PRESS RELEASE

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Media Contact:
ASHG Press Office
240-281-2386
press@ashg.org

New Genetics Research Findings Expand Our Understanding of How Human Populations Have Evolved

Researchers Present Latest Evolution and Population Genetics Study Results at The American Society of Human Genetics 59th Annual Meeting in Honolulu

BETHESDA, MD – October 22, 2009 – Thousands of the world's top scientists and clinicians in the human genetics field will convene to present their latest research at the 59th Annual Meeting of The American Society of Human Genetics (ASHG) in Honolulu, Hawaii, on October 20-24, 2009.

ASHG is the primary professional membership organization for human genetics specialists worldwide, representing nearly 8,000 researchers, academicians, clinicians, genetic counselors, nurses and others with a special interest in the field. The Society's Annual Meeting is the world's largest gathering of human genetics professionals and a forum for renowned experts in the field to share their latest research results.

At the ASHG 2009 Annual Meeting, a number of scientific presentations will provide information about important new research findings that advance our current understanding of human evolutionary history and population genetics.

ASHG will also host a press briefing session to highlight three scientific presentations at the 2009 Annual Meeting that report findings on the latest advances in evolution and population genetics research addressing questions of where humans originated, and how far we have come in evolutionary history.

ASHG invites members of the media to attend this press briefing titled, *"New Genetics Research Findings Expand Our Understanding of How Human Populations Have Evolved,"* which will be held on Thursday, October 22, 2009, from 12:00-1:00 p.m. (HST) in the ASHG Press Briefing Room, located on the third level of the Hawaii Convention Center (Room #319A). Members of the press who cannot attend this event in person can register to view the webcast of this session that will be posted online about two to three hours after the event has ended (*see section in red below for more information about the webcast).

The presentations included in this press briefing will report on: new research methods that will allow for a more uniform and representative sampling of human genetic diversity in populations around the world; a study of the complex genetic history of human populations in East Africa (a traditionally underrepresented group) that provides a first look into how the genetic ancestry of East Africans correlates with – and is influenced by – geographic and linguistic differences; as well as the exciting discovery of a new gene that plays a major role in human vocal development and speech. The speakers and abstracts featured in this press briefing will include the following:

- Lynn Jorde, PhD – *"Toward a More Uniform Sampling of Human Genetic Diversity: A Survey of Worldwide Populations by High-density Genotyping"*
- Sarah Tishkoff, PhD – *"Complex Genetic History of Human Populations in East Africa"*
- Raymond Clarke, PhD – *"Tospeak, a Novel Gene Involved in Human Vocal Development and Speech, Identified by Australian-based Research Team"*

Brief summaries of the major research findings reported in these three abstracts are included below:

Toward a More Uniform Sampling of Human Genetic Diversity: A Survey of Worldwide Populations by High-density Genotyping

Many studies of human genetic variation have been based on restricted samples of human populations (focusing, for example, on Northern Europe, Eastern Asia and sub-Saharan Africa). Consequently, differences among groups of human populations have sometimes been exaggerated, and traditional "racial" groups are perceived to be more discrete and well-defined than they really are.

To provide a more complete picture of human diversity, Lynn Jorde, PhD, professor and chairman of the Department of Human Genetics at the University of Utah School of Medicine, and his research team have begun to focus on studying previously un-sampled portions of the world. The current research reports new high-density genotype data (250,000 to 1,000,000 DNA variants) on several hundred individuals from regions such as Western, Central, and Southern Asia and the Middle East.

"High-throughput genotype data are useful for making inferences about human evolutionary history on a fine scale. However, populations sampled to date are distributed unevenly across the globe, and some areas, such as Western and Southern Asia, have rarely been sampled in large-scale studies," said Dr. Jorde.

Using standard measures of genetic differentiation on a total sample of more than 1,300 individuals, Jorde's team found that the average differences among continental populations are reduced by at least one-third when using a more complete human diversity sample.

Their research also shows how these data can be used to infer patterns of ancient human migration and genetic drift. For example, more detailed analyses of Eurasian populations suggest complex genetic structure in populations from Central and South Asia – including samples from Kyrgyzstan, Mongolia, Nepal, Pakistan and India. These results suggest high rates of gene flow in Central Asia, consistent with the position of this region as a cross-road between East and West.

"Our research results provide insight into the patterns of human genetic variation and ancient migration of populations in previously under-sampled regions of the world," said Dr. Jorde. "In addition, genotypes generated in this study from previously under-represented populations can serve as a resource for future studies of human genetic variation."

Complex Genetic History of Human Populations in East Africa

Results from disparate fields of research indicate that modern Homo sapiens originated in Africa around 200,000 years ago, and that East Africa is the likely source of migration of modern humans out of Africa within the past 100,000 years. However, Africa – especially East Africa – has not been well studied for human genetic diversity compared to non-African populations, probably due to the fact that DNA samples from many regions of Africa are currently not available.

Sarah Tishkoff, PhD, Associate Professor in the Departments of Genetics and Biology at the University of Pennsylvania School of Medicine, and her team of researchers sought to remedy this gap in the current genetics research on this population. The goal of their study was therefore to sample 1,500 individuals from

previously unstudied East African populations in order to characterize their mitochondrial DNA (mtDNA) and Y chromosome genetic variation.[1] These data were then compared to independently collected data of the same populations from 1,327 variable sites in the nuclear genome. The data was used to gain insight into patterns of genetic diversity, to construct past relationships of East African populations to each other and to other African populations, and to clarify historical demographic events such as population expansion, contraction and migration that these populations might have experienced.

"Our research indicates that there is significant correlation between genetic and geographic/linguistic distances among East African populations," said Dr. Tishkoff. "However, the genetic correlation with geography is stronger than it is with linguistics."

"Overall, the correlations between genetic versus geographic/linguistic variation are stronger for autosomal and paternal lineage data than for maternal lineages," she explained. "In addition, our research results indicate that paternal and maternal lineage distributions seem to cluster geographically, and for some lineages, linguistically."

The current research findings also suggest that two major migration events within the past 5,000 years have had a major influence on extant patterns of genetic variation in East Africa – the migration of Bantu-speaking populations from Central/West Africa across sub-Saharan Africa, and the migration of pastoralist populations from Sudan and Ethiopia.

Tospeak*, a Novel Gene Involved in Human Vocal Development and Speech, Identified by Australian-based Research Team*

An international team of scientists based in Australia has made an important recent breakthrough by identifying a novel gene involved in vocal development and the human capacity for speech. Because the gene was disrupted in a large family with a severe vocal disorder, the researchers named this new gene '*tospeak*.'

A team of medical scientists, headed by Raymond Clarke, PhD, Principal Scientific Officer at St. George Clinical School of Medicine, University of New South Wales (NSW) in Sydney, Australia, screened the affected family for changes in their DNA. The researchers investigated blood and DNA samples from the family using the latest techniques in gene analysis.

The most exciting breakthrough in their research came when Clarke's group discovered that the *tospeak* gene was unique to primates. Most of the human genome contains genes that are older (i.e., conserved over generations) and can also be found in other mammals, including the mouse. However, the *tospeak* gene is a relatively young gene that is only found in primates. Further excitement came when the group discovered that the *tospeak* gene has a special control region, known as a promoter, which is only found in humans.

"The discovery that a unique and more powerful human gene/promoter was disrupted in this vocally impaired family is of particular interest to the field of evolutionary genetics, since humans are the only creatures that have developed the capacity to speak," said Dr. Clarke.

Clarke provided the following example as a comparison to help explain this new discovery: "Unlike *GDF6*, a bone protein gene which has existed since the dawn of vertebrate evolution, the *tospeak* gene is only found in primates. The best indication of the role of *tospeak* in human vocal development is that it was the only gene disrupted in a large family with a severe vocal disorder, altered composition of the vocal cords, and malformation of the voice box."

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[1] The types of markers used in genetic ancestry testing may be *patrilineal*, as is the case with tests that use Y chromosome markers, or *matrilineal*, as is the case with tests using mitochondrial DNA (mtDNA) markers.

"*Tospeak* is a very large gene found in primates that overlaps two other neighboring genes. While we still don't know exactly how these genes function, we believe that these three genes probably act together to influence human vocal development through the regulation of gene transcription, the biochemical composition of the vocal cords, and the anatomical structure of the voice box which underlie the human capacity for speech," Clarke said. "However, research is currently underway to determine exactly how *tospeak* functions in regulating the human capacity for speech."

PRESS BRIEFING SESSION & WEBCAST INFORMATION

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For those members of the press who wish to view the session remotely via webcast, please note that the online webcasts of all three ASHG Press Briefing Sessions held at this year's Annual Meeting, will *NOT* be webcast live, in real time. To view the *archived* webcast recording of this event, please click on the link below (no sooner than) two to three hours after the press briefing session has ended *(i.e., ~4:00 p.m. HST / 9:00 p.m. EST) on Thursday, October 22, 2009*:
http://www.ashg.org/populationgenetics/

Please direct all media inquiries to ASHG Press Office, ASHG Communications Manager, via e-mail at press@ashg.org, or by phone at 240-281-2386.

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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