# **GENETICS**

Emeritus: (Professor) Leonard Herzenberg

Interim Chair: John Pringle

Professors: Russ Altman, Gregory Barsh, Michele Calos, Stanley Cohen, Ronald Davis, Andrew Fire, Uta Francke, Margaret Fuller, Mark Kay, Stuart Kim, Joseph Lipsick, John Pringle, Matthew Scott, Tim Stearns

Associate Professors: Laura Attardi, James Ford, Arend Sidow, Anne Villenueve, Douglas Vollrath

Assistant Professors: Julie Baker, Anne Brunet, Julien Sage, Man-Wah Tan, Hua Tang

Professor (Research): Leonore Herzenberg

Associate Professors (Research): J. Michael Cherry, Zijie Sun

Assistant Professor (Research): Gavin Sherlock

Courtesy Professor: Hank Greely Consulting Professor: David Cox

Mail Code: 94305-5120 Phone: (650) 723-3335

Email: genetics-info@genome.stanford.edu Web Site: http://genetics.stanford.edu

Courses offered by the Department of Genetics have the subject code GENE, and are listed in the "Genetics (GENE) Courses" section of this bulletin.

## GRADUATE PROGRAMS IN GENETICS

## MASTER OF SCIENCE IN HUMAN GENETICS AND GENETIC COUNSELING

The University requirements for the M.S. are described in the "Graduate Degrees" section of this bulletin.

The Department of Genetics offers an M.S. in Human Genetics and Genetic Counseling, which is accredited by the American Board of Genetic Counseling. This program prepares students to practice in the healthcare profession of genetic counseling. The program is a full time two-year program, and accepts students to begin the program only in Autumn quarter. Students must be admitted directly into this program, and cannot automatically transfer from the Ph.D. programs within the department. While courses are oriented primarily towards genetic counseling students, they may also be taken by medical students, other graduate students, residents or postdoctoral fellows, and (with permission) undergraduates.

The degree requires the completion of clinical rotations and an approved research project. Students must also complete required course work (GENE 271-286), several additional required courses (MED 250A, MED 255, DBIO 201, and GENE 238), and are encouraged to take 3-4 elective courses of their choice. Faculty members include members of the Stanford faculty from Genetics, Pediatrics, Obstetrics, Pathology, Developmental Biology, Biomedical Ethics, Law and Psychology, and practicing genetic counselors and clinical geneticists in various medical centers across the Bay Area.

Applications are due in December (see web site) for admission in the following Autumn Quarter. Applicants should demonstrate a combination of academic preparation, exposure to genetic counseling, and counseling and/or laboratory experiences. Exposure to persons with disabilities or chronic illness is also helpful. Additional information about the program is at http://www.med.stanford.edu/genetic-counseling.

## DOCTOR OF PHILOSOPHY IN GENETICS

University requirements for the Ph.D. degree are described in the "Graduate Degrees" section of this bulletin.

The Ph.D. program in the Department of Genetics offers graduate students the opportunity to pursue a discipline that encompasses both a set of tools and a coherent way of thinking about biology and medicine. All major areas of genetics are represented in the department, including human genetics (molecular identification of Mendelian traits and the pathophysiology of genetic disease, gene therapy, genetic epidemiology, analysis of complex traits, and human evolution), and application of model organisms such as

bacteria, yeast, flies, worms, or mice to basic questions in biomedical research. The department is especially strong in genomic and bioinformatic approaches to genome biology and evolution, and includes several genome-scale databases such as the Saccharomyces Genome Database (SGD), the Stanford Microarray Database (SMD), and the Pharmacogenetics and Pharmacogenomics Knowledge Base (PharmGKB), and, administered through the Department of Biochemistry, the Stanford Genome Technology Center (SGTC).

Exposure to the intellectual scope of the department is provided by laboratory rotations, dissertation research, advanced courses in genetics and other areas of biomedical science, seminar series, journal clubs, and an annual three-day retreat of faculty, students, postdoctoral fellows, and staff scientists. Emphasis is placed on interactions and collaborations among students, postdoctoral students, and faculty within the department and throughout the campus.

During their first year, graduate students in the department take graduate courses and sample areas of research by doing rotations in three or four laboratories. At the end of the first three quarters, students may select a laboratory in which to do their dissertation research. While the dissertation research is generally performed in one laboratory, collaborative projects with more than one faculty member are encouraged. In addition to interacting with their faculty preceptor, graduate students receive advice regularly from other faculty members who serve as members of their dissertation committee. Study for the Ph.D. generally requires between four and five years of graduate work, most of which is focused on dissertation research.

Students are generally enrolled in the program to receive the Ph.D. degree, although a limited number of M.D. candidates can combine research training in genetics with their medical studies. Ph.D. candidates who have passed the qualifying exam in the second year can opt to receive the M.S. as a terminal degree

There are opportunities for graduate students to teach in graduate-level and professional-school courses. In addition, students have the opportunity to participate in educational outreach activities coordinated by the department, which include opportunities to interact with secondary school students and teachers, lay groups, and local science museums.

Students who have recently received a bachelor's, master's, M.D., or Ph.D. degree in related fields may apply for graduate study. Prospective students must have a background in biology, mathematics, physics, and chemistry. Decisions for admission are based on comparison of the relative merits of all the candidates' academic abilities and potential for research and the department's interest in promoting a diverse learning environment. Interviews take place in late February or early March and successful applicants are offered admission by early spring. Students who wish to pursue a combined M.D./Ph.D. degree are considered for admission into the graduate program in the department after they have been admitted to the M.D. program in the School of Medicine.

Students begin graduate studies in Autumn Quarter. Prospective students are encouraged to start the application process early to ensure that they are able to submit a complete application by the December deadline. All students accepted into the Ph.D. program in the Department of Genetics are provided with full tuition and a stipend. Two training grants from the National Institutes of Health provide major support for the graduate training program in the department. Other student support is provided by departmental funds and from research grants, both federal and private, of the faculty. In addition, a number of graduate students are funded by fellowships, including those from the National Science Foundation and the Stanford Graduate Fellows program.

# GENETICS (GENE) COURSES

For information on graduate programs in the Department of Genetics, see the "Genetics" section of this bulletin. Additional courses in or related to genetics are included in the deaprtments of Biology, Biochemistry, Developmental Biology, Microbiology and Immunology, Neuroscience, Biomedical Informatics, and Structural Biology.

## UNDERGRADUATE COURSES IN GENETICS

## GENE 109Q. Genomics: A Technical and Cultural Revolution (S,Sem Same as BIOMEDIN 109Q.) Stanford Introductory Seminar. Preference to sophomores. For non-science majors. Concepts of

genomics, high-throughput methods of data collection, and computational approaches to analysis of data. The social, ethical, and economic implications of genomic science. Students may focus on computational or social aspects of genomics.

3 units, Win (Altman, R)

## **GENE 199. Undergraduate Research**

Students undertake investigations sponsored by individual faculty members. Prerequisite: consent of instructor.

1-18 units, Aut (Staff), Win (Staff), Spr (Staff), Sum (Staff)

## **GRADUATE COURSES IN GENETICS**

Primarily for graduate students; undergraduates may enroll with consent of instructor.

## **GENE 202. Human Genetics**

Theoretical and experimental basis for the genetics of human health and disease. Molecular, chromosomal, biochemical, developmental, cancer, and medical genetics, emphasizing the last. Clinical case discussions. Prerequisites: biochemistry; basic genetics.

4 units, Aut (Ormond, K; Hudgins, L)

## **GENE 203. Advanced Genetics**

(Same as BIO 203, DBIO 203.) For graduate students in Bioscience programs; may be appropriate for graduate students in other programs. The genetic toolbox. Examples of analytic methods, genetic manipulation, genome analysis, and human genetics. Emphasis is on use of genetic tools in dissecting complex biological pathways, developmental processes, and regulatory systems. Faculty-led discussion sections with evaluation of papers. Students with minimal experience in genetics should prepare by working out problems in college level textbooks.

4 units, Aut (Stearns, T; Barsh, G; Sidow, A)

## **GENE 206. Epigenetics**

(Same as PATH 206.) For graduate students; undergraduates by consent of instructor. Mechanisms by which phenotypes not determined by the DNA sequence are stably inherited in successive cell divisions. From the discovery of position-effect variegation in Drosophila in the 20s to present-day studies of covalent modifications of histones and DNA methylation. Topics include: position effect, gene silencing, heterochromatin, centromere identity, genomic imprinting, histone code, variant histones, and the role of epigenetics in cancer. Prerequisite: background in genetics and molecular biology.

2 units, alternate years, not given this year

## **GENE 211. Genomics**

Genome evolution, organization, and function; technical, computational, and experimental approaches; hands-on experience with representative computational tools used in genome science; and a beginning working knowledge of PERL.

3 units, Win (Cherry, J; Myers, R; Sidow, A; Sherlock, G)

## **GENE 212. Introduction to Biomedical Informatics Research** Methodology

(Same as BIOE 212, BIOMEDIN 212, CS 272.) Hands-on software building. Student teams conceive, design, specify, implement, evaluate, and report on a software project in the domain of biomedicine. Creating written proposals, peer review, providing status reports, and preparing final reports. Guest lectures from professional biomedical informatics systems builders on issues related to the process of project management. Software engineering basics. Prerequisites: 210, 211 or 214, or consent of instructor.

3 units, Aut (Altman, R; Cheng, B; Klein, T)

## **GENE 214. Representations and Algorithms for**

Computational Molecular Biology (Same as BIOE 214, BIOMEDIN 214, CS 274.) Topics: algorithms for alignment of biological sequences and structures, computing with strings, phylogenetic tree construction, hidden Markov models, computing with networks of genes, basic structural computations on proteins, protein structure prediction, protein threading techniques, homology modeling, molecular dynamics and energy minimization, statistical analysis of 3D biological data, integration of data sources, knowledge representation and controlled terminologies for molecular biology, graphical display of biological data, machine learning (clustering and classification), and natural language text processing. Prerequisites: programming skills; consent of instructor for 3 units.

3-4 units, Spr (Altman, R)

## **GENE 215. Frontiers in Biological Research**

(Same as BIOC 215, DBIO 215.) Literature discussion in conjunction with the Frontiers in Biological Research seminar series hosted by Biochemistry, Developmental Biology, and Genetics in which distinguished investigators present current work. Students and faculty meet beforehand to discuss papers from the speaker's primary research literature. Students meet with the speaker after the seminar to discuss their research and future direction, commonly used techniques to study problems in biology, and comparison between the genetic and biochemical approaches in biological research.

1 unit, Aut (Harbury, P: Tan, M; Villeneuve, A), Win (Harbury, P; Tan, M; Villeneuve, A)

## **GENE 221. Current Issues in Aging**

(Same as DBIO 221, NENS 221.) Current research literature on genetic mechanisms of aging in animals and human beings. Topics include: mitochondria mutations, insulin-like signaling, sirtuins, aging in flies and worms, stem cells, human progeria, and centenarian studies. Prerequisite: GENE 203

2 units, Spr (Kim, S; Brunet, A; Rando, T)

#### **GENE 222. Method and Logic in Experimental Genetics**

For graduate students only. How experimental strategies are applied to biological questions irrespective of discipline boundaries. Examples include purifying activities from complex mixtures, localizing molecules in space and time, discovering macromolecular interactions, inferences from sequence similarity, using structure to elucidate function, and applying genomics to biological problems. Weekly discussion of two representative papers selected by faculty and a student presentation of a third paper which illustrate principles of biochemistry and cell and molecular biology, and the historical context of important scientific advances.

3 units, Win (Baker, J; Calos, M)

#### GENE 233. The Biology of Small Modulatory RNAs

(Same as MI 233, PATH 233.) Open to graduate and medical students. How recent discoveries of miRNA, RNA interference, and short interfering RNAs reveal potentially widespread gene regulatory mechanisms mediated by small modulatory RNAs during animal and plant development. Required paper proposing novel research.

2 units, alternate years, not given this year

## GENE 235. C. Elegans Genetics

Genetic approaches to C. elegans, practice in designing experiments and demonstrations of its growth and anatomy. Probable topics include: growth and genetics, genome map and sequence, mutant screens that start with a desired phenotype, reverse genetics and RNAi screens, genetic duplications, uses of null phenotype non-null alleles, genetic interactions and pathway analysis, and embryogenesis and cell lineage. Focus of action, mosaic analysis, and interface with embryological and evolutionary approaches.

2 units, alternate years, not given this year

#### **GENE 238.** Current Concepts and Dilemmas in Genetic **Testing**

(Same as INDE 238.) Issues arising from the translational process from research to commercialization. Diagnostic inventions and applications, community implications, newborn screening, cancer genetics, and pharmacogenomics. Guest experts. For M.D., biomedical graduate, and genetic counseling students.

2 units, Spr (Tobin, S; Schrijver, I; Cowan, T; Magnus, D)

## **GENE 244. Introduction to Statistical Genetics**

Statistical methods for analyzing human genetics studies of Mendelian disorders and common complex traits. Probable topics include: principles of population genetics; epidemiologic designs; familial aggregation; segregation analysis; linkage analysis; linkagedisequilibrium-based association mapping approaches; and genomewide analysis based on high-throughput genotyping platforms. Prerequisite: STATS 116 or equivalent or consent of instructor.

3 units, alternate years, not given this year

## **GENE 245. Computational Algorithms for Statistical Genetics**

(Same as STATS 345.) Computational algorithms for human genetics research. Topics include: permutation, bootstrap, expectation maximization, hidden Markov model, and Markov chain Monte Carlo. Rationales and techniques illustrated with existing implementations commonly used in population genetics research, disease association studies, and genomics analysis. Prerequisite: GENE 244 or consent of instructor.

2-3 units, Spr (Tang, H; Zhang, N)

#### **GENE 260. Supervised Study**

Genetics graduate student lab research from first quarter to filing of candidacy. Prerequisite: consent of instructor.

1-18 units, Aut (Staff), Win (Staff), Spr (Staff), Sum (Staff)

## **GENE 271. Human Molecular Genetics**

For genetic counseling students, graduate students in genetics, medical students, residents, and postdoctoral fellows interested in the practice of medical genetics. Gene structure and function; the impact of mutation and polymorphism as they relate to developmental pathways and health and human disease; population based genetics; approaches to the study of complex genetic conditions, and gene therapy, proteomics, stem cell biology, and pharmacogenetics. Undergraduates require consent of instructor and a basic genetics course.

4 units, Aut (Ormond, K; Francke, U)

## GENE 272. Introduction to Medical Genetics

For genetic counseling students, graduate students in human genetics, medical students, residents, and fellows; undergraduates with consent of instructor. Principles of medical genetics including taking a family history, modes of inheritance, and mathematical principles of medical genetics (Bayes theorem, population genetics). An additional paper is required for 3 units.

2-3 units, Aut (Hudgins, L; Ormond, K)

#### **GENE 273. Introduction to Clinical Genetics Testing**

For genetic counseling students, graduate students in genetics, medical students, residents, and postdoctoral fellows; undergraduates with consent of instructor. Principles of cytogenetic, molecular, and biochemical laboratory analysis. How to select the appropriate laboratory for testing and laboratory quality assurance, including the CLIA process. An additional paper is required for 3 units.

2-3 units, Aut (Ormond, K; Cowan, T; Cherry, A; Schrijver, I)

#### GENE 274A. A Case Based Approach to Clinical Genetics

For genetic counseling students, graduate students in genetics, medical students, residents, and post-doctoral fellows. Case-based scenarios and guest expert lectures. Skills in case preparation, management, and presentation.

2 units, Win (Staff)

## **GENE 274B.** A Case Based Approach to Clinical Genetics

For genetic counseling students, graduate students in genetics, medical students, residents, and post-doctoral fellows. Case-based scenarios and guest expert lectures. Skills in case preparation, management, and presentation.

2 units, Spr (Hudgins, L; Ormond, K)

## **GENE 275. Role Play and Genetic Counseling Observations**

Observation includes genetic counseling sessions in prenatal, pediatric, and cancer settings, and medical genetics procedures and testing settings.

2 units, Aut (Ormond, K)

## **GENE 276. Genetic Counseling Clinical Rotations**

For genetic counseling students only. Supervised clinical experiences. May be repeated for credit. Prerequisite: GENE 275.

4-7 units. Aut (Ormond, K), Win (Ormond, K), Spr (Ormond, K), Sum (Ormond, K)

## **GENE 278. Prenatal Genetic Counseling**

Internet-based course for genetic counseling students, graduate students in genetics, medical students, residents, and postdoctoral fellows; genetic counseling students should take this course in conjunction with their initial prenatal genetics rotation. Topics include prenatal genetic screening and diagnosis in the first and second trimesters, ultrasound, teratology, and genetic carrier screening.

1 unit, Aut (Ormond, K), Win (Ormond, K), Spr (Ormond, K), Sum (Ormond, K)

#### GENE 279. Pediatric and Adult Genetic Counseling

Internet-based course for genetic counseling students, graduate students in genetics, medical students, residents, and postdoctoral fellows; genetic counseling students should take this course in conjunction with their initial general genetics rotation. Topics include: common genetic conditions; assessment of child development and medical history in the context of a genetic workup; dysmorphology; development of a differential diagnosis; and resources for case management and family support.

1 unit, Aut (Ormond, K), Win (Ormond, K), Spr (Ormond, K), Sum (Ormond, K)

## GENE 280. Metabolic Genetic Counseling

Internet-based course for genetic counseling students, graduate students in genetics, medical students, residents, and postdoctoral fellows; genetic counseling students should take this course in conjunction with their metabolic genetics rotation. Overview of metabolic diseases; common pathways; diagnosis, management, and treatment of metabolic disorders; and newborn screening.

1 unit, Aut, Win, Spr, Sum (Ormond, K)

## **GENE 281. Cancer Genetic Counseling**

Internet-based course for genetic counseling students, graduate students in genetics, medical students, residents, and postdoctoral fellows; genetic counseling students should take this course in conjunction with their initial cancer genetics rotation. Topics include: cancer cytogenetics and genetic principles; diagnosis and management of common cancer genetic syndromes; predictive testing; psychology of cancer genetic counseling; and topics recommended by ASCO guidelines.

1 unit, Aut (Ormond, K), Win (Ormond, K), Spr (Ormond, K), Sum (Ormond, K)

## **GENE 282. Genetic Counseling Research Seminar**

For genetic counseling students only. Facilitated discussions on identifying a topic and mentor for genetic counseling departmental research projects. Corequiste: GENE 299.

2 units, Win (Staff)

## **GENE 282. Genetic Counseling Research Seminar**

For genetic counseling students only. Facilitated discussions on identifying a topic and mentor for genetic counseling departmental research projects. Corequiste: GENE 299.

2 units, Win (Ormond, K)

#### **GENE 283. Genetic Counseling Research**

Investigations sponsored by individual faculty members. Students complete an approved research project. May be repeated for credit. Prerequisite: GENE 282.

1-8 units, Aut (Ormond, K), Win (Ormond, K), Spr (Ormond, K), Sum (Ormond, K)

## **GENE 284. Medical Genetics Seminar**

Presentation of research and cases. Students enrolling for 2 units also attend and report on external seminars. May be repeated for credit.

1-2 units, Aut (Ormond, K), Win (Ormond, K), Spr (Ormond, K)

## **GENE 285A.** Genetic Counseling Seminar

Year-long seminar primarily for genetic counseling students. Autumn: basics of medical communication; crosscultural and disability sensitive communication about genetics, and principles of providing genetic counseling. Winter: the impact of chronic illness and genetic disease in a developmental manner. Spring: applying therapeutic counseling approaches to the practice of genetic counseling. Undergraduates may enroll in Autumn Quarter with consent of instructor.

2-3 units, Aut (Ormond, K)

## **GENE 285B. Genetics Counseling Seminar**

Year-long seminar primarily for genetic counseling students. Autumn: basics of medical communication; crosscultural and disability sensitive communication about genetics, and principles of providing genetic counseling. Winter: the impact of chronic illness and genetic disease in a developmental manner. Spring: applying therapeutic counseling approaches to the practice of genetic counseling. Prerequisite: 285A.

2-3 units, Win (Ormond, K)

#### **GENE 285C. Genetic Counseling Seminar**

Year-long seminar primarily for genetic counseling students. Autumn: basics of medical communication; crosscultural and disability sensitive communication about genetics, and principles of providing genetic counseling. Winter: the impact of chronic illness and genetic disease in a developmental manner. Spring: applying therapeutic counseling approaches to the practice of genetic counseling. Prerequisite: 285 A/B.

2-3 units, Spr (Ormond, K)

## **GENE 286A. Advanced Genetic Counseling Seminar**

For genetic counseling students only. Psychosocial issues associated with genetic counseling cases through cases that students have seen throughout their training. Professional development topics including: the expanding roles of genetic counselors; billing, reimbursement, and licensing; the role of genetic counseling in the changing healthcare system; the incorporation of genetics into all areas of medicine and public health; and implications of direct-to-consumer genetic testing. Prerequisites: GENE 285 A,B,C and 276.

2 units, Aut (Ormond, K)

## GENE 286B. Advanced Genetic Counseling Seminar

Continuation of GENE 286A. For genetic counseling students only. Psychosocial issues associated with genetic counseling cases through cases that students have seen throughout their training. Professional development topics including: the expanding roles of genetic counselors; billing, reimbursement, and licensing; the role of genetic counseling in the changing healthcare system; the incorporation of genetics into all areas of medicine and public health; and implications of direct-to-consumer genetic testing. Prerequisites: GENE 285 A.B.C and 276.

2 units, Win (Ormond, K)

#### **GENE 286C. Advanced Genetic Counseling Seminar**

Continuation of 286A/B. For genetic counseling students only. Psychosocial issues associated with genetic counseling cases through cases that students have seen throughout their training. Professional development topics including: the expanding roles of genetic counselors; billing, reimbursement, and licensing; the role of genetic counseling in the changing healthcare system; the incorporation of genetics into all areas of medicine and public health; and implications of direct-to-consumer genetic testing. Prerequisites: GENE 285 A.B.C and 276.

2 units, Spr (Ormond, K)

## **GENE 299. Directed Reading in Genetics**

Prerequisite: consent of instructor.

1-18 units, Aut (Staff), Win (Staff), Spr (Staff), Sum (Staff)

## **GENE 399. Graduate Research**

Investigations sponsored by individual faculty members. Prerequisite: consent of instructor.

1-18 units, Aut (Staff), Win (Staff), Spr (Staff), Sum (Staff)