



## PRESS RELEASE

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## Next-Generation Genome Sequencing Methods Reveal New Findings on Evolution and Population Genetics

*Researchers present study results at ICHG/ASHG 2011 Meeting that reveal new insights on genetic ancestry of ancient and modern human populations and their evolutionary history*

BETHESDA, MD – September 29, 2011 – The world's top scientists and clinicians in the human genetics field will gather to present their latest research findings at the [12th International Congress of Human Genetics \(ICHG\) / 61st Annual Meeting of the American Society of Human Genetics \(ASHG\)](#), which will be held on October 11-15, 2011, at the Montreal Convention Center (Palais des Congrès) in Montreal, Quebec, Canada. A number of scientific presentations at the ICHG/ASHG 2011 Meeting will provide information about important new research findings that expand our current understanding of human evolutionary history and population genetics.

ASHG will be hosting a press briefing session (which will also be made available to the media as an online webcast) to highlight four presentations of interest on this topic at the ICHG/ASHG 2011 Meeting that report on exciting new evolution and population genetics research findings that have resulted from the use of next-generation genome sequencing technologies and novel methods of analyzing gene sequence data. The findings presented in this press briefing will shed light on the genetic ancestry of ancient and modern human populations (including one presentation on the genomic reconstruction of an ancient, extinct population), how they have evolved, and new insights into their evolutionary history. The abstracts featured in this press briefing will also address new findings that have come from tracing genetic admixture (i.e., genetic mixing) among members of different populations, and a new method that allows researchers to decipher relationships among individuals in a population with extreme accuracy by estimating recent shared ancestry based on their sequence data.

ASHG invites members of the media to attend this press briefing session entitled, “*Next-Generation Genome Sequencing Methods Reveal New Findings on Evolution and Population Genetics*,” which will be held on Thursday, October 13, 2011, from 3:00-4:00 p.m. in the ICHG/ASHG Press Briefing Room, located in Room 518A on the fifth floor of the Montreal Convention Center. [To view the online webcast recording of this press briefing event, please go to the following link: <http://www.ashg.org/ICHG2011/EvoPopulationGenetics>](http://www.ashg.org/ICHG2011/EvoPopulationGenetics) (\*see section below for more information about this webcast).

After a brief introduction by session moderator and President of ASHG, [Dr. Lynn Jorde, PhD](#), each of the following four press briefing speakers will discuss their abstracts presented at the ICHG/ASHG 2011 Meeting on important new evolution and population genetics research findings that have only recently been made possible through the latest advances in next-generation genome sequencing technologies and methods:

- **Lynn B. Jorde, PhD (Moderator)** – [“Using Whole-genome Data to Reveal Distant Relationships Between Individuals by Analysis of Genomic Segments Shared Identically by Descent”](#)
- **Lalji Singh, PhD** – [“Genetic Diversity in Indian Populations and its Health Implications”](#)
- **Carlos D. Bustamante, PhD** – [“Genomic Reconstruction of an Extinct Population from Next-Generation Sequence Data: Insights from the Taino Genome Project”](#)
- **David E. Reich, PhD** – [“Denisova Genetic Admixture and the First Modern Human Population Dispersals into Southeast Asia and Oceania”](#)

Brief summaries of the content covered in these four presentations are included in the section below:

### **[Using Whole-genome Data to Reveal Distant Relationships Between Individuals by Analyzing Genomic Segments Shared Identically by Descent](#)**

**[Lynn B. Jorde, PhD](#)**, *ASHG President; Chair and Professor, Dept. of Human Genetics, University of Utah School of Medicine*

Accurate estimation of recent shared ancestry is important in genetics, evolution, medicine, conservation biology, and forensics. Chromosomal segments shared by two individuals due to identity by descent (IBD) from a recent shared ancestor can be estimated from whole-genome data. The information gathered from the analysis of IBD segments allows the detection and accurate estimation of genetic relationships between pairs of individuals as distant as fifth cousins.

**[Lynn B. Jorde, PhD](#)**, and his research team at the University of Utah established a new, more accurate method to estimate familial relationships from whole-genome data using [Estimation of Recent Shared Ancestry \(ERSA\)](#) – a novel piece of software that was developed by his lab ([Huff, Witherspoon, et al. \*Genome Research\*, 2011](#)). The research team applied ERSA in an analysis of 1000 Genomes Project sequence data and Human Genome Diversity Project (HGDP) SNP data to identify relationships among the sampled individuals. In their analysis, hundreds of previously unknown relationships were identified with high confidence in the 1000 Genomes Project samples, and several thousand were detected in the HGDP samples. First- and second-cousin pairs were identified in nearly all 1000 Genomes population samples, as well as in some of the larger HGDP sample sets. Numerous more distant relationships were observed in all samples. In some HGDP samples (e.g., Amazonian groups, Mexican Pima and Maya, Siberian Yakut, Pakistani Kalash, and the Melanesian samples), all pairs of individuals were at least third cousins or more closely related. In samples from Pakistan, the Bedouin, Druze, Palestinian and Mozabite from the Middle East, and in the Basque HGDP sample, more than half of relationships were third-cousin or closer. However, other HGDP population sample sets showed lower levels of relatedness, such that only 5-30% of relationships were at the third-cousin level or closer (e.g., the African Mandenka and Biaka Pygmy, the Makrani, Hazara and Balochi of Pakistan, and Italian Sardinians). In the majority of 1000 Genomes Project samples, and in the HGDP Han Chinese and Pakistani samples, fewer than 1% of pairs of individuals were third cousins or closer.

The findings from the current analysis demonstrate that relationships between individuals from different population samples are uncommon and nearly always distant. The relationships that were found between individuals in this study reflect what is known about recent demographics and mating patterns in these populations. The new method of IBD estimation using the ERSA software greatly expands the relationships that can be estimated from genetic data, and with such precise and detailed estimation of relationships it will be possible to draw stronger and more exact inferences about recent effective population sizes and mating patterns. In addition, this method can be applied in forensic settings to help to identify missing persons through DNA comparisons with their known relatives.

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### **[Genetic Diversity in Indian Populations and its Health Implications\\*](#)**

**[Lalji Singh, PhD](#)**, *Bhatnagar Fellow, Centre for Cellular and Molecular Biology (CCMB), Council of Scientific and Industrial Research (CSIR), Hyderabad, India; Vice Chancellor, Banaras Hindu University, Varanasi, India*

As the structure, sequence, and function of the human genome are defined, scientists are using that information to understand the origins of populations as well as the origins of specific diseases. Researchers studying the evolution of human populations with respect to the origins and frequency of genetic diseases have recently made major advances, and have greatly increased our understanding of inherited disorders. [Lalji Singh, PhD](#), Director of the Centre for Cellular and Molecular Biology at the Council of Scientific and Industrial Research (CSIR) in India, will be presenting an overview of his research on “[Genetic Diversity in Indian Populations and its Health Implications](#)” in a Plenary Session titled, “[Diseases, Populations and Evolution: From the Cellular to the Continental](#),” at the ICHG/ASHG 2011 Meeting.

In recent years, maps of human genetic variation have expanded our understanding of the diversity of populations across the world, yet India has been largely unrepresented until now. India represents one of the most genetically diverse populations in the world. There are 4,635 anthropologically well-defined populations with little or no gene flow between them. Differences between these groups reflect geographic, caste, and other cultural barriers to interbreeding. Genetic variations among these groups can also be related to differences in the distribution and frequency of specific genetic disorders.

In an effort to address this research gap, Singh and his colleagues analyzed genetic variation in 132 Indian samples from 25 different population groups that represent all the major language families, social groups, and geographical regions in India. They found strong evidence for the existence of two ancient and genetically divergent populations that are ancestral to most Indian groups today, namely: the Ancestral North Indians (ANI) and the Ancestral South Indians (ASI). The ANI are genetically close to Middle Easterners, Central Asians, and Europeans, while the ASI are not close to any group outside the subcontinent. However, they show genetic affinities with the Andamanese, who are thought to be the first modern human population that migrated out of Africa.

Following the ANI and ASI admixture, many groups experienced periods of genetic isolation from each other for thousands of years. In scientific parlance this is called a “founder event.” It has medical implications for Indian populations, because recessive hereditary diseases are likely to be common in populations descended from so few ‘founder’ individuals. This helps to explain why the incidence of genetic diseases among Indians is different from the rest of the world. For example, an ancient deletion of 25 base pairs in the cardiac myosin-binding proteins-C gene (MYBPC3) is associated with heritable cardiomyopathies, as well as with an increased risk of heart failure. Its prevalence is high (~4 %) in the general populations from the Indian subcontinent. However, this mutation is completely absent among people from the rest of the world. It is important to carry out a systematic survey of the different Indian population groups to identify which ones descend from the strongest founder events. Further studies of these groups should lead to the rapid discovery of genes that cause devastating hereditary diseases, which will in turn aid in prevention and clinical care of affected individuals and their families who are at risk.

*[\*Presented in ICHG 2011 Plenary Session: [“Diseases, Populations and Evolution: From the Cellular to the Continental”](#)]*

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### **[Genomic Reconstruction of an Ancient, Extinct Population Using Next-Generation Sequence Data: Insights from the Taíno Genome Project](#)**

[Carlos D. Bustamante, PhD](#), Professor, Department of Genetics, Stanford University School of Medicine

The first Native American people encountered by Europeans across the Caribbean were the indigenous pre-Columbian inhabitants of the Bahamas, Greater Antilles, and Lesser Antilles; they were given the collective name “Taínos” by the arriving Spaniards. One hundred years after this initial contact, the Taíno population was effectively eliminated due to war, slavery, suicide, hunger, and disease. Today, the ancestral legacy of the Taínos is found in traces of their genomes still present in the current inhabitants of the Caribbean islands. An incredible opportunity to recover these ancestral lineages came about when 35 Puerto Rican trios were included in the sampling plan of the 1,000 Genomes Project. Due to the complex history of Puerto Rico, the genomes of its inhabitants show three-way genetic admixture including ancestry from European, African, and Taíno origins.

A team of researchers led by [Carlos D. Bustamante, PhD](#), a Professor in the Department of Genetics at Stanford University School of Medicine, analyzed the genomes of the Puerto Rican samples from the 1,000 Genomes Project data to infer the ancestral origin of each locus along the genome for all individuals. By examining patterns in this genomic data, the researchers hoped to gain genetic insights into the Taíno population and a deeper understanding of the colonial history of Puerto Rico. Bustamante and his colleagues were able to infer various aspects of the demographic history by analyzing the distribution of lengths of Taíno ancestry “tracts,” or chromosomal segments, combined with recent migrant ancestry. By analyzing the lengths and frequencies of contiguous ancestry tracts, Bustamante’s research team was able to infer various aspects of demographic history on the island. For example, they found that Taíno ancestry tracts are relatively short with lengths that are exponentially distributed, which implies that the Taíno ancestry that is present today in the Puerto Rican population is due to a single pulse of admixture from approximately 17 generations ago. On the other hand, both European and African ancestry tract length distributions show an abundance of long tracts, which is suggestive of continued African and European migration to the island. This finding is consistent with historical accounts of colonization. Furthermore, the researchers found that there is a strong correlation in European ancestry proportions in husbands and wives, which might be due to assortative mating. The results of their analysis also show higher proportions of Taíno ancestry on the X chromosome, which is likely due to sex-biased migration.

Using their ancestry inference data coupled with high-throughput genome sequencing data allowed Bustamante’s team to examine additional research questions. For example, they used patterns of sequence

variation observed in regions of homozygous ancestry, where both the maternal and paternal chromosomes have the same ancestral origin, to infer the time to the most recent common ancestor and effective population sizes for each source population. This information might be particularly interesting for the Taino source population, as there has been some dispute about the population size at the time of European contact. Also, although the average proportion of the genome that is of Taino origin is small, the location of these Taino ancestry tracts is different in each individual. Thus, when considering all Puerto Rican individuals together, a large proportion of the genome is overlapped by one or more chromosomes of Taino origin, making it possible for Bustamante's team to assemble a Taino consensus genome sequence that includes any variation unique to this population. In summary, Bustamante's team was able to identify Taino-specific genomic variation, cataloging what remains of this lost ancestral lineage. This is the first known reconstruction of the genomic variation of an extinct human population using modern sequence data.

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### **[Genetic Analysis of Archaic Hominin DNA Reveals Ancient Migration Patterns of the First Modern Humans to Populate Southeast Asia and Oceania\\*](#)**

[David E. Reich, PhD](#), Professor, Department of Genetics, Harvard Medical School

The history of the earliest arrival of modern humans in Southeast Asia and Oceania from Africa has been a source of unresolved controversy. Archaeological evidence supports either a single wave of settlement or, alternatively, multiple waves of settlement, the first leading to the initial peopling of Southeast Asia and Oceania. Recent research findings indicate that ancestors of New Guineans and Bougainville Islanders have inherited about 4-6% of their ancestry from Denisovans, an ancient hominin group from Siberia. This finding provides insights for understanding the earliest human migrations to the region. This information could resolve the current controversy over the number of waves of settlement that occurred. However, in previous studies only a sparse sampling of populations from Southeast Asia and Oceania were analyzed.

A group of researchers led by [David Reich, PhD](#), a Professor in the Department of Genetics at Harvard Medical School, analyzed genome-wide data from an additional 33 populations from south Asia, Southeast Asia, and Oceania for traces of genetic admixture that matches up with the Denisova from Siberia. Reich and his colleagues then used the results of their analysis to infer the history of human migration(s) to this part of the world. The group's study results are published in the October 2011 issue of ASHG's scientific journal, *The American Journal of Human Genetics (AJHG)*, in a paper titled, "[Denisova Admixture and the First Modern Human Dispersals into Southeast Asia and Oceania](#)."\*

The results of Reich's analysis indicate that Aboriginal Australians, Near Oceanians, Polynesians, Fijians, east Indonesians, and Mamanwa) populations have all inherited genetic material from the Denisovans of Siberia, but mainland East Asians, western Indonesians, and the Jehai and Onge groups from Malaysia and Andaman Islands have not. These results suggest that the gene flow from Denisovans occurred into the common ancestors of New Guineans, Australians, and Mamanwa, but not into the ancestors of the Jehai and Onge. The results also suggest that relatives of present-day East Asians were not in Southeast Asia when the gene flow occurred, rather they descended from later migrations. The finding that descendants of the earliest inhabitants of Southeast Asia did not all harbor genes from the Siberian Denisovans is inconsistent with a history in which the Denisova populated mainland Asia and then spread over Southeast Asia, leading to all of the earliest modern human inhabitants in these areas.

Overall, Reich's study findings suggest that Southeast Asia was settled by modern humans in multiple waves: the first wave produced the ancestors of present-day Onge, Jehai, Mamanwa, New Guineans, and Australians (some of whom interbred with Denisovans), and a second wave contributed much of the ancestry of present-day East Asians and Indonesians. Furthermore, their findings also suggest that archaic Denisovans must have lived over an extraordinarily broad geographic and ecological range that extended from Siberia to tropical Asia.

[\***CITATION:** [Reich et al., Denisova Admixture and the First Modern Human Dispersals into Southeast Asia and Oceania, The American Journal of Human Genetics \(2011\), doi:10.1016/j.ajhg.2011.09.005](#)]

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### **PRESS BRIEFING SESSION & WEBCAST INFORMATION**

ASHG invites members of the media to attend this press briefing at the ICHG/ASHG 2011 Meeting on the latest advances in evolution and population genetics research, which will be held on Thursday, October 13, 2011, from

3:00-4:00 p.m. in the ICHG/ASHG Press Briefing Room (located in Room 518A on the fifth level of the Montreal Convention Center). To register onsite as a member of the media, you must bring your press credentials\* with you to the ICHG/ASHG Press Office in Room 518B (Level 5) upon arrival at the Montreal Convention Center in order to obtain a press badge.

Please note that the online webcast recordings of ASHG's press briefing events at the ICHG/ASHG 2011 Meeting will *not* be webcast live. **To view the online webcast of this press briefing, click on the link below about an hour after the event has ended** (the webcast will be posted on ASHG's Web site at the following URL by 5:00 p.m. on Thursday, Oct. 13, 2011): <http://www.ashg.org/ichg2011/evopopulationgenetics/>.

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### **About The American Society of Human Genetics**

Founded in 1948, the [American Society of Human Genetics \(ASHG\)](http://www.ashg.org) 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 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 [ASHG Annual Meeting](#) and in the [American Journal of Human Genetics \(AJHG\)](#); (2) advance genetic research by advocating for research support; (3) educate current and future genetics professionals, health care providers, advocates, policymakers, educators, students, and the 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 our Web site at: <http://www.ashg.org>.

### **About The International Congress of Human Genetics**

The [12th International Congress of Human Genetics \(ICHG\)](http://www.ichg2011.org), which is sponsored by the American Society of Human Genetics (ASHG) in partnership with the International Federation of Human Genetics Societies (IFHGS), is the foremost meeting of the worldwide human genetics community, and it is held every five years. In 2011, the 12th ICHG Meeting will be the major focal point and platform for the world's most exciting research, and attendance at this year's meeting is anticipated to be more than 7,000 delegates from over 60 countries around the world. For more information about the ICHG Meeting, go to: <http://www.ichg2011.org>.

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*\*Please note that all press registrants will be required to provide appropriate media credentials and identification to ASHG Press Office staff before receiving a press badge to access meeting events.*

### **ASHG EXPERTS & PRESS BRIEFING SPEAKERS AVAILABLE FOR INTERVIEW**

Please contact Kristen Long via e-mail at [press@ashg.org](mailto:press@ashg.org), or call 301-634-7346.