

GENETIC ANALYSIS

of Complex Human Diseases

GENETIC ANALYSIS OF COMPLEX HUMAN DISEASES is a comprehensive four-day course directed toward physician-scientists and other medical researchers. The course will introduce state-of-the-art approaches for the mapping and characterization of human inherited disorders with an emphasis on the mapping of genes involved in common and genetically complex disease phenotypes. The overall focus is a broad-based understanding of the problems and solutions to the design and execution of disease gene mapping projects using Human Genome Project resources.

GOALS OF THIS COURSE

The primary goal of this course is to provide participants with an overview of approaches to identifying genes involved in complex human diseases. At the end of the course, participants should be able to identify the key components of a study team, and communicate effectively with specialists in various areas to design and execute a study. Learning objectives for the course include:

- ▶ To instruct participants about the necessary steps and procedures used in ascertaining, collecting, and databasing pedigree, demographic, family history, environmental risk factor and clinical information for genetic disease mapping studies. The impact of genetic research on patients and their families will also be discussed.

- ▶ To provide background information in the basic techniques of linkage analysis. The discussion will include problems and confounding issues that commonly arise.

- ▶ To provide an introduction to the various strategies, designs, and methods of analysis needed to dissect the genetic basis of common and genetically complex (e.g. multifactorial or polygenic) traits. Examples are drawn from successful applications in human genetic disease. Discussions will include current approaches to both qualitative and quantitative trait phenotype assignment, methods of analysis, interpretation, follow-up and refinement of the preliminary linkage and/or association data, investigation of power, examination of heterogeneity, and gene/gene and gene/environment interactions. This course will not include any bench or "wet" laboratory experience.

- ▶ To introduce newly evolving "genome-wide" study methodologies from the laboratory and statistical analysis perspectives including SNP-based (single nucleotide polymorphism) genome-wide association mapping and "genomic convergence" (integration of linkage, association, expression, and gene function data).

- ▶ To incorporate discussion of the participants' individual research interests. Participants will be encouraged to bring preliminary information and/or data for both formal and informal group discussion and instructor consultation.

2012 COURSE TOPICS

Topics will be covered through lectures by course faculty, informal discussions of particular examples from the literature, and discussions of students' own research projects and experience.

Basics of Population Genetics

- ▶ Evaluating evidence of a genetic component to the trait
- ▶ Modes of inheritance, penetrance and trait expression
- ▶ Allele frequencies and Hardy-Weinberg equilibrium
- ▶ Linkage vs. association, linkage disequilibrium

Phenotype definition

- ▶ Strategies for collecting phenotypic data
- ▶ Defining a phenotype for study
- ▶ Intermediate phenotypes

Study designs

- ▶ Candidate gene studies
- ▶ Genome-wide studies
- ▶ Family-based versus population based study designs

Tools and technologies

- ▶ Human genome project sequence/HapMap data
- ▶ Single nucleotide polymorphism detection
- ▶ Copy number variation analysis
- ▶ Large-scale sequencing

Association studies and linkage disequilibrium

- ▶ Family-based and population based designs
- ▶ Control of population structure
- ▶ Admixture mapping
- ▶ Genome-wide association

Quantitative traits

- ▶ Linkage analysis
- ▶ Association analysis

Evaluating functional relevance of trait loci

- ▶ Bioinformatics approaches
- ▶ Molecular approaches

Complex patterns and data mining

- ▶ Gene-gene interaction Gene-environment interaction
- ▶ "Genomic convergence": integration of different data streams

COST

LOCAL / DAY PARTICIPANTS:

\$500

These costs include all conference fees, continental breakfast, and lunch each day and a Welcome reception/dinner.

VISITING / OVERNIGHT PARTICIPANTS:

\$800

These costs include all conference fees, four nights lodging at the Miami Beach Resort (Sunday through Thursday morning), continental breakfast, lunch each day and a Welcome reception/dinner.

KEY DATES

JANUARY 31, 2012:

Deadline for receipt of completed application. Space is limited so apply early. Applications will be reviewed upon receipt. To ensure a place in the course, applications should be submitted ASAP!

FEBRUARY 29, 2012:

Final selection and notification of class participants, scholarship recipients, and alternates.

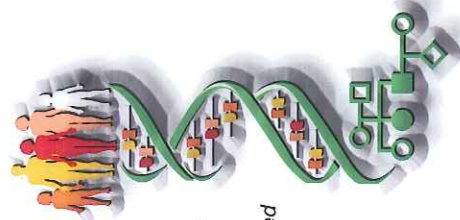
MARCH 31, 2012:

Deadline for confirmation and receipt of all conference fees.

NOTE: There will be a penalty assessed for cancellation after this point. A full description of the cancellation policy is available upon request.

MAY 21-24, 2012:

Course dates (arrival May 20, 2012)



COURSE APPLICATION

APPLICATION DEADLINE

JANUARY 31, 2012

Participation in the course will be dependent on completion of an application form that describes the applicant's background and research interests. Space is limited so apply early. Applications will be reviewed upon receipt.

We recommend that all participants have a graduate level course in genetics, molecular biology or the equivalent. Participants must provide a brief statement describing their research interests, their reason for taking the course, and their long-term objective in relation to the course curriculum. This information will be used to select a highly motivated participant group. *Underrepresented minorities, women and persons with disabilities are strongly encouraged to apply.*

Download the application for the

GENETIC ANALYSIS OF COMPLEX HUMAN DISEASES

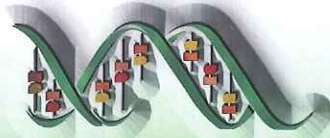
course, from our website at:
<http://hig.med.miami.edu/educational-programs/analysis-of-complex-human-diseases/course-application>

For additional information, please contact:

Lourdes Acosta
Course Administrative Co-Coordinator
Telephone: 1-877-686-6444
higcourses@med.miami.edu

SCHOLARSHIP & FELLOWSHIP Information

A limited number of scholarships and fellowships are available for registered students. Scholarship selection will be based on the strength of the individual applicants.



This course is co-organized by the

UNIVERSITY OF MIAMI
John P. Hussman Institute for Human Genomics
and the

VANDERBILT UNIVERSITY
Center for Human Genetics Research

Organizer

William K. Scott, Ph.D.

Professor of Human Genetics
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Co-Organizers

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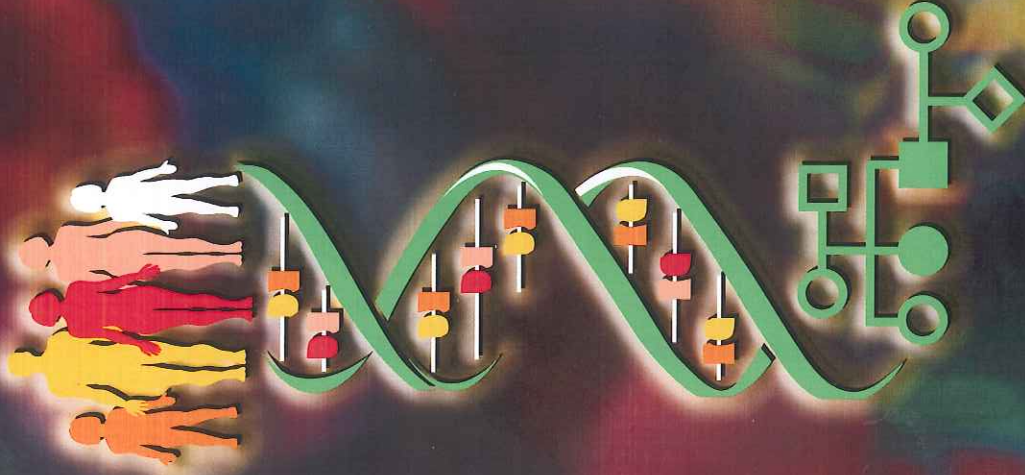
Marylyn D. Ritchie, Ph.D.

Director, Center for Systems Genomics
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May 21-24 2012

Miami Beach Resort
Miami Beach, Florida