

# Illumina Technology Workshops

**Workshop #1:** Empowering New Science through Advances in Next-Generation Sequencing and Genotyping Technologies

Thursday, October 22, 12:00 p.m.–1:00 p.m.

Convention Center, Room: Emalani (#320)

Lunch will be served

*New Technology Advancements from Illumina Including Genome Analyzer Sequencing Developments and GWAS Scalability Roadmap*

Illumina, Inc.

*Genomic Variation and Disease*

David Altshuler, M.D., Ph.D., Broad Institute of Harvard and MIT

*Cancer Genome Sequencing*

Elaine Mardis, Ph.D., Washington University

**Workshop #2:** Accelerating Genetic Research with New Solutions for DNA Analysis

Thursday, October 22, 3:00 p.m.–4:30 p.m.

Convention Center, Room: Emalani (#320)

Refreshments will be served

*Practical Applications of Illumina Technology Today*

Illumina, Inc.

*Impact on Workflow and Throughput: Improvements from the HumanOmni1-Quad BeadChip and GoldenGate Indexing™ Assay*

Kim Doheny, Ph.D., CIDR

*Illumina Cancer Research Update*

David Bentley, Ph.D., Illumina

*Applying Genome Analyzer Sequencing Innovations for Expanded Applications*

Illumina Customer Presentation

# Illumina Product Promotions

## Human iSelect® HD Custom Arrays Promotion

For a limited time only, customers can take advantage of discounted pricing on Illumina's iSelect custom genotyping arrays. Take your research farther – design your own content for GWAS follow-up studies and even include SNPs from the 1,000 Genomes Project.

## VeraCode® ADME Core Panel Promotion

Investigate pharmacogenetic variability associated with drug response and disposition precisely, using the VeraCode ADME Core Panel. Enjoy preliminary pre-order pricing on all orders placed before the end of 2009. Ask your local account manager for details.

# 'Power of the Genome' Challenge

Visit us at booth #516 to receive your key to unlock the 'Power of the Genome' Challenge. Your goal is to find the FOUR hidden bases by following clues you can discover at Illumina events at ASHG. Once you've found all FOUR bases, return to the Illumina booth to claim your prize!

# Sponsored Event

## **Concurrent Invited Sessions II – SESSION 9**

The 1,000 Genomes Project and Medical Genetic Uses of Sequencing

Wednesday, October 21, 9:30 a.m.–11:30 a.m

Convention Center, Hall I

Sponsored by Illumina

For more information on Illumina events and activities at ASHG, please visit [www.illumina.com/ashg](http://www.illumina.com/ashg)

# Illumina Technology Expert Sessions

Meet our experts and discover the latest in Illumina technology.

Located at the Ala Moana Hotel in Suite 3317

## Wednesday, October 21

### Genome-Wide Association Studies (GWAS)

9:30 a.m.–10:15 a.m.

Learn about the latest technology for GWAS, including an overview of the new HumanOmni1-Quad BeadChip. Topics include the selection of tagSNPs, content targeted towards regions of copy number variation, information from the 1,000 Genomes Project, and genotype imputation. A Q&A session will follow.

### Cytogenetics

10:15 a.m.–11:00 a.m.

As the adoption of SNP-based arrays continues to grow for rapid whole-genome cytogenetic analysis, new questions surface about workflow management and data analysis. In this session, attendees will hear from an expert panel about their experience addressing these issues and the results of their research using SNP-based arrays for comprehensive cytogenetic analysis.

### VeraCode ADME

11:00 a.m.–11:45 a.m.

Understanding genetic variability associated with drug response or disposition is a key step toward personalized medicine. Although the gene products involved in metabolizing and transporting drug molecules have been extensively described and studied, the past and ongoing discoveries of polymorphisms in these genes raise a need for molecular tools that will enable rapid, comprehensive, and accurate assessment of pharmacogenetic profiles. In this session we describe a new genotyping assay platform for pharmacogenetic profiling: VeraCode ADME Core Panel for use on the BeadXpress® Reader.

### Sequencing Simplified

1:15 p.m.–2:00 p.m.

Find out about the latest product developments for the Illumina Genome Analyzer, including an overview on recent advancements in chemistry, software, and hardware. It's not just more data, it's better data.

### Illumina Informatics

2:00 p.m.–2:45 p.m.

Learn about new sequence analysis products and solutions for the Genome Analyzer. Topics include IlluminaCompute, a set of pre-configured, ready-to-deploy hardware offerings capable of processing data from multiple Genome Analyzers, as well as a new Technical Note on *de novo* assembly using Genome Analyzer reads, and a review of the Illumina-Connect ecosystem of third-party software providers. A Q&A session will follow.

## Thursday, October 22

### Custom Genotyping

9:30 a.m.–10:15 a.m.

Illumina offers a broad portfolio of custom genotyping products, powered by robust assays and covering a wide range of multiplex and sample throughput options. For high-plex needs, iSelect products provide researchers with the ability to design content ranging from 3K to 200K markers. For low- to mid-plex needs, the diversity of the GoldenGate® products is ideal for cost-effective follow-up studies without concern for variable sample quality.

### Cytogenetics

10:15 a.m.–11:00 a.m.

As the adoption of SNP-based arrays continues to grow for rapid whole-genome cytogenetic analysis, new questions surface about workflow management and data analysis. In this session, attendees will hear from an expert panel about their experience addressing these issues and the results of their research using SNP-based arrays for comprehensive cytogenetic analysis.

## Targeted Resequencing

11:00 a.m.–11:45 a.m.

Find out about the latest sequencing developments from Illumina and how targeted resequencing is easily performed on the Genome Analyzer. An overview of validated enrichment technologies will be given.

## Sequencing Simplified

1:30 p.m.–2:15 p.m.

Find out about the latest product developments for the Illumina Genome Analyzer, including an overview on recent advancements in chemistry, software, and hardware. It's not just more data, it's better data.

## Illumina Informatics

2:15 p.m.–3:00 p.m.

Learn about new sequence analysis products and solutions for the Genome Analyzer. Topics include IlluminaCompute, a set of pre-configured, ready-to-deploy hardware offerings capable of processing data from multiple Genome Analyzers, as well as a new Technical Note on *de novo* assembly using Genome Analyzer reads, and a review of the Illumina-Connect ecosystem of third-party software providers. A Q&A session will follow.

## VeraCode ADME

3:00 p.m.–3:45 p.m.

Understanding genetic variability associated with drug response or disposition is a key step toward personalized medicine. Although the gene products involved in metabolizing and transporting drug molecules have been extensively described and studied, the past and ongoing discoveries of polymorphisms in these genes raise a need for molecular tools that will enable rapid, comprehensive, and accurate assessment of pharmacogenetic profiles. In this session we describe a new genotyping assay platform for pharmacogenetic profiling: VeraCode ADME Core Panel for use on the BeadXpress Reader.

Friday, October 23

## iScanSQ System

9:30 a.m.–10:15 a.m.

Join us to learn more about the benefits of integrating microarrays and next-generation sequencing on Illumina's iScan. Topics include system capabilities, highlights, and example projects. A Q&A session will follow.

## Sequencing Simplified

10:15 a.m.–11:00 a.m.

Find out about the latest product developments for the Illumina Genome Analyzer, including an overview on recent advancements in chemistry, software, and hardware. It's not just more data, it's better data.

## Gene Expression and Regulation Sequencing

11:00 a.m.–11:45 a.m.

Come hear about the latest applications of the Genome Analyzer, including an early look at the upcoming Universal RNA kit. Topics include Illumina's current and future RNA sequencing applications and reduced representation bisulfite sequencing for DNA methylation profiling. A Q&A session with Illumina product managers will follow.

# Illumina Customer Presentations

At ASHG, there are ~300 presentations featuring data generated using Illumina's next-generation sequencing and array platforms across a broad range of applications. Be sure to visit the poster and platform sessions listed below and ask researchers about their experience using Illumina products.

POSTER SESSION WEDNESDAY, OCTOBER 21, 2009 9:30 AM–3:30 PM			
Program No.	Title	Poster No.	Lead Author
379	Identification of a candidate locus in familial systemic lupus erythematosus by homozygosity mapping.	37	Kitamura, A
387	Homozygosity mapping in patients with malformations of cortical development.	45	Bilguvar, K
411	Transcriptional profile of the embryonic myosin heavy chain gene MYH3 responsible for two congenital distal arthrogyrosis syndrome subtypes.	69	Beck, AE
475	Pursuit of additional 'Cohesinopathy' loci involved in human developmental disorders.	133	Deardorff, M
501	Next generation technologies for gene discovery in autosomal dominant retinitis pigmentosa.	159	Daiger, SP
571	Statistical methods for estimating the rate of de novo mutation at FGFR3 in sperm samples from healthy men using high-throughput sequencing.	229	Pfeifer, SP
573	Identification of rare variants for ulcerative colitis and celiac disease by high-throughput sequencing.	231	Szperl, A
627	Population structure in Brazilian and other worldwide human populations revealed by SNP arrays.	285	Giolo, SR
651	Exploring the genetic basis of human uniqueness.	309	Noonan, JP
655	Use of 96-plex indexing and Illumina sequencing to analyse the worldwide population genetics of a polymorphism associated with human muscle function.	313	MacArthur, DG
693	Developmental elevation of neutrophils is associated with increased Serpina-1 expression in a rat model of innate airway hyperresponsiveness.	351	Carpe, N
697	A novel glaucoma locus identified in Dandie Dinmont Terrier dog breed.	355	Ahonen, SJ
721	Fine mapping of Hirschsprung's disease loci in 9q31.	379	Cherny, SS
753	SPAG16 gene variants are associated with fasting glucose and insulin sensitivity.	411	Li, WD
771	Development of methods for resequencing of genes using indexed DNA samples and the SureSelect target enrichment system.	429	Kenny, EM
787	Integration of human and mouse genome-wide association studies (GWAS) to identify genetic determinants for lung development and asthma.	445	Berndt, A
809	Linkage and association studies of joint morbidity from rheumatoid arthritis.	467	Min, K
829	Genome-wide screen for common and rare copy number variation in metabolic syndrome.	487	Kristiansson, K
833	The genetic association of copy number variation and Type 1 diabetes.	491	Qu, HQ
839	Genetics of gene expressions under perturbation of environments.	497	Lai, S
845	COPACETIC, a genome-wide association study on chronic obstructive pulmonary disease (COPD).	503	Smolonska, J
849	Comparative haplotypic congruence analysis of Addison's Disease and Type 1A diabetes.	507	Baschal, EE
857	Replication of 5q22.1 region for intraocular pressure in Mongolian population by linkage and association analyses. The GENDISCAN Project.	515	Lee, M
901	Genome-wide analysis of gene-gene interaction.	559	Zhu, Y
947	Variants in the TNFAIP3 locus form three risk haplotypes associated with SLE in Koreans.	605	Bates, JS
953	GWAS of systemic lupus erythematosus on a Chinese population in Hong Kong.	611	Yang, W
957	Genome-wide association study of migraine with aura in a large international consortium sample identifies first SNP and CNV associations affecting migraine susceptibility.	615	Anttila, V
963	Genome wide search for genes controlling risk to non-syndromic oral clefts in a consortium case-parent trio study.	621	Beaty, TH
977	Combining Genome Wide linkage and association reveals new candidate gene loci for Celiac Disease.	635	Naluai, AT
985	Genomewide association study of age of onset of asthma in the Childhood Asthma Management Program.	643	Forno, E
987	Evidence for significant allele frequency variation as a function of age within and across GWAS control datasets detected using PLINK and MAP.	645	Hamza, TH
989	Genome-wide association study of serum lipoprotein(a) concentration identifies multiple associated SNPs in addition to the kringle repeat.	647	Hosseini, SM
995	GWA study of a genetic subisolate identifies variants in C7 and STAT3 predisposing to multiple sclerosis and suggest a positive selection of a "protective"haplotype outside Africa.	653	Leppä, V
999	The POCEMON (Point-Of-Care MONitoring and Diagnostics for Autoimmune Diseases) project. Building a Lab-On-Chip centered on Rheumatoid Arthritis and Multiple Sclerosis.	657	Lupoli, S

POSTER SESSION WEDNESDAY, OCTOBER 21, 2009 9:30 AM-3:30 PM

Program No.	Title	Poster No.	Lead Author
1007	Analysis of GWAS data reveals another locus for nicotine dependence.	665	Rice, JP
1025	Association of the Protocadherin gene cluster on chromosome 5q31 with extreme obesity in the NHLBI Family Heart Study (FamHS).	683	Borecki, IB
1057	Structural variant discovery in a liver cancer-related 39Mb region using capture-sequencing approach.	715	Lin, CH
1079	Genetic variation in apoptosis genes and susceptibility to non-Hodgkin Lymphoma.	737	Schuetz, J
1085	The application of SNP-based array CGH for the detection of novel cancer susceptibility genes in multi-cancer families.	743	Chenevix-Trench, G
1119	Cell cycle genes and ovarian cancer susceptibility - a tagSNP analysis.	777	Cunningham, J
1167	de novo methylation in DLBCL frequently occurs proximal to genes that are poorly expressed in normal progenitor cells.	825	Wang, X
1177	Identification of regulatory genetic variation that affects drug response in childhood acute lymphoblastic leukemia (ALL).	835	Kiialainen, A
1269	Genome-wide allelic imbalance in prostate cancer. Comparison of aggressive and non-aggressive disease.	927	Tai, Y
1393	Detection of copy number variations (CNV) in patients with mental retardation using high density SNP microarrays.	1051	Rivera-Brugues, N
1445	Diagnosis of recessive disorders using a genome-wide SNP microarray.	1103	Crowley, M
380	Disease mutation detection by next generation sequencing platforms.	38	Wang, H
382	Hunting down the last Meckel syndrome genes in the Finnish population.	40	Tallila, J
422	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS). Expanding the genotype in the French-Canadian population.	80	Thiffault, I
520	Copy number variation analysis in patients with malformations of cortical development.	178	Ozturk, AK
536	Brachydactyly caused by a frameshift mutation in CHSY1.	194	Merriman, B
594	Gorillas out of the mist. Unravelling the fog surrounding another genome.	252	Yngvadottir, B
608	Improved resolution of the human Y-chromosomal phylogeny using targeted next-generation sequencing.	266	Ayub, Q
610	Genetic differences of the Northern and Southern Han Chinese.	268	Chen, CH
624	Analysis of genomic admixture in Costa Rica population.	282	Wang, Z
626	Genome-wide patterns of population structure and admixture among Hispanic/Latino populations.	284	Bryc, K
638	Genetics of celiac disease in the Saharawi population.	296	Romanos, J
648	Homing in on signals of positive selection in the human genome by large-scale re-sequencing.	306	Hu, M
698	Genome-wide linkage and association mapping of hip dysplasia using dog as a model organism.	356	Zhao, K
770	Evaluation of pooled sequence variation in SERPINE2 using the Illumina Genome Analyzer.	428	Cho, MH
776	Characterization of candidate loci for Autism Spectrum Disorders.	434	Rossi, M
798	Detection of excess rare damaging variants in surfactant associated genes among infants with Respiratory Distress Syndrome using pooled next generation sequencing.	456	Wegner, D
802	A pathway approach to decoding Multiple Sclerosis reveals several new associated loci.	460	Zuvich, R
808	TNFR2 associated with hand OA and evidence of quantitative trait loci (QTL) on chromosome 16. The CARRIAGE family study.	466	Chen, HC
832	Comprehensive metabolic trait association analysis using gene expression and single nucleotide polymorphisms.	490	Inouye, M
834	Functional classification based on gene expression data indicates that T2D genes play a role in liver, subcutaneous adipose tissue, and visceral adipose tissue.	492	Wolfs, MGM
846	Studies of genetic and phenotypic variations under environmental stimulation.	504	Zhou, X
912	Integrative genomics. Environmental influences on expression quantitative trait loci.	570	Tantisira, KG
914	Transcriptomic epidemiology of human apolipoprotein E variation.	572	Drigalenko, E
956	Bone density GWAS in premenopausal white women and replication in other cohorts.	614	Koller, DL
966	Meta-analysis of six large type 1 diabetes cohorts identifies multiple loci.	624	Bradfield, JP
978	Testing association with a reference control population. Evaluation of case-control matching strategies.	636	Babron, MC
980	Genome-wide association study of lipid related phenotypes in the Avon Longitudinal Study of Parents and Children (ALSPAC).	638	Boustred, CR
984	A genome-wide approach for the identification of shared susceptibility loci.	642	Ellinghaus, D
986	Interim analysis from a genome-wide association study of childhood obesity identifies FTO as strongly associated with the trait.	644	Grant, SFA
988	Combination of case-control and family-based asthma genome-wide association results using probands from a single population.	646	Himes, BE
996	Genome-wide association study of asthma identifies RAD50-IL13 and HLA-DR/DQ regions.	654	Li, X
1018	Genome wide association study of non-diabetic end stage renal disease in African Americans using pooled DNA.	676	Bostrom, MA

**POSTER SESSION WEDNESDAY, OCTOBER 21, 2009 9:30 AM–3:30 PM**

Program No.	Title	Poster No.	Lead Author
1028	A combined genome-wide association scan and proteomics approach for the identification of genetic modifiers of pseudoexfoliation syndrome.	686	Burdon, K
1040	Association of chromosome 8q variants with prostate cancer risk in Caucasian, Hispanic, and African American men.	698	Beuten, J
1058	A large scale genetic association study of esophageal adenocarcinoma risk.	716	Liu, C
1070	Association of FPGS and MLL variants with childhood acute lymphoblastic leukemia in different ethnicities.	728	Piwkham, D
1080	Capture sequencing of chromosome 4q21.3-25 region frequently lost in the hepatocellular carcinoma.	738	Su, J
1082	Identification of a novel lung tumor suppressor candidate at the lung cancer susceptibility locus 6q23-25.	740	Wilson, IM
1088	Variation at 8q24 and 9p24 and risk of epithelial ovarian cancer.	746	White, KL
1092	Familial haematological malignancies. The search for a causative gene continues.	750	Tegg, EM
1154	Profiling critical genetic mutations in clinical tumor samples.	812	MacConaill, L
1168	Digital counting of gene copy number in clinical settings.	826	Ishikawa, S
1234	Identification of copy number variable regions (CNVRs) associated with risk of prostate cancer in Mexican-Americans.	892	Blackburn, A
1280	Characterization of myelodysplastic syndromes and acute myeloid leukemia with isodicentric X chromosome.	938	Paulsson, K
1378	The effect of large de novo chromosomal deletions on gene expression.	1036	Ghahramani Seno, M
1384	A novel 1.5 Mb de novo non-recurrent deletion at 14q23 detected in autism spectrum disorders.	1042	Ma, DQ
1386	Parental origin bias in de novo CNVs detected in autism probands.	1044	Shah, N
1412	Comparison of genome-wide array technologies for the detection of copy-number variants.	1070	Montpetit, A
1430	Comprehensive preimplantation genetic diagnostics (PGD) by single cell SNP microarrays.	1088	Kearns, WG

**PLATFORM SESSION THURSDAY, OCTOBER 22, 2009 7:00 AM–9:00 AM**

Program No.	Title	Location	Lead Author
47	A statistical approach for identifying differentially methylated CpG sites with DNA methylation array data.	Room 313	Conneely, KN
67	Development and validation of a disease targeted SNP-based array for clinical personal genomic profiling.	Room 323	Willis, AS
8	Sequencing cancer genomes.	Hall I	Nelson, S
18	Genome-wide combined linkage/association scan localizes two QTLs influencing human caudate nucleus volume.	Ballroom A	Glahn, D
38	1st generation genome-wide map of cis-regulatory SNPs in human cells.	Ballroom C	Pastinen, T
9	The complete genome sequence of a human melanoma.	Hall I	Cheetham, RK
19	Genome-wide association of human amygdala volume identifies a QTL at IRX2.	Ballroom A	Charlesworth, J
29	Expression quantitative trait loci (eQTL) analysis of subcutaneous human adipose tissue from Body Mass Index (BMI)-discordant sibpairs.	Ballroom B	Walley, A
40	Cis-regulatory variants exert stable effects upon environmental perturbation in human primary cells.	Ballroom C	Grundberg, E
21	Genome-wide association study in partial epilepsies.	Ballroom A	Kasperaviciute, D
41	"Simple" disorders, complex traits. A search for genetic modifiers in neurofibromatosis type 1.	Ballroom C	Pemov, A
12	Detection of tumor-specific somatic mutations by transcriptome sequencing of a cytogenetically normal acute myeloid leukemia.	Hall I	Benet-Pages, A
42	Genome-wide association reveals master eQTL regulators of microRNA expression variation in human fibroblasts.	Ballroom C	Borel, C
14	Aberrant microRNA expression in retinoblastoma by high-throughput next generation sequencing.	Hall I	Chao, E
15	The Cancer Genome Atlas study on serous ovarian carcinoma. Preliminary analysis of copy number alterations.	Hall I	Winckler, W

**POSTER SESSION THURSDAY, OCTOBER 22, 2009 7:00 AM–3:30 PM**

Program No.	Title	Poster No.	Lead Author
1452	"Universal" carrier screening of recessive genetic disorders by next generation sequencing.	1	Kingsmore, S
1488	Identification of a putative region associated with primary microcephaly through SNP array analysis in a consanguineous family.	37	Wiszniewska, J
1636	Genome-wide methylation analysis in women with trisomic pregnancies.	185	Hanna, CW
1670	A two-stage genome-wide association study of young-onset hypertension in Han Chinese population of Taiwan.	219	Chiang, KM
1684	Avoiding the high Bonferroni penalty in genome-wide association studies.	233	Gao, X
1712	Shared controls for genome-wide association studies.	261	Mukherjee, S
1718	A HMM-based optimal Multiple Testing procedure for GWAS.	267	Wei, Z
1758	Using a local extension of HapMap 3 to raise SNP imputation quality and to identify CNV tag-SNPs in population isolates.	307	Ripatti, S
1772	Impact of sample pretesting in a high through-put genotyping facility.	321	Marosy, B

POSTER SESSION THURSDAY, OCTOBER 22, 2009 7:00 AM–3:30 PM

Program No.	Title	Poster No.	Lead Author
1782	Assessing the accuracy of ancestry reported in a biorepository linked to electronic medical records for genetic association studies.	331	Ritchie, MD
1798	Genetic Mechanisms for Venous Thromboembolism (VTE).	347	Heit, J
1828	SAFARI. A software package for genome-wide interaction studies.	377	Ding, B
1864	Genomewide linkage and association study of onset age of LRRK2-related Parkinson's Disease.	413	Latourelle, JC
1878	Multiple loci associated with prostate cancer susceptibility on 8q24.	427	Amin Al Olama, A
1910	Bayesian network model for SNP calling in shallow sequencing with haplotype based prior.	459	Zhi, D
1922	A software tool for scoring CNVs and runs of homozygosity using linkage results.	471	Karlsson, R
1930	Performance of population stratification correction. A simulation study using observed genotypes from a large European Reference Panel.	479	Genin, E
1972	Genome-wide linkage analysis for genetic causes of Macular Telangiectasia Type 2.	521	Parmalee, NL
1994	Genome-wide association study of cataract in the Marshfield Personalized Medicine Research Project as part of the eMERGE network.	543	Torstenson, ES
2024	Integrated next-generation sequencing and array based analysis of genetic modifiers in Velocardiofacial Syndrome (VCFS).	573	Urban, A
2028	Testing epilepsy candidate genes for association to autism in a genome wide association study dataset.	577	Cuccaro, M
2054	Novel copy number variation in autism.	603	Thomson, SA
2062	Replication of a novel Autism peak region in two ethnically diverse populations.	611	Jaworski, J
2082	The role of the mitochondria in autism.	631	Conroy, Y
130	Genome-wide association study for susceptibility genes contributing to familial Parkinson disease.	1905	Pankratz, N
131	Genome-wide association study of Parkinson's disease by using 550K SNP array.	1907	Satake, W
132	High-density linkage screen identifies potential dementia loci in the Amish.	1911	Davis, AC
133	Title: Identification and validation of novel regions associated with sporadic amyotrophic lateral sclerosis (ALS).	1914	Huang, J
134	A genome-wide association study of 1,000 Han-Chinese bipolar I cases vs. 1,000 controls residing in Taiwan.	1915	Lee, MTM
135	Whole-genome mapping identifies multiple quantitative trait loci (QTL) for novel cardiovascular disease biomarkers.	2006	Shah, S
136	The use of genome-wide eQTL associations to identify novel genetic pathways involved in complex traits.	2245	Min, JL
137	Association of interferon regulatory factor 5 (IRF5) polymorphisms with systemic sclerosis (SSc).	2249	Gourh, P
138	Identification of modifier genes for cutaneous malignant melanoma (CMM) and dysplastic nevi (DN) in melanoma families with and without CDKN2A mutations.	2492	Yang, R
139	Genome-wide analysis of microsatellite mutation in humans.	2608	Coghlan, A
140	Comparison of genome-wide recombination intensity from pedigree/linkage data to population recombination rates estimated from SNP genotype data in the European-American population.	2614	Wang, X
2090	Allelic spectrum of rare mutations within CNTNAP family members in autism spectrum disorders and related phenotypes.	639	Szelinger, S
2106	Clinical characterization and mapping of a late-onset painful recessive sensory neuropathy.	655	Putorti, ML
2164	Identification of novel early-onset progressive cerebellar ataxia gene in Finnish Hound dogs.	713	Kyöstilä, K
2180	Genome-wide association of cerebrovascular and neurodegenerative quantitative MRI traits in alzheimer disease.	729	Parrado, A
2182	Genome-wide association study identifies common variants on two novel loci, -synuclein, and upstream LRRK2 as genetic risks for Parkinson's disease.	731	Satake, W
2186	A genome-wide association study of obsessive-compulsive disorder.	735	Stewart, E
2190	Genome-wide screen for rare copy-number variants in Amyotrophic Lateral Sclerosis.	739	Blauw, H
2218	X-linked dystonia-parkinsonism, effects of disease-specific single nucleotide change 3 (DSC3) within the TAF1/DYT3 multiple transcript system in a cell system.	767	Muller, U
2226	Association of polymorphisms in transcription factor genes with variable bronchodilator response among asthmatics.	775	Duan, QL
2246	Genome-wide association study of response to warfarin in a UK prospective cohort.	795	Bourgeois, S
2266	A genome-wide association study on opioid analgesic sensitivity in patients undergoing painful cosmetic surgery.	815	Nishizawa, D
2270	Human_CVD 50K SNP array analysis in the Drug-Induced Arrhythmia Risk Evaluation (DARE) study.	819	Dalageorgou, C
2320	Massively parallel sequencing in pediatric cardiomyopathy patients.	869	Ware, SM
2334	Searching for a gene on chromosome 18q influencing systolic blood pressure in Mexican Americans.	883	Rutherford, S
2344	Next-generation-diagnostics of heterogeneous monogenic disorders using high-throughput-sequencing. The promising example of Hypertrophic Cardiomyopathy.	893	Fokstuen, S
2358	A genome-wide study of common and rare variants for myocardial infarction and related traits in South Asians, the INTERHEART study.	907	Do, R

**POSTER SESSION THURSDAY, OCTOBER 22, 2009 7:00 AM–3:30 PM**

Program No.	Title	Poster No.	Lead Author
2362	Genome-wide copy number analysis to identify candidate genes for thoracic aortic aneurysms and dissections.	911	Prakash, S
2364	Multiethnic genetic association study of carotid intima-media thickness using a targeted cardiovascular SNP microarray.	913	Lanktree, MB
2366	Longitudinal genome-wide association of cardiovascular disease risk factors in the Bogalusa Heart study.	915	Smith, EN
2368	Follow up genome-wide association study of intracranial aneurysms identifies novel risk loci and confirms previous findings.	917	Yasuno, K
2370	Coronary ARtery Disease Genome-wide Replication And Meta-Analysis (CARDIoGRAM)—Design of a prospective meta-analysis of 14 genome-wide association studies.	919	König, IR
2372	A genome-wide association study identifies new loci for ACE activity. Potential implications for response to ACE inhibitor.	921	Chung, C
1621	Prenatal diagnosis and therapy development in harlequin ichthyosis.	170	Rajpopat, S
1705	Comprehensive haplotype association tests for genome wide data of myopia in Chinese schoolchildren.	254	Fan, Q
1761	A new statistic in evaluating imputation reliability.	310	Lin, P
1787	Families versus unrelateds. An empirical comparison of gene mapping power based on genetic analysis of expression profile data.	336	Göring, HHH
1791	Genomic and genealogic investigation of the French Canadian founder population structure.	340	Roy-Gagnon, MH
1795	From association to disease risk prediction. An optimistic view from genome-wide association studies on type 1 diabetes.	344	Zhang, H
1881	SNP association study of Behcet's disease to unravel the genetic susceptibility and pathoetiology.	430	Kurata, R
1943	Base calling for resequencing chips.	492	Wang, W
1971	Genomewide identity by descent and homozygosity mapping reveals candidate regions for genetic susceptibility to obsessive-compulsive and tic disorders.	520	Nurmi, E
1977	Multi-stage genome-wide association study uncovers genetic factors influencing the P300 event-related brain potential.	526	Zlojutro, M
1981	A genome-wide association scan localizes a QTL influencing Epstein-Barr virus infection to the major histocompatibility complex region.	530	Rubicz, R
1983	Linkage analysis using 370K chips in two sets of Retinitis Pigmentosa pedigree samples.	532	Cheng, F
1989	Genome-wide linkage analysis using SNP chip on a Korean family with late-onset nonsyndromic hearing loss.	538	Won, HH
2011	RGS4 gene in Schizophrenia. Case-control and family-based studies, drug response and Tardive Dyskinesia.	560	Hirata, Y
2025	Common Variant (CV) and rare deletion in the tyrosine hydroxylase gene contribute to Parkinson disease risk.	574	Wang, L
2031	Replication of highly associated polymorphisms and candidate genes in an independent dataset for late-onset Alzheimer disease.	580	Gallins, PJ
2047	Further investigation of IMMP2L, LRRN3 and DOCK4 as potential candidate genes for autism susceptibility.	596	Sykes, NH
2057	Linkage analysis of plasma dopamine -hydroxylase activity in families segregating schizophrenia.	606	Cubells, JF
2071	New generation sequencing for the identification of novel variant late-infantile neuronal ceroid lipofuscinosis (vLINCL) genes.	620	Kousi, M
2077	Rare variant discovery using pooled DNA for next generation sequencing; Multiple rare alleles of the ATP-binding cassette transporter A1 (ABCA1) gene as risk factors for Alzheimer's disease.	626	Lupton, MK
2083	Exploring mitochondrial variation in Autism.	632	Gilbert, JR
2099	Gene expression provides evidence that cell adhesion is involved in the etiology of schizophrenia.	648	Evgrafov, O
2103	Association studies of Autism spectrum disorders in populations of European ancestry.	652	Holt, R
2111	De novo mutations in autism affected subjects from a large cohort screening of polymorphisms in genes IL1RAPL1 and IL1RAPL2 by pooling and next generation sequencing.	660	Pruzin, JJ
2171	DISC1 conditioned genome-wide association study of psychosis proneness in a large birth cohort.	720	Tomppo, L
2183	Genome-wide association study of Gilles de la Tourette Syndrome in European ancestry samples and four population isolates.	732	Scharf, JMI
2185	Genome-wide association study of schizophrenia in a Chinese population.	734	So, HC
2187	Genome-wide patterns of genetic variation in individuals with autism spectrum disorders from Croatia.	736	Wang, L
2193	Genome-wide association analysis of sleep length reveals new genes and pathways controlling sleep homeostasis.	742	Ollila, HM
2201	A unified theory of Autism revisited. Linkage evidence points to the IL1RAPL1 gene using a high-risk subset of AGRE families.	750	Allen-Brady, K
2221	Gene expression, aging and Alzheimer's disease.	770	Avramopoulos, D
2225	Glycogen synthase kinase 3 polymorphisms and weight change during clozapine treatment.	774	Souza, RP
2247	Validation and application of a broad-based ADME genotyping assay in research and clinical trials.	796	Brown, AMK

**POSTER SESSION THURSDAY, OCTOBER 22, 2009 7:00 AM–3:30 PM**

Program No.	Title	Poster No.	Lead Author
2261	Variant discovery by targeted resequencing of “high-value” regions in drug pathways.	810	Lam, ET
2275	Association between common genetic variants in the NOS1AP (CAPON) gene in patients experiencing amiodarone-induced ventricular arrhythmias.	824	Jamshidi, Y
2317	Large scale candidate gene analysis of lipid traits in the Pakistan risk of myocardial infarction study and comparison with participants of European descent.	866	Saleheen, D
2361	Genome-wide association study on HDL cholesterol level in the Marshfield Personalized Medicine Research Project as part of the eMERGE network.	910	Turner, SD
2373	A genome-wide association study identifies a potential locus near PKHD1 that might increase aldosterone renin ratio in Taiwanese young-onset hypertension patients.	922	Kuo, TY
2419	Genetic influences on asthma susceptibility in the developing lung.	968	Mandeville, I
2431	Molecular investigation of an autism risk region on chromosome 12.	980	Cukier, HN
2433	Comprehensive targeted re-sequencing of a 150kb contiguous genomic region of chromosome 5p14.1 associated with autism.	982	Griswold, AJ
832	Using Drosophila and mammalian model systems to study the effect of MeCP2 on signaling pathways.	490	Inouye, M
2435	Functional classification based on gene expression data indicates that T2D genes play a role in liver, subcutaneous adipose tissue, and visceral adipose tissue.	984	Gamliel, A

**PLATFORM SESSION FRIDAY, OCTOBER 23, 2009 7:00 AM–9:30 AM**

Program No.	Title	Location	Lead Author
78	Genomewide joint tests reveal associations and gene by environment interactions in multiplex Autism families.	Ballroom A	Lu, A
81	Next generation sequencing of chromosome 5q33 and Xp22 in the PIC schizophrenia sample.	Ballroom A	Deng, A
93	Genome-wide analysis identifies alpha-2 catenin (CTNNA2) as a risk factor for diabetes complicating cystic fibrosis (CF).	Ballroom B	Blackman,, SM

**PLATFORM SESSION FRIDAY, OCTOBER 23, 2009 1:00 PM–3:30 PM**

Program No.	Title	Location	Lead Author
167	Genomic profiling of Human LINE-1 variation.	Ballroom C	Ewing, AD
177	Simultaneous genotype calling and haplotype phase inference improves genotype accuracy and reduces false positive associations for genome-wide association studies.	Room 313	Browning, B
180	Genome wide association study of age-related macular degeneration identifies TIMP3 and HDL-associated alleles as new susceptibility loci.	Room 313	Chen, W
161	Genetic variation based on SNPs, CNVs and RoHs in European populations..	Ballroom B	Metspalu,A
156	Assessing copy number variation in 1,184 individuals from 11 world populations using multiple SNP/CNV array platforms.	Ballroom A	Nemesh, J
216	Exon capture and large-scale sequencing of 828 ciliopathy candidate genes in patients with nephronophthisis, Senior Loken-, and Joubert syndrome.	Room 311	Otto, E

**POSTER SESSION FRIDAY, OCTOBER 23, 2009 7:00 AM–12:00 PM**

Program No.	Title	Poster No.	Lead Author
2587	Familial congenital mirror movements maps to chromosome 18q21.1-q22.1.	103	Srouf, M
2621	Proximal interstitial 4q deletion associated with macrocephaly, plagiocephaly, ptosis, intraventricular hemorrhage, hypotonia, and growth and global development delays.	137	Zambrano, R
2683	Hypertrophic cardiomyopathy multigene analysis by next generation sequencing.	199	Dames, S
2693	ALFRED. A resource for research and teaching.	241	Rajeevan, H
2695	Combination of genotyping platforms to develop a haplotype map in Mexican Mestizo and Amerindian populations.	243	Richard, G
2699	Comparison of next gen sequencing technologies and bioinformatic analysis tools—Case studies on different projects.	247	Stangier, K
2701	Data storage & archiving for high-throughput genotyping.	281	Watkins, L
2709	On design of deep-sequencing experiments.	289	Bashir, A
2727	Copy number variation in candidate regions in extended families with autism spectrum disorder.	339	Salyakina, D
2729	Detection of copy number events in patients with eye development disorders using SNP arrays and multiple detection algorithms.	341	Ragoussis, I
2739	Genome-wide analysis of copy number alterations in colorectal cancer.	383	Gornick, M
2751	Genome-wide analysis of structural variations in different ethnic groups.	427	Kumar, V
2755	Copy number variants in myopia of Singapore Chinese schoolchildren.	431	Li, YJ
2759	Data quality control in detecting copy number variations using Illumina SNP arrays.	435	Lu, E
2773	Comparing CNV detection methods for SNP arrays.	481	Winchester, LM

**POSTER SESSION FRIDAY, OCTOBER 23, 2009 7:00 AM–12:00 PM**

Program No.	Title	Poster No.	Lead Author
2783	Mobile Element Scanning (ME-Scan). Complete ascertainment and genotyping of transposons in large population samples using targeted high-throughput sequencing.	523	Witherspoon, DJ
2791	A workbench for measuring transcription using next-generation sequencing.	531	Busby, M
2793	Using integrative genomics to develop novel diagnostic phenotypes for asthma severity.	533	Howrylak, JA
2815	Mapping regulators of the transcriptional response to hypoxia in human cells.	619	Mohr, AJ
2817	Genetic identification, replication, and functional fine-mapping of expression quantitative trait loci in primary human liver tissue.	621	Cooper, GM
2825	The impact of racial differences in epidemiologic studies of gene expression.	629	Sharma, S
2835	Measurement of allelic expression in postmortem brain samples using Illumina Goldengate assay.	703	Mantripragada, KK
2845	Fine mapping of association signals with haplotype association analysis in CGEMS when a functional variant is known.	745	Gonzalez Bosquet, J
2857	Linkage analysis and comprehensive resequencing of the whole exon-captured genomic DNAs toward identification of causative genes for hereditary neurodegenerative diseases.	757	Fukuda, Y
2859	Whole genome amplification and screening methods of formalin-fixed paraffin-embedded tissues for selective hybridization sequencing on Illumina's Genome Analyzer II sequencer.	759	DeFelice, M
2865	Detection of small indels from next generation sequencing data by Bayesian evaluation of alternative haplotypes.	797	Albers, C
2905	Using next generation sequencing technologies to identify variants in the neuroligin pathway associated with autism spectrum disorder.	837	Steinberg, KM
2907	Targeted enrichment of genomic loci using custom microarrays for massively parallel sequencing.	839	Corneveaux, JJ
2911	Multiple sample pooling and next-generation sequencing to determine RET sequence variation within a population.	843	Margraf, RL
2915	Whole exome resequencing in a multigenerational pedigree with hereditary spastic paraplegia.	847	Züchner, S
2957	Methylation profile of placentas with confined mosaicism for trisomy 16.	889	Diego-Alvarez, D
2959	Epigenetic associations with triglyceride response to a high-fat meal. A pilot study of the Genetics of Lipid Lowering Drugs and Diet Network (GOLDN) study.	891	Wojczynski, MK
2963	Genome wide methylation analysis in Beckwith Wiedemann syndrome reveals CpG methylation alterations at several imprinted loci.	895	Choufani, S
2981	The relationship of DNA methylation with age, gender and genotype in twins and healthy controls.	913	Ophoff, RA
2985	Identification of tissue-specific methylation signatures in the 2nd trimester fetus and placenta.	917	Robinson, W
2642	A t(5q;17q) in a patient with acampomelic campomelic dysplasia.	158	Vernon, H
2682	Informatic sample handling processes. A high throughput genotyping facility's workflow for sample information tracking.	198	Craig, B
2730	Cnstream. A method for the identification and genotyping of copy number polymorphisms using Illumina microarrays.	342	Alonso, A
2744	Definitive SNP/CNV haplotyping of Asian genomes using DNAs derived from complete hydatidiform moles.	388	Hayashi, K
2748	An accurate genome-wide map of copy number variants of NA10851, a widely used reference sample for array CGH.	392	Ju, Y
2804	Gene expression profiling of human whole blood samples with the Illumina DASL assay.	576	Winn, ME
2818	Cis-association mapping of expression trait loci underlying the transcriptional response induced by TLR7 activation in lymphoblastoid cell lines.	622	Biswas, S
2844	Three individual human genome sequences.	712	van Velkinburgh, J
2852	Resequencing all unique X chromosome exons to identify genetic variants contributing to autism susceptibility.	752	Mondal, K
2856	Taybi-Linder syndrome (MOPD I/III) maps to a 3Mb interval on chromosome 2q.	756	Ederly, P
2860	Genomic Approaches to Understanding PGC-1 $\alpha$ Regulated Metabolism.	760	Charos, A
2882	Population genomics of human gene expression using next-generation RNA sequencing.	814	Montgomery, SB
2896	Identification of novel sequence variants in FMR1 in developmentally delayed males.	828	Collins, SC
2900	Exon capture approaches for gene discovery in callosal agenesis.	832	Li, J
2906	Discovery and analysis of short sequence variants from population resequencing of two genes in the human genome.	838	Bansal, V
2910	Comprehensive identification of SNPs, indels and structural variants by sequence capture and deep re-sequencing of large genomic regions in pooled DNA samples.	842	Hunt, KA
2914	Population-based detection of rare, surfactant associated gene variants using pooled sample next generation sequencing.	846	Heins, H
2924	Association of Biomolecular Resource Facilities (ABRF). Advancing human genetics through research, communication, and education.	856	Hunter, T

**POSTER SESSION FRIDAY, OCTOBER 23, 2009 7:00 AM–12:00 PM**

Program No.	Title	Poster No.	Lead Author
2926	Optimization of library preparation methods for next-generation sequencing.	858	Scott, AF
2930	Genetic analysis of single cells.	862	Fan, JB
2948	The detailed genomic and epigenetic characterization of human 15q11-13 in an autism endophenotype family.	880	Gregory, SG
2950	Feasibility for detecting epigenetic abnormalities in autism brain.	882	Person, R
2954	Comprehensive DNA methylation analysis in neuronal and non-neuronal cells.	886	Iwamoto, K
2956	Genomic methylation variation correlated with metabolic syndrome-related phenotypes.	888	Carless, MA
2966	Genome-wide DNA methylation patterns associate with genetic and gene-expression variation.	898	Bell, JT
2978	Studying the extent and function of epigenetic variation in twins.	910	Lyle, R
2988	Identification of epipolymorphisms in the human placenta.	920	Yuen, R

**PLATFORM SESSION SATURDAY, OCTOBER 24, 2009 10:00 AM–12:00 PM**

Program No.	Title	Location	Lead Author
217	Identification of multiple novel genetic variants which predispose to prostate cancer by genome-wide association in the PRACTICAL consortium	Ballroom A	Kote-Jarai, Z
278	Genome-wide reprogramming of promoter DNA methylation in the derivation of human induced pluripotent stem cells	Room 311	Fan, G
259	Large CNVs in an isolated population show enrichment in neuropsychiatric diagnoses including an enrichment of three schizophrenia associated CNVs overlapping developmental genes	Room 316	Pietilainen, OPH
279	The retinoblastoma (RB1) gene is imprinted	Room 311	Kanber, D
260	Copy number variation discovery in autism spectrum disorder.	Room 316	Pagnamenta, A
261	Copy-number variation reveals both known and new susceptibility loci for autism spectrum disorders in a population isolate.	Room 316	Rehnström, K
271	A large-scale replication study identifies novel risk loci for Systemic Lupus Erythematosus.	Room 323	Gateva, V
242	Shared genomic segment analysis. A powerful method for detecting rare risk variants.	Ballroom C	Knight, S
273	A comprehensive GWAS follow-up in Multiple Sclerosis identifies novel risk loci.	Room 323	McCauley, JL
283	Genomewide ChIP-sequencing reveals that the ATR-X Syndrome protein influences allele-specific gene Expression through its interaction with guanine-rich VNTRs.	Room 311	Law, MJ
244	High resolution detection of identity by descent with linkage disequilibrium modelling.	Ballroom C	Browning, SR
274	Novel risk variants in a genome wide scan of pediatric onset Inflammatory Bowel Disease.	Room 323	Imielinski, M
235	Prospective comparison of 6 genome wide array genomic hybridization platforms for the detection of copy number variants in mental retardation.	Ballroom B	Tucker, T
311	Common genetic variation in UGT8 is associated with musical ability.	Room 313	Lee, S
304	Development of a pharmacogenetic test that predicts response to inhaled corticosteroids in asthma patients.	Ballroom C	Wu, AC
289	A haplotype in the MSRA gene confers decreased risk of meconium ileus in cystic fibrosis.	Ballroom A	Henderson, LB
321	Common variants in the TSLP/WDR36 region are associated with Eosinophilic Esophagitis.	Room 316	Annaiah, K
289	Genome-wide CNV association study identifies a rare deletion on chromosome 6p25 associated with osteoporotic fractures.	Ballroom B	Estrada, K
307	Genome-wide association study identifies novel genomic regions associated with drug-induced Long QT Syndrome.	Ballroom C	Roden, DM
315	Identification of multiple functional PLTP locus SNPs and their effects on linkage evidence for other chromosomes.	Room 313	Rosenthal, EA
317	Using high density 1meg SNP genotyping arrays to determine the lower size for anomalous contiguous homozygosity as a methodology for generating candidate genes in the NIH Undiagnosed Disease Program.	Room 313	Markello, T
236	Identifying the genetic underpinnings associated with healthspan.	Room 316	Murray, SS

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