

UCSD Center For Human Genetics/Genomics Formed To ID Novel Diagnostics And Therapies

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A Center for Human Genetics/Genomics has been established at the University of California, San Diego (UCSD) with a strong scientific base designed to translate the promise offered by knowledge of the human genome into new medical tools for diagnosis and treatment geared to each individual's specific genetic makeup.

Established in UCSD Health Sciences, which includes the School of Medicine and School of Pharmacy and Pharmaceutical Sciences, the Center brings together UCSD faculty from multiple campus departments and schools including medicine, pharmacy, biology, biostatistics and mathematics.

Taking advantage of the unparalleled opportunities presented by this post-genomic era, the Center "will help us better manage human disease by understanding its genetic basis," said Edward W. Holmes, M.D., UCSD Vice Chancellor for Health Sciences and Dean, UCSD School of Medicine.

"At UCSD, we are fortunate to have extraordinary strength, breadth and depth of research talent," Holmes added. "Because most problems and research questions in biological and biomedical sciences require the implementation of a coordinated genetic and genomic approach, our new Center transcends departmental boundaries to bring together scientists from various disciplines to make modern genetics and genomic research a primary focus at UCSD."

Palmer Taylor, Ph.D., Associate Vice Chancellor for Health Sciences and Dean of the School of Pharmacy and Pharmaceutical Sciences, noted that the Center's "strong scientific base will define the role genetics plays in human disease and the efficacy of pharmacotherapy."

Traditionally, genetics has meant the study of inheritance of characteristics that lead to individual differences or even disease. Often it involves the investigation of the function of a specific gene and its effects in the body. The continuum of recent findings regarding human genes has led to the term genomics, which is the study of the function and interactions of all genes in the DNA. The new UCSD Center for Genetics/Genomics will embrace gene studies for both single-gene diseases such as Huntington's disease, and those illnesses caused by multiple genes, such as schizophrenia.

"The Human Genome Project has led to an understanding about how human genes are organized. This has paved the way for more sophisticated efforts to understand how genes work and why genes do what they do. This is what we are investigating," said the Center's director, Anthony Wynshaw-Boris, M.D., Ph.D., UCSD professor of pediatrics and medicine, and chief of the genetics division in the Department of Pediatrics. "If we can identify what it is about defective genes that causes them to create disease, then we can develop strategies to potentially correct those defects."

The strong interdisciplinary nature of the new Center provides its primary focus on understanding human genetic disease, and translating that knowledge into new medical tools for diagnosis and treatment. Animal models will be used to investigate gene function and to test new approaches prior to human investigation. With

access to high-throughput genetic technologies and the huge volume of data produced by these technologies, efficient statistical organization and analysis is vital. Many of these analyses will be done in coordination with staff at the San Diego Supercomputing Center, a world leader in computational biosciences.

Joining Wynshaw-Boris are Center co-directors Daniel O'Connor, M.D., UCSD professor of medicine, and Nicholas Schork, Ph.D., UCSD professor of psychiatry and biostatistics. The research interests of the director and co-directors are examples of UCSD's strength in genetics/genomics.

Wynshaw-Boris focuses on understanding the genetic and biochemical pathways that are disrupted in human genetic disease during prenatal and early childhood, resulting in birth defects. By understanding what goes wrong during development in tissues such as the brain, his research group hopes to prevent tragic malformations and mental retardation. Formerly with the National Human Genome Research Institute, Wynshaw-Boris was cited as one of the nation's top researchers by *Discover* magazine in 1998 for his creation of mice lacking a key developmental gene. In subsequent studies, he discovered a gene he named NUDEL, which helps newborn neurons move to their correction position in the developing brain. (See http://health.ucsd.edu/news/2000_12_15_Neuron.html).

O'Connor is a national leader in pharmacogenomics, which is another name for individualized medicine, where therapy is tailored to a person's specific genetic profile. His team is investigating whether subtle genetic variations among patients with high blood pressure (hypertension) can predict treatment effectiveness. (See http://health.ucsd.edu/news/2001/09_04_OConnor.html)

Schork is a statistical geneticist with expertise in the design and analysis of large-scale genetic studies. To gain a comprehensive understanding of the genetic basis of complex traits and diseases, his laboratory develops mathematical, statistical and computational models and analytical tools that can be applied to genetic studies. In addition, much of his work considers how one can integrate information from different studies and study designs in order to draw inferences about the complexities that underlie disease and phenotypic expression. Considered a leader in this new and growing research field, Schork is the statistical genetics director for some of the largest human genetics initiatives in the world, including consortia devoted to understanding the genetic basis of longevity, cardiovascular disease, schizophrenia and drug response.

The Center for Human Genetics/Genomics, which will have a strong link with the UCSD Center for Molecular Genetics, will be located on the fourth floor of the new School of Pharmacy and Pharmaceutical Sciences building when it is completed in 2005. Several new faculty will be recruited there to complement current UCSD Health Sciences investigators who utilize sophisticated genetics and genomics technologies and research models. Other activities of the Center will include an integrated website with cross-referenced lists of participating faculty and resources, areas of interest, and contact information; a national genetics/genomics meeting at UCSD as well as monthly seminars for faculty and students; and the development of new curriculum to reflect a move to interdisciplinary training for UCSD undergraduates and graduate students. News Media Contact: Sue Pondrom (619) 543-6163