the exam, the committee will ask questions only from those topics.

**REQUIRED QUESTIONS:** Fundamental Genetics; Population and Evolutionary Genetics; Statistics in Genetics

**FOUR OPTIONAL TOPICS:** 1) Mapping; 2) Genetic Architecture of Human Phenotype; 3) Gene Regulation and Human Phenotypes; 4) Study Design and Statistical Data Analysis

The purpose of this exam is to provide practice in oral presentations and discussion as well as to demonstrate the ability to think critically.

The CSAC chooses three faculty for each examining committee from departmental faculty. The names of the examining committee are provided to the student one week prior to the exam.

Based upon the student's performance, the Preliminary Exam committee recommends one of the following options to the CSAC:

- Pass Unconditionally
- Pass conditionally, with written answers to a question(s) required. Answers should be submitted within two weeks. The student will then meet again with the Exam committee to defend their answers
- Pass conditionally, with further coursework, or comparable training experience (e.g., course TAship), required in one or more areas.
- Fail, with the recommendation that the student retake the examination within the quarter.
- Fail, with the recommendation that the student leave the program (if this is a retake of the examination).

The CSAC then meets to consider the examining committee’s recommendation, taking into account the student's overall academic performance as well as his or her performance on the examination. If a student who fails the exam is allowed to retake it, a new committee containing at least one member of the original preliminary examining committee and at least one new member will be selected by the CSAC.

**The Qualifying Examination**

The Qualifying Examination (QE) evaluates a student's ability to propose and defend a doctoral thesis research plan. Upon successful completion of the exam, the QE committee becomes the student's doctoral advisory committee. Students must have the endorsement of their research advisor in order to stand for the QE. In the event that a research advisor declines to endorse a student for the QE, the CSAC will review the student's record and determine if that student will be allowed to seek a new research advisor or be asked to leave the program.
After the student chooses a research advisor and defines their project, the student, in consultation with the research advisor, formulates a list of four or five prospective qualifying exam committee members (including the student's advisor) and submits the petition to the CSAC for approval. It is not uncommon for the CSAC to recommend adding or changing the composition of the committee to broaden the overall expertise of the committee. Final decisions on committee membership are made by agreement between the CSAC, the research advisor and the student.

In addition to approving the initial thesis advisory committee, the CSAC must also approve replacements when members of a doctoral committee resign. In the event that more than one member of a doctoral committee resigns, the CSAC will meet to consider the circumstances that led to the resignations and decide on an appropriate course of action. Possible courses of action include (but are not limited to) replacement of doctoral committee members, formation of a new doctoral committee or reconsideration of the student's qualifications for candidacy.

The QE committee members will continue to serve as an advisory committee throughout the course of the student’s doctoral research. This doctoral committee will be chaired by a member other than the student's advisor. The function of the doctoral committee is to monitor the student's progress and to assist the student in the development of the dissertation research. For this reason, the choice of the members of the doctoral committee should be based on their knowledge and expertise in the area of the student's research. In the event the student chooses to work with a member of the faculty who does not have an appointment in the Human Genetics, the student must petition the CSAC for approval. At least two members of the doctoral committee, including the chair, must have primary appointments in the Department of Human Genetics.

It is important to note that the QE is not a thesis defense. It does not require preliminary results although, if available, they may be used.

The exam tests the student’s ability to:
- Choose a topic. That is, to formulate an important biological question;
- Propose a coherent set of avenues to answer the question;
- Summarize critically the current literature on that topic; and
- Describe a series of experiments, taking into account possible pitfalls and therefore alternative approaches.

The written proposal should be modeled after an NIH postdoctoral grant application and it should be organized as follows:

Specific Aims (no more than 1 page): describe at least three specific aims and the broad, long-term objectives of the proposed research to test a stated hypothesis.

Research Strategy (no more than 6 pages): This section should be further divided into:
1) Significance of the proposed studies, including background information, and 2) Approach (including preliminary studies, if any) to provide experimental support of the proposed hypothesis. This is not a place for trivial experimental details.

All proposals must have a title sheet (not included in page limits) and the author’s name on each page. Pages must be numbered.

**Annual Doctoral Committee Meetings**

Within twelve months of passing the QE, the student should schedule a meeting with their doctoral committee. All students should meet at least once a year with their doctoral committee and present a brief (up to three pages) written report of their research as a basis for discussion. The report must be distributed at least one week prior to the scheduled meeting.

After the fourth year, a minimum of two meetings per year may be required, with at least three members of the doctoral committee present at each meeting. These meetings help to ensure that students are making adequate progress toward the completion of their research and to provide the student with a broader base of expertise on which to seek help and advice. They also strengthen the student’s acquaintance with faculty other than the research
advisor, providing a basis for future letters of recommendation.

With doctoral committee approval, a student may prepare a dissertation (Penultimate Meeting). Following each meeting the chair of the doctoral committee prepares a summary of the meeting. This summer is distributed to the student, the doctoral committee members and the Chair of the HG Graduate Program.

**Penultimate Meeting**

The doctoral committee should be convened six months before a student expects to receive their degree to indicate their agreement that the student is nearing completion of their research and to arrange for subsequent approval that the student may begin writing the dissertation.

In general, the mentor and other members of the advisory committee strive to minimize the possibility of an unsuccessful thesis defense via thoughtful and straightforward advice to the candidate. The penultimate meeting is particularly important in this regard. Permission to write should not be granted if more than one member of the committee lacks confidence that the thesis will be acceptable. The written report from the penultimate meeting should contain a fairly detailed description of any additional work that needs to be completed prior to submission of the thesis. This list should be limited to a small number of minor items. If, in the judgment of the advisory committee, substantial work is needed prior to the defense, an additional meeting should be scheduled to review such progress before permission to write is granted.

**Dissertation – Public and Private Defense**

The final steps in earning the PhD are writing the dissertation, publicly presenting it, and defending it to the committee. In all three of these items, the candidate should demonstrate that 1) they have made an independent contribution to scientific knowledge in their field, and 2) they have the qualifications to be awarded the doctoral degree, including mastery of the conceptual and technical foundations of their field; knowledge of relevant prior work; ability to identify, articulate and justify significant scientific questions; capacity to devise and execute scientific strategies to answer those questions; rigor in analysis and interpretation of data; and the ability to draw valid scientific conclusions and articulate their significance for the field.

The university has strict rules concerning the preparation of the dissertation. Detailed information can be obtained from the Dissertation Office located on the 1st floor of the Regenstein Library, Room 100B. Visit the Dissertation Office website which has the most current information about upcoming deadlines, required forms, etc.

**The dissertation.** The doctoral dissertation should report original research for which the student was primarily responsible. It should contribute well supported new knowledge on a significant subject for the relevant scientific field. The dissertation should be of sufficient quality and rigor to pass stringent peer review and therefore contribute valid knowledge to the scientific literature. The thesis research should represent a contribution equivalent to at least two, and preferably three, published or publishable research papers in the peer-reviewed literature; it should also contain a scholarly introduction that justifies the significance and strategy of the research and places it in the context of current knowledge, as well as a conclusion that discusses limitations of the work, significance and implications of the findings, and potential directions for future research.

**Collaborative work.** The dissertation must represent the candidate’s work as an individual, but most modern biology is collaborative. Projects to which others have contributed may be included, so long as the candidate’s contribution represents that of a first or co-first author of the reported work. That is, the candidate must have played a lead role in the conception, development, execution, and interpretation of the project, and must have been the primary contributor to the writing of the chapter. If others (including the advisor) have contributed to any of these aspects of a chapter, the specific contributions made by the candidate and by each contributor must be specifically described.

**Thesis Defense.** The defense consists of two portions, a public presentation and a private session with their doctoral committee members immediately following the public presentation. In the public presentation, the candidate presents the research on which the claim to the doctoral degree is based. It should justify the significance of the question(s) the candidate has addressed and the scientific strategy used. The candidate should present the experiments, analyses, and conclusions that represent the dissertation’s major contributions in sufficient detail to
persuade an audience of scientific peers. If time constraints prevent a detailed treatment of all aspects of the thesis, it is acceptable to provide an overview of the thesis' scope and a detailed treatment of a portion of the research. A scholarly conclusion reflecting on the significance of the work, its limitations, and future directions should be included. A brief statement of acknowledgements is very appropriate; lengthy reflections and statements of personal gratitude should be made elsewhere. The audience will have an opportunity to ask questions of the candidate, who should be prepared to respond in a constructive and scholarly manner.

During the private doctoral committee members portion of the defense, the candidate should be prepared to discuss and defend all aspects of the work reported in the thesis.

**Master's Degrees – Transitional and Terminal**
The Department of Human Genetics does not admit students directly into an MS program. Master’s degrees are awarded only to students who are enrolled in the PhD program.

**Transitional Master’s Degrees**
Upon completing all course requirements with a “B” average and successfully passing the Preliminary and Qualifying Examinations HG students will receive a Transitional MS degree. However, the Transitional MS Degree will only be issued once the student has successfully defended their thesis (not after QE).

**Terminal Master’s Degrees**
For a student who decides not to complete their PhD candidacy, or who loses PhD candidacy status, but has completed all course requirements with a “B” average and has successfully passed the Preliminary Examination may be eligible for a Terminal Master’s degree. The CSAC makes final decisions with respect to the granting of Master’s degree

**Seminars**
In addition to formal courses, there are many regularly scheduled research seminars that help keep students current on new developments in human genetics and related disciplines.

All students are expected to attend the Department of Human Genetics events, class schedules permitting.

Seminars are held on Wednesdays at noon. Check the [Events Calendar](#) on the Human Genetics website for the most current schedule.

- **The Department of Human Genetics Seminar Series** features a research talk by a visitor from outside the University of Chicago.
- **The Department of Human Genetics Journal Club** is a presentation of a current journal article of current relevance to the field of human genetics research. Presentations are made by pre-doctoral students in consultation with a faculty member.
- **The Department of Human Genetics Work-in-Progress** is a presentation by a pre- or post-doctoral researcher in a Human Genetics’ faculty member’s lab. The seminar allows members of the department to stay abreast of faculty research projects throughout the department.

Students are also encouraged also to attend Seminars, Journal Clubs and Work-in-Progress events sponsored by the [Committee on Genetics, Genomics and Systems Biology](#).

**FINANCIAL SUPPORT**
The Department of Human Genetics attempts to ensure that all students registered in the PhD program are provided with adequate financial aid. Financial support is guaranteed to all incoming students for their first four years, subject to satisfactory academic performance. Support for subsequent years of study is subject to the student's satisfactory research progress, as determined by the faculty sponsor, the doctoral committee and the Division of Biological Sciences.
Sources of Support

Students receive tuition plus a stipend. The various sources of support include, but are not limited to:

- Divisional Funding
- NIH Training Grants
- External Fellowships
- University Fellowship
- Research Assistantships

Payment of Stipend Checks

Divisional funding and NIH checks are paid in equal quarterly installments at the beginning of each quarter and cover the calendar year. Taxes are owed on, but not deducted, from these stipend checks (see section on “Taxes” below). Research Assistant Type B (RA Type B) and Research Assistant Type A (RA Type A) students are paid on a monthly basis. Taxes will be deducted from the RA Type-B checks.

Taxes

Graduate student stipends are taxable by Illinois and the Federal government. Students on fellowships and NIH training grant support must calculate and pay estimated taxes several times a year.

The following IRS forms provide information on determining what portion of your stipend is taxable and when to pay taxes you owe: Tax Benefits for Education, PUB 970; US Tax Guide for Aliens, PUB 519 and US Tax Treaties, PUB 901. IRS form 520 provides information on determining what portion of your stipend is taxable and how and when to pay taxes you owe. These forms are available from the IRS. Regenstein Library also carries tax forms particularly after January 1st. For additional information see: http://www.irs.gov/Individuals/Students

Supplies and Research Expenses

In general, costs of research supplies and equipment are covered by grants or contracts held by the faculty member in whose laboratory you are working. Limited funds for supplies are available on training grants, and are disbursed on an annual pro-rated basis, directly to the laboratories in which trainees are working.

Travel to Scientific Meetings

Attendance at scientific meetings is an important part of the educational process. Should you wish to apply for support, check with the source of your funding, (your Research Advisor, or training grant or fellowships). When making your request, please supply the following information: purpose of meeting and relevance to the research; title, place and time of the meeting; title and authors of paper being presented; and amount requested for travel, registration fees, food and lodging. The Department of Human Genetics is not able to provide financial support to students for scientific meetings

REGISTRATION

General Information

Approximately one week before the dates designated for registration, the Graduate Education Administrator will contact students via email informing them of the dates and times to register online. If a student does not register for their courses prior to the deadline, they will be charged a late registration fee of $100.

Special registration procedures have been established for the first year students in the Autumn quarter. During Orientation week, first year students will meet with the HG Program Chair and CSAC to finalize their Autumn courses and map out a program of study for the first year. The Graduate Education Administrator will meet with first year students to assist with their Autumn registration. If necessary, second year students also will meet with members of the CSAC to review their progress in the preceding year and to discuss further degree requirements.
Residency Status

All students are in one of three levels of residency, depending on the number of quarters they have been registered at the University. The three levels and the number of corresponding registration units are:

**Scholastic Residence (SR) Years 1-4:** Students in SR are eligible for all benefits associated with full time student status at the University, such as the student health plan, university housing, student loans and loan deferment.

**Advanced Residence (AR) Years 5-12:** Students in AR are eligible for all benefits associated with full time student status at the University, such as the student health plan, university housing, student loans and loan deferment.

**Extended Residence (ER) - Years 12 and beyond:** Students in ER are entitled to use of the library, email accounts, networked access, and faculty contact, but not to other benefits or facilities.

Leave of Absence

During Scholastic and Advanced Residence a student may, if necessary, apply for a Leave of Absence from the PhD program to be approved by the CSAC and the Dean and Director of Graduate Affairs. Only students in good academic standing will be granted a Leave of Absence. Student may also apply for a Medical Leave of Absence. For additional information, contact the Graduate Education Administrator.

Should the need arise, student may also apply for a Medical Leave of Absence approved by the CSAC and the Dean and Director of Graduate Affairs. Talk to your Graduate Education Administer for additional information.

Additional information for the University of Chicago Policies and the Biological Sciences Division policies governing students is available on the OGPA website

Pro-Forma Registration

Students in Advanced Residence, whose dissertation research requires residence away from Chicago, may register pro-forma. A fee per quarter is assessed and keeps the student in full-time registration for purposes of reporting to outside agencies such as to defer student loans. Pro-forma status establishes a good faith relationship between the student and the University. The following regulations apply:

- Pro-forma registration is approved for only one academic year at a time and the maximum pro-forma enrollment allowed is eight quarters
- Applications for pro-forma registration must be approved in writing by the Department of Human Genetics Program Chair, whose signature means that the student's work away from Chicago is recognized as essential to the dissertation. Normally, students applying for pro-forma status will have been admitted to candidacy and have had dissertation topics approved.
- An applicant for renewal of pro-forma status must show the Department of Human Genetics Program Chair that good use has been made of the time already spent "on location" and that additional time is essential to completing the original task. Renewals of pro-forma status must be approved by the Office of Graduate and Postdoctoral Affairs.
- A student on pro-forma status may not be gainfully employed for more than 19 hours a week.
- Pro-forma students may not use the facilities of the University or the time of its faculty, except for progress reports that may be required by the students' program.
- The Registrar will certify that a pro-forma student is duly registered at the University to any agency requiring such certification.
- The fact that a registration is pro-forma will be noted on the student's academic record.
- Pro-forma registrations do not count toward satisfying a student's residence requirements toward a degree.

MISCELLANEOUS INFORMATION

Curriculum and Student Affairs Committee
This faculty committee is responsible for advising all students during the first year of graduate study or until a research advisor has been chosen. Each student will be assigned a member of the committee to serve as temporary advisor during this time and to aid in selecting courses and arranging lab rotations. This committee conducts a quarterly review of each student's course performance and administers the preliminary examination. Members of the CSAC meet with first-year students after each quarter to discuss any issues concerning the first-year curriculum or other topics of concern.

**Student Representatives**

The Human Genetics graduate program have student representatives to represent HG student concerns as needed at the quarterly HG quarterly faculty meetings. At any time should a student representative have an agenda item for one of these meetings, they should contact the HG Program Chair to request they be added to the agenda for the next meeting. Each representative has a two year term, with one representative new each year, and the other tenured by one year. Each summer, once a representative’s two year term has expired, a new representative is appointed. Student representatives also participate in Molecular Biosciences Cluster events, such as Orientation, Retreat, and Recruitment planning. These representatives are volunteers who are interested in participating and contributing to these events. The Student Representative will ask for volunteers each year. Students interested in becoming a student representative should contact the Graduate Education Administrator.

**Molecular Biosciences Retreat**

The Molecular Biosciences Annual Retreat provides an opportunity for students, post-docs, and faculty to meet in a pleasant, informal setting to learn about the various research programs of the various research laboratories. The program consists of several sessions of presentations by students and post-docs. Each session is chaired by a faculty member. The Retreat is held annually in the Autumn quarter.

**ID & Privileges Office**

The ID & Network Privileges Office, located in the lobby of Regenstein Library (1100 E. 57th Street), is a joint venture between the Library and IT services. They offer a variety of services to the University community. Their main services include: UChicago Cards, Library access and privileges and Passport photos.

**Bursar's Office**

The Bursar's Office, located at 6030 S. Ellis Avenue. The University Cashier (In-Person Cash and Check Payments) is located at 5525 S. Ellis (55th and Ellis Parking Structure). Students may contact the Bursar’s Office for information on:

- Tuition Inquiries & Bursar Restrictions: 773/702-7086
- Other Information: 773/702-8000
- For additional information go to: http://bursar.uchicago.edu/

**Student Wellness**

**Student Health Services** provides health care to all registered students in the University. It is funded by a mandatory quarterly Student Services Fee. Payment of this fee allows the student access to the University’s Student Health Services. Some specialized and emergency care is not covered, nor does the fee include the cost of outside referrals, laboratory tests, and hospitalizations.

**University Student Health Insurance Plan (USHIP)** In addition to participation in Student Health Services, all students are REQUIRED to carry a health insurance plan (either university student health insurance or comparable insurance) to cover the costs of hospitalization, outpatient diagnostic and surgical procedures, laboratory tests and catastrophic illness. Charges for university insurance are assessed for each of three quarters (Autumn, Spring, Winter); there is no charge for coverage for the Summer Quarter. Additional information can be found on the, University Student Health Insurance Plan (USHIP) website
Students with comparable group insurance coverage through a parent, spouse, or their own policy may request that participation in the university program be waived. However, they must cover the cost of alternative health insurance out of their own pocket.

**Student Counseling Services**

*UChicago Student Wellness* is committed to promoting the mental health and well-being of UChicago undergraduate and graduate students by providing accessible, high-quality, culturally sensitive mental health services. We also provide outreach and consultation to the University community. All of their services are covered by the *Student Services Fee*, and there is no additional cost for students to access our services. Call 773.702.9800 to make an appointment with a therapist. Additional links and information can be found on the [Student Wellness](#) website.

For location, hours, how to make appointments and additional information please see the [Student Wellness](#) website.

**Student Disability Services**

To ensure the intellectual richness of research and education, the University of Chicago seeks to provide an environment conducive to learning, teaching, working, and conducting research that values the diversity of its community. The University strives to be supportive of the academic, personal, and work-related needs of each individual and is committed to facilitating the full participation of students with a disability in the life of the University.

Student Disability Services works to provide resources, support, and accommodations for all students with disabilities and works to remove physical and attitudinal barriers, which may prevent their full participation in the University community.

Contact Student Disability Services directly for general questions about accommodations for University classes, programs or activities, please contact them at: 773-702-6000, or via email:

- General: disabilities@uchicago.edu
- Exams: proctor@uchicago.edu
- Notetaking: notetake@uchicago.edu
- Alternative Format Text: text.sds@uchicago.edu

Please see below for additional important and helpful University of Chicago links

- [Human Genetics Graduate Program Resources Page](#)
- [CNetID account assistance](#)
- [Computing Facilities](#)
- [Copying, Printing & Scanning](#)
- [Gerald Ratner Athletics Center](#)
- [Office of International Affairs](#)
- [Safety and Security](#)
- [Transportation & Parking](#)
- [TransLoc](#)
- [UGo NightRide Shuttles](#)
Chicago at Large

Chicago is a fantastic city for cultural pursuits including museums, music, theatre, and dining out. The Chicago Symphony Orchestra, the Lyric Opera, jazz and blues clubs, The Goodman Theater, and off-loop theatres are all excellent. Both inexpensive ethnic restaurants and expensive special-occasion restaurants abound.

Chicago Area Festivals, Exhibits and Special Events Websites

For information on outdoor concerts, cultural and neighborhood festivals, art fairs and other special events in the Chicagoland area visit the following websites:

The Chicago Convention and Tourism Bureau: www.choosechicago.com

Special Events Management: www.chicagoevents.com

The Chicago Park District: http://www.chicagoparkdistrict.com


Metromix: http://chicago.metromix.com/

The Chicago Reader: http://www.chicagoreader.com


Block Club Chicago: https://blockclubchicago.org/

The Magnificent Mile: http://www.themagnificentmile.com/

The Promontory: http://promontorychicago.com/

Ravinia: http://www.ravinia.org

The Chicago Symphony Orchestra: https://cso.org/

Chicago Architecture Center: http://www.architecture.org/

The Museum of Broadcast Communications: https://www.museum.tv/

The Museum of Science and Industry: http://www.msichicago.org

The Field Museum: http://www.fieldmuseum.org

Illinois Holocaust Museum & Education Center: https://www.ilholocaustmuseum.org/

The International Museum of Surgical Science: https://imss.org/

The Alder Planetarium: http://www.adlerplanetarium.org
John G. Shedd Aquarium: http://www.sheddaquarium.org
The Art Institute: http://www.artic.edu
Kohl’s Children Museum: http://www.kohlchildrensmuseum.org
Lincoln Park Zoo: http://www.lpzoo.com
Brookfield Zoo: http://www.brookfieldzoo.org
Navy Pier: http://www.navypier.com
Broadway in Chicago: http://www.broadwayinchicago.com
The League of Chicago Theatres: http://www.chicagoplays.com
The Goodman Theatre: http://www.goodmantheatre.org/
The Looking Glass Theatre: https://lookingglasstheatre.org/
Theater Wit: http://www.theaterwit.org/
The Second City: http://www.secondcity.com/
Steppenwolf Theatre: http://www.steppenwolf.org
The Chicago Botanic Gardens: http://www.chicago-botanic.org
The Morton Arboretum: http://www.mortonarb.org
Chicago Public Library: https://www.chipublib.org/
Chicago Sport and Social Club: http://www.chicagosportandsocialclub.com/
Chicago Athlete: http://www.mychicagoathlete.com/
Fleet Feet Sports: http://www.fleetfeetchicago.com
Divvy Bikes – Bike Sharing System: http://divvybikes.com/
Forest Preserves of Cook County: https://www.chipublib.org/
Illinois State Parks: https://www.dnr.illinois.gov/recreation/Pages/default.aspx
Starved Rock State Park: http://www.starvedrockstatepark.org/
Indiana State Parks: http://www.in.gov/dnr/parklake/
Indiana Dunes: http://www.indianadunes.com/
Wisconsin State Parks: http://dnr.wi.gov/topic/parks/
Wisconsin Bike Trails: [http://dnr.wi.gov/topic/parks/activities/bike.html](http://dnr.wi.gov/topic/parks/activities/bike.html)
HUMAN GENETICS EMPIRICAL TRACK COURSES

REQUIRED COURSES FIRST YEAR CURRICULUM

A complete list of courses is available on the Academic Information System and Human Genetics Graduate Program website


THREE [3] 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 47500 Genetic Mechanisms from Variation to Evolution. Spitz, Novembre. This course provides a graduate-level introduction to enduring questions regarding the fundamental processes by which genetic information is inherited, regulated, and transformed into organismal phenotypes and how these mechanisms shape and interact with evolutionary processes. We will describe different strategies, including new genome analysis and engineering technologies and statistical/computational principles, that can be used to study the complex, multi-layered organization of genomes, their interactions with varying environments, and ultimately, their evolution. Autumn.

HGEN 46900 Human Variation and Disease. Di Rienzo, Berg, Novembre, 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 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 used 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

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

HGEN TBD Quantitative Genetics for the 21st Century. Berg, Dahl. In this course we will describe opportunities and pitfalls in genetic studies of complex traits, evolutionary parameters, and group differences. We will review 20th century quantitative genetics work on evolution and animal breeding. Then we will describe how these methods were repurposed for complex human diseases. Finally, we will discuss the recent trend of applying these tools to complex social traits, emphasizing how they can potentially improve health but also how naive uses can amplify nongenetic factors. 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.

OR

ECEV 35600 Population Genetics I. Kreitman, 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

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.

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.

BSDG 40100 Section 11 Non-Thesis Research / BSDG 40200 Section 11 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 THREE [3] ELECTIVE COURSES

ADDITIONAL ELECTIVE COURSES TO CHOOSE FROM TO FULFILL 3 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. 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. 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.

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. 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.
HUMAN GENETICS COMPUTATIONAL TRACK - COURSES

A complete list of courses is available on the Academic Information System and Human Genetics Graduate Program website.


THREE [3] REQUIRED COURSES

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. Autumn, Winter.

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.

HGEN 48800 Fundamentals of Computational Biology: Algorithms and Applications. Chen, He. 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.

PLUS THREE [3] CORE ELECTIVES CHOSEN FROM THE FOLLOWING LIST

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.

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
**HGEN 47500 Genetic Mechanisms from Variation to Evolution. Spitz, Novembre.** This course provides a graduate-level introduction to enduring questions regarding the fundamental processes by which genetic information is inherited, regulated, and transformed into organismal phenotypes and how these mechanisms shape and interact with evolutionary processes. We will describe different strategies, including new genome analysis and engineering technologies and statistical/computational principles, that can be used to study the complex, multi-layered organization of genomes, their interactions with varying environments, and ultimately, their evolution. Autumn.

OR
**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
**ECEV 35600 Population Genetics I. Kreitman, 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. 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 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
**BCMB 32200 Biophysics of Biomolecules. Sosnick.** This course covers the properties of proteins, RNA, and DNA, as well as their interactions. We emphasize the interplay between structure, thermodynamics, folding, and function at the molecular level. Topics include cooperativity, linked equilibrium, hydrogen exchange, electrostatics, diffusion, and binding. Spring

OR
**HGEN 46900 Human Variation and Disease. Di Rienzo, Berg, Novembre, 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.

OR
**HGEN TBD Quantitative Genetics for the 21st Century. Berg, Dahl.** In this course we will describe opportunities and pitfalls in genetic studies of complex traits, evolutionary parameters, and group differences. We will review 20th century quantitative genetics work on evolution and animal breeding. Then we will describe how these methods were repurposed for complex human diseases. Finally, we will discuss the recent trend of applying these tools to complex social traits, emphasizing how they can potentially improve health but also how naive uses can amplify nongenetic factors. 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 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.*

**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*

**BSDG 40100 Section 11 Non-Thesis Research / BSDG 40200 Section 11 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 TWO [2] ADDITIONAL ELECTIVES CHOSEN FROM THE FOLLOWING LIST**

**STAT 34300 Applied Linear Statistical Methods.** This course introduces the theory, methods, and applications of fitting and interpreting multiple regression models. Topics include the examination of residuals, the transformation of data, strategies and criteria for the selection of a regression equation, nonlinear models, biases due to excluded variables and measurement error, and the use and interpretation of computer package regression programs. The theoretical basis of the methods, the relation to linear algebra, and the effects of violations of assumptions are studied. Techniques discussed are illustrated by examples involving both physical and social sciences data. *Autumn*

**OR**

**STAT 37790 Topics in Statistical Machine Learning** "Topics in Statistical Machine Learning" is a second graduate level course in machine learning, assuming students have had previous exposure to machine learning and statistical theory. The emphasis of the course is on statistical methodology, learning theory, and algorithms for large-scale, high dimensional data. The selection of topics is influenced by recent research results, and students can take the course in more than one quarter. *Autumn*

**OR**

**ECEV 32000 Introduction to Scientific Computing for Biologists.** The course will cover basic concepts in computing for an audience of biology graduate students. The students will receive basic training in the use of version control systems, databases and regular expressions. They will learn how to program in python and R and how to use R to produce publication-grade figures for their manuscripts, and how to typeset scientific manuscripts and theses using LaTeX. All the examples and exercises will be biologically motivated and will make use of real data. The approach will be hands-on, with lecturing followed by exercises in class. *Winter*

**OR**

**STAT 30900. Mathematical Computation I: Matrix Computation.** This is an introductory course on numerical linear algebra, which is quite different from linear algebra. We will be much less interested in algebraic results that follow from axiomatic definitions of fields and vector spaces but much more interested in analytic results that
hold only over the real and complex fields. The main objects of interest are real- or complex-valued matrices, which may come from differential operators, integral transforms, bilinear and quadratic forms, boundary and coboundary maps, Markov chains, correlations, DNA microarray measurements, movie ratings by viewers, friendship relations in social networks, etc. Numerical linear algebra provides the mathematical and algorithmic tools for analyzing these matrices. Topics covered: basic matrix decompositions LU, QR, SVD; Gaussian elimination and LU/LDU decompositions; backward error analysis, Gram-Schmidt orthogonalization and QR/complete orthogonal decompositions; solving linear systems, least squares, and total least squares problem; low-rank matrix approximations and matrix completion. We shall also include a brief overview of stationary and Krylov subspace iterative methods; eigenvalue and singular value problems; and sparse linear algebra.  

**OR**

**BIOS 20186 Fundamentals of Cell and Molecular Biology.** This course is an introduction to molecular and cellular biology that emphasizes the unity of cellular processes amongst all living organisms. Topics are the structure, function, and synthesis of nucleic acids and protein; structure and function of cell organelles and extracellular matrices; energetics; cell cycle; cells in tissues and cell-signaling; temporal organization and regulation of metabolism; regulation of gene expression; and altered cell functions in disease states.  

**Autumn**

**OR**

**BIOS 20187 Fundamentals of Genetics.** The goal of this course is to integrate recent developments in molecular genetics into the structure of classical genetics with an emphasis on recent advances in genetics and genomics. Topics include Mendelian inheritance, genotype-phenotype relationships, linkage analysis, modern gene mapping techniques, gene expression, model systems genetics and analysis of genetic pathways.  

**Autumn, Spring**

**OR**

**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.**

**OR**

**STAT 32950 Multivariate Statistical Analysis: Applications and Techniques.** This course focuses on applications and techniques for analysis of multivariate and high dimensional data. Beginning subjects cover principal component analysis, factor model, canonical correlation, multi-dimensional scaling, discriminant analysis, clustering, and common techniques of dimension reduction. Further topics on statistical learning for high dimensional data and complex structures include penalized regression models (LASSO, ridge, elastic net), sparse PCA, independent component analysis, Gaussian mixture model, and Expectation-Maximization methods. Theoretical derivations will be presented with emphasis on motivations, applications, and hands-on data analysis.  

**Spring**

**OR**

**ECEV 42900 Theoretical Ecology.** An introduction to mathematical modeling in ecology. The course will begin with linear growth and Lotka-Volterra models, and proceed to partial differential equations. The course's perspective will emphasize numerical computations and fitting models to data.  

**Winter**

**OR**

**STAT 24610 Pattern Recognition** This course treats statistical models and methods for pattern recognition and machine learning. Topics include a review of the multivariate normal distribution, graphical models, computational methods for inference in graphical models in particular the EM algorithm for mixture models and HMM's, and the sum-product algorithm. Linear discriminative analysis and other discriminative methods, such as decision trees and SVM's are covered as well.  

(Spring)
OR

STAT 30210 Bayesian Analysis and Principles of Statistics  This course continues the development of Mathematical Statistics, with an emphasis on Bayesian analysis and underlying principles of inference. Topics include Bayesian Inference and Computation, Frequentist Inference and interpretation of p values and confidence intervals, Decision theory, admissibility and Stein’s paradox, the Likelihood principle, Exchangeability and De Finetti’s theorem, hierarchical modelling, multiple comparisons and False Discovery Rates. The mathematical level will generally be at that of an easy advanced calculus course. We will assume familiarity with standard statistical distributions (e.g., Normal, Poisson, Binomial, Exponential), with the laws of probability, expectation, conditional expectation, etc., and exposure to common statistical concepts such as p values and confidence intervals. Familiarity with the R statistical language will also be expected, and homework assignments will include programming problems in R. (Spring)

OR

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

OR

STAT 37710 Machine Learning. This course provides hands-on experience with a range of contemporary machine learning algorithms, as well as an introduction to the theoretical aspects of the subject. Topics covered include: the PAC framework, Bayesian learning, graphical models, clustering, dimensionality reduction, kernel methods including SVMs, matrix completion, neural networks, and an introduction to statistical learning theory. Spring

For additional information please view the Doctoral Training in Computational Genomics website