PRESS RELEASE

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GENOME-WIDE ASSOCIATION STUDIES AND THE 1000 GENOMES PROJECT: IMPLICATIONS AND CLINICAL APPLICATIONS

Renowned Geneticist Francis Collins, M.D., Ph.D., to Moderate Press Briefing at The American Society of Human Genetics 58th Annual Meeting

BETHESDA, MD – November 9, 2008 – *Francis Collins, M.D., Ph.D.*, world-renowned author, physician-geneticist and former director of the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH), will moderate a press briefing session on *"Genome-Wide Association Studies and the 1000 Genomes Project: Implications and Clinical Applications"* at the 58th Annual Meeting of The American Society of Human Genetics (ASHG) in Philadelphia, Pa., on Thursday, November 13, 2008 at 4:30 p.m. This press briefing session will also be available online via live webcast for credentialed members of the media who wish to view this event remotely *(see press briefing session and webcast details below)*.

Within the past couple of years, genetics researchers have been working together and sharing data in a collaborative effort advancing genome-wide association studies (GWAS) to identify common genetic factors that influence health and disease. The NIH defines GWAS as "any study of genetic variation across the entire human genome that is designed to identify genetic associations with observable traits (such as blood pressure or weight), or the presence or absence of a disease or condition." When combined with data from clinical studies, the information gained from whole genome analyses "offers the potential for increased understanding of basic biological processes affecting human health, improvement in the prediction of disease and patient care, and ultimately the realization of the promise of personalized medicine."

Genome-wide analyses have had a significant impact on advancing our understanding of the link between genes and health in a very short time span. The first generation of GWAS found well over 100 genetic loci associated with a variety of common diseases, and many more such studies are ongoing or in the design phase. Challenges still remain, however, and it is clear that larger studies, along with the aggregation of research results from multiple studies, will be crucial in finding genetic variants with small effect. A central question, then, is how to move from GWAS that detect genes associated with common human diseases, to methods that will allow researchers to gain a clearer understanding of the genes that are directly involved in causing these conditions. There is still much work to be done beyond these preliminary steps before the research findings gained from genome-wide analyses of common genetic diseases can be translated into safe and effective clinical treatments that will benefit patients.

ASHG's press briefing session moderated by Dr. Collins features the following panel of distinguished speakers who will be addressing the benefits and challenges of genome wide analyses, including their impact on health care practice, and the ethical, legal, and social implications involved:

- *David Altshuler, M.D., Ph.D.* – "The 1000 Genomes Project"

- *Kathy Hudson, Ph.D.* – “GWA Studies’ Impact on Society: An Overview of Ethical, Legal and Social Implications and the Influence on Public Attitudes About Genetic Research”
- *David Valle, M.D.* – "Translating GWAS Data: Bridging the Gap Between Scientific Discovery, Clinical Research and Health Care Practice”

The first two press briefing speakers listed above will be giving talks based on work they will be presenting in scientific sessions at the ASHG 2008 Annual Meeting. Brief descriptions of the content – including a summary of the major research findings that will be reported in these two presentations – are included in the section below:

THE 1000 GENOMES PROJECT

The first generation of genome-wide association studies (GWAS) have demonstrated that systematic studies of genetic variation offer a general approach to mapping novel loci contributing to common human diseases and medical phenotypes. Only a few such genes are causal mutation(s), however, and most of the heritability of common diseases remains unexplained. Thus, the next steps in genetic mapping require a more complete picture of single nucleotide and structural variants across human populations, and methods to discover rare variants with high specificity and accuracy.

In this press briefing event, David Altshuler, M.D., Ph.D., Associate Professor of Genetics and Medicine at Harvard Medical School, will provide an overview of the rationale, design, data analysis, and initial results of the 1000 Genomes Project, a two-year initiative that will sequence the genomes of 1000 or more people to create the most detailed and medically-useful map of human genome variation. A variety of next-generation sequencing technologies are being employed and compared by the international public-private consortium of multidisciplinary research teams participating in the Project, which include sequencing centers, biotechnology companies, statistical and population geneticists, and ethicists, among others.

Dr. Altshuler will also discuss the progress and results of three pilot projects that are the current focus of the 1000 Genomes Project Consortium. These research initiatives aim to: 1.) evaluate strategies to identify variants by combining data across multiple samples; 2.) obtain high quality reference data for the genomes of a small number of individuals, and to evaluate the benefits and costs of high coverage whole genome sequencing of two parent-offspring (i.e., mother, father and child) trios; and 3.) explore new strategies and technologies for detailed sequencing of approximately 1,000 genes from 1,000 individuals.

PUBLIC ATTITUDES ABOUT LARGE COHORT GENETIC RESEARCH

Genetic researchers agree that the nexus of genes, lifestyle, and environment is likely to yield many discoveries about the nature of many common, complex diseases, but unraveling the various factors contributing to disease would require studying large numbers of people over long periods of time.

Kathy Hudson, Ph.D., will address a recently completed public consultation project conducted by the Genetics and Public Policy Center for NHGRI. The two-year, nationwide project gauged public attitudes toward a proposed federal decade-long, large-cohort study on the overlapping effects of genetics and environment on health. She will discuss the Center’s findings, which are based on a number of focus groups, interviews and town halls conducted in five major cities (Philadelphia, PA; Portland, OR; Phoenix, AZ; Jackson, MS; and Kansas City, MO), as well as a national survey of more than 4,600 U.S. residents.

The survey and other consultation methods analyzed public support for and willingness to participate in a large-cohort study, and explored study burden, compensation, and preferences for receiving health information (including research results) as factors that might influence public support. The town halls and focus groups also identified issues related to consent and accountability that reflect differences in understanding of informed consent by potential study participants versus members of the research community.

PRESS BRIEFING SESSION & WEBCAST INFORMATION

ASHG invites members of the media to attend this press briefing session on ***"Genome-Wide Association Studies and the 1000 Genomes Project: Implications and Clinical Applications,"*** which will be held on Thursday, November 13, 2008 from 4:30-5:30 p.m. in the ASHG Press Briefing Room (Room 307A), located on the third level of the Pennsylvania Convention Center.

To view the archived webcast recording of this event, please click on the link below:
http://hosted.mediasite.com/hosted4/Viewer/?peid=4544181a99d9490887db45328f1732cB/

Please direct all other media inquiries to ASHG Press Office, ASHG Communications Manager, via e-mail at press@ashg.org, or by phone at .

ABOUT THE AMERICAN SOCIETY OF HUMAN GENETICS

Founded in 1948, The American Society of Human Genetics (ASHG) is the primary professional membership organization for human genetics specialists worldwide. The nearly 8,000 members of ASHG include researchers, academicians, clinicians, laboratory practice professionals, genetic counselors, nurses and others involved in or with a special interest in human genetics.

The Society's mission is to serve research scientists, health professionals and the public by providing forums to: (1) share research results through the Annual Meeting and in *The American Journal of Human Genetics* (*AJHG*); (2) advance genetic research by advocating for research support; (3) educate future genetics professionals, health care providers, advocates, teachers, students and the general public about all aspects of human genetics; and (4) promote genetic services and support responsible social and scientific policies. For more information about ASHG, please visit http://www.ashg.org/.

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