PRESS BRIEFINGS
ASHG 57th Annual Meeting
October 24-27, 2007
Room 24B

Wednesday, October 24, 10:00 AM. Pre-conference press briefing with Drs. Elizabeth Hauser, Hope Northrup and Michael Lovett – all members of the 2007 Program Committee . They will discuss meeting content and scientific highlights from their perspective.

Wednesday, October 24 at 4:30 PM. **Rick Guidotti, former fashion photographer** whose clients included ALL the major fashion magazines, is now directing his energy and talent to redefining beauty through Positive Exposure. His compelling photographs and passionate presentation celebrating genetic differences in kids and adults from around the globe will warm your hearts and souls. Guidotti, who founded Positive Exposure, a nonprofit organization dedicated to challenging the notion of what is typically considered beautiful and applauding human diversity, will highlight his work for members of the press and meeting registrants.

Thursday, October 25 at 9:00 AM. Roundtable on Ethical Issues in Genomic Screening and Profiling: As the technology for predictive screening and genetic testing has expanded to include the landscape of the entire genome, new ethical dilemmas have been raised. A roundtable discussion will be devoted to debating some of the most pressing issues. Drs. Cecile Janssens, Dept. of Public Health, Erasmus University Medical Center, Rotterdam, the Netherlands will describe how the prospect of genomic personalized medicine, whereby unique genomic profiles for prevention and treatment are created raises different ethical issues than traditional genetic testing for monogenic disorders. As efforts to use a single assay to test for large numbers of known disease causing mutations are advanced, Drs. John Belmont and James Lupski, Baylor College of Medicine, will discuss concerns about distributing information on deleterious mutations in individuals and populations. Despite advances in testing, the clinical implications of genomic hybridization test results are not always clear. However, there is clear consensus in the ethical community that patients have a right to receive their research test results, and a strong desire in the clinical community to also learn the results. Ms. Shelin Adam and Dr. Jan Friedman, University of British Columbia, will pose some of the compelling issues involved when children are the subject of genetic research. Ms. Gail Javitt, Genetics and Public Policy Center, Washington, DC, will address the public policy and oversight implications of genomic profiling. At the same time, it is crucial that protections be implemented early in this process. Dr. Amy McGuire, Baylor College of Medicine, will speak to the challenge of implementing federal policies to protect identifiable genetic information.

Thursday, October 25 at 11:00 AM. Unleashing the Power of Pharmacogenomics: Elucidation of the human genome has enhanced our knowledge of the genetic determinants of disease. This has broad application in predicting the prognosis of disease progression and in individualizing therapy, based on each patient's unique genomic profile. No longer a mere promise on the horizon, several examples of both types of clinical applications are provided. The presence of a mutated or deleted ATM gene is indicative of vastly different prognoses, depending upon the disease. Individual susceptibility to ionizing radiation induced apoptosis in the general population also appears to be genetically determined, as described by Dr. Annette Schmitz, Institute of Cellular and Molecular Radiobiology, Fontenay-aux-roses, France. The results provide further arguments for personalization of radiotherapy and for the study of susceptibility to various types of cancer. Outcomes for the treatment of depression are also influenced by genetic factors, since polymorphisms in serotonin receptors affect response to antidepressants. Dr. John Kelsoe and Ms. Amelia Kotte, Univ. California, San Diego, will discuss how polymorphisms in the NTRK2 and HTR2A genes expand on findings from functional MRI studies examining response to both cognitive behavioral therapy and treatment with selective serotonin reuptake inhibitors in reflecting common neurobiological mechanisms of psychotherapy and pharmacotherapy. Similarly, as described by Dr. Erik. Toonen, Radboud University, Netherlands, IF gene expression profiling of rheumatoid arthritis patients can predict who will respond favorably to treatment with anti-tumor necrosis factor, therapy that is only effective in 60% of patients, these findings would be encouraging.

Over…

Thursday, October 25 at 1:00 PM. Use of Genome Wide Association (GWA) Scans to Detect Molecular Determinants of Autoimmunity: The increasing incidence of autoimmune disease heightens the urgency to understand the molecular mechanisms involved in autoimmunity. Towards this end, several research groups are focusing their efforts on specific common autoimmune diseases. Dr. Ann Begovich, Celera Diagnostics, will begin by presenting the immunologic framework for the presentations. Drs. Anne Bowcock and Peter Liu, Washington University, St. Louis, will describe new susceptibility loci identified by GWA scans for psoriasis and psoriatic arthritis that play pivotal roles in inducing inflammation. She will also discuss the role of the RUNX family of transcription factors in inducing the inflammation found in psoriasis, rheumatoid arthritis, and systemic lupus erythematosus (SLE). How this knowledge can be used to prevent autoimmune and inflammatory disease has been the focus of their most recent work. In SLE, a single nucleotide polymorphism (SNP) screen has implicated specific regions of the major histocompatibility complex (MHC), as described by Dr. Lisa Barcellos, Univ. of California, Berkeley, and mutations in the TREX1 gene, as described by Dr. Min Ae Lee-Kirsch, Technische Universitat Dresden. Her findings in SLE patients are not based on a genomewide SNP screen but rather a deep resequencing approach. Dr. Bowcock will also discuss some of the caveats in using SNP associations to determine the functional consequences of complex diseases. Dr. Mark Daly, Mass. General Hospital, will, on behalf of an international consortium of researchers, discuss the dramatic progress in Crohn's disease genetics where multiple GWA studies have, in combination, resulted in the identification of more than 20 significant associations.

Friday, October 26 at 11:00 AM. Unraveling the Genetic Determinants of Autism: Expansion of the spectrum of autism disorders to include Asperger's syndrome has added a level of complexity to the interplay of genetic and environmental risk factors that contribute to the pathogenesis of autism. At the same time, the rising incidence of autism has heightened the urgency of determining the factors that predispose to it. Three research groups have used genomic analysis to examine the genetic basis for autism. Drs. Simon Gregory, Duke Center for Human Genetics, and Margaret Pericak-Vance, Miami Institute for Human Genomics, will describe the use of microarrays to identify chromosome rearrangements in autism families that lead to copy number variants (CNVs) and deletions in the oxytocin receptor gene, and in the transcription factor MAFF which regulates it. Drs. Aravinda Chakravarti and Dan Arking, The Institute of Genetic Medicine at Johns Hopkins, will discuss identification in autism families of a single SNP on chromosome 7q35, located in an intron of the contactin-associated protein-like 2 gene, which encodes a member of the neurexin superfamily. Analysis of CNVs in a large cohort of autism patients revealed two sub-domains of clinical features related to early development that are associated with duplications or deletions in two distinct genes, as described by Dr. Ping-I Lin, Univ. Maryland School of Medicine. A variety of approaches will be discussed by Dr. Thomas Insel, National Institutes of Mental Health.

Friday, October 26, 4:00 PM. Successful Treatment of Genetic Disorders: Several promising new treatments for genetic disorders are being tested in humans or mouse models of human genetic disease. Dr. Francis Collins and Mr. Brian Capell, NHGRI, report the successful amelioration of cardiovascular disease in a mouse model of Hutchinson-Gilford progeria syndrome, the most dramatic form of human premature aging which results in death at a mean age 13 from heart attack or stroke. The dose-dependent administration of tipifarnib, a farnesyltransferase inhibitor, resulted in prevention of the cardiovascular phenotype. Furthermore, preliminary data suggests that tipifarnib can also reverse this phenotype in mice that are allowed to reach the age of 6 or 9 months prior to beginning treatment. Cystagon, an FDA-approved drug for use in pre-transplant cystinosis patients, has been successfully used to decrease morbidity and mortality in patients with nephropathic cystinosis, a lysosomal storage disorder, as described by Dr. William Gahl, NHGRI. Based on its success, they recommend newborn screening for this disease, and earlier diagnosis and treatment of the disease in adults. Dr. Aartsma-Rus, Dr. Judith van Deutekom and Prof. Dr. GertJan van Ommen, Center for Human and Clinical Genetics, Leiden University, reports the success of localized intramuscular injection of anti-sense oligoribonucleotides in restoring dystrophin expression in 4 patients with Duchenne muscular dystrophy.