# HUMAN AND MOLECULAR GENETICS (HGEN)

#### HGEN 501. Introduction to Human Genetics. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Enrollment by undergraduates students requires permission of instructor. Basic knowledge of genetics is recommended. Provides a comprehensive examination of the fundamentals of human genetics. Explores topics including Mendelian and non-Mendelian inheritance, pedigree analysis, cytogenetics, aneuploid syndromes, cancer, gene structure and function, epigenetics, gene expression, biochemical genetics, and inborn errors of metabolism.

#### HGEN 502. Advanced Human Genetics. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Prerequisite: HGEN 501 or equivalent. Enrollment restricted to graduate students. A comprehensive study of the principles of specific areas in human genetics. Explores topics including quantitative genetics, genetic epidemiology, gene mapping, animal models, the characterization of complex disease, diagnostic testing and genetic counseling.

## HGEN 510. Classic Papers in Human Genetics. 1 Hour.

Semester course; 1 lecture hour. 1 credit. Enrollment restricted to graduate students in the School of Medicine. This course surveys the seminal discoveries in the discipline of human genetics and introduces students to reading, understanding, discussing, critiquing and presenting original journal articles.

#### HGEN 511. Human Cytogenetics. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Prerequisite: HGEN 501. A discussion of recent advances in human cytogenetics. Topics covered will include chromosome banding techniques and ultrastructure, meiosis, numerical and structural abnormalities, fragile sites, cancer cytogenetics, methodology for linkage studies, and population cytogenetics. Clinical cases are used to illustrate the application of special diagnostic methodologies.

# HGEN 517. Introduction to R Programming for Statistical Genetics. 1 Hour.

Semester course; 1 lecture hour. 1 credit. Open only to graduate students or by permission of course director. This course is to provide and introduction to statistical programming in R. Lectures will provide the fundamentals for efficient handling and exploration of common data set structures in genetic and biomedical sciences.

#### HGEN 525. Practice of Genetic Counseling. 3 Hours.

Continuous courses; 3 lecture hours. 3-3 credits. Enrollment restricted to genetic counseling master's students. Provides context for practice of genetic counseling through literature review and practical techniques. Places specific emphasis on pregnancy and childhood evaluation, interviewing techniques, social and ethical issues, including fieldwork in prenatal, general genetics and specialty clinics.

# HGEN 526. Practice of Genetic Counseling. 3 Hours.

Continuous courses; 3 lecture hours. 3-3 credits. Enrollment restricted to genetic counseling master's students. Provides context for practice of genetic counseling through literature review and practical techniques. Places specific emphasis on pregnancy and childhood evaluation, interviewing techniques, social and ethical issues, including fieldwork in prenatal, general genetics and specialty clinics.

# HGEN 527. Medical Genetics. 3 Hours.

Continuous courses; 3 lecture hours. 3-3 credits. Prerequisite: HGEN 525-526 or permission of instructor. Enrollment restricted to genetic counseling master's students. Provides medical information and principles of human genetic disease with specific emphasis on the molecular basis of Mendelian disorders, disorders of sexual development, assessment of dysmorphic features, and the genetics of common diseases. Emphasizes the use of all available resource materials in genetics.

#### HGEN 528. Medical Genetics. 3 Hours.

Continuous courses; 3 lecture hours. 3-3 credits. Prerequisite: HGEN 525-526 or permission of instructor. Enrollment restricted to genetic counseling master's students. Provides medical information and principles of human genetic disease with specific emphasis on the molecular basis of Mendelian disorders, disorders of sexual development, assessment of dysmorphic features, and the genetics of common diseases. Emphasizes the use of all available resource materials in genetics.

#### HGEN 600. Clinical Genetics. 3 Hours.

Semester course; 1 lecture and 4 laboratory hours. 3 credits. May be repeated for credit. Enrollment is restricted to students in the genetic counseling master's program. Practical experience in the genetic counseling clinic and on ward rounds. Includes collection and analysis of family histories, genetic counseling and introduction to genetic nosology. Graded as S/U/F.

# HGEN 601. Research in Genetic Counseling. 2 Hours.

Semester course; 1.5 lecture and .5 thesis hours. 2 credits. Enrollment restricted to genetic counseling graduate students only. Students must have chosen their research project adviser, with whom they will meet throughout the semester, prior to enrolling. Provides a comprehensive examination of the fundamentals of research relevant for the scientific advancement of the genetic counseling field. Explores topics including developing a research question; conducting literature reviews; designing a research project; working with the institutional review board; and collecting, analyzing and interpreting data. Students will develop and deliver a research proposal orally and in writing.

#### HGEN 602. Genetic Models of Disease. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Understanding the molecular basis of human disease states is a major focus for biomedical research. This course will train students to investigate molecular-genetic mechanisms of disease using four genetic model organisms: the nematode C. elegans, the fruit fly Drosophila melanogaster, the teleost zebrafish Danio rerio and the mouse Mus musculus, which serve as important laboratory models for human diseases and facilitate the elucidation of the underlying molecular mechanisms.

#### HGEN 603. Mathematical and Statistical Genetics. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Prerequisites: HGEN 611; and BIOS 543 and BIOS 544 or HGEN 651 and HGEN 652. Provides an introduction to the rudiments of theoretical and applied mathematical population genetics including the segregation of genes in families, genetic linkage and quantitative inheritance. Emphasizes the methods used in the analysis of genetic data.

# HGEN 605. Experimental Methods in Human Genetics. 1-3 Hours.

Semester course; 2-6 laboratory hours. 1-3 credits. Restricted to students in the M.S. or Ph.D. programs in human genetics. Provides hands-on experience with the experimental methods that are used to carry out research in specific areas of human genetics prior to beginning thesis/ dissertation research. Graded S/U/F.

# HGEN 606. Introduction to Clinical Genetics. 1 Hour.

Semester course; 1 lecture hour. 1 credit. Prerequisite: open only to graduate students in human genetics programs or by permission of instructor. Provides an overview of medical genetics and counseling practice for non-genetic counseling students, including orientation to the translational side of research genetics and contemporary practice of clinical genetics. Graded S/U/F.

#### HGEN 607. Processes in Genetic Counseling I. 1 Hour.

Semester course; 1 practicum hour. 1 credit. Enrollment restricted to students in the genetic counseling program. Training in the ability to recognize the psychological and social processes affecting counselor-patient interactions. Graded as pass/fail.

#### HGEN 608. Processes in Genetic Counseling II. 1 Hour.

Semester course; 1 practicum hour. 1 credit. Prerequisite: HGEN 607. Enrollment restricted to second-year students in the genetic counseling program. Further training in the ability to recognize the psychological and social processes affecting counselor-patient interactions. Graded as pass/fail.

#### HGEN 609. Clinical Genomics. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Enrollment is restricted to graduate students and residents with undergraduate degrees in an area related to genetics, biology or psychology. Provides an overview of modern genetic and genomic diagnostic testing. Explores topics including genomic variation, epigenetics, modern methodologies, applications of testing, data interpretation including variant classification, and the benefits and limitations of testing. Crosslisted as: PATH 609.

#### HGEN 610. Current Literature in Human Genetics. 1 Hour.

Semester course; 1 lecture hour. 1 credit. Enrollment is restricted to graduate students. Provides directed experience in critiquing, understanding and presenting current literature on a focused topic in human genetics. Graded as S/U/F.

# HGEN 611. Data Science I. 3 Hours.

Semester course; 3 lecture hours. 3 credits. This course will introduce students to tools and techniques from the discipline of data science that support efficient and reproducible scientific computing. Students will gain hands-on experience developing complete data analysis projects based on real-world datasets. Lessons will cover the primary tasks that comprise most analyses: data management/acquisition, cleaning, reshaping, manipulation, analysis and visualization, as well as strategies for arranging these constituent parts into cohesive workflows that are verifiable, easily repeatable and consistent with best practices for reproducible computational research. This course will focus on the statistical programming language R but no programming background is necessary. Crosslisted as: OVPR 611.

#### HGEN 612. Data Science II. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Prerequisite: HGEN 611/ OVPR 611. This course builds upon the material introduced in the prerequisite course by providing instruction on advanced techniques for working with data and producing highly reproducible data products. The learning path focuses on the fundamentals of both machine learning and the creation of production-ready web applications as two highly marketable skills for future data scientists. Project-based assignments culminate in students creating their own applications that take advantage of tidymodel principles to automate machine-learning workflows, visually communicate knowledge with interactive graphics and using Git and OSF for project management. The guiding principle of the course is that the these products of research should be open and accessible to all members of a project team for maximum impact. This course will continue the use of the statistical programming language R with a focus on advanced tidyverse functions for data wrangling and statistical model development. Crosslisted as: OVPR 612.

#### HGEN 614. Pathogenesis of Human Genetic Disease. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Enrollment is restricted to graduate students. Surveys the mechanisms and varieties of human gene mutations resulting in human genetic disease and emphasizes different investigational disorders using current scientific literature.

#### HGEN 615. Techniques in Genetic Counseling. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Enrollment restricted to students in the M.S. in Genetic Counseling program or by permission of the instructor. Provides theory and context for interviewing as well as counseling skills required for genetic counseling practice. Literature and practical techniques utilized to acquire skills. There is significant reliance on live in-class role play scenarios to exercise and demonstrate emerging skills. Additional deconstruction of taped master genetic counselor role plays aids in the understanding and evaluation of theory and skill to be acquired. Emphasis is on understanding and developing the verbal and non-verbal skills required for effective genetic counseling practice.

# HGEN 616. Cultural Diversity in Genetic Counseling. 1 Hour.

Semester course; 1 lecture hour. 1 credit. Enrollment is restricted to students in the M.S. in Genetic Counseling program. This class explores topics related to providing genetic counseling to individuals from diverse backgrounds. Students learn skills for working with in-person and phone interpreters and practice applying these skills. Students will receive instruction in how to provide care for individuals from diverse spiritual backgrounds and the role that hospital chaplains can serve in helping families dealing with grief and crisis. Students are led in discussion to begin to recognize the unique health issues that are encountered by marginalized populations, including transgender and LGBTQ+. Students will also learn about health disparities among different cultural backgrounds and learn to recognize personal biases and ways to avoid countertransference. This course will use readings from peerreviewed literature to emphasize concepts presented in class. Graded as satisfactory/unsatisfactory.

# HGEN 617. Genetic Analysis of Complex Traits. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Prerequisite: introductory biostatistics or permission of instructor. Introduces the theory and practice of analysis of complex human traits. Provides a solid grounding in the fundamental concepts, study designs and analytical strategies for this evolving and important area.

## HGEN 618. Evidence Based Genetic Counseling. 1 Hour.

Semester course; 1 seminar hour. 1 credit. May be repeated for a maximum of two credits. Enrollment is restricted to student in the M.S. in Genetic Counseling program. The course will provide oral and visual communication skills in presenting clinical information and research in the field of genetic counseling; a review of current literature to inform evidence-based practices in genetic counseling and clinical genetics; and oral and written competency in the transmission of complex genetic information to an educationally heterogeneous audience. This course will inform best practices for succinct and informative clinical case presentations and contribute to students' appreciation of the established and emerging best practices in genetic counseling, including ethical, legal and social issues in healthcare, counseling techniques, genetic testing technologies, differential diagnoses, medical management guidelines, therapies, and clinical trials. Presentations will foster critical thinking skills in the clinical environment by providing opportunities to discuss, question, and adopt evidence-based practices in clinical genetics published in current literature. Graded as satisfactory/unsatisfactory.

#### HGEN 619. Quantitative Genetics. 3 Hours.

Semester course; 3 lecture hours. 3 credits. The effects of genes and environment on complex human traits with emphasis on: Genetic architecture and evolution; nongenetic inheritance; mate selection; developmental change; sex-effects; genotype-environment interaction; resolving cause from effect; design of genetic studies, statistical methods and computer algorithms for genetic data analysis.

#### HGEN 620. Principles of Human Behavioral Genetics. 3 Hours.

Semester course; 3 lecture hours. 3 credits. The theory of genetic and nongenetic transmission considered in relation to the design, analysis, and interpretation of studies to identify the principal genetic and environmental causes of behavioral variation. Included will be analysis of intelligence, personality, social attitudes, and psychiatric disorders.

## HGEN 622. Cancer Genetic Counseling. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Prerequisite: HGEN 501 or permission of instructor. Provides a background in as well as the most current information relevant to cancer genetics and cancer genetic counseling. Includes instruction in basic science and genetic and psychosocial aspects of cancer, with an emphasis on familial and hereditary cancers.

# HGEN 651. Statistics for Genetic Studies I. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Teaches students statistical methods for multidisciplinary research, specifically presenting the mathematical components that underlie statistical analysis and including probability theory, statistical distributions, inference and linear models.

#### HGEN 652. Statistics for Genetic Studies II. 3 Hours.

Semester course; 3 lecture hours. 3 credits. Prerequisite: HGEN 651. Builds upon the quantitative statistical methods from prerequisite course. Students will learn the mathematical components that underlie statistical analysis with a focus on maximum-likelihood methods and structural equation modeling. These components provide the necessary foundation for the advanced statistical genetic methods for understanding how genetic and environmental factors impact the development of psychiatric and substance abuse disorders.

#### HGEN 690. Genetics Research Seminar. 1 Hour.

Semester course; 1 lecture hour. 1 credit. Selected topics in genetics presented by students and staff.

# HGEN 691. Special Topics in Genetics. 1-4 Hours.

1-4 credits. Lectures, tutorial studies, library assignments in selected areas of advanced study or specialized laboratory procedures not available in other courses or as part of the research training.

# HGEN 692. Special Topics. 1-4 Hours.

Semester course; 1-4 variable hours. 1-4 credits. Lectures, tutorial studies, library assignments in selected areas of advanced study or specialized laboratory procedures not available in other courses or as part of the research training. Graded as S/U/F.

## HGEN 697. Directed Research in Genetics. 1-15 Hours.

1-15 credits. Research leading to the M.S. or Ph.D. degree and elective research projects for other students.