Human Genetics Ph.D. Program Handbook

Rationale

Genetics is the study of variations in and transmission of hereditary material from generation to generation and how this information is translated into biological function. Genetics utilizes many methods ranging from molecular to population genetics techniques. Because of the fundamental impact of genetic variation on biological function, genetics has become a significant unifying theme for research in the biological and biomedical sciences and can serve as a focus for the study of virtually all biological processes and systems. Genetics plays an ever-increasing role in the elucidation of the cellular and molecular mechanisms of human disease and birth defects, and in their prevention, diagnosis, and therapy. In addition to using genetics to study biomedical questions posed by other fields, genetics encompasses an important set of intrinsic questions as to how the information content of a set of relatively simple molecules can be translated into complex systems and organisms, how variation at the molecular level can cause phenotypic differences among individuals, and how this variation within and among populations can be used to explain differences in disease prevalence.

In recent years, human genetics, as a major subfield of genetics, has contributed significantly to our understanding of disease processes. This explosive growth in our knowledge is an outcome of genetic analysis and the rapid technological advances fostered by the Human Genome Project and will continue to increase over the foreseeable future. The Ph.D. Program in Human Genetics (HGEN) has already enabled Vanderbilt University to become a key player in this explosion of knowledge, both nationally and internationally, by attracting and training the best students interested in human genetic research and serving as a focus for the recruitment of new faculty interested in training students in genetics.

The goal of the Ph.D. Program in HGEN is to encourage the training of students to explore questions motivated by genetic research in general and particularly as they apply to human disease. This curriculum will also teach students both within the program and in other disciplines how to use the tools of genetics to answer a variety of important biological questions. The program provides a cohesive program that unifies human genetic research at Vanderbilt and provides common direction and vision for investigators interested in training graduate students in genetics.

Training Objective

The overall goal of the HGEN Ph.D. degree program is to provide students with a solid foundation for a career in human genetics research and teaching. Training is available in genetic analysis of humans and model systems that contribute to our understanding of human disease. The training combines a required set of basic courses intended to ground students in the fundamentals of genetic analyses and the basics of human genetics, a set of elective courses designed to meet individual needs, and a rigorous research experience that will contribute to the field of genetics. Students completing the requirements of the Ph.D. program in HGEN will have demonstrated mastery of knowledge in genetics and contributed substantial and original scientific knowledge to the field.

The specific objectives are:

1. To provide graduate training and research on the genetic basis of human disease and variation
2. To provide students with an integrated and comprehensive academic curriculum designed for a concentrated program of study in human genetics
3. To ensure that the faculty will interact synergistically in their efforts to provide students with firsthand knowledge of emerging concepts and “state of the art” technology in the rapidly evolving field of human genetics

The Ph.D. program will provide students with options for the study of human genetics from several perspectives, ranging from population genetics and genetic epidemiology to molecular genetics/genomics of model organisms and disease states, and will build on the training that students receive in the admitting programs including the

Interdisciplinary Graduate Program in the Biomedical Sciences (IGP), the Quantitative and Chemical Biology Program (QCB), the Medical Scientist Training Program (MSTP), and directly admitted students.

It is important to note that the intent of the training program is not to train students who only use one or a few techniques of genetics to address a wide variety of biological questions, but to train students who address questions fundamental to the discipline of human genetics. These areas include: 1) the characterization and implications of genetic variation; 2) the transmission of genetic information within and across populations; 3) genomic structure and function; 4) how genetic variation is translated into phenotypes; and 5) the genetic basis of human disease and phenotype.

Relationship to the Interdisciplinary Graduate Program in the Biomedical Sciences (IGP) and Other Admitting Programs

Students may enter the HGEN Ph.D. Program from the IGP, QCB, and Medical Scientist Training Program (MSTP) programs. All graduate students entering these programs are admitted uncommitted to a specific department or Ph.D. program. Each umbrella program has its own initial course requirements, all of which are acceptable to the HGEN Ph.D. Program. Students will enter the HGEN Ph.D. Program at the end of the first year of the IGP or QCB programs. MSTP students will enter the HGEN Ph.D. Program at an equivalent time in their training.

Acceptance into the HGEN Ph.D. program depends on satisfactory performance (B or better in all classes) in the admitting program. Acceptance will also depend on the recommendation of a thesis advisor who is part of the HGEN program faculty.

Academic Curriculum for the Genetics Ph.D.

Ph.D. students in HGEN are required to complete a minimum of 29 credit hours of formal coursework (combined credits from required and elective courses). Students will take a minimum of 6 hours of didactic classes per semester during their first two years of study. It is expected that, during the second year, at least one semester will exceed this minimum to complete the required courses prior to year 3 of study. The electives will come from an approved list of advanced genetics courses (see below). In individual cases, other courses approved by the Director of Graduate Studies (DGS) and a student's committee can serve as electives. The choice of these courses will be based on the individual student's research interests. Other specific needs of students can be met with the electives, or in very rare cases, students with the support of their mentors can petition to replace **one** required course with another one suited to their research needs. Courses from other relevant programs (ex. Molecular Physiology & Biophysics, Biomedical Informatics, Biostatistics, and Epidemiology) may be taken for credit towards the HGEN Ph.D. program with permission from the HGEN DGS and the course instructor.

Course summaries are included in this document (Appendix I).

Required Courses for All Students

*Genetics Interest Group (GIG) (HGEN 8335) (1)
Human Genetics I (HGEN 8340) (3)
Tutorials in Human Genetics I (HGEN 8370) (1)
Human Genetics II (HGEN 8341) (3)
Tutorials in Human Genetics II (HGEN 8371) (1)

*Refer to Additional Requirements section for more information

Advanced Genetics Course Electives

Biobank Study Design (HGEN 8391) (3)
Practical Python Programming and Algorithms for Data Analysis (HGEN 8394) (3)

Academic Performance

All students must maintain an overall B (3.0) grade point average (GPA) in their didactic coursework. Student progress will be monitored by the DGS. If a student's GPA drops below 3.0, they will be placed on academic probation. If the GPA remains below 3.0 after a second semester, the Oversight Committee will evaluate the student's overall performance and may recommend either further remedial action or dismissal from the program. If the GPA remains below 3.0 for a third semester, the student will likely be dismissed from the program unless there are extraordinary circumstances. Continuation in the program would then require the unanimous decision of the Oversight Committee.

Any financial support received from training grants or the Ph.D. program is contingent upon maintaining an overall GPA of 3.0 and taking a full course load each semester.

The Dissertation Committee

The Dissertation Committee is composed of the advisor, at least two other members of the HGEN program faculty, and at least one faculty member from outside the HGEN program, who may be from a different institution. One of these members, other than the student's advisor, must be nominated as the Chair of the Committee. The Chair must be a HGEN faculty member. The DGS will serve as an *ex officio* member of the committee if they are not an official member of the committee. Thus, committees should include at least 4 faculty members, including the mentor but excluding *ex officio* members. A committee may include additional members if deemed appropriate by the student, research mentor, and the DGS. The student and dissertation advisor propose the composition of the Dissertation Committee the Graduate School who sends it to the HGEN DGS to approve. The student must also use the Kuali system for reporting Dissertation Committee Members to the Graduate School. Instructions for system use can be found in Appendix II.

The Dissertation Committee serves as a working team to help the student in several ways, including offering suggestions about experimental or analytical techniques and design and providing continual encouragement to be innovative and take risks – characteristics that are crucial to long-term success in research. The Dissertation Committee also serves as a resource to help guide the development of the student's career. However, the research mentor is primarily responsible for guidance of the student's research and training. It is important that the Dissertation Committee be carefully selected with consideration of the scientific training, intellectual interests, and research activities of each Committee member. The diversity of intellectual activity that will be present in a student's research project should be reflected in the composition of the Dissertation Committee. The Dissertation Committee is crucial to the trainee's research progress and professional advancement, and thus its composition should be based on sound scholarship and service to the student.

Ph.D. Qualifying Exam

Academic Requirements and Timeline

To qualify for candidacy, a student must complete all the required first year courses with an average GPA $\geq$ 3.0 and must be in good academic standing (GPA $\geq$ 3.0). The Qualifying Examination (QE) will be taken before or at approximately the beginning the third year of graduate school for those entering from the IGP or QCB programs, and the fourth year for MSTP students. Delays in scheduling the QE must be approved by the DGS.

Exam Overview

The QE has two components: a written proposal, and a 90-minute oral defense of the proposal. The proposal defense will be in front of a faculty committee (Dissertation Committee) that has expertise on the thesis topic and can adequately judge the student's knowledge of the topic and their ability to carry out the project.

The Dissertation Proposal

The written document is based on the format of an NIH F31 predoctoral fellowship application (parts and page limits here: tinyurl.com/yyegcs7r). However, the submitted document **only** needs **these** parts:

- **Project Summary/Abstract** (~ 400 words)
- **Specific Aims** (1-page limit. Usually similar to project summary but with more detail and the aims described more specifically)
- **Research Strategy** (6-page limit, **including** all figures, but **not** counting bibliography)
- **Bibliography** (no limit)

Process

The student must use the Kuali system for scheduling the QE with the Graduate School. Instructions for system use can be found in Appendix II.

The student must turn in the full, complete proposal to the examining committee at least **two weeks** prior to the student's exam date. All examining committee members will then indicate to the committee chair whether the proposal is sufficiently adequate so that the oral exam can proceed as scheduled. If two or more examining committee members indicate that the proposal is inadequate to such an extent that the exam cannot proceed, then the DGS should be informed and the student must revise the proposal and reschedule their exam.

The student's supervisor will not be present during the QE. A member of the HGEN program leadership must attend the exam, if not already in attendance as a member of the Dissertation Committee.

There will be brief time at the beginning of the meeting (15 minutes) where the student leaves the room for the examining committee to introduce themselves to one another and discuss the proposal. The student will present key details of their proposal, and the committee members will ask question for 90 minutes. At the end of the scheduled time, the student will leave the room, and the committee will confer to determine the result and fill out the SACS survey (15 minutes). The Chair will lead the proceedings and prepare the letter from the committee summarizing the results of the exam. The result of the exam will then be communicated to the student by the Chair and HGEN representative. Forms will then be signed and filed with the Graduate School.

Evaluation of Examinations

The overall aim of the QE is to ensure that the student has a sufficient knowledge of the proposed research and to judge if the proposed research has a reasonable chance of success in a time period appropriate for a PhD thesis. Students will have 2 attempts to pass the QE.

There will be three possible outcomes of the QE:

1. Pass
2. Conditional Pass – Specific conditions and time requirements to meet the conditions will be determined by each committee with approval of the DGS. Multiple serious deficiencies should be classed as a "Fail". Conditions should not be so onerous that they could not be reasonably completed within 1 month after the exam.
3. Fail

At the end of the exam, the committee will confer in private to reach consensus on the outcome. If consensus cannot be reached, a majority vote of the committee will decide the outcome.

In the case of failure, the examining committee, with approval of the DGS, will determine the date of the second examination. Failure to pass a second examination will result in dismissal from the Ph.D. program. In this case, a plan for a terminal master's degree may be developed.

On satisfactory completion of the examination, the student will be admitted to candidacy.

Training in the Responsible Conduct of Research (RCR)
It is essential that all scientific inquiry be performed in a responsible and ethical manner. To that end, all incoming graduate students at Vanderbilt are required to attend initial RCR training. To reinforce and enhance this training over the course of the Ph.D. investigations, the program both encourages and require additional documented RCR training. No student will be allowed to graduate without documented exposure to RCR training at least twice per year. Such training may take many different forms. A partial listing includes:

- Documented attendance at the annual BRET RCR symposium.
- Documentation in the summary letter of RCR discussions during thesis committee meetings
- Attendance at the mini-retreat and the full retreat, where RCR issues will be discussed

Additional Requirements
First-year students in the HGEN Ph.D. degree program will be required to obtain one hour of credit for attending GIG. The one hours of credit will be given at the end of two consecutive semesters of satisfactory attendance at GIG.

In addition to attending and participating in the discussions at GIG, all students are required to present their research progress at GIG once per year. The presentation will consider of a 15–40-minute talk depending on year in program, followed by questions from the audience. These experiences serve to train students in oral presentation of their research and to provide increased input from the Vanderbilt genetics community regarding the research project.

Participation in journal clubs available on campus which relate to the student's research project is strongly encouraged.

Trainees studying off campus
In rare circumstances, it may be necessary for a student to spend extended periods of time off-campus. This is acceptable, but extra care must be taken to maintain a high level of exposure to appropriate training experiences. In such a circumstance, the trainee and mentor must write a brief but detailed (~ one page) request for off-campus training. Requests will only be considered for trainees who have completed their first year of HGEN required courses and have passed their QE exam. The request should provide the following information:

- Reason for the request, including the location and length of time away from Vanderbilt
- Proof of good standing in the HGEN program and the Graduate School
- Details of the opportunities available for formal and informal training and experiences that are equivalent to those available at Vanderbilt including:
    - Regular seminars/discussion groups/journal clubs equivalent to GIG
    - Availability of other seminars and training opportunities
    - If necessary, arrangement for clinical rotation
- Method of documentation that these training experiences are being fulfilled
- Procedures to ensure that all the program requirements are being met (e.g. regular committee meetings, etc.)

The request must be approved by the DGS and Program Director before the trainee physically leaves Vanderbilt.

Length of Training
Students and advisors should aim for completion of graduate studies within a period of three years after passing their QE. Most students will be able to meet this expectation. All students are expected to graduate within six

years of matriculating as graduate students at Vanderbilt. If this time limit is unlikely to be met, the student will be required to submit a formal petition to the Oversight Committee to grant an extension. The petition must include an explanation for the inability to complete training within six years and a projected time for degree requirement completion. If an extension is recommended by the Oversight Committee, the DGS will petition the Dean of the Graduate School for the extension.

Thesis Defense

The Dissertation Committee is responsible for administering the final Ph.D. examination and will determine whether the candidate has presented an acceptable thesis. Thesis committee members are also strongly encouraged to provide written comments on the student's thesis research proposal at the first committee meeting. A summary of all committee meetings will be written by the student and approved by the committee. Copies of the summary will be sent to all committee members and the DGS and become a part of each student's permanent record. The Chair of the Dissertation Committee will inform the DGS in writing of the results of the final examination, including completion of any required revisions. If all other requirements are satisfied, the DGS will notify the Dean of the Graduate School that the student has completed the requirements for the Ph.D. degree.

Candidates for the Ph.D. degree in HGEN must present an acceptable dissertation that adds to or modifies what was previously known. The requirements of the Graduate School, as described in the Graduate School Bulletin, must be followed when preparing the thesis. Professional achievement must also be evident and should include the presentation of research work at a national meeting(s). Prior to the thesis defense the student should have at least one first authored publication (published or in press) in a peer reviewed scientific journal.

Students must schedule and report their defense to the Graduate School using the Kuali system. Instructions for system use can be found in Appendix II.

Faculty

Faculty with primary appointments at Vanderbilt and with an appointment in the Graduate School are eligible for appointment to the HGEN Ph.D. Program faculty. Faculty are chosen based on their area of research expertise and their willingness to train students in the prescribed program. The program Oversight Committee will review the Curriculum Vitae of all interested faculty and determine their eligibility based on criteria given below:

1. Area of research interest
2. An active research laboratory with peer-reviewed support, or start-up funds
3. A track record in training graduate students
    a. For early-career faculty, potential to train students effectively will be considered sufficient to become members of the training faculty. However, these faculty will be periodically counseled on the responsibility and duties of a thesis advisor
4. Active participation in Ph.D. related courses, seminars, or other program activities

Administrative Structure

The Ph.D. Program will be administered by the Program Director and the DGS in close consultation with an Oversight Committee. Additionally, the Oversight Committee will evaluate student program based on reports from the Dissertation Committees.

The DGS will have primary responsibility for overseeing all aspects of the Ph.D. program. They will do so with the help of the Oversight Committee. The Program Director and the DGS will be the official spokespersons for the Ph.D. program and will serve as its representatives in matters related to University policy and programs. The DGS will be responsible for the maintenance of high standards in the academic program, including the continuous evaluation of all required and elective courses in the program, the qualifications and diversity of the faculty. The DGS will initiate and coordinate recruitment activities and will be also responsible for identifying and applying for (or assisting others in applying for) internal and external support for graduate training in HGEN.

In addition, the DGS will be responsible for monitoring the progress of each student throughout their training. The DGS will be responsible for explaining the program requirements to the students and monitoring their performance in coursework. They will also serve as a student advocate when personal problems arise and in case of faculty irresponsibility or misconduct. Students will be encouraged to present issues regarding the program and/or courses directly to the DGS. However, they may also choose to do so through a representative of the Human Genetics Student Association (HGSA, described below). In cases where there is an issue with the DGS students should consult directly with the Program Director or a member of the Oversight Committee.

The Oversight Committee will also serve multiple roles. Included in its roles are:

1. An advisory committee on student-related issues, including monitoring student progress, performance, and welfare. However, it should be noted that the Committee will not provide the kind of individualized attention that the Dissertation Committee is required to do. The Committee's purpose instead is to make sure that the goals of the program are met in a more general sense
2. A committee on policy changes for the program. This will include the addition of and removal of courses from the program and the addition of tracks to the program. Changes made to the program will not be retroactive and students who entered the program prior to changes will follow the requirements that were in place at the time of their admission. In some cases (at the discretion of the Program Director, the DGS and the Oversight committee) students may have the option of choosing to follow new or old guidelines. Prior to any changes input from the entire faculty of the program will generally be solicited
3. Selection and evaluation of program faculty based on the criteria above.

The Oversight Committee will represent a broad spectrum of Ph.D. program faculty representing all areas of human genetics included in the training program and will be capable of advising the Program Director and the DGS on all program and research related topics pertinent to student training.

Human Genetics Student Association

The graduate students have formed a HGSA. The HGSA serves to formalize student input for internal and external matters, to promote communication among students of the program, and to promote and welcome potential students to the graduate program. The HGSA adopts its own by-laws and elects its own officers following these by-laws. It also appoints a representative to the Oversight Committee. The HGSA is also responsible for inviting one outside speaker to Vanderbilt each year. They identify which individuals to invite and make all arrangements for the visits with the help of the Program Director.

The roles and responsibilities within the HGSA include, but are not limited to:

- President
    - Open to current HGSA officers. In the case none of the current officers are interested in serving, this position will also be open to anyone.
    - Set goals for HGSA vision and impact
    - Attend monthly Graduate Student Council meetings and relay information to HGSA
    - Present at IGP/QCB recruiting events
    - Work with Vice President to coordinate invited speaker visit
    - Work with Social Chair to order swag
- Vice President
    - Chair of Curriculum Committee (see below)
    - Organize mock QEs for students
    - Report curriculum feedback to faculty representatives
    - Work with President to coordinate invited speaker visit
    - Update HGEN student handbook if necessary
- Secretary

    - Schedule and record minutes for HGSA meetings
    - Monitor and update HGSA budget
    - Work with Social chair to advertise social events
    - Work with President and Social Chair to order swag
- Social Chair/Committee
    - Organize social events
    - Work with secretary to advertise social events
    - Run social media page
    - Notify students regarding upcoming events
    - Work with President and Secretary to order swag
    - Communicate with Vanderbilt Genetics Institute FUN Committee and retreat committee
- Education Chair/Committee
    - Organize lunch and learn events
- Mentorship Chair
    - Develop and maintain system to connect older students to younger students as peer mentors

Human Genetics Student-led Curriculum Committee
The HGEN Curriculum Committee is composed of one Committee Chair and up to 3 members. The Chair of the Committee is also the Vice President of the HGSA. The Curriculum Committee's responsibilities include:

- Collecting and organizing course and QE feedback
- Monitor and disseminate information regarding curriculum/program requirements
- Updating catalog of course descriptions
- Organize mock QE for students preparing to take their exams
- Harmonizing information within the HGEN student handbook

Financial Support
Stipends and tuition allowances are awarded to students on the basis of academic merit. Stipend levels are set by the Biomedical Research and Training (BRET) Office in consultation with the department chairs. Stipends are considered taxable income.

If students are in the IGP, their first two semesters of support are provided by funds from the Graduate School. Following the IGP year, multiple avenues of support will be available. For United States citizens or permanent residents, training grant slots may be available, in which case most of the tuition is covered and the remained by tuition remission. For international students, support will come from research grants with tuition remission. Alternative sources of support include faculty research grants for all students and individual fellowships from extramural sources. Financial support for senior students (years 4 and above) will be the responsibility of the thesis advisor and the advisor's academic department. Financial support may be withdrawn from a student whose cumulative GPA is less than 3.0 at the end of two semesters or whose performance is deemed otherwise inadequate as described elsewhere in this document.

Appendix I.
Kuali System – See Attached Document

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