

# HUMAN GENETICS AND GENOMICS, PHD

## Ph.D. Program

The Johns Hopkins Human Genetics and Genomics Training Program provides training in all aspects of human genetics and genomics relevant to human biology, health and disease.

Advances in human genetics and genomics continue at an astounding rate and increasingly they are being integrated into medical practice. The Human Genetics and Genomics Program aims to educate highly motivated and capable students with the knowledge and experimental tools that will enable them to answer important questions at the interface between genetics and medicine. Ultimately, our trainees will be the leaders in delivering the promise of genetics to human health.

The overall objective of the Human Genetics and Genomics program is to provide our students with a strong foundation in basic science by exposure to a rigorous graduate education in genetics, genomics, molecular biology, cell biology, biochemistry and biostatistics as well as a core of medically-related courses selected to provide knowledge of human biology in health and disease.

This program is also offered as training for medical students in the combined MD/PhD program (<https://e-catalogue.jhu.edu/medicine/medical-students/md-phd-combined-degree/>). Students apply to the combined program at the time of application to the MD program.

## Research Facilities

Research laboratories are well equipped to carry out sophisticated research in all areas of genetics. The proximity to renowned clinical facilities of the Johns Hopkins Hospital, including the Department of Genetic Medicine, and Oncology Center provides, faculty and students with access to a wealth of material for study. Our computer, data storage and library facilities are excellent. Laboratories involved in the Human Genetics and Genomics Program span Johns Hopkins University; consequently, supporting facilities are extensive.

## Financial Aid

The program is supported by a training grant from the National Institute of General Medical Sciences. These fellowships, which are restricted to United States citizens and United States permanent residents, cover tuition, healthcare insurance and a stipend during year one. For non-training grant eligible students, tuition and healthcare is also covered. Once a student has joined a thesis lab, all financial responsibilities belong to the mentor and are covered until completion of studies. Students are encouraged, however, to apply for fellowships from outside sources (e.g., the National Science Foundation, Fulbright Scholars Program, Howard Hughes Medical Institute) before entering the program.

## Vivien Thomas PhD Scholars at JHU

The Vivien Thomas Scholars Initiative (VTSI) is an endowed fellowship program at Johns Hopkins for PhD students in STEM fields. It provides full tuition, stipend, and benefits while also providing targeted mentoring, networking, community, and professional development opportunities. Students who have attended eligible institutions, including historically Black colleges and universities (HBCUs), community colleges, and regional institutions in Maryland, Virginia, and the District of Columbia

for undergraduate study, are eligible to apply. More information about the VTSI program is available at this link: <https://provost.jhu.edu/about/vivien-thomas-scholars-initiative/>. To be considered for the VTSI, all application and supplementary materials must be received by December 1st.

## Admission

Applicants for admission should show a strong academic foundation with coursework in biology, chemistry and quantitative analysis. Applicants are encouraged to have exposure to lab research or to data science. A bachelor's degree from a qualified college or university will be required for matriculation. GREs are no longer required.

The Human Genetics and Genomics site (<https://www.hopkinsmedicine.org/institute-genetic-medicine/education-training/predoctoral-human-genetics/>) has up-to-date information on "How to Apply (<https://www.hopkinsmedicine.org/som/education-programs/graduate-programs/admissions/>)." For questions not addressed on these pages, please access the contact information listed on the program page: Human Genetics and Genomics Training Program | Johns Hopkins Department of Genetic Medicine (<https://www.hopkinsmedicine.org/institute-genetic-medicine/education-training/predoctoral-human-genetics/>).

## Program Requirements

The program's course requirements are listed below.

Additionally, the program requires the "OPTIONS" Career Curriculum offered by the Doctoral Life Design Studio (DLDS). OPTIONS is designed to provide trainees with the skills for career building and the opportunity for career exploration as well as professional development training.

Human Genetics and Genomics trainees also take a two-week course in July at the Jackson Labs in Bar Harbor, Maine entitled "Human and Mammalian Genetics and Genomics: The McKusick Short Course" which covers a variety of topics from basic principles to the latest developments in mammalian genetics. The faculty numbers about 50 and consists roughly in thirds of JAX faculty, Hopkins faculty and "guest" faculty comprising outstanding mammalian geneticists from other US universities and around the world.

Trainees must complete three research rotations before deciding on their thesis lab. Presentations are given after the first and third rotation. Additionally, students must fulfill an ethics requirement by (1) completing the Introduction to Responsible Conduct of Research course in their first year (listed in the course table below) and (2) starting in year 3, attending at least two Research Integrity Colloquia lectures per year.

During their fourth year, students present their work to other students, faculty and staff at our Monthly Science and Pizza meeting.

Our trainees participate in weekly journal clubs and seminars in the Department of Genetic Medicine as well as others throughout the School of Medicine.

At the end of the second year, trainees take their Doctoral Board Oral Examination. Annual thesis committee meetings must be held following successful completion of this exam. For the first committee meeting, the student must prepare a five page NIH formatted proposal; for all subsequent meetings, a progress report must be submitted. These are sent to the committee members prior to the meeting. Also prior to the meeting, the student must complete an Individual Development Plan (IDP). The form includes questions on the student's research and

progress, professional development, mentoring relationship with their advisor, overall lab/research team environment and an evaluation of competencies. At this time, the advisor also completes their form. They will then discuss both reports. Their action plan is shared with their thesis committee members.

All students are required to TA for one School of Medicine course (at least one quarter) during year three. These courses include all courses required of the HGG program as well as courses offered by other programs/ departments.

Our students must take a minimum of four electives, one of which must provide computational/statistical training. These elective courses may be from any program, if relevant to thesis work and must be approved in advance by program directors.

Average time for completion is 5.5 years.

## Core Courses

Code	Title	Credits
ME.710.700	Advanced Topics in Human Genetics	1.5
ME.710.748	Introduction to Rigor and Reproducibility in Research	1.5
ME.710.745	Evolving Concepts of the Gene	5
ME.800.811	Introduction to Responsible Conduct of Research	1
ME.710.746	Human Genetics Boot Camp	2
ME.110.728	Cell Structure and Dynamics	1.5
ME.260.709	Molecular Biology and Genomics	1.5
ME.710.802	Research Rotations <sup>1</sup>	1-18
ME.710.800	Independent Research <sup>2</sup>	1 - 18
ME.710.747	Systems, genes and mechanisms in disease	3
ME.710.744	Genomic Technologies: Tools for Illuminating Biology and Dissecting Disease	1.5
ME.710.740	Understanding Genetic Disease	0.5
ME.360.728	Pathways and Regulation	2

<sup>1</sup> Repeat year 1 in fall, spring, and summer.

<sup>2</sup> Repeat year 2 and above in fall, spring, and summer.

## Learning Outcomes

Graduates from the Human Genetics and Genomics program pursue careers in academia, medicine, industry, teaching, government, law, as well the private sector. Our trainees are encouraged to explore the full spectrum of professional venues in which their training may provide a strong foundation. Driven by curiosity and a desire for excellence, our trainees stand out as leaders in the chosen arenas of professional life. They are supported in the development of their career plans by a program faculty and administration who are dedicated to their success, and by a myriad of support networks across the Johns Hopkins University, many of which are provided by the Professional Development Career Office of the School of Medicine.

## Core competencies in Human Genetics and Genomics

### 1. Basic knowledge of human genetics and genomics principles

All graduates should be able to:

- Demonstrate robust understanding of fundamental principles in Human Genetics and Genomics.
- Demonstrate understanding of modes of phenotypic inheritance, including the basic principles of Mendelian and Multifactorial inheritance.
- Understand the strategies utilized and values of identifying the genetic variants and genes responsible for genetic disease.
- Explain the differences between and consequences of Somatic and Germline variation.
- Explain genetic mosaicism and its role in biology and disease.
- Demonstrate a fundamental understanding of heritability and how it can be calculated.
- Demonstrate a fundamental understanding of variation types; how they arise; and their potential roles in modifying (or not) biological function/disease risk.
- Describe the composition of the human genome (coding versus noncoding) and how variation in these components and their epigenetic marks can contribute to individuality and impact disease risk.
- Describe the fundamental principles and differences between pol1, pol2, and pol 3 genes and their transcription.
- Explain how our understanding of gene structure and regulation of gene expression has evolved over time.
- Identify and discuss common disorders of major human organ systems, and the potential role of genetic variation in each.
- Grasp the fundamental approaches used in clinical genetic analyses to define inheritance patterns, seek to identify causative and validate identified variation.
- Grasp the fundamental approaches used in population genetics to define the role of common variation in common disease risk, and the implicit challenges in connecting variation to biological mechanisms.
- Demonstrate familiarity with common genomic methodologies.
- Explain biochemical and cell biological processes important for function.

### 2. Critical thinking

All graduates should be able to:

- Analyze primary literature and identify strengths and shortcomings of the methods employed.
- Construct testable hypotheses and design experiments to challenge these hypotheses.
- Identify orthogonal approaches that can strengthen their conclusions.

### 3. Quantitative analysis

All graduates should be able to:

- Appropriately process and apply analytical techniques and statistical tests to their data.

- Access and utilize large biobanks containing genotypic and phenotypic data to formulate and answer questions regarding the genetic contribution to phenotypes.
- Determine the power of their analyses and the technical and biological replication required for statistical significance.
- Create figures that effectively communicate results.
- Critically evaluate and interpret quantitative data.
- Identify and use computational tools in their research.
- Appropriately document and store corresponding versions of code generated/edited in the process of their studies.

#### 4. Effective written/oral communication skills

All graduates should be able to:

- Organize their oral and written scientific communications to effectively transmit: 1. The significance of topic, 2. The relevant background material to place the topic in context, 3. The knowledge gap to be addressed, including the manner and the appropriateness of the strategies chosen to address the knowledge gap, 5. Define what they expect of successful completion of the experiment independent of the observations.
- Orally present scientific material in a clear and effective manner.
- Articulate contributions to their work by others.

#### 5. Career-specific skills

All students should have the opportunity to develop additional skills required to translate their PhD training in Human Genetics and Genomics into a successful career. For example: Biotechnology/Pharmaceutical internships; curriculum design for teachers; or financial analysis/patent law internships for consulting. Students will develop a plan with their mentor and thesis advisory committee based on their Individual Development Plan and through the OPTIONS program career curriculum (required in the HGG program) and other programs offered by the Doctoral Life Design Studio, as well as other available resources.

#### 6. Self-motivated learning/scientific inquiry

All graduates should be able to:

- Independently explore and assimilate existing literature in their field of interest.
- Identify and engage expert guidance when needed.

#### 7. Discipline-specific research skills

All graduates should be able to:

- Develop hypothesis driven research questions founded on their current studies.
- Conduct discipline-specific experimental techniques with appropriate controls and analysis.
- Troubleshoot and solve emergent problems.
- Explain how their studies may impact human health.

#### 8. Citizenship

All graduates should demonstrate:

- A fundamental understanding of research ethics.
- The ability to work collaboratively with others.
- Leadership skills necessary to effectively train and supervise others.

#### 9. Completion of a PhD quality thesis

Unifying all theses is the expectation that the thesis assembled by the graduate should report work of sufficient quantity and quality to constitute a significant contribution to the field of genetics. Therein, it should:

- Formulate a robust Problem and Research Plan and pose an insightful question.
- It should describe and employ Material and Methods which are appropriate to address the question and reflect implementation by the candidate.
- It must reflect the personal contribution by the candidate that is consistent with completion of a doctoral thesis and acknowledge the contribution of others to their work (where appropriate).
- The findings and results must be robustly generated and analyzed and constitute a significant contribution to the field.
- The candidate must provide a well-rounded discussion/conclusion of their work, placing their observations in the context of the field.