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NHGRI Shares Wealth of Science at ASHG Meeting

Research from the National Human Genome Research Institute (NHGRI) will be featured prominently at the American Society of Human Genetics (ASHG) annual meeting in Philadelphia from November 11-15, 2008. From the exhibitor floor to the plenary halls, NHGRI's participation will point to both its range of outstanding research and its leading role in genetics and genomics. Among the NHGRI-led research to be showcased at the ASHG meeting are preliminary results from a large, international study exploring a possible genetic connection between a rare disease, Gaucher disease, and the risk of a more common disorder, Parkinson disease.

"NHGRI investigators will present a wide range of impressive research results at this year's ASHG meeting," said NHGRI Scientific Director Eric Green, M.D., Ph.D. "I am pleased by our scientific contributions and for this outstanding opportunity to engage with numerous colleagues from around the world who are committed, as we are, to advancing genetic and genomic research."

On Thursday, Ellen Sidransky, M.D., a senior investigator in NHGRI's Medical Genetics Branch, will present preliminary results of a meta-analysis that examined the frequency of mutations in the glucocerebrosidase gene (*GBA*) in more than 5,000 patients with Parkinson disease and matched controls. The study was a collaborative effort involving investigators from 15 different centers world-wide. Glucocerebrosidase is an enzyme involved in the breakdown of a fatty substance, called glucocerebroside, which can harm the spleen, liver, lungs, bone marrow and, in some cases, the brain if not properly degraded.

People with two mutated copies of the *GBA* gene suffer from Gaucher disease, a rare, inherited metabolic disorder. In the past, it was thought that people who carry just one mutated *GBA* gene were unaffected. However, in recent years, Dr. Sidransky's laboratory and other research groups have completed small studies suggesting that carriers of mutated *GBA* may have an increased risk of developing Parkinson disease.

To capitalize on those intriguing early clues, Dr. Sidransky was recruited in March 2007 to coordinate an international meta-analysis of all data on the frequency of *GBA* mutations gathered during studies of Parkinson disease at 15 centers around the world. Preliminary results from that meta-analysis show that *GBA* mutation carriers have a nearly six-fold greater risk of developing Parkinson disease compared to people who have no mutated copies of the *GBA* gene. According to Dr. Sidransky, *GBA* mutations appear to be the most frequent genetic variant associated with Parkinson disease reported to date. "These findings are another example of how studies of a rare genetic disease can provide a window into more common, complex disorders," Dr. Sidransky said.

Dr. Sidransky is among many NHGRI researchers poised to deliver platform presentations or present scientific posters during the ASHG meeting.

General presentation sessions get underway on Wednesday, when NHGRI scientists will moderate three sessions. Lawrence Brody, Ph.D., a senior investigator in the Genome Technology Branch, will moderate a session about how genetic testing for common diseases will impact preventive medicine; Adam Felsenfeld, Ph.D., program director for the Large-Scale Sequencing Program, will introduce presentations on the promises of medical sequencing; and Francis Collins, M.D., Ph.D., former

director of NHGRI, will moderate a session on open models of data access for genome-wide association studies.

Also on Wednesday, NHGRI Acting Director Alan Guttmacher, M.D., will highlight the collection of family health history as a tool for genomic medicine, and other NHGRI researchers will offer presentations on population-based genetics and understanding the utility of genetic testing for common health conditions. Featured at Wednesday's poster session will be NHGRI presentations on: clinical genetics, including the NIH Undiagnosed Diseases Program; metabolic disorders, from Chediak-Higashi syndrome to idiopathic nephrocalcinosis; Mendelian disorders, from hereditary inclusion body myopathy to Gaucher disease; polygenic disorders, from cholesterol to insulin disorders; and genetic epidemiology, including an array of analyses related to genome-wide association study.

Along with Dr. Sidransky's talk, Thursday's offerings from NHGRI researchers include platform presentations on high-density haplotype mapping in dog, cattle and human genomes; translational aspects of the ClinSeq study that involves high-throughput genomic sequencing; a look at the diversity of the human skin microbiome; a study of a liver disorder known as congenital hepatic fibrosis; identification of genetic variations that play a role in breast cancer; and changes that have occurred on chromosome X during human migration out of Africa.

Many of the NHGRI poster presentations on Thursday focus on the role of genetics in a diverse range of disorders, including autism, lung cancer, scoliosis and dental abnormalities. There will also be a poster from NHGRI researchers describing their work exploring sialic acid as a treatment for inherited muscle and kidney disease, as well as one discussing the possibility that a rare genetic disease, Hermansky-Pudlak syndrome, may serve as a useful model for studying pulmonary fibrosis.

Friday will feature platform presentations from NHGRI researchers on their research looking at what motivates people to participate in whole-genome sequencing studies and on research that examined the age of onset for Neimann-Pick C disease. The third day of posters will include findings of NHGRI investigators related to type 2 diabetes, prostate cancer, polycystic kidney disease, a common birth defect known as holoprosencephaly and a number of rarer disorders, such as Ellis van Creveld syndrome (a rare genetic disorder affecting bone growth).

The ASHG meeting concludes on Saturday with a series of NHGRI platform presentations on prognosis and treatment of Hermansky-Pudlak syndrome; retroviral-mediated gene therapy to rescue mice with a severe organic acidemia; fixed-trait mapping of chondrodysplasia, a sometimes crippling deformity in domestic dogs; and possible treatments for the most dramatic form of premature aging, Hutchinson-Gilford Progeria syndrome. Dr. Collins will also moderate a special plenary session on genome-wide association studies.

ASHG is the primary professional membership organization for human genetics specialists worldwide, representing 8,000 researchers, academicians, clinicians, genetic counselors, nurses and others with a special interest in this area. This year's meeting, which is the group's 58th annual scientific gathering, will feature a total of 286 platform presentations, 2,329 posters and 26 invited sessions.

Participation from the National Human Genome Research Institute (NHGRI)
Read NHGRI abstracts for each ASHG presentation.

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