

# GENETIC COUNSELING (GENC)

## **GENC 6010 Advanced Genetics and Genomics**

This course will provide an extensive survey of molecular genetics including molecular and cellular biology and the regulation of gene expression. Applications to human clinical genetics and genetic counseling through discussion of relevant case studies will be incorporated. The class will be primarily lecture-based with some small group work. In addition to exams, students will prepare and present a teaching tool to describe a complex topic learned in the course to individuals with limited knowledge of molecular genetics. The content of this course will provide the foundation for understanding the detailed topics covered in additional courses in the genetic counseling master's program including Foundations in Clinical Genetics and Genomic Technologies, Cancer Genetics, Reproductive and Developmental Genetics, and Medical Genetics.

Fall

1 Course Unit

## **GENC 6020 Mechanisms of Disease**

Primary concepts in this course introduce the fundamental principles of pathophysiology, including normal and abnormal embryological development. Pathophysiology and development are covered by major organ systems. Lectures and class exercises will review normal as well as abnormal physiology in all systems. The focus of the course will be on understanding the pathophysiology of human genetic diseases. The coursework serves as the background for understanding specific diseases as they present in clinical genetics and research that has led to improved therapy and treatment for patients. The class will be primarily lecture-based using case-based learning. Students will give formal presentations on the pathophysiology of specific disorders.

Fall

1 Course Unit

## **GENC 6030 Introduction to Genetic Counseling**

Genetic counseling students will be oriented to the profession through a review of the history of genetic counseling and basic tenets of the field. Topics covered include medical terminology, pedigree construction, the structure of genetic counseling sessions, discussion of the scope of practice and disability rights, instruction in obtaining family pedigrees, review of genetic testing practices, and the National Society of Genetic Counselors' Code of Ethics. Students will gain an appreciation for the lived experience of individuals with genetic diseases by reading memoirs, reviewing online support groups, watching videos, and performing role plays. The course will provide experiential learning about advocacy groups for patients and their families through first-hand interviews. Students will engage in role play scenarios throughout the course.

Fall

1 Course Unit

## **GENC 6040 Reproductive and Developmental Genetics**

A variety of topics in reproductive genetics will be covered in this course, including a review of human development emphasizing the genetic basis of these complex events and how teratogenic factors lead to abnormal development. The common indications for prenatal genetic counseling, such as advanced maternal age, first-trimester screening, NIPS, ultrasound anomalies, diagnostic procedures, prenatal complications, and prenatal therapies will be reviewed. Through the completion of assignments and discussion, students will demonstrate their abilities to think critically through cases, assess risk, and provide patient-centered care. The course will discuss psychosocial issues surrounding fertility counseling as well as perinatal bereavement and palliative care.

Spring

1 Course Unit

## **GENC 6110 Cancer Genetics**

In this course, students will gain an understanding of cancer genetics. Topics covered include an overview of cancer biology and clinical oncology; diagnostics and predictive testing in cancer, including familial mutations in BRCA 1 and 2, and panel testing; cancer risk assessment; psychosocial aspects of cancer genetic testing and counseling. Students will examine specific inherited disorders predisposing individuals to malignancies and counseling for familial cancers. Students will learn how to conduct a genetic counseling session for inherited cancer syndromes such as HBOC and Lynch syndrome and to identify when genetic testing panels are indicated. In addition, the course will cover somatic cancer NGS technology and its relevance in cancer genetic counseling.

Spring

1 Course Unit

## **GENC 6120 Genetic Counseling Theory and Practice I**

Students will learn how to structure genetic counseling sessions including the competencies of establishing rapport, contracting, and eliciting patient goals. The teaching of active listening and interviewing skills guides students in responding empathically to patient concerns. Students use role play to practice skills and demonstrate their ability to organize a genetic counseling session. Students will learn chart documentation and practice letter writing. Each student will be responsible for presenting a Genetic Counseling Practice Guideline and leading a class discussion. Through role play and exploration of current practice students learn to adapt genetic counseling skills for alternate service delivery models. Students will gain an appreciation of appropriate professional interactions and expand their understanding of the impact of genetic disease on families. At the end of the semester, all students complete a genetic counseling session with a standardized patient at the Experiential Learning and Assessment Center at the Perelman School of Medicine. The highly skilled staff ensures safe, measurable, and authentic learning with qualified standardized patients.

Spring

1 Course Unit

**GENC 6130 Foundations of Clinical Genetics and Genomic Technologies**

This course introduces the key concepts and skills required for a genetic counselor in both the genetics clinic and the clinical genetics laboratory. In this course, students will understand the importance of collaborative efforts by clinical teams in the clinic and laboratory that lead to identification of the correct genetic diagnosis for a patient. Students will learn the principles of pedigree review, particularly how to apply Mendelian genetics, as well as the approach to clinical evaluation of a patient that leads to proper genetic test selection. Case-based learning will help students understand the complexities of genetic diagnoses and the importance of obtaining an accurate medical, surgical, prenatal, developmental, family, and social history, as well as how to write a standard chart note. They will also learn how genetic and molecular principles contribute to the etiology, clinical features, and expression of genetic conditions. They will appreciate how the natural history and variable expressivity of select genetic diagnoses influence differential diagnoses, interpretation of dysmorphology, choices of genetic testing, and test report interpretation. Common genetic testing methodologies and associated limitations will be introduced, particularly the concepts and tools used to analyze and interpret data from genomic level next-generation sequencing (NGS). Working in groups, students will analyze a simulated clinical NGS case, which will allow them to experience the process of clinical correlation, variant interpretation, and results reporting and provide hands-on opportunities to utilize relevant resources to systemically review and classify variants. Ethical principles that guide clinical care and genetic testing will be highlighted and incorporated throughout the course.

Fall

1 Course Unit

**GENC 6140 Introduction to Genetic Counseling Research**

This course will equip students to begin a genetic counseling thesis project by the summer term. Students will be exposed to research methodologies and project management skills that are necessary to successfully complete the thesis requirement of the master's program. This course will introduce students to scientific writing, critical interpretation of scientific literature, and a range of qualitative and quantitative research methods. Topics covered include identifying literature gaps, developing study aims, establishing a study timeline, IRB submission, participant recruitment, data collection and analysis, time management, writing, and manuscript submission. Students will be required to obtain CITI training and to establish a project proposal and timeline in concert with their primary thesis advisor and program mentor. This course includes lectures, group work, and assignments that support project initiation and timely manuscript development. Most class days are structured with a lecture followed by a workshop; students are expected to come with tasks and questions prepared to make the most of workshop time.

Spring

1 Course Unit

**GENC 6200 Medical Genetics I**

The first semester of this year-long course presents a broad overview of concepts in Medical Genetics including natural history, management and counseling strategies for major pediatric and adult genetics diagnoses and syndromes, focusing on Genomic Medicine and Pediatric Genetics. Topics covered include autism, ophthalmogenetics, hearing loss, psychiatric genetics, genodermatoses, immune genetics, pediatric cancer genetics, bone marrow failure syndromes, differences of sex development, craniofacial disorders, skeletal dysplasias and imprinting disorders. Students will be exposed to future directions of clinical genetics and genetic counseling and its impact on the management and treatment of common conditions. Students will actively engage in Clinical Case Presentations and a Journal Club session. Course directors will attend each class, and guest lecturers with relevant clinical expertise will be invited to share their clinical knowledge.

Fall

0.5 Course Units

**GENC 6210 Genetic Counseling Theory and Practice II**

This class teaches students the basic tenets of counseling theory and how they can be applied to genetic counseling sessions. Theories covered include person-centered counseling emphasizing genuineness, empathy, and positive regard. Narrative Medicine will be covered in detail, and students will be required to digitally record a narrative medicine interview with an individual or parent of an individual with a genetic disease. The recordings are analyzed and reviewed. Standardized patient assessments will be performed to monitor students' acquisition of more advanced counseling skills. Students will learn the integration of process in the genetic counseling session and how to address sensitive patient issues such as sharing challenging news and working with angry patients. They will learn to use active listening and value-free communication to manage a genetic counseling session in a culturally responsive manner.

Fall

1 Course Unit

**GENC 6220 Biochemical Genetics**

This course presents a broad overview of concepts of inborn errors of metabolism as well as detailed reviews of specific conditions so that students will learn to distinguish the basic descriptors of a wide variety of metabolic conditions, including cardinal features, biochemical signatures, genetic etiologies, inheritance patterns, and available treatments. Students will also understand the basis for, and implications of, newborn screening for both metabolic and other disorders. Students will learn the methods available for diagnosis, genetic and tissue-based testing, and the various forms of treatment for the disorder, from nutrition therapy and transplants to clinical trials. Approaches to appropriately counsel families with a wide range of metabolic conditions and to identify at-risk family members are included in this course. Course directors will attend each class, and guest lecturers with relevant clinical expertise will be invited to share their clinical knowledge.

Fall

0.5 Course Units

**GENC 6230 Ethical Issues in Genetic Counseling**

In this introductory course, we will review ethical, legal and social issues that arise within the field of genetic counseling. This course is designed to educate students about bio-ethical frameworks and how to construct a moral argument. Lectures will focus on providing background information that applies to specific sub-specialties within genetics such as reproductive, rare diseases, and research. Further, we will analyze challenging scenarios to develop ethical reasoning skills, with a focus on practical solutions that integrate legal and social considerations. Class time will include didactic lectures with extensive discussion, in addition to assignments that apply the concepts reviewed in class.

Fall

1 Course Unit

**GENC 6300 Medical Genetics II**

The second semester of this year-long course presents a broad overview of concepts in medical genetics including natural history, management and counseling strategies for major pediatric and adult genetics diagnoses and syndromes. Topics covered this semester include single gene disorders, bleeding and clotting disorders, hemoglobinopathies, adult liver disease, connective tissue disorders, cardiovascular genetics, psychiatric genetics, neurogenetic conditions such as epilepsy, movement disorders, ataxias, Huntington disease, and ALS, as well as classic genetic conditions identified by the organ system involved, such as cystic fibrosis. Students will learn the methods available for genetic testing, diagnosis, and treatment of genetic conditions. Students will be exposed to future directions of clinical genetics and genetic counseling and its impact on the management and treatment of common conditions. Therapeutic approaches and different modalities to treating genetic diagnoses will be reviewed, including approved gene therapy treatments as well as investigational therapies and clinical trials. Students will actively engage in Clinical Case Presentations and a Clinical Research Presentation. Course directors will attend each class, and guest lecturers with relevant clinical expertise will be invited to share their clinical knowledge.

Spring

Prerequisite: GENC 6200

1 Course Unit

**GENC 6310 Genetic Counseling Theory and Practice III**

This course provides students with advanced counseling skills such as advanced empathy and confrontation. Students will explore counseling theories such as solution-focused brief therapy, cognitive behavioral therapy, and family systems models. Counselor issues such as self-disclosure and countertransference will be discussed in detail. Additional skills and techniques from the counseling literature will be practiced in role plays to illustrate the usefulness of various counseling techniques in clinical genetics. This course includes an experiential component for practicing techniques utilizing standardized patients. Students will analyze their standardized patient recordings and identify missed opportunities and areas where they need to focus their counseling skills. Each student will analyze one of their clinical cases utilizing at least two theories discussed in class.

Spring

1 Course Unit

**GENC 6400 Professional Development I**

The goal of this seminar is to increase awareness of the many facets of genetic counseling as a career. The genetic counseling profession is rapidly evolving as new diagnostic technologies and treatments are developed. Skills learned by genetic counselors are applicable to many different job settings. Knowledge of these alternative ways in which counselors may work increases a student's ability to successfully pursue a career. Members of the genetics community from Philadelphia and the surrounding area provide lectures describing the current state of the profession. Selected topics include genetic counseling for a diverse population, the intersection between palliative care and genetic counseling, and how to manage a clinical genetic counseling practice with issues surrounding billing reimbursement and credentialing. This seminar also helps prepare students for the demands of the genetic counseling profession by focusing on emotional well-being through a process group facilitated by a professional counselor. Students prepare a career development plan reflecting on the essential components of an "ideal job" and develop a curriculum vitae and cover letters.

Fall

0 Course Units

**GENC 6450 Professional Development**

This course aims to increase awareness of the many facets of genetic counseling as a career. The students learn how to obtain a license and register to take the American Board of Genetic Counseling Certification Examination. Members of the genetics community from Philadelphia and the surrounding area provide lectures describing the profession's current state. Students explore professional issues by reviewing the National Society of Genetic Counselors' position statements and legislative initiatives. Students will prepare for the demands of the genetic counseling profession by focusing on emotional well-being through a process group facilitated by a professional counselor. Students present in journal club on a paper discussing current genetic counseling trends. During the semester, students write Op-Ed pieces exploring contemporary issues that may impact patient care or the genetic counseling profession, identify community resources for adults with neurodevelopmental disabilities, prepare abstracts of their thesis research for submission to a conference and learn test taking strategies for the ABGC certification exam.

Spring

0 Course Units

**GENC 6500 Genetic Counseling Rounds I**

Once a week, students participate in discussion of clinical cases and recent journal literature. Through their participation, students gain familiarity with the process of reviewing clinical material and providing peer supervision. Presenting clinical and scientific information for group discussion in conferences and patient rounds is an important skill for genetic counselors. This seminar provides these experiences in a supportive environment and thus permits skill building in this area. This activity introduces students to a broad range of diagnoses and methods of managing a case. Discussions of case organization and tailoring a presentation to a specific patient is another aspect of this seminar.

Fall

0 Course Units

**GENC 6550 Genetic Counseling Rounds II**

In this continuation of GENC 6500, students participate in weekly discussions of clinical cases and recent journal literature. Through their participation, students gain familiarity with the process of reviewing clinical material and providing peer supervision. Presenting clinical and scientific information for group discussion in conferences and patient rounds is an important skill for genetic counselors. This seminar provides these experiences in a supportive environment and thus permits skill building in this area. This activity introduces the students to a broad range of diagnoses and methods of managing a case. Discussions of case organization and tailoring a presentation to a specific patient is another aspect of this seminar.

Spring

Prerequisite: GENC 6500

0 Course Units

**GENC 6700 Internship**

In addition to course-based degree requirements, students participate in internships related to the field of genetic counseling. These internships begin in the students' first term. Internships give students opportunities to learn in varying practice-based environments including clinical and laboratory settings, as well as placements in a research and/or advocacy capacity with a specific organization or research protocol. Students document their experiences online daily and provide written summaries of their experiences for the program leadership to review. Students attend their internships once a week. Clinical supervisors evaluate the students in each of the placements.

0 Course Units

**GENC 6701 Internship**

In addition to course-based degree requirements, students participate in internships related to the field of genetic counseling. These internships begin in the students' first term. Internships give students opportunities to learn in varying practice-based environments including clinical and laboratory settings, as well as placements in a research and/or advocacy capacity with a specific organization or research protocol. Students document their experiences online daily and provide written summaries of their experiences for the program leadership to review. Students attend their internships once a week. Clinical supervisors evaluate the students in each of the placements.

1 Course Unit

**GENC 6800 Clinical Internship**

The clinical internships during the second year consist of four clinical placements in genetics clinics. Students will have a minimum of 20 days in each specialty. This experiential course will expose genetic counseling students to the genetic counselors' role in different clinical settings and to the broad range of diagnoses and methods of effectively managing patient care. Clinical supervision is provided in a supportive environment that permits skill building as students take on more roles throughout the course of the year. Students will have exposure to cases representing the wide range of diagnoses and indications seen in a clinical genetics practice and will collect at least 50 participatory encounters to satisfy the ACGC requirement. Certified genetic counselors will supervise all students for cases to be acceptable for the core case log book. Students will log each case for program review, including self-reflection about their performance. All students will be evaluated by their clinical supervisors and will prepare case analyses for the program leadership to review. The course is required for all second-year students.

Fall, Spring, and Summer Terms

Prerequisite: GENC 6700

0 Course Units

**GENC 7101 Research Design and Methodologies**

This course focuses on study design and proposal development of research projects relevant to clinical genetics and genetic counseling, encompassing translational, clinical, and behavioral projects. Covered topics include identifying a research topic, aims, study design, background, methods, quantitative and qualitative methods, biostatistics, and proposal writing. IRB submission and feasibility issues will also be addressed. This course will include online asynchronous lectures, online group discussions, weekly readings, and a draft of the research proposal as the final assignment. In addition, there will be synchronous weekly hour-long online group meetings to discuss research progress. Students will implement course lessons by developing and executing a research thesis project during the one-year program.

Fall

1 Course Unit

**GENC 7102 Ethical, Legal, and Social Implications of Genetics**

This course acquaints students with the long-standing field of research on the ethical, legal, and social implications (ELSI) of genetics and genomics. The course highlights NHGRI's ELSI program and guides robust discussion of contemporary ELSI topics. National experts offer lectures on race and ancestry in relation to biomedical research and practice, disability perspectives on genetics, public health genomics, and research bioethics with an emphasis on biobanks. Students are expected to complete reading assignments, case analyses, and participate in online discussion. Synchronous sessions will be held occasionally throughout the course.

Fall

1 Course Unit

**GENC 7103 Manuscript & Grant Writing**

This comprehensive course will focus on the fundamentals of preparing a manuscript for publication and writing and submitting grant proposals. Manuscript writing training will include scientific writing skills, crafting a narrative, writing an effective abstract, introduction compared to discussion sections, data visualization techniques, how to identify the appropriate journal for submission, guidance for authorship, and details about article submission, the peer review process, revisions, and response to reviewers and editors. Grant writing training will include the mechanics of proposal writing and aspects of "grantmanship" as students identify appropriate sources of grant funding, use narrative tools to create informative, persuasive and engaging proposals, creation of Aims that outline the goals of the grant, and support of Aims with well thought out research design. Grant submission process such as creation of facility and budget sections will be discussed. This course will include online asynchronous lectures, online group discussions, weekly readings, and a final draft of the research manuscript as the final assignment. There will be synchronous weekly hour-long online group meetings to discuss research progress.

Spring

1 Course Unit

**GENC 7104 Biomedical Informatics for Genetic Counselors**

In this course, students will develop a broad understanding of the field of biomedical informatics. The field of Biomedical Informatics (BMI) considers a variety of approaches and methods drawn from the computational, quantitative, and qualitative sciences. This introductory course provides an introduction to many research topic areas for the field with emphasis on the multidisciplinary nature of biomedical informatics.

1 Course Unit

**GENC 9900 Thesis**

This is an independent study course with a strong emphasis on project and time management. Students develop a project timeline and complete project tasks such as collecting data, analyzing data, presenting research progress, and composing a final manuscript, typically for submission to an academic journal. Students are responsible for meeting regularly with their primary thesis mentor and program advisor to complete the project and associated manuscript in a timely fashion. Course requirements include reporting on regular study meetings, meeting interim deadlines for partial and full manuscript drafts, and making use of resources supplied in the associated Canvas community. Students are also expected to submit an abstract for a professional meeting, poster presentation at an annual symposium, and a platform presentation to members of the genetic counseling program and broader community. Students are strongly encouraged to submit their manuscript for publication and are required to develop a clear publication plan and study materials deposit with their primary mentors before graduating.

Spring

1 Course Unit