

Session Information

Session Title: Evolutionary and Population Genetics **Session Type:** Poster

Session Location: Exhibit Hall, Lower Level South, Moscone Center **Session Time:** Wed 10:00AM-4:30PM

Abstract Information

Program Number: 3377W **Presentation Time:** Wed, Nov 7, 2012, 2:15PM-3:15PM

Keywords: Evolutionary and Population Genetics, KW140 - population genetics, KW141 - population structure, KW159 - SNP analysis/discovery, KW081 - genomic methodologies, KW110 - microarrays

Abstract Content

The GenoChip: a new tool for genetic anthropology. *S. Wells¹, E. Greenspan², S. Staats², T. Krahn², C. Tyler-Smith³, Y. Xue³, S. Tofanelli⁴, P. Francalacci⁵, F. Cucca⁶, L. Pagani⁷, L. Jin⁸, H. Li⁸, T. G. Schurr⁹, J. B. Gaieski⁹, C. Melendez⁹, M. G. Vilar⁹, A. C. Owings⁹, R. Gomez¹⁰, R. Fujita¹¹, F. Santos¹², D. Comas¹³, O. Balanovsky¹⁴, E. Balanovska¹⁴, P. Zalloua¹⁵, H. Soodyall¹⁶, R. Pitchappan¹⁷, G. Arun Kumar¹⁷, M. F. Hammer¹⁸, B. Greenspan², E. Elhaik¹⁹* 1) Mission Programs, National Geographic Society, Washington, DC; 2) Family Tree DNA, Houston, TX; 3) Wellcome Trust Sanger Institute, Hinxton, UK; 4) University of Pisa, Italy; 5) University of Sassari, Italy; 6) National Research Council, Monserrato, Italy; 7) University of Cambridge, UK; 8) Fudan University, Shanghai, China; 9) University of Pennsylvania, Philadelphia, PA; 10) CINVESTAV, Mexico City, Mexico; 11) University of San Martin de Porres, Lima, Peru; 12) Federal University of Minas Gerais (UFMG), Belo Horizonte, Brazil; 13) Pompeu Fabra University, Barcelona, Spain; 14) Russian Academy of Medical Sciences, Moscow, Russia; 15) Lebanese American University, Byblos, Lebanon; 16) University of the Witwatersrand, Johannesburg, South Africa; 17) Chettinad Academy of Research and Education, Chennai, India; 18) University of Arizona, Tucson, AZ; 19) Johns Hopkins University, Baltimore, MD.

Background: The Genographic Project is an international effort aimed at charting human history using genetic data. The project is non-profit and non-medical, and through the sale of its public participation kits it supports cultural preservation efforts in indigenous and traditional communities. To extend our knowledge of the human journey, interbreeding with ancient hominins, and modern human demographic history, we designed a genotyping chip optimized for genetic anthropology research. **Methods:** Our goal was to design, produce, and validate a SNP array dedicated to genetic anthropology. The GenoChip is an Illumina HD iSelect genotyping bead array with over 130,000 highly informative autosomal and X-chromosomal SNPs ascertained from over 450 worldwide populations, ~13,000 Y-chromosomal SNPs, and ~3,000 mtDNA SNPs. To determine the extent of gene flow from archaic hominins to modern humans, we included over 25,000 SNPs from candidate regions of interbreeding between extinct hominins (Neanderthal and Denisovan) and modern humans. To avoid any inadvertent medical testing we filtered out all SNPs that have known or suspected health or functional associations. We validated the chip by genotyping over 1,000 samples from 1000 Genomes, Family Tree DNA, and Genographic Project populations. **Results:** The concordance between the GenoChip and the 1000 Genomes data was over 99.5%. The GenoChip has a SNP density of approximately (1/100,000) bases over 92% of the human genome and is highly compatible with Illumina and Affymetrix commercial platforms. The ~10,000 novel Y SNPs included on the chip have greatly refined our understanding of the Y-chromosome

phylogenetic tree. By including Y and mtDNA SNPs on an unprecedented scale, the GenoChip is able to delineate extremely detailed human migratory paths. The autosomal and X-chromosomal markers included on the GenoChip have revealed novel patterns of ancestry that shed a detailed new light on human history. Interbreeding analysis with extinct hominids confirmed some previous reports and allowed us to describe the modern geographical distribution of these markers in detail. **Conclusions:** The GenoChip is the first genotyping chip completely dedicated to genetic anthropology with no known medically relevant markers. We anticipate that the large-scale application of the GenoChip using the Genographic Project's diverse sample collection will provide new insights into genetic anthropology and human history.

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