

The American Society of Human Genetics
55th Annual Meeting

■ Program Addendum ■

■ Special Notice ■

Due to security reasons and overcrowding in session rooms in previous years, **badges must be worn and visible at all times** and will be checked at all scientific sessions, including the Social Issues and Education Sessions on Wednesday morning. Individuals who have not registered or who have lost or forgotten their badges will not be admitted to sessions or to the exhibit and poster areas. Registrants who lose badges may obtain a replacement badge for a charge of \$5.

■ C. W. Cotterman Award Winners ■

Eric Jorgenson and Mingyao Li. Their winning papers are:

Ethnicity and Human Genetic Linkage Maps. Eric Jorgenson, Hua Tang, Maya Gadde, Mike Province, Mark Leppert, Sharon Kardia, Nicholas Schork, Richard Cooper, D. C. Rao, Eric Boerwinkle, Neil Risch (2005) *Am J Hum Genet* 76:276-290.

Joint Modeling of Linkage and Association: Identifying SNPs Responsible for a Linkage Signal. Mingyao Li, Michael Boehnke, Gonçalo R. Abecasis (2005) *Am J Hum Genet* 76:934-949.

■ Education Grant Acknowledgment ■

The American Society of Human Genetics gratefully acknowledges an education grant from **Merck Research Laboratories** to support the 55th annual ASHG meeting.



■ Program Updates ■

■ TUESDAY, October 25

Asterisks (*) next to the times of events denote meetings that the organizers have indicated are open to all ASHG meeting registrants. Events without an asterisk are limited to invitees or those who pay required fees.

Ancillary Event Added:

2:00 PM-3:30 PM **ICHG Program Committee Meeting**, Convention Center, Ken Knight Board Room

Ancillary Events Changed:

9:00 AM-12 Noon **ABGC Working Groups Meeting** **MOVED TO Grand America Hotel, Venice Room**

10:00 AM-12:00 Noon **ABMG Item Writing Workshop** **MOVED TO** Grand America Hotel, Imperial C

*1:00 PM-2:30 PM **Global Organisation for Lysosomal Diseases Annual General Meeting**, Hilton Hotel, Alpine Ballroom. **TIME AND LOCATION CHANGED TO** 12:30 PM-2:00 PM, Little America Hotel, Arizona Room

Ancillary Event Cancelled:

6:30 PM-8:30 PM **Exploring the Mechanisms of Disease Using Affymetrix® DNA Analysis Products** (originally scheduled on Thursday, October 27)

■ WEDNESDAY, October 26

Asterisks (*) next to the times of events denote meetings that the organizers have indicated are open to all ASHG meeting registrants. Events without an asterisk are limited to invitees or those who pay required fees.

Ancillary Events Added:

5:00 PM-7:00 PM **ACMG Panel Discussion: Promoting Safe and Effective Genetic Testing Services**, Grand America Hotel, Hermitage Room

6:30 PM-8:00 PM **ASHG Information and Education Subcommittee on Undergraduate Education**, Convention Center, Ken Knight Board Room

7:00 PM-9:00 PM **ESHG Board Meeting**, Convention Center, Room 151A/B

Poster Changed:

1598/W Candidate gene analysis of myopia. Correct spelling of author names: M. E. Cooper, S. E. O'Brien, K. S. Zadick, J.C. Murray, M.L. Marazita, D. O. Mutti

Posters Cancelled:

908/W A transcriptional function for SRY in mammalian sex determination. V. Harley

1295/W Genomewide survey of gene copy number change spanning over 20 million years of human and primate evolution. J. Sikela

1736/W The promoter polymorphism of HMOX1 and cardiovascular disease. J. Pohorence Ferguson

(Wednesday, October 26, continued)

Posters Added:

1667A/W A high density SNP panel in the MHC region. L. Galver, S. S. Murray, Illumina, Inc., San Diego, CA.

The major histocompatibility complex (MHC) is a ~4 Mb gene-dense region of the human genome on Chromosome 6p21. There are over 160 RefSeq genes in this region of which ~40% encode proteins involved in immune defense including the human leukocyte antigen (HLA) membrane glycoproteins involved in recognition of T lymphocytes. Since the classical HLA loci represent a minority of genes found in the MHC region, it is likely that many disease-causing mutations may actually reside outside one of the classical HLA genes. Therefore, since almost every autoimmune and inflammatory disorder is studied in this genomic region, we have developed a panel of 2,390 SNPs that can be used as an efficient method for fine mapping the MHC region for identification of genes associated with disease phenotypes. In addition, this panel can be used to discriminate between causal alleles and variation that is in linkage disequilibrium (LD) with causal alleles. Content was chosen based on SNPs being within 10kb of coding sequences of genes in the MHC region, spanning from ret finger protein (RFP) to motilin (MLN). There is at least 1 SNP within 10kb of 159 RefSeq genes, representing 94% of all RefSeq genes in the MHC region (average 11 SNPs per gene). In addition, SNPs were chosen based on being a tag SNP in a Caucasian population and/or based on even spacing throughout the MHC region (average spacing is 2kb). Haplotype structure and diversity have also been investigated in the 3 HapMap plates (average block size = 16.2 kb, 15.8kb, 14 kb in the CEU, CHB/JPT, and YRI populations, respectively; average number of haplotypes = 223, 215, and 207 in the CEU, CHB/JPT, and YRI populations, respectively). Given the MHC region is one of the most difficult regions of the genome to interrogate, this panel of SNPs offers a novel resource for genetic studies in the MHC region.

1667B/W Whole genome genotyping on BeadChips using ASPE and SBE. K. L. Gunderson, F. J. Steemers, W. Chang, G. Lee, C. Tsan, D. Bullis, J. Musmacker, L. Zhou, S. Murray, P. C. Ng, K. Kuhn, D. Barker, R. Shen. Illumina, Inc., San Diego, CA.

We have developed a whole genome genotyping (WGG) assay that combines hybridization capture of whole genome amplified products with array-based allele-specific primer extension (ASPE) to score captured SNP targets (Gunderson et al., Nat Genet. 37:549-554, 2005). This assay design combines inherently unlimited multiplexing potential with relatively unconstrained SNP selection. We have deployed this assay on our high-density BeadChip platform to create an exon-centric genotyping array containing over 100,000 assays including over 23,000 assays directly in transcripts. The average SNP spacing is 28 kb with a median spacing of 13 kb. We assessed genotyping quality by analyzing 138 samples selected from among European (CEU), Han Chinese/Japanese (CHB/JPT), and Yoruba (YRI) populations. The overall call rate was 99.4% (over 14 million calls) with a reproducibility of 99.99% and Mendelian Inconsistency of 0.014%. The complete genotyping process from sample preparation through

hybridization, washing, extension, and staining have been automated through the use of robotics and capillary gap flow cells. In addition to ASPE-WGG, we have developed an alternative array-based primer extension assay using single base extension (SBE). SBE doubles the information content on the array since only a single bead type per assay rather than two is employed. This assay effectively allows over 250,000 SNP to be placed on our current BeadChip substrate. We tested the feasibility of WGG-SBE by designing assays to the biallelic HapMap QC SNPs (723) compatible with our two-color readout. We found an assay conversion rate of 89.9% (650/723), a call rate of 99.85%, reproducibility of 100%, Mendelian Inconsistencies of 0.016%, and concordance with the GoldenGate® assay of 99.98%. These results indicate that SBE is a highly robust and accurate genotyping assay with the same inherent scalability features as the current ASPE-WGG assay. Furthermore, based upon these feasibility studies, we have implemented SBE in the development of our Phase I and Phase II HapMap tag SNP panels containing >250,000 and 500,000 SNPs, respectively.

1667C/W Construction of a Phase I HapMap Tag SNP Panel. S. S. Murray, P. C. Ng, L. Galver, K. L. Gunderson, D. Barker, R. Shen. Illumina, Inc., San Diego, CA.

Data generation for Phase I of the International HapMap Project is complete, and results for over 1 million SNPs genotyped in four populations are now publicly available (www.hapmap.org). We are currently developing a panel of >250,000 SNP loci chosen from the Phase I HapMap data. We use a novel assay to interrogate this large number of SNPs efficiently and accurately on a single slide. The novel assay uses a single tube whole genome amplification step that does not require PCR or any reduction of genome complexity, and uses a single base extension reaction (Gunderson et al., Nat Genet. 37:549-554, 2005; Gunderson et al., personal communication). A maximally informative set of tag SNPs were derived from the CEPH population and their utility will be assessed in the Han Chinese, Japanese, and Yoruba populations. Tag SNPs were chosen using algorithms utilizing the linkage disequilibrium statistic r^2 (Carlson et al., 2004; deBakker, <http://www.broad.mit.edu/mpg/tagger/>). Using these algorithms, >95% of all common polymorphisms in the Phase I data set (>775,000 SNPs with MAF > 0.05) are either directly assayed or exceed a threshold level of association with a tag SNP that is assayed. A higher density of tag SNPs within 10 kb of genes or in evolutionarily conserved regions were chosen by using a more stringent r^2 threshold in these genomic regions. In addition, we have included ~10,000 nsSNPs and >1,000 tag SNPs chosen from a 2 kb map of SNPs across the MHC region. The average spacing between SNP loci is ~10kb (median ~6 kb; 90th percentile ~22kb). Genomic coverage will also be assessed by comparing all common variation in ten 500kb genomic regions characterized in the HapMap project to the tag SNPs chosen in this panel within the same genomic regions. This panel of tag SNPs will provide a valuable resource for whole genome genotyping studies and utilizes the vast information generated from the Phase I HapMap project.

■ THURSDAY, October 27

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Ancillary Events Added:

1:00 PM-2:00 PM **European Journal of Human Genetics**, Convention Center, Ken Knight Board Room

1:00 PM-2:00 PM **Lunchtime Symposium: Whole Genome Amplification Using Phi29 DNA Polymerase**. Sponsored by GE Healthcare. Grand America Hotel, Audubon Room

7:00 PM-10:00 PM **Neurofibromatosis Symposium**, Grand America Hotel, Grand Ballroom A

Ancillary Events Changed:

6:30 PM-8:30 PM **How to Use the HapMap Data. TITLE AND REGISTRATION REQUIREMENT CHANGED TO HapMap Tutorial: How to Use the HapMap Data.** Registration by October 14 is required (limited to the first 1000 registrants)

7:00 PM-9:00 PM **Johns Hopkins Institute of Genetic Medicine Alumni Reception MOVED TO** Grand America Hotel, Grand Salon

8:00 PM-10:00 PM **University of Chicago Reception MOVED TO** Grand America Hotel, Imperial Ballroom C

9:00 PM-11:00 PM **Case Western University & University Hospitals of Cleveland Reception. MOVED TO** Grand America Hotel, Murano Room

Ancillary Event Cancelled:

6:30 PM-8:00 PM **Successful Microarray Data Analysis: Presentation & Reception**

Invited Session Presentation Changed:

Session 30. The Spliceosome and Human Disease

11:30 AM **PRESENTATION TITLE CHANGED TO:** Mechanisms of mis-regulated alternative splicing in development and disease. Tom A. Cooper

Poster Added:

2514/T Mapping small human segmental chromosomal gains and losses at 2-25 Kb resolution with a whole-genome oligonucleotide array CGH platform. R. R. Selzer¹, T. A. Richmond¹, M. J. Walter², R. R. Ries², T. J. Ley², P. S. Eis¹. ¹R&D Department, NimbleGen Systems Inc., Madison, WI, ²Siteman Cancer Center, Washington University Medical School, St. Louis, MO.

Microarray-based comparative genomic hybridization (array CGH) methods have been widely used to investigate chromosomal abnormalities associated with cancer.¹ The level of resolution for whole-genome array CGH analysis has improved.² However, BAC array CGH resolution is inherently limited by the large size of the BAC clones. We will present array CGH data using an oligonucleotide-based array CGH platform that contains 390K unique probes per array.³ A whole-genome, tiling-path array design format was used to systematically map copy number changes within genes and in intergenic regions. The probes were designed to be isothermal (target Tm = 76C) and varied in length from 45 to 85 nucleotides to enable detection of copy number changes in both AT- and GC-rich regions in the genome. Analysis was performed with full complexity genomic DNA samples in a two-color detection format.

In collaboration with the Genomics of Acute Myeloid Leukemia Program Project Grant, we are mapping "submicroscopic" segmental chromosomal gains and losses in AML samples that are not detected with standard cytogenetic studies or array CGH using 1 Mb BAC arrays.

A set of 23 age and ethnically matched "cancer-free" control samples are being used to annotate the "normal" copy number polymorphisms (CNPs)^{4,5}. While the initial study on the cancer-free samples utilized a single whole-genome array with 25 Kb resolution, the high number of small-sized events suggested the presence of even smaller-sized CNPs. Thus, we analyzed a subset of the cancer-free samples on a 2 Kb resolution whole-genome 8-array set and detected many CNPs in the 2-25 Kb size range. Data from both sets of experiments will be presented at the meeting.

¹Albertson, D. G. and Pinkel, D. (2003) *Hum. Mol. Genet.* **12**, R145-R152; ²Ishkanian, A.S. et al. (2004) *Nat. Genet.* **36**:299; ³Selzer, R.R. et al. (2005) *Genes, Chromosomes and Cancer*, Epub Aug 1; ⁴Sebat, J. et al. (2004) *Science* **305**:525; ⁵van Ommen, G. (2005) *Nat. Gen.* **37**:333.

Poster Changed:

564/T Brain MRI findings in a fetus carrying an oligophrenin-1 (OPHN1) mutation and in his carrier mother. **CORRECT AUTHOR LISTING:** F. Mochel, N. Boddaert, P. Sonigo, S. Romano, M. Rio, N. Philip, F. Brunelle, C. Beldjord, P. de Lonlay, A. Munnich, S. Lyonnet

Posters Cancelled:

474/T MSI colorectal cancer is characterized by decreased expression of CDK2-AP1. Z. Yuan

1689/T Cross-species microarray analysis identifies Prolactin as a candidate hypothalamic signaling molecule for sexual orientation in sheep. S. Bocklandt

2268/T HEXA gene as a model of single cell genetic testing: credibility, precision, implications. K. Dotan

■ **FRIDAY, October, 28**

Curbstone Consultations, Hall D

Genetic Counselors available:

1:30 PM–2:30 PM

Cancer: Amy Roberson
Elizabeth Hoodfar
Talia Donenberg
Susan Hassad
Christine Miller

Adult Non-Cancer:

2:30 PM–3:30 PM

Ethics: Kelly Taylor
Michelle Fox

Prenatal:

Sarah Noblin
Janet Williams

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Ancillary Event Added:

12:30 PM-1:30 PM **CodeLink Bioarray System: Flexible System Solutions for Innovative Array-based Application.**

Sponsored by GE Healthcare. Grand America Hotel, Murano Room

Ancillary Event Changed:

12:30 PM-1:30 PM **Whole Genome Amplification and Advanced Methods in PCR. TITLE CHANGED TO Application of Whole Genome Amplification to Clinical Testing Using Chromosomal Microarray Analysis.** Convention Center, Room 253A

Ancillary Event Cancelled:

*12:30 PM-1:30 PM **Ampliflour, a Loci-Independent Method for SNP Genotyping and Measuring Gene Expression Levels**

Platform Session Changed:

Session 46. Therapy for Genetic Disorders. 8:00 AM-10:30 AM Salt Palace Convention Center, Ballroom E-H, **CO-MODERATOR CHANGED TO Thomas C. Markello**, Children's National Medical Center, Washington, DC, (replacing Cary O. Harding)

Posters Changed:

472/F Ultra-deep sequencing of EGFR from lung carcinoma patients reveals low abundance drug response mutations. **PRESENTER CHANGED TO J. F. Simons** instead of M. Egholm

1150/F Family history tools for underserved and underrepresented communities. **PRESENTER CHANGED TO J. O'Leary** instead of N. T. Robinson

1738/F Epicardial coronary artery spasm and microvascular angina are differentially influenced by *PON1* A632G polymorphism in the Japanese. **PRESENTER CHANGED TO a co-author** instead of G. Koike

Poster Cancelled:

1231/F Towards a new paradigm: Redefining race, ethnicity and underserved communities in light of genetic advances. L. Wise

■ **Exhibitor Updates** ■

■ **Exhibitor Information Changed**

Exhibitor	<u>Booth Number</u>
■ Ambry Genetics	1026

INFORMATION CHANGED TO:
100 Columbia #200
Aliso Viejo, CA 92656
Tel: (949) 900-5500 or (866) 262-7943
Fax: (949) 900-5501
E-mail: mhamilton@ambrygen.com

■ CYCLERtest-BIOplastics	1130
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COMPANY NAME CHANGED TO:
BIOplastics-CYCLERtest, Inc.

■ Operon Biotechnologies, Inc.	1101
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INFORMATION CHANGED TO:
Tel: (256) 704-8200 or (800) 688-2248
Fax: (256) 704-8189
E-mail: oligo-us@operon.com
Operon Biotechnologies, Inc., is the global market leader in high throughput synthesis and design of synthetic DNA oligonucleotides and array-ready oligo sets (AROS™), driven by incomparable customer service and consistent quality designed using cutting edge technologies.

■ PSS Bio Instruments, Inc.	817
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INFORMATION CHANGED TO:
6052 Industrial Way, Suite H
Livermore, CA 94551
Tel: (925) 960-9182
Fax: (925) 960-9184
Provides fully automated nucleic acid isolation instruments, **Magtration System** with common kit for whole blood, tissue, bacteria and virus. Kits for DNA, RNA and Plasmid are now available.

■ Vysis, An Abbott Company	522, 524
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COMPANY NAME AND INFORMATION CHANGED TO:
Abbott Molecular
1300 E. Touhy Ave.
Des Plaines, IL 60018
Tel: (224) 361-7000 or (800) 553-7042
Fax: (224) 224-7578

■ **Exhibitors' Booths Moved**

■ City of Hope Clinical Molecular Diagnostic Laboratory , formerly in booth 1227, moved to	507
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■ OpenHelix, LLC , formerly in booths 1219, 1221, moved to	701, 703
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■ **Exhibitors Added After Program Guide Publication**

■ Ocimum Biosolutions	1221
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8765 Guion Road, Suite G
Indianapolis IN 46268
Tel: (317) 228-0600 Fax: (317) 228-0700
E-mail: subash@ocimumbio.com
URL: <http://www.ocimumbio.com>
Ocimum Biosolutions is a life sciences R&D enabling company. Microarrays division provides custom and catalog OciChips. BioIT provides products for biotech/pharma industries including LIMS and Bioinformatics products like Biotracker, Toxchek, Pharmatracker, Genchek, OptGene, Genowiz and iRNAchek. Contract services division provides molbio services: GMO testing, DNA extractions, fermentation and gene synthesis.

■ Oregon Health & Science University Clinical Genetic Laboratories	1219
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3181 SW Sam Jackson Park Rd., MP-350
Portland OR 97239
Tel: (503) 494-5400 or (888) 375-4636
Fax: (503) 494-6922
URL: <http://www.ohsu.edu/genetics>
Oregon Health & Science University Laboratories have provided genetics testing for almost 40 years. The Molecular Diagnostic Center tests for genetic and infectious diseases. The Cytogenetics Laboratory provides high-resolution chromosome analysis, molecular cytogenetic diagnosis, and research studies. The Biochemical Genetics Laboratory diagnoses and monitors patients with inborn errors of metabolism.

■ SCHOTT NEXTERION	1229
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Hattenbergstrasse 10
Mainz 55122
Germany
Tel: 49 (0) 6131-66-25036
Fax: 49 (0) 6131-66-1916
E-mail: info.nexterion@schott.com
URL: <http://www.us.schott.com./nexterion>

■ **Exhibitor Cancelled**

■ Chemicon International, Inc.	701/703
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Salt Palace Convention Center ASHG Food Service Areas

The following areas will offer “fast food” options. Hours and prices are subject to change.

Quik Quisine

Tuesday/Wednesday, 7:30 AM–3:00 PM

Thursday–Saturday, 7:30 AM–3:00 PM

Fresh Gourmet Sandwiches - \$6.75–\$9; Fruit/Veggie Trays - \$5
Pastries/Yogurt - \$2 each; Muffins/Bagels - \$2.50 each; Donuts - \$1.25
Soft Drinks - \$2.50; Coffee/Tea - \$2 each

Exhibit Hall Concessions, Hall A & Hall C

Tuesday, 9:30 AM–3:00 PM (Hall C only)

Wednesday–Friday, 9:30 AM–3:00 PM (Halls A & C)

Fresh Sandwiches - \$4.50–\$6.50; Hot Dogs - \$2.50–\$4; Pizza - \$6–\$6.75
Salads - \$5–7; Soups - \$3.50
Pastries/Yogurt - \$2 each; Muffins/Bagels - \$2.50 each; Donuts - \$1.25
Soft Drinks - \$2.50 each; Coffee/Tea - \$2 each; Beer - \$5
Pretzels/Popcorn - \$2 each

CAFFE LATTE

Exhibit Hall: Wednesday–Friday, 10:30 AM–3:00 PM

Registration Area: Wednesday–Saturday, 7:30 AM–11:00 AM

serving a variety of specialty drinks, coffees and hot teas