PROGRAM AT-A-GLANCE

Registration and Program Pick-up Hours:

Tuesday: 10:00am-7:00pm
Wednesday: 7:00am-5:00pm
Thursday: 7:30am-5:00pm
Friday: 7:30am-5:00pm
Saturday: 7:30am-10:30am

Schedule of ASHG Scientific Sessions and Events

All meeting rooms are located in the Moscone Center unless otherwise indicated. (*) Asterisk denotes events that are by pre-registration only. Otherwise, attendance may be assumed to be open to all scientific registrants

Tuesday, November 6		
4:00pm-4:30pm	1. Presidential Address: The Scientist as a Citizen of the World	Hall D, Lower Level North
4:30pm-6:30pm	2. Plenary Abstract Presentations	Hall D, Lower Level North
7:00pm -8:30pm	ASHG Opening Mixer and Trainee Mixer-within-a-Mixer	Marriott Marquis Hotel Yerba Buena 7/8/9, Lower B2 Level
Wednesday, November 7		
8:00am-10:00am	**Concurrent Invited Session I (3-10):**	
	3. Implementing Next-Generation Sequencing as a Clinical Test	Hall D, Lower Level North
	4. Assessing the Pathogenicity of Genetic Variants: Translating in Vitro and in Silico Advances to the Clinic	Gateway Ballroom 103, Lower Level South
	5. Gene Regulatory Change: The Engine of Human Evolution?	Room 135, Lower Level North
	6. Insights into Human Demography and Selection from Full Genome Sequencing	Room 134, Lower Level North
	7. Age-Related Macular Degeneration—GWAS and Beyond: Guiding Light for the Complex Neurodegenerative Diseases	Gateway Ballroom 104, Lower Level South
	8. "Yes Virginia, Family Studies Really Are Useful for Complex Traits in the Next-Generation Sequencing Era" (session in honor of Dr. Robert Elston's contributions to human genetics in the year of his 80th birthday)	Room 124, Lower Level North
	9. Surveying Customer Responses to Personal Genetic Services	Room 132, Lower Level North
	10. Metabolism, Metals, and Neurodegeneration: Toward Enhanced Understanding of Disease Mechanisms and Rational Therapeutics	Room 130, Lower Level North
10:00am-4:30pm	Exhibits and Posters Open	Exhibit Hall, Lower Level South
10:30am-12:45 pm	**Concurrent Platform Session A (11-19):**	
	11. Genetics of Autism Spectrum Disorders	Hall D, Lower Level North
	12. New Methods for Big Data	Gateway Ballroom 103, Lower Level South
	13. Cancer Genetics I: Rare Variants	Room 135, Lower Level North
	14. Quantitation and Measurement of Regulatory Oversight by the Cell	Room 134, Lower Level North
	15. New Loci for Obesity, Diabetes, and Related Traits	Gateway Ballroom 104, Lower Level South
	16. Neuromuscular Disease and Deafness	Room 124, Lower Level North
	17. Chromosomes and Disease	Room 132, Lower Level North
	18. Prenatal and Perinatal Genetics	Room 130, Lower Level North
	19. Vascular and Congenital Heart Disease	Room 123, Lower Level North
12:45pm-2:15pm	Lunch Break, Open Viewing for Posters and Exhibits	Exhibit Hall, Lower Level South

*12:45pm-2:15pm	Trainee-Mentor Luncheon (Advance ticket purchase required.)	Room 303/305, Esplanade Level South
*12:45pm-2:15pm	Clinical Interpretation of Cytogenomic Arrays: Tools & Resources (Advance ticket purchase required.)	Room 304/306, Esplanade Level South
*12:45pm-2:15pm	Discovering Biological Data at NCBI (Advance ticket purchase required.)	Room 307, Esplanade Level South
2:15pm-4:15pm	**Poster Session I** (Wednesday Poster Authors Present)	Exhibit Hall, Lower Level South
4:30pm-6:30pm	20. Invited Presidential Symposium: Gene Discovery and Patent Law: Present Experience in the U.S. and in Europe	Hall D, Lower Level North
*7:00pm-8:30pm	Interactive Workshop on the UCSC Genome Browser for Intermediate/Advance User (Advance ticket purchase required.)	Room 304/306, Esplanade Level South
*7:00pm-8:30pm	Social Media + Scientists = Success: Strategies for Using Social Media to Benefit Your Research, Your Career and Your Connections (Advance ticket purchase required.)	Room 310, Esplanade Level South
Thursday, November 8		
7:00am-4:30pm	Posters Open	Exhibit Hall, Lower Level South
8:00am-10:00am	**Concurrent Invited Session II (21-28):**	
	21. Mendelian Randomization: Using Genetic Variants to Inform Causality in Observational Epidemiology	Room 135, Lower Level North
	22. Common and Rare CNVs: Genesis, Patterns of Variations and Human Diseases	Hall D, Lower Level North
	23. Advancing Gene Therapy to the Clinic: Molecular Medicines Come of Age	Gateway Ballroom 104, Lower Level South
	24. RNA Splicing in Human Development, Diseases and Natural Variation	Room 124, Lower Level North
	25. Genomic Medicine: ELSI Goes Mainstream	Room 132, Lower Level North
	26. Model Organism Genetics, Human Biology and Human Disease	Gateway Ballroom 103, Lower Level South
	27. Next-Generation Sequencing in Isolated Populations: Opportunities for Accelerated Gene Discovery in Complex Traits	Room 134, Lower Level North
	28. Transforming Medical Student Education in Genetics and Genomics: How Do We Improve Health and Individualize Care through Medical School Genetic and Genomic Curricula?	Room 130, Lower Level North
10:00am-4:30pm	Exhibits Open	Exhibit Hall, Lower Level South
10:30am-12:45pm	**Concurrent Platform Session B (29-37):**	
	29. Next-Generation Sequencing: Methods and Applications	Hall D, Lower Level North
	30. Genetics and Intellectual Disability	Gateway Ballroom 103, Lower Level South
	31. GWAS from Head to Toe	Room 135, Lower Level North
	32. Cardiovascular Genetics: GWAS and Beyond	Room 134, Lower Level North
	33. Clinical Genetics: Mutations, Mutations and Syndromes	Gateway Ballroom 104, Lower Level South
	34. Cancer Genetics II: Clinical Translation	Room 124, Lower Level North
	35. Ethical, Legal, Social and Policy Issues	Room 132, Lower Level North
	36. Chipping Away at Autoimmune Disease	Room 130, Lower Level North
	37. Metabolic Disease Discoveries	Room 123, Lower Level North

12:45pm-2:15pm	Lunch Break, Open Viewing for Posters and Exhibits	Exhibit Hall, Lower Level South
*12:45pm-2:15pm	Diagnostic Challenges: Review and Discussion of Unique Cases, Rare and Unknown Cases (Advance ticket purchase required.)	Room 303/305, Esplanade Level South
2:15pm-4:15pm	**Poster Session II** (Thursday Poster Authors Present)	Exhibit Hall, Lower Level South
4:30pm-6:45pm	**Concurrent Platform Session C (38-46):**	
	38. A Sequencing Jamboree: Exomes to Genomes	Hall D, Lower Level North
	39. Admixture and Demography	Gateway Ballroom 103, Lower Level South
	40. Analysis of Multilocus Systems	Room 135, Lower Level North
	41. Genes Underlying Neurological Disease	Room 134, Lower Level North
	42. Cancer Genetics III: Common Variants	Gateway Ballroom 104, Lower Level South
	43. Genetics of Craniofacial and Musculoskeletal Disorders	Room 124, Lower Level North
	44. Tools for Phenotype Analysis	Room 132, Lower Level North
	45. Therapy of Genetic Disorders	Room 130, Lower Level North
	46. Pharmacogenetics: From Discovery to Implementation	Room 123, Lower Level North
*7:00pm-8:30pm	Galaxy 101: Data Integration, Analysis and Sharing (Separate registration required.)	Room 304/306, Esplanade Level South
*7:00pm- 9:30pm	Trainee Development Program and Networking: Science and Public Policy: Why Should Scientists Care About and Become Active in Public Policy Involving Science? (Separate registration required.)	Room 309, Esplanade Level South
Friday, November 9		
7:00am-4:30pm	Posters Open	Exhibit Hall, Lower Level South
8:00am-10:15am	**Concurrent Platform Session D (47-55):**	
	47. Structural and Regulatory Genomic Variation	Hall D, Lower Level North
	48. Neuropsychiatric Disorders	Gateway Ballroom 103, Lower Level South
	49. Common Variants, Rare Variants, and Everything in-Between	Room 135, Lower Level North
	50. Population Genetics Genome-Wide	Room 134, Lower Level North
	51. Endless Forms Most Beautiful: Variant Discovery in Genomic Data	Gateway Ballroom 104, Lower Level South
	52. Clinical Genetics: Complex Mechanisms and Exome-Discovery	Room 124, Lower Level North
	53 From SNP to Function in Complex Traits	Room 132, Lower Level North
	54. Genetic Counseling and Clinical Testing	Room 130, Lower Level North
	55. Mitochondrial Disorders and Ciliopathies	Room 123, Lower Level North
10:00am-4:30pm	Exhibits Open	Exhibit Hall, Lower Level South
10:30am-11:15am	56. Gruber Genetics Prize Award Presentation and Rosalind Franklin Young Investigator Award Announcement	Hall D, Lower Level North
11:15am-11:45am	57. William Allan Award Presentation	Hall D, Lower Level North
11:45am-12:45pm	58. Membership and Business Meeting	Hall D, Lower Level North
12:45pm-2:15pm	Lunch Break, Open Viewing for Posters and Exhibits	Exhibit Hall, Lower Level South
*12:45pm-2:15pm	Mock Study Section Workshop (Advance ticket purchase required.)	Room 303/305, Esplanade Level South

*12:45pm-2:15pm	Working with High-Throughput Data and Data Visualization (Separate advance registration required.)	Room 304/306, Esplanade Level South
2:15pm-4:15pm	**Poster Session III** (Friday Poster Authors Present)	Exhibit Hall, Lower Level South
4:30pm-6:45pm	**Concurrent Platform Session E (59-67):**	
	59. Genome Structure and Variation	Hall D, Lower Level North
	60. Advances in Neurodegenerative Disease	Gateway Ballroom 103, Lower Level South
	61. Missing Heritability, Interactions and Sequencing	Room 135, Lower Level North
	62. Exome Sequencing Uncovers Etiology of Mendelian Disease	Room 134, Lower Level North
	63. Transcriptional Regulation, Variation and Complexity	Gateway Ballroom 104, Lower Level South
	64. Epigenetics	Room 124, Lower Level North
	65. Advances in Ocular Genetics	Room 132, Lower Level North
	66. Cancer Genetics: Somatic Variants	Room 130, Lower Level North
	67. Developmental Insights into Human Malformations	Room 123, Lower Level North
*7:00pm-8:30pm	Ensembl Web-Based Genomic Tools Workshop for Intermediate/Advance Users (Advance ticket purchase required.)	Room 304/306, Esplanade Level South
*7:00pm-9:00pm	Drama, Discourse and Genomics: IRBs to Ifs—An Interactive Play (Advance ticket purchase required.)	Room 300, Esplanade Level South
Saturday, November 10		
8:00am-8:20am	68. Award for Excellence in Human Genetics Education	Hall D, Lower Level North
8:20am-8:40am	69. Victor A. McKusick Leadership Award Presentation	Hall D, Lower Level North
8:40am-8:45am	70. *AJHG* C.W. Cotterman Awards Announcement	Hall D, Lower Level North
8:45a-8:55am	71. Charles J. Epstein Trainee Awards for Excellence in Human Genetics Research: Announcement of Winners	Hall D, Lower Level North
8:55am-9:20am	72. Curt Stern Award Presentation	Hall D, Lower Level North
9:40am-11:40am	**Concurrent Invited Session III (73-80):**	
	73. Returning Results from Large-Scale Sequencing: Where the Rubber Meets the Road	Gateway Ballroom 103, Lower Level South
	74. Genomic Approaches to Mendelian Disorders	Hall D, Lower Level North
	75. Emerging Applications of Identity by Descent Segment Detection	Gateway Ballroom 104, Lower Level South
	76. The Functional Consequences of microRNA Dysregulation in Human Disease	Room 134, Lower Level North
	77. Centralizing the Deposition and Curation of Human Mutations	Room 132, Lower Level North
	78. Stem Cells and Personalized Medicine	Room 135, Lower Level North
	79. Should Noninvasive Prenatal Diagnosis Augment or Replace Current Prenatal Screening and Diagnosis?	Room 124, Lower Level North
	80. Selection Signatures and the Genetics of Autoimmunity and Infectious Diseases	Room 130, Lower Level North
12:00pm-1:00pm	81. Closing Plenary: Human Genetics 2012 and Beyond: Present Progress and Future Frontiers	Hall D, Lower Level North

Tuesday, November 6
4:00 PM–4:30 PM

SESSION 1 – ASHG Presidential Address: The Scientist as a Citizen of the World
Hall D, Lower Level North, Moscone Center
Presenter:
Mary-Claire King
ASHG 2012 President,
Univ. of Washington

One of the most fulfilling features of our lives as scientists is to act as citizens of the world. We both take this citizenship for granted and take its responsibilities seriously. In one generation, this role has evolved dramatically, as scientists travel far more widely and as modern communication tools enable us to remain in contact with our colleagues worldwide. In my presidential address, I will specify some elements of the scientific life that lead naturally to world citizenship. Then I will focus in particular on how the intellectual structure of human genetics enables us to be particularly effective world citizens. I will also discuss migration as a force in our lives, as well as in human evolution, and will try to define the concept of "home" for a scientist. Finally, I will suggest that scientific goals are both practical and idealistic, and that we should celebrate both.

Tuesday, November 6
4:30 PM–6:30 PM

SESSION 2 – Plenary Abstract Presentations
Hall D, Lower Level North, Moscone Center
Moderator: Joel N. Hirschhorn, 2012 Program Committee Chair Boston Children's Hosp., Harvard Med. Sch. and Broad Inst.

1/4:30 **A novel molecular and functional mechanism predisposing to ototoxicity.** B. Wollnik, E. Pohl, N. Offenhäuser, A. Uzumcu, F. J. Kersten, A. K. Rzadzinska, O. Uyguner, B. Lorente, G. Nürnberg, M. Emiroglu, H. Kayserili, I. del Castillo, P. Nürnberg, T. Moser, C. Kubisch, K. P. Steel, P. P. Di Fiore, H. Kremer, Y. Li.

2/4:50 **Genome-wide comparison of genetic and epigenetic regulatory mechanisms in primates.** Y. Gilad, A. Pai, R. Pique-Regi, C. Cain, J. Degner, N. Lewellen, K. Michelini, J. Pritchard.

3/5:10 **Multidisciplinary and translational task force for neonatal genomics.** E. E. Davis, A. Sabo, N. C. Oien, S. H. Katsanis, H. Cope, K. Sheets, A. Sadeghpour, K. McDonald, M. Kousi, J. R. Willer, J. Kim, S. Dugan-Rocha, D. M. Muzny, A. Ashley-Koch, E. Hauser, M. Hauser, J. Sun, J. Kurtzberg, A. Murtha, B. Boyd, W. B. Gallentine, R. Goldberg, M. T. McDonald, R. A. Gibbs, M. Angrist, C. M. Cotten, N. Katsanis.

4/5:30 **Genome-wide identification and functional analysis of distant-acting craniofacial enhancers.** C. Attanasio, Y. Zhu, M. J. Blow, A. S. Nord, V. Afzal, B. Hallgrimsson, D. FitzPatrick, H. Morrison, E. M. Rubin, L. A. Pennacchio, A. Visel.

5/5:50 **Translational cis-regulation of gene expression in human genome: The effect of human single nucleotide polymorphisms.**Q . Li, A. Makri, Y. Lu, L. Marchand, R. Grabs, M. Rousseau, H. Ounissi-Benkalha1, H. Qu, C. Polychronakos.

6/6:10 **Lessons learned from the NHLBI-Exome Sequencing Project.** S. M. Leal on behalf of NHLBI Exome Sequencing Project.

The Opening Mixer and Trainee-Mixer-within-a-Mixer will follow the plenary session at the Marriott Marquis Hotel, Yerba Buena Ballroom. For those interested, large screens will display the U.S. election updates after the Plenary session has concluded.

Taking photographs or sound recordings in all meeting rooms is strictly prohibited. Thank you for your cooperation.

Wednesday, November 7: Concurrent Invited Session I (3-10)

8:00 AM–10:00 AM

Hall D, Lower Level North	Gateway Ballroom 103, Lower Level South	Room 135, Lower Level North	Room 134, Lower Level North	Gateway Ballroom 104, Lower Level South	Room 124, Lower Level North	Room 132, Lower Level North	Room 130, Lower Level North
SESSION 03 – Implementing of Next-Generation Sequencing as a Clinical Test Co-Moderators: Nazneen Aziz, Col. of American Pathologists, Lexington, MA; and Ira Lubin, Ctr. for Dis. Control and Prevent.	**SESSION 04 – Assessing the Pathogenicity of Genetic Variants: Translating in Vitro and in Silico Advances to the Clinic** Co-Moderators: Marc S. Greenblatt, Univ. of Vermont; and Sean V. Tavtigian, Univ. of Utah	**SESSION 05 – Gene Regulatory Change: The Engine of Human Evolution?** Co-Moderators: James P. Noonan, Yale Univ. Sch. of Med.; and Nadav Ahituv, UCSF	**SESSION 06 – Insights into Human Demography and Selection from Full Genome Sequencing** Co-Moderators: Jeffrey M. Kidd, Univ. of Michigan; and Carlos D. Bustamante, Stanford Univ.	**SESSION 07 – Age-Related Macular Degeneration—GWAS and Beyond: Guiding Light for the Complex Neurodegenerative Diseases** Co-Moderators: Anand Swaroop, NEI/NIH; and Hemin Chin, NEI/NIH	**SESSION 08 – "Yes Virginia, Family Studies Really Are Useful for Complex Traits in the Next-Generation Sequencing Era" (session in honor of Dr. Robert Elston's contributions to human genetics in the year of his 80th birthday)** Co-Moderators: Michael A. Province, Washington Univ. in St. Louis; and Francoise Clerget-Darpoux, INSERM U781, Univ. Paris V, France.	**SESSION 09 – Surveying Customer Responses to Personal Genetic Services** Moderator: J. Scott Roberts, Univ. of Michigan	**SESSION 10 – Metabolism, Metals, and Neurodegeneration: Toward Enhanced Understanding of Disease Mechanisms and Rational Therapeutics** Co-Moderators: Stephen G. Kaler, NICHD/NIH; and Susan J. Hayflick, Oregon Hlth. & Sci. Univ.
08:00 am Challenges of introducing NGS in the clinical laboratory. **S. Richards.**	08:00 am Innovative in vitro and in vivo assays to investigate the function of mismatch repair gene variants in Lynch syndrome. **N. de Wind.**	08:00 am Chromatin profiling of human embryonic tissues identifies regulatory elements with human-specific developmental functions. **J. P. Noonan.**	08:00 am The effect of out-of-Africa migrations on the distribution of deleterious alleles in diverse human genomes. **B. M. Henn.**	08:00 am The bigger the better: Searching for novel loci for age-related macular degeneration in a large consortium effort. **I. Heid.**	08:00 am Whole genome sequencing in large pedigrees for the identification of human QTLs. **J. Blangero.**	08:00 am Interpretomics: Using studies of DTC testing and the return of research results to shape the interpretation of personal whole genomic sequence data. **D. Kaufman.**	08:00 am Alzheimer disease and the metal hypothesis. **R. E. Tanzi.**
08:15 am Addressing the fundamentals: NGS validation and implementation in a clinical setting. **M. Hegde.**	08:30 am Analysis of splicing abnormalities to define pathogenic variants in cancer susceptibility genes. **A. B. Spurdle.**	08:25 am Many human accelerated regions are developmental enhancers. **K. S. Pollard.**	08:30 am Genetic adaptations to new environments in humans. **A. Di Rienzo.**	08:30 am From genetic association to causal alleles by resequencing and exome arrays: The stage after GWAS. **G. Abecasis.**	08:30 am Linkage and association information should be considered as complementary and not redundant. **F. Clerget-Darpoux.**	08:30 am Impact of DTC genomic testing at long-term follow-up. **C. S. Bloss.**	08:30 am Neurodegeneration with brain iron accumulation. **S. J. Hayflick.**
08:45 am Proficiency testing, quality control and development of reference material for NGS clinical testing. **E. Lyon.**	09:00 am Integrating in silico with in vitro, statistical, and phenotype data to classify missense variants: A paradigm that is ready for translation to the clinic. **S. V. Tavtigian.**	08:50 am Linking human and mammalian genotypes to phenotype. **G. Bejerano.**	09:00 am Insights into selective sweeps and diversity from thousands of sequenced genomes. **R. Hernandez.**	09:00 am An integrated hypothesis of the development and progression of age-related macular degeneration based upon available genetic and biological data. **G. S. Hageman.**	09:00 am Power to find rare causal variants in pedigrees. **M. A. Province.**	09:00 am Rendering population differences meaningful: A study of consumer interpretation of genetic diversity. **S. S-J. Lee.**	09:00 am Friedreich ataxia and diseases of iron sulfur cluster assembly. **T. A. Rouault.**
09:00 am Development of accreditation standards for laboratories offering NGS as a clinical test. **N. Aziz.**	09:20 am CAGI: The Critical Assessment of Genome Interpretation, a community experiment to evaluate phenotype prediction. **S. E. Brenner.**	09:15 am The role of repetitive elements in driving human and mammalian genome regulation. **D. Odom.**	09:30 am A genomic view of the demographic and adaptive history of African pygmies. **L. Quintana-Murci.**	09:30 am An updated recipe for Mendel's pea soup. **M. A. Pericak-Vance.**	09:30 am Whither human genetics? **R. C. Elston.**	09:30 am The role of personal genomic testing companies in research: A panel discussion featuring industry and academic perspectives. **J. Mountain et al.**	09:20 am Neurodegeneration and disorders of copper transport. **S. G. Kaler.**
09:30 am Lessons from the clinic—What's next? **H. Jacob.**	09:40 am International collaborations to establish standards for classifying genetic variants and to disseminate results. **M. S. Greenblatt.**	09:40 am Evidence of regulatory turnover in the human lineage revealed by comparing mammalian constraint, human diversity, and biochemical activity. **M. Kellis.**					09:40 am Exploring the link between glucocerebrosidase mutations and Parkinson disease. **E. Sidransky.**

During the meeting, attendees are encouraged to post thoughts on exciting scientific or clinical advances heard at a session or workshop and on challenges that the field will face by using **hashtag #ASHG2012** or by posting on the ASHG Facebook page. The posts will be monitored and may be discussed at the Closing Symposium on Saturday.

Twitter: **@Genetics Society,** #ASHG2012

Facebook: **https://www.facebook.com/GeneticsSociety**

Follow ASHG on Twitter before and during the week of the 2012 Meeting to get the latest updates, tips, news, and announcements.

RECORDED PRESENTATIONS AVAILABLE AFTER THE MEETING
Invited Sessions, Award Presentations, and the Plenary Sessions will be recorded and posted on the Web site after the meeting.

These recordings are free to meeting registrants.
Visit the ASHG Web site in early December to view these presentations.

Visit the Exhibits
Wednesday, Thursday and Friday: 10:00 am – 4:30 pm

Posters Open
Wednesday: 10:00am – 4:30 pm
Thursday: 7:00 am – 4:30 pm
Friday: 7:00 am – 4:30 pm

Wednesday, November 7: Concurrent Platform (abstract-driven) Session A (11-19)

10:30 AM–12:45 PM

	Hall D, Lower Level North	Gateway Ballroom 103, Lower Level South	Room 135, Lower Level North	Room 134, Lower Level North	Gateway Ballroom 104, Lower Level South	Room 124, Lower Level North	Room 132, Lower Level North	Room 130, Lower Level North	Room 123, Lower Level North
	SESSION 11 – Genetics of Autism Spectrum Disorders Co-Moderators: Fuki M. Hisama, Univ. of Washington; and Dan E. Arking, Johns Hopkins Sch. of Med.	**SESSION 12 – New Methods for Big Data** Co-Moderators: Elizabeth Marchani, Univ. of Washington; and Rita Cantor-Chiu, UCLA Sch. of Med.	**SESSION 13 – Cancer Genetics I: Rare Variants** Co-Moderators: Ellen L. Goode, Mayo Clin. Col. of Med.; and John D. McPherson, Ontario Inst. for Cancer Res., Canada	**SESSION 14 – Quantitation and Measurement of Regulatory Oversight by the Cell** Co-Moderators: Richard M. Myers, HudsonAlpha Inst. for Biotechnol., Huntsville, AL; and Jeffrey C. Barrett, Wellcome Trust Sanger Inst., U.K.	**SESSION 15 – New Loci for Obesity, Diabetes, and Related Traits** Co-Moderators: Jose Florez, Massachusetts Gen. Hosp.; and Richa Saxena, Massachusetts Gen. Hosp.	**SESSION 16 – Neuromuscular Disease and Deafness** Co-Moderators: Anthony Antonellis, Univ. of Michigan; and Thomas Glover, Univ. of Michigan	**SESSION 17 – Chromosomes and Disease** Co-Moderators: Christa Lese Martin, Emory Univ.; and Blake Ballif, Signature Genomics, Spokane, WA	**SESSION 18 – Prenatal and Perinatal Genetics** Co-Moderators: Natalie Blagowidow, Harvey Inst. for Human Genet., Baltimore; and David Chitayat, Mount Sinai Hosp., Toronto, Canada	**SESSION 19 – Vascular and Congenital Heart Disease** Co-Moderators: Amy Roberts, Children's Hosp. Boston; and Bart L. Loeys, Univ. of Antwerp., Belgium
10:30	**7** Genetic epidemiology of autism spectrum disorder in a cohort of over 11,000 affected sibships and half-sibships: Evidence of genetic and maternal environmental effects. **N. Risch et al.**	**16** The detection, structure and uses of haplotype identity in population genetic data. **D. Xifara et al.**	**25** Exome sequencing of more than 6,700 samples and the study of genetic susceptibility to common cancer. **A. Kiezun et al.**	**34** ChipEnrich: Gene set enrichment testing for ChIP-seq data. **R. P. Welch et al.**	**43** A genome-wide association analysis of early-onset severe obesity: The SCOOP project. **E. Wheeler et al.**	**52** The TRK-fused gene is mutated in hereditary motor and sensory neuropathy with proximal dominant involvement (HMSN-P). **H. Ishiura et al.**	**61** Characterization of de novo copy-number variations in two subjects with a constitutional "CNV mutator" phenotype. **P. Liu et al.**	**70** Lessons learned from next-gen cytogenetics: Whole genome sequence-based prenatal diagnosis of apparently balanced de novo chromosome rearrangements. **Z. Ordulu et al.**	**79** Heterozygous germline mutations in a prototypical TGFβ repressor cause Shprintzen-Goldberg syndrome with aortic aneurysm. **A. J. Doyle et al.**
10:45	**8** Identifying inherited autism mutations using whole exome sequencing. **T. W. Yu et al.**	**17** Inferring and sequencing the founding bottleneck of Ashkenazim. **I. Pe'er et al.**	**26** Exome sequencing of families severely affected with breast cancer suggests eight new candidate genes: *ATR, BAP1, CHEK1, GEN1, KANK4, OBSL1, RAD51B* and *TP53BP1.* **C. H. Spurrell et al.**	**35** Enhanced exome sequencing to capture genome-wide common variants. **I. C. R. M. Kolder et al.**	**44** Mapping obesity traits using an integrated 'omics' approach in adipose tissue from female twins. **A. K. Hedman et al.**	**53** Mutation in the autophagy-related *TECPR2* gene causes hereditary spastic paraparesis. **D. Oz-Levi et al.**	**62** Associations between gene expression and phenotypes in 16p11.2 rearrangements. **E. Migliavacca et al.**	**71** The use of chromosome microarray analysis as a first-line test in pregnancies with a priori low risk for detection of submicroscopic chromosomal abnormalities. **F. Fiorentino et al.**	**80** Loss-of-function mutations in *TGFB2* cause Loeys-Dietz syndrome: Towards solving the TGFβ paradox in aortic aneurysmal disease. **B. Loeys et al.**
11:00	**9** Identical by descent filtering in extended families reveals novel autism genes detected by exome sequencing. **H. N. Cukier et al.**	**18** Statistical methods for association test of rare variants using summarized data without individual-subject information. **Q. Zhang et al.**	**27** Rare variants in *XRCC2* as breast cancer susceptibility alleles. **F. S. Hilbers et al.**	**36** Complete HIV-1 genomes from sequencing single molecules: Simple and complex samples. **M. P. S. Brown et al.**	**45** Whole exome sequencing identifies candidate causal genes for severe insulin resistance. **F. Payne et al.**	**54** Spinal muscular atrophy associated with progressive myoclonic epilepsy is caused by mutations in *ASAH1.* **J. Melki et al.**	**63** De novo triplication can arise from a duplication of the 17p12 region and confers a severe Charcot-Marie-Tooth, type 1A phenotype. **V. Gelowani et al.**	**72** The challenge of preconceptional, preimplantation, and prenatal genetic diagnoses of mitochondrial DNA disorders. **J. Steffann et al.**	**81** Genetic dissection of aortic disease in the Marfan syndrome. **F. Ramirez et al.**
11:15	**10** The discovery and validation of genes recurrently disrupted in autism spectrum disorders. **B. J. O'Roak et al.**	**19** Testing for rare variant associations in the presence of missing data. **P. Livermore Auer et al.**	**28** *HOXB13* is a susceptibility gene for prostate cancer: Results from the International Consortium for Prostate Cancer Genetics. **K. Cooney et al.**	**37** DeTCT pipeline: A software pipeline for the analysis of transcript count data. **J. A. Morris et al.**	**46** Exome analysis in 8,232 Finnish men identifies novel loci and low-frequency variants for insulin processing and secretion. **J. R. Huyghe et al.**	**55** Genetic variants in chromatin modifying genes cause D4Z4 hypomethylation, *DUX4* expression, and contraction-independent facioscapulohumeral muscular dystrophy (FSHD2). **D. G. Miller et al.**	**64** A long, non-coding RNA from the Prader-Willi critical region forms a subnuclear cloud and recruits transcriptional activating complexes to the Snord116 locus in postnatal neurons. **W. T. Powell et al.**	**73** The incidence and spectrum of genomic imbalance in products of conception: New insights from SNP microarray analysis of 2,400 miscarriage specimens. **B. Levy et al.**	**82** Bicuspid aortic valve, aortic coarctation and patent ductus associated with *MATR3* disruption in human and mouse. **F. Quintero-Rivera et al.**
11:30	**11** Rare complete human knockouts: Population distribution and significant role in autism spectrum disorders. **E. T. Lim et al.**	**20** Quantitative trait locus analysis for next-generation sequencing with the functional linear models. **M. Xiong et al.**	***29** Parkinson disease and melanoma: A common genetic pathway linked to PARKIN inactivation. **N. Soufir et al.**	**38** Fast genome-wide QTL association mapping with pedigrees. **H. Zhou et al.**	**47** Global genomic and transcriptomic variation in human pancreatic islets reveals novel loci associated with type 2 diabetes and related traits. **J. Fadista et al.**	**56** Unexpected extension of the phenotype spectrum associated with *SMAD3* mutations in aneurysms-osteoarthritis syndrome. **M. Aubart et al.**	**65** Molecular analysis of patients whose clinical features overlap the 22q11.2 deletion syndrome. **S. Saitta et al.**	**74** Noninvasive whole-genome sequencing of a human fetus. **J. O. Kitzman et al.**	**83** Identification of the cause of blue rubber bleb nevus syndrome. **J. Soblet et al.**
11:45	**12** Exome-based discovery of CNVs in simplex autism families. **N. Krumm et al.**	**21** A rapid and powerful method for protein-protein interaction- and pathway-based association analysis in genome-wide association studies. **M. Li et al.**	**30** Exome sequencing in families at high risk for lymphoid malignancies. **L. R. Goldin et al.**	**39** Discovering SNPs regulating human gene expression using allele specific expression from RNA-seq data. **E. Eskin et al.**	**48** Identification of a novel genome-wide significant association with type 2 diabetes risk in Mexican and Mexican Americans. **A. L. Williams et al.**	**57** Whole-exome sequencing for autosomal recessive non-syndromic deafness: 93% of known genes covered and *OTOGL* and *SLITRK6* are novel genes. **M. Tekin et al.**	**66** Mouse model implicates *GNB3*copy number in a novel childhood obesity syndrome. **I. S. Goldlust et al.**	**75** Spina bifida risk is conferred by multiple polymorphisms in folate one-carbon pathway genes. **D. Gilbert et al.**	**84** Identifying genetic determinants of congenital heart defect in Down syndrome. **M. R. Sailani et al.**
12:00	**13** Delta catenin (*CTNND2*): Genetics and function of a novel autism gene. **T. Turner et al.**	**22** Statistics for X-chromosome association. **U. Ozbek et al.**	**31** Rare allelic forms of *PRDM9* associated with childhood leukemia. **J. Hussin et al.**	**40** Association of genetic variation affecting exon skipping to disease susceptibility. **Y. Lee et al.**	**49** Discovery and fine-mapping of type 2 diabetes susceptibility loci through trans-ethnic meta-analysis. **A. Mahajan et al.**	**58** Whole exome sequencing and more to unravel the genetics and genotype-phenotype correlations for deafness. **H. Kremer et al.**	**67** Modeling neurogenesis impairment in Down syndrome using induced pluripotent stem cells from monozygotic twins discordant for trisomy 21. **Y. Hibaoui et al.**	**76** Bioinformatics approach for identifying the genetic contributions to preeclampsia. **A. Uzun et al.**	**85** Transcriptome-wide decreased variation in gene expression of Down syndrome fibroblasts: Selection or canalization? **K. Popadin et al.**
12:15	**14** Novel hotspots of recurrent copy number variation associated with autism spectrum disorder. **S. Girirajan et al.**	**23** Joint association analysis of pleiotropy SNPs using GWAS summary statistics. **R. M. Salem et al.**	**32** De novo mutation of the TGF beta family in early-onset ovarian cancer. **I. Tournier et al.**	**41** Haplotype-based variant detection and interpretation enables the population-scale analysis of multi-nucleotide sequence variants. **E. Garrison et al.**	**50** *TCF7L2* genetic variation is associated with impaired incretin effect and lower glucagon. **B. Chamarthi et al.**	**59** A mutation in Ca^{2+} binding protein 2, expressed in cochlear inner hair cells, causes autosomal recessive hearing impairment. **I. Schrauwen et al.**	**68** Discovery and interpretation of balanced chromosomal aberrations in neurodevelopmental abnormalities and prenatal diagnostics. **M. E. Talkowski et al.**	**77** Antenatal spectrum of CHARGE syndrome in 40 fetuses with *CHD7* mutations. **M. Legendre et al.**	**86** Mutations in *OLFML2B* within the QT interval associated region 1q23.3 disturb cardiac repolarization, predispose to Long-QT syndrome and to sudden infant death syndrome. **A. Pfeufer et al.**
12:30	**15** Cluster analysis defines subgroups of phenotypic expression for autism spectrum disorders. **O. J. Veatch et al.**	**24** Multivariate regression-based analysis of relative abundance data in metagenomics. **O. Libiger et al.**	**33** Somatic activating mutations in *PIK3CA* cause progressive segmental overgrowth. **M. J. Lindhurst et al.**	**42** eQTL analysis identifies novel associations between genotype and gene expression in the human intestine. **B. Kabakchiev.**	**51** Novel locus including *FGF21* is associated with dietary macronutrient intake. **A. Y. Chu et al.**	**60** Comprehensive diagnosis for hearing loss using personal genomics: The first 100 cases. **E. Shearer et al.**	**69** Predisposition of acrocentric short arm fusions due to nuclear location, nucleolar disorganization, and telomere-induced DNA damage. **K. M. Stimpson et al.**	**78** Genetic normalization of day-3 embryos: Results from two independent preimplantation genetic screening laboratories. **P. Brezina et al.**	**87** The impact of inherited genetic variants associated with lipid profile, hypertension, and coronary artery disease on the risk of intracranial and abdominal aortic aneurysms. **F. N. G. van 't Hof et al.**

* Presenting Author Change

Thursday, November 8: Concurrent Invited Session II (21-28)

8:00 AM–10:00 AM

Room 135, Lower Level North	Hall D, Lower Level North	Gateway Ballroom 104, Lower Level South	Room 124, Lower Level North	Room 132, Lower Level North	Gateway Ballroom 103, Lower Level South	Room 134, Lower Level North	Room 130, Lower Level North
SESSION 21 – Mendelian Randomization: Using Genetic Variants to Inform Causality in Observational Epidemiology Co-Moderators: David M. Evans, Univ. of Bristol, U.K.; and Lyle J. Palmer, Univ. of Ontario, Canada	**SESSION 22 – Common and Rare CNVs: Genesis, Patterns of Variations and Human Diseases** Co-Moderators: Chack Yung Yu, Nationwide Children's Hosp. and The Ohio State Univ.; and Edward J. Hollox, Univ. of Leicester, U.K.	**SESSION 23 – Advancing Gene Therapy to the Clinic: Molecular Medicines Come of Age** Moderator: Beverly Davidson, Univ. of Iowa	**SESSION 24 – RNA Splicing in Human Development, Diseases and Natural Variation** Co-Moderators: David E. Symer, The Ohio State Univ. Comprehen. Cancer Ctr.; and Richard A. Padgett, Lerner Res. Inst., Cleveland	**SESSION 25 – Genomic Medicine: ELSI Goes Mainstream** Co-Moderators: Wylie Burke, Univ. of Washington; and James P. Evans, Univ. of North Carolina at Chapel Hill	**SESSION 26 – Model Organism Genetics, Human Biology and Human Disease** Co-Moderators: Phil Hieter, Univ. of British Columbia; and Hal Dietz, Johns Hopkins Univ. Sch. of Med.	**SESSION 27 – Next-Generation Sequencing in Isolated Populations: Opportunities for Accelerated Gene Discovery in Complex Traits** Co-Moderators: William K. Scott, Univ. of Miami; and Jeffrey R. O'Connell, Univ. of Maryland Baltimore	**SESSION 28 – Transforming Medical Student Education in Genetics and Genomics: How Do We Improve Health and Individualize Care through Medical School Genetic and Genomic Curricula?** Co-Moderators: Joann N. Bodurtha, Johns Hopkins Univ.; and Joan Scott, NCHPEG, Lutherville, MD
08:00 am Mendelian randomization: Overcoming the limitations. **G. D. Smith.**	08:00 am CNVs engaged in immune complex handling and autoimmune diseases: Complement C4 and immunoglobulin Fc-gamma receptors. **C. Y. Yu.**	08:00 am Safety and efficacy of AAV-mediated gene transfer to liver for severe hemophilia B. **K. High.**	08:00 am Functional consequences of minor spliceosomal snRNA mutations in human development and natural variation. **D. E. Symer.**	08:00 am Views of patients, parents of patients, and clinicians toward whole genome sequencing for clinical care management. **A. A. Lemke.**	08:00 am Budding yeast: Lessons from yeast applied to the study of human genetic diseases of protein traffic. **R. Schekman.**	08:00 am Using low-pass whole genome sequencing to create a reference population for genome imputation in an isolated population: Examples from the SardiNIA study. **S. Sanna.**	08:00 am Genes to Society—3 years of implementation. **D. Valle.**
08:30 am Utilizing multiple variants to improve Mendelian randomization studies. **B. Pierce.**	08:30 am Human lineage-specific CNVs: DUF1220 domain copy number linked to cognitive disease and brain evolution. **J. M. Sikela.**	08:25 am Safety and efficacy after AAV2 re-administration in subjects with congenital blindness due to RPE65 mutations. **J. Bennett.**	08:25 am Multicopy snRNA genes and neurodegeneration. **S. L. Ackerman.**	08:30 am My46: An innovative web-based approach to managing and returning results from exome and whole genome sequencing. **H. K. Tabor.**	08:30 am The nematode worm: Mechanisms regulating aging in worms and man. **C. Kenyon.**	08:30 am Fine-mapping linkage of age-related traits using whole-exome sequencing in a midwestern Amish population sample. **W. K. Scott.**	08:30 am The Vermont Integrated Curriculum: The UVM experience. **L. Burke.**
09:00 am Application of Mendelian randomization analyses in prospective studies from Denmark. **A. Tybjærg-Hansen.**	08:55 am TAR: A mixed genomic disorder caused by a low-frequency regulatory SNP combined with a 1q21.1 microdeletion. **W. H. Ouwehand.**	08:45 am Advancing gene therapy for ADA-SCID and beyond. **M-G. Roncarolo.**	08:50 am Understanding the chemical mechanisms and biological implications of splicing reactions. **R. A. Padgett.**	09:00 am Returning "actionable" results to family members in a pancreatic cancer biobank: Views of probands and family members. **B. Koenig.**	09:00 am The zebrafish: Zebrafish heart development and function. **D. Stainier.**	09:00 am The many-of-few: The power of genetic isolates for discovery and function of rare variants. **J. R. O'Connell.**	08:55 am Effecting change: Building a genetics curriculum that supports the physicians of tomorrow. **L. Potocki.**
09:30 am Mendelian randomization for HDL levels and implications for clinical risk prediction. **B. F. Voight.**	09:20 am Genetic and environmental risk factors for de novo CNVs. **T. W. Glover.**	09:10 am Gene therapy for the leukodystrophies. **N. Cartier.**	09:15 am Overlaying RNA maps onto human disease. **R. B. Darnell.**	09:20 am Approaches and attitudes on return of WGS/WES results. **K. Ormond.**	09:30 am The laboratory mouse: Mouse models of glaucoma and retinal ganglion cell loss. **S. John.**	09:30 am Studying rare variants in the Genetic Research in Isolated Populations program. **C. van Duijn.**	09:20 am Lessons learned from the introduction of personalized genotyping into a medical school curriculum. **L. Demmer.**
	09:40 am Frequency estimation of low-level somatic mosaicism for pathogenic CNVs. **P. T. Stankiewicz.**	09:30 am AAV gene therapy for childhood onset neurological disease caused by lysosomal enzyme deficiencies. **B. Davidson.**	09:40 am "Seq-ing" the Myotonic Dystrophy Transcriptome. **E. Wang.**	09:40 am Next steps in development of best practices for use of genome sequencing in clinical care. . **A. McGuire.**			09:40 am Personal genotyping in a medical school curriculum on genomics and personalized medicine. **K. Salari.**

American Society of Human Genetics October 22-26 2013 BOSTON

Invited Proposals Now Being Accepted:

Deadline, December 5, 2012

Visit the Exhibits
Wednesday, Thursday and Friday: 10:00 am – 4:30 pm

Posters Open
Wednesday: 10:00am – 4:30 pm
Thursday: 7:00 am – 4:30 pm
Friday: 7:00 am – 4:30 pm

Thursday, November 8: Concurrent Platform (abstract-driven) Session B (29-37)

10:30 AM–12:45 PM

	Hall D, Lower Level North	Gateway Ballroom 103, Lower Level South	Room 135, Lower Level North	Room 134, Lower Level North	Gateway Ballroom 104, Lower Level South	Room 124, Lower Level North	Room 132, Lower Level North	Room 130, Lower Level North	Room 123, Lower Level North
	SESSION 29 – Next-Generation Sequencing: Methods and Applications Co-Moderators: John S. Witte, UCSF; and Priya Duggal, Johns Hopkins Bloomberg Sch of Publ. Hlth.	**SESSION 30 – Genetics and Intellectual Disability** Co-Moderators: Roger Reeves, Johns Hopkins Univ. ; and Heidi Rehm, Harvard Univ.	**SESSION 31 – GWAS from Head to Toe** Co-Moderators: Erik Ingelsson, Karolinska Inst., Sweden; and Nora Franceschini, Univ. of North Carolina at Chapel Hill	**SESSION 32 – Cardiovascular Genetics: GWAS and Beyond** Co-Moderators: Cristen J. Willer, Univ. of Michigan; and Panagiotis Deloukas, Wellcome Trust Sanger Inst., U.K.	**SESSION 33 – Clinical Genetics: Mutations, Mutations and Syndromes** Co-Moderators: Nathaniel Robin, Univ. of Alabama at Birmingham; and Anne Slavotinek, UCSF	**SESSION 34 – Cancer Genetics II: Clinical Translation** Co-Moderators: Robert Pilarski, The Ohio State Univ.; and Stephen Thibodeau, Mayo Clin.	**SESSION 35 – Ethical, Legal, Social and Policy Issues** Co-Moderators: Maureen Smith, Northwestern Univ.; and Neil Lamb, HudsonAlpha Inst. for Biotechnol., Huntsville, AL	**SESSION 36 – Chipping Away at Autoimmune Disease** Co-Moderators: Judy H. Cho, Yale Univ.; and Soumya Raychaudhuri, Brigham and Women's Hosp.	**SESSION 37 – Metabolic Disease Discoveries** Co-Moderators: Kimberly Chapman, Children's Natl. Med. Ctr.; and Hans Andersson, Tulane Univ. Med. Ctr.
10:30	**88** The value of population-specific reference panels for genotype imputation in the age of whole-genome sequencing. **C. Fuchsberger et al.**	**97** Diagnostic exome sequencing in patients with intellectual disability of unknown cause. **J. de Ligt et al.**	**106** Androgenetic alopecia: Identification of four new genetic risk loci and evidence for the contribution of WNT-signaling to its etiology. **S. Heilmann et al.**	**115** Coronary artery disease loci identified in over 190,000 individuals implicate lipid metabolism and inflammation as key causal pathways: Evidence for independent signals in many of the risk loci. **S. Kanoni et al.**	**124** Baraitser-Winter syndrome: Delineation of the phenotypicspectrum in a large series of molecularly defined patients. **A. Verloes et al.**	**133** Clinical implementation of a cancer care model based on comprehensive molecular profiling of tumor-normal pairs. **J. C. Taylor et al.**	**142** Newborn screening for cystic fibrosis: Preliminary results on the false positive experience. **C. J. Barg et al.**	**151** Immunochip: Redefining the genetic architecture of multiple sclerosis. **J. McCauley.**	**160** Mutations in *DDHD2* cause recessive spastic paraplegia with intellectual disability, thin corpus callosum and periventricular white matter hyperintensities. **A. P. M. de Brouwer et al.**
10:45	**89** Fast and accurate 1000 Genomes imputation using summary statistics or low-coverage sequencing data. **B. Pasaniuc et al.**	**98** C-terminal deletions of the *AUTS2* locus cause distinct syndromic features and cognitive impairment. **E. Voorhoeve et al.**	**107** A polymorphism in human estrogen-related receptor beta is associated with early indications of hearing loss from acoustic overload in young adult musicians. **V. C. Henrich et al.**	**116** Genome-wide association study in Han Chinese identifies four new susceptibility loci for coronary artery disease. **D. Gu et al.**	**125** Three novel mutations in *MED12* cause Ohdo syndrome Maat-Kievit-Brunner type. **A. T. Vulto-van Silfhout et al.**	**134** Clinical implementation of single nucleotide polymorphism microarrays in pediatric cancer and non-malignant hematologic disorders. **X. Lu et al.**	**143** Conflicting views on newborn and infant genetic screening: Perspectives of relatives of children with genetic conditions causing developmental delay and parents of healthy children. **S. A. Metcalfe et al.**	**152** Dense genotyping of candidate genes indentifies 16 new susceptibility loci in ankylosing spondylitis. **A. Cortes et al.**	**161** Lipidomics of Gaucher disease: Substrate composition and nature is dependent on tissue/region and acid β-glucosidase mutations: Phenotypic implications. **Y. Sun et al.**
11:00	**90** Accurate haplotype estimation using phase informative sequencing reads. **O. Delaneau et al.**	**99** Autism traits in the RASopathies. **I. Corbin et al.**	**108** Dissection of polygenic variation for human height into individual variants, specific loci and biological pathways from a GWAS meta-analysis of 250,000 individuals. **T. Esko et al.**	**117** Discovery of 63 novel loci and refinement of known loci associated with lipid levels. **C. Willer et al.**	**126** Heterogeneity of mutational mechanisms and modes of inheritance in auriculo-condylar syndrome. **C. Gordon et al.**	**135** A prospective clinical trial to evaluate DNA sequencing as a diagnostic tool to guide cancer therapy. **A. M. K. Brown et al.**	**144** Do research participants really want to know? The Seattle Colorectal Cancer Family Registry experience on the return of research genetic test results. **M. Laurino et al.**	**153** Dense fine-mapping study identifies novel disease loci and implicates coding and non-coding variation in primary biliary cirrhosis risk. **J. Z. Liu et al.**	**162** Sterol precursors induce Niemann-Pick C disease phenotypes in Smith-Lemli-Opitz syndrome causing defective LDL-cholesterol utilization that is corrected by imino-sugars. **C. A. Wassif et al.**
11:15	**91** An LD-based method for genotype calling and phasing using low-coverage sequencing reads and a haplotype scaffold. **A. Menelaou et al.**	**100** Identification of novel recessive mutations in genes for intellectual disability. **B. De Vries et al.**	**109** Genome-wide association studies meta-analysis for fracture risk points to loci related to hormonal and neurological pathways: The GEFOS Consortium. **L. Oei et al.**	**118** The Kaiser Permanente/UCSF Genetic Epidemiology Research Study on Adult Health and Aging: Genome-wide association study of plasma HDL and LDL and treatment response in over 100,000 subjects. **T. J. Hoffmann et al.**	**127** Genetic heterogeneity of Myhre syndrome. **C. Le Goff et al.**	**136** Whole genome sequencing of a highly aggressive melanoma identified *BRAF L597* mutants associated with sensitivity to MEK inhibitors. **Z. Zhao et al.**	**145** The student-athletes' knowledge of sickle cell trait and the impact of mandatory genetic testing. **N. Lovick et al.**	**154** Fifteen novel psoriasis susceptibility loci: Disease-specific signals highlight the role of innate immunity. **L. C. Tsoi et al.**	***163** Glucose kinetics in subjects with MELAS syndrome: Interim results. **A. El-Hattab et al.**
11:30	***92** Mixed functional linear model for sequence-based quantitative trait association studies unifying population and family study designs. **Y. Zhu et al.**	**101** Causal de novo SNVs, indels and CNVs in children with undiagnosed developmental disorders. **M. Hurles et al.**	**110** Genetic landscape of the red blood cell. **J. C. Chambers et al.**	**119** Genome-wide screen with 1000 Genomes imputed data identifies 19 new lipid loci and new variants with stronger effects in previously known loci. **I. Surakka et al.**	**128** Seven novel families with ADCL favor clinical and molecular homogeneity. **C. Bodemer et al.**	**137** Identification of novel mechanisms of drug resistance in *BRCA1*-deficient cancer by exome and RNA sequencing. **K. K. Dhillon et al.**	**146** Impact of direct-to-consumer pharmacogenomic testing. **C. S. Bloss et al.**	**155** MHC fine-mapping in celiac disease reveals structural basis of HLA-gluten interaction. **J. Gutierrez-Achury et al.**	**164** Phenylbutyrate therapy for pyruvate dehydrogenase complex deficiency. **R. Ferriero et al.**
11:45	**93** Rare variant extensions of the transmission disequilibrium test detects associations with autism exome sequence data. **Z. He et al.**	**102** Making headway with the molecular and clinical definition of rare genetic disorders with intellectual disability. **M. H. Willemsen et al.**	**111** Discovery and fine-mapping of serum protein loci through trans-ethnic meta-analysis. **A. P. Morris et al.**	**120** Rare coding variation and risk for myocardial infarction: an exome chip study of ~6,000 cases and controls. **R. Do.**	***129** Comprehensive clinical and molecular analysis of 12 families with type I recessive cutis laxa. **S. Hadja Rabia et al.**	**138** *BRCA1* and *BRCA2* mutational spectrum in a normal population: Implications for clinical diagnostics and incidental findings. **E. Ruark et al.**	**147** Impact of genomic risk for type 2 diabetes on health behaviors. **S. B. Haga et al.**	**156** Host-microbe interactions shape genetic risk for inflammatory bowel disease. **J. Barrett.**	**165** Etiologies for neurocognitive delays in argininosuccinic aciduria. **A. Erez et al.**
12:00	**94** Methods for noninvasive prenatal determination of fetal genomes. **M. W. Snyder et al.**	**103** *MBD5* dosage affects multiple neurodevelopmental pathways in common with other genetic syndromes. **S. V. Mullegama et al.**	**112** The Kaiser Permanente/UCSF Genetic Epidemiology Research Study on Adult Health and Aging: A genome-wide association study of telomere length in a multi-ethnic cohort of 100,000 subjects. **M. Kvale et al.**	**121** High exome mutational burden in 58 African Americans with persistent extreme blood pressure. **K.-D. H. Nguyen et al.**	**130** M694V mutation in Armenian-Americans: A ten-year retrospective study of *MEFV* mutations testing for familial Mediterranean fever at UCLA. **F. S. Ong et al.**	**139** Targeted re-sequencing of 10 ovarian cancer candidate genes in 2,240 cases and 355 controls. **H. Song et al.**	**148** African American attitudes toward exome and whole genome sequencing. **J. Yu et al.**	**157** Sequencing-based and multiplatform genome-wide association study for multiple sclerosis and type 1 diabetes in Sardinians. **I. Zara et al.**	**166** Enzyme substitution therapy for phenylketonuria delivered orally using a genetically modified probiotic: Proof of principle. **J. Christodoulou et al.**
12:15	**95** Associating complex traits with rare variants identified by NGS: Improving power by a position-dependent kernel approach. **U. Bodenhofer et al.**	**104** Exome sequencing in X-linked intellectual disability family assess the role of the *KIAA2022* gene in the etiology of intellectual disability. **M. Rio et al.**	**113** Heritability of the variation in aging in two longitudinal family cohort studies: SardiNIA/Progenia Study and Framingham Heart Study. **J. Bragg-Gresham et al.**	**122** Chipping a hole-in-one from the FAIRE way: Use of post-GWAS fine-mapping genotyping arrays for functional variant discovery. **A. J. P. Smith et al.**	**131** Clinical features of individuals with Floating-Harbor syndrome due to mutations in *SRCAP*. **S. M. Nikkel et al.**	**140** Enhanced detection of low-level mosaic mutations in *RB1* gene in sporadic unilateral RB by ion torrent semiconductor sequencing: Risk of second cancer. **Z. Chen et al.**	**149** Personalized health literacy in the age of personalized medicine: Results from a deliberative public engagement exercise. **B. J. Wilson et al.**	**158** Admixture mapping for asthma in Latinos identifies additional heritable risk factors from genome-wide meta-analysis data. **C. R. Gignoux et al.**	**167** A new inborn error of manganese metabolism caused by mutations in *SLC30A10*, a newly identified human manganese transporter. **K. Tuschl et al.**
12:30	**96** The Kaiser Permanente/UCSF Genetic Epidemiology Research Study on Adult Health and Aging: Demographic and behavioral influences on telomeres and relationship with all-cause mortality. **C. Schaefer et al.**	***105** Biallelic mutations of a ubiquitin-ligase-encoding gene cause an Ohdo-like intellectual disability syndrome. **B.B. Lina et al.**	**114** Over 250 novel associations with human morphological traits. **N. Eriksson et al.**	**123** Strong association of one carbon metabolism genes with stroke and change in post-methionine load homocysteine levels in the Framingham Heart and Vitamin Intervention for Stroke Prevention studies. **S. R. Williams et al.**	**132** A prospective natural history study of *DICER1*-related familial pleuropulmonary blastoma syndrome shows incomplete penetrance, pleiotropy and variable expressivity. **D. R. Stewart et al.**	**141** Risk of colorectal cancer for monoallelic and biallelic *MUTYH* mutation carriers. **A. K. Win et al.**	**150** Dynamics, definitions and discrepancies: Public perspectives on the systematic collection and use of family health history in routine health care. **H. Etchegary et al.**	**159** Deep exome sequencing of psoriasis identified new association signals contribute by INDELs, CNVs and rare SNPs. **X. Jin et al.**	**168** Combined methylmalonic acidemia and homocystinuria, cblC type: A prospective clinical protocol focusing on neurologic and neurodevelopmental parameters in a cohort of pre-school children diagnosed on expanded newborn screening. **J. D. Weisfeld-Adams et al.**

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	SESSION 38 – A Sequencing Jamboree: Exomes to Genomes Co-Moderators: Nancy Cox, Univ. of Chicago; and Orli Bahcall, *Nature Genetics*	**SESSION 39 – Admixture and Demography** Co-Moderators: Katarzyna Bryc, Harvard Univ.; and John Novembre, UCLA	**SESSION 40 – Analysis of Multilocus Systems** Co-Moderators: Brendan Keating, Univ. of Pennsylvania; and Laura Almasy, Texas Biomed. Res. Inst., San Antonio	**SESSION 41 – Genes Underlying Neurological Disease** Co-Moderators: Stylianos Antonarakis, Univ. of Geneva Med. Sch.; and Laurie Ozelius, Mount Sinai Med. Sch.	**SESSION 42 – Cancer Genetics III: Common Variants** Co-Moderators: Kathleen Cooney, Univ. of Michigan; and Meredith Yeager, NCI/NIH	**SESSION 43 – Genetics of Craniofacial and Musculoskeletal Disorders** Co-Moderators: Irini Manoli, NHGRI/NIH; and Siddharth Prakash, Univ. of Texas Hlth. Sci. Ctr. at Houston	**SESSION 44 – Tools for Phenotype Analysis** Co-Moderators: Donna Maglott, NCBI/NIH; and Gregory E. Crawford, Duke Univ.	**SESSION 45 – Therapy of Genetic Disorders** Co-Moderators: Cynthia J.R. Curry, UCSF; and Brunhilde Wirth, Univ. of Cologne, Germany	**SESSION 46 – Pharmacogenetics: From Discovery to Implementation** Co-Moderators: Toni Pollin, Univ. of Maryland Sch. of Med.; and Edward Ramos, NHGRI/NIH
4:30	**169** Whole-exome sequencing of 10,000 type 2 diabetes cases and controls from five major ancestry groups. **T. M. Teslovich et al.**	**178** Differential relatedness of African Americans to populations within West Africa. **K. Bryc et al.**	**187** Gene-based epistasis analysis in genome-wide association studies. **L. Ma et al.**	**196** *TMTC4*: A novel candidate gene for callosal development. **L. Fernandez et al.**	**205** Identification of 23 novel prostate cancer susceptibility loci using a custom array (the iCOGS) in an international consortium, PRACTICAL. **R. Eeles et al.**	**214** Next-generation sequencing detects mutations in *ISPD* as a common cause of Walker-Warburg syndrome with defective glycosylation of σ-dystroglycan. **T. Roscioli et al.**	**223** PRIMUS: Pedigree Reconstruction and Identification of the Maximum Unrelated Set. **J. Staples et al.**	**232** Pathogenic exon-trapping by SVA retrotransposon and rescue in Fukuyama muscular dystrophy. **M. Taniguchi et al.**	**241** Pharmacogenomics, ancestry and clinical decision making for global populations. **E. Ramos et al.**
4:45	**170** Rare and low frequency coding variants are associated with LDL cholesterol levels: Findings from the NHLBI Exome Sequencing Project. **L. A. Lange et al.**	**179** Fine scale population genetic structure of African Americans. **E. Y. Durand et al.**	**188** Building human phenotype networks from shared genetic risk variants. **C. Darabos et al.**	**197** *CLK2* missense mutation in a family with pontocerebellar hypoplasia type 7. **V. R. C. Eggens et al.**	**206** Large-scale genotyping identifies more than 40 novel breast cancer susceptibility loci. **K. Michailidou et al.**	**215** The identification of a novel gene identified by exome sequencing reveals the upstream components of the RAS/MAPK signaling pathway involved in Noonan syndrome. **H. Yntema et al.**	**224** Pharmacogenoinformatics: Novel approach of in silico drug designing based on genetic variation of *MDR1* gene involved in statin resistance. **A. Munshi et al.**	**233** Hematopoietic stem cell transplantation for adolescent and adult onset cerebral X-linked adrenoleukodystrophy. **T. Matsukawa et al.**	**242** Cell line profiling in oncology (CELLO) as a discovery platform for systematic identification of genetic and genomic biomarkers of drug sensitivity. **J. Zhong et al.**
5:00	**171** Exome sequencing of extreme phenotypes identifies *DCTN4* and *CAV2* as modifiers of chronic *Pseudomonas aeruginosa* infection in cystic fibrosis. **M. J. Emond et al.**	**180** A model-based approach for analysis of spatial structure in genetic data. **W. Yang et al.**	**189** Incorporating network dynamics to prioritize genes through genome-wide association studies. **L. Hou et al.**	**198** Missense mutations in *ITPR1* cause autosomal dominant congenital nonprogressive spinocerebellar ataxia. **J. Warman Chardon et al.**	**207** Genome-wide association study in *BRCA1* mutation carriers identifies novel loci associated with breast and ovarian cancer risk. **A. C. Antoniou et al.**	**216** *DYNC2H1* mutations are commonly found in Jeune asphyxating thoracic dysplasia (JATD) without extraskeletal features while *IFT140* mutations cause JATD with renal involvement. **M. Schmidts et al.**	**225** A general, integrated variant prioritization method for rapid determination of disease causing mutations from next-generation sequencing data. **B. D. O'Fallon et al.**	**234** Treating Pelizaeus-Merzbacher disease with clinically applicable compounds, curcumin and chloroquine: Preclinical studies. **K. Inoue et al.**	**243** Screening of the *TPMT* gene before thiopurine treatment results in a lower leucopenia occurrence in patient with inflammatory bowel disease. **M. J. H. Coenen et al.**
5:15	**172** A high resolution study of type 2 diabetes genetic architecture through whole-genome sequencing of 2850 European individuals: The GoT2D Study. **J. Flannick et al.**	**181** People of the British Isles: An analysis of the genetic contributions of European populations to a UK control population. **S. Leslie et al.**	**190** Large-scale multi-phenotype meta-analysis evaluates pleiotropic effects at *FADS1* and *GIPR* loci. **V. Lagou et al.**	**199** *Vps37A* causes a novel form of complex hereditary spastic paraparesis. **T. Falik-Zaccai et al.**	**208** Identification of the first locus to modify breast cancer risk specifically in *BRCA2* mutation carriers. **K. Kuchenbaecker et al.**	**217** Dominant missense mutations in *ABCC9* cause Cantú syndrome. **G. van Haaften et al.**	**226** Visually integrating and exploring high throughput phenome-wide association (PheWAS) results using PheWAS-view and PhenoGram. **S. A. Pendergrass et al.**	**235** Systemic L-threo-dihydroxyphenylserine corrects neurochemical abnormalities in a mouse model of Menkes disease. **S. Kaler et al.**	**244** PGRNseq: A new sequencing-based platform for high-throughput pharmacogenomic implementation and discovery. **A. S. Gordon et al.**
5:30	**173** Mapping quantitative traits with integrated whole exome/genome/array panel in individuals of European descent. **X. Sim et al.**	**182** The applicability of the Balding-Nichols model to a dataset of over 100,000 Brazilian individuals. **R. V. Rohlfs et al.**	**191** Building and assessing protein-protein interaction networks from genome-wide association results in cancer. **L. T. Hiraki et al.**	**200** Genome-wide association study identifies two novel susceptibility loci for musician's dystonia. **K. Lohmann et al.**	**209** Fine-scale mapping and functional analysis of the breast cancer 11q13 (CCND1) locus. **M. Ghoussaini et al.**	**218** Reduced dosage of ERF causes complex craniosynostosis in humans and mice, and links ERK1/2 signaling to regulation of osteogenesis. **S. R. F. Twigg et al.**	**227** PhenoDB: A new web-based tool for the collection, storage and analysis of phenotypic features. **A. Hamosh et al.**	**236** Response to VPA therapy in SMA patients is concordant from blood to neurons and influenced by CD36. **B. Wirth et al.**	**245** Genetic variation in the *GRK4* gene associates with susceptibility to hypertension and response to angiotensin receptor blockade. **M. White et al.**
5:45	**174** Whole genome sequence analyses describe the genetic architecture of complex traits: The Cohorts for Heart and Aging Research in Genetic Epidemiology Consortium. **A. C. Morrison et al.**	**183** Rare genetic variants in deep sequencing of neutral regions from a homogeneous population refine models of recent explosive human population growth. **A. Keinan et al.**	**192** A smoothed functional principal component analysis for pathway analysis with next-generation sequencing data. **J. Zhao et al.**	**201** Autosomal recessive axonal neuropathy with neuromyotonia: A novel disease entity caused by mutations in *HINT1*. **J. Baets et al.**	**210** Three independent loci within the TERT-CLPTM1L locus associated with telomere length and risk of breast and ovarian cancer. **G. Chenevix-Trench et al.**	**219** Mutations in the multidomain protein MEGF8 identify a new subtype of Carpenter syndrome associated with defective lateralization. **D. L. Lloyd et al.**	**228** A novel metabolomics analysis workflow provides new biological insights into the genetic basis of human metabolic variation. **H. Dharuri et al.**	**237** Melatonin, a new biomarker reflecting brain serotonin metabolism in individuals with phenylketonuria: Evaluation of large neutral amino acid therapy by a randomized, double-blind crossover study. **S. Yano et al.**	**246** Genome-wide discovery of drug-dependent human liver enhancers. **R. P. Smith et al.**
6:00	**175** Genome sequencing and analysis in autism spectrum disorder. **S. Walker et al.**	**184** Estimating human population sizes using the coalescent with recombination. **S. Sheehan et al.**	**193** Variants in exons and in transcription factors affect gene expression in trans. **A. Kreimer et al.**	**202** De novo gain of function *KCNT1* channel mutations cause seizures and developmental delay in malignant migrating partial seizures of infancy. **G. Barcia et al.**	**211** Statistical fine mapping of regions containing melanoma susceptibility genes identified through genome-wide association studies. **J. H. Barrett et al.**	**220** Increased frequency of *FBN1* variants in adolescent idiopathic scoliosis. **J. G. Buchan et al.**	**229** Integration of large-scale gene annotation, electronic medical records, and incidence data to produce phenotype-specific posterior probabilities to aid interpretation of genome-wide variant data. **I. M. Campbell et al.**	**238** Beyond cholesterol: Antioxidant treatment for patients with Smith-Lemli-Opitz syndrome. **E. Elias et al.**	**247** Genome-wide association study of vancomycin pharmacokinetics using a de-identified biorepository. **S. L. Van Driest et al.**
6:16	**176** Deep whole genome sequencing in pedigrees illuminates the contribution of low frequency and private mutations to the genetic architecture of metabolic quantitative traits. **A. K. Manning et al.**	**185** Reconstructing historical contributions to modern gene pools using the sequentially Markovian coalescent conditional sampling distribution. **A. Platt et al.**	**194** The continuation of theory by other means: ForSim as a forward simulator for improved understanding of the genetic architecture of complex traits and its evolution. **K. M. Weiss et al.**	**203** Investigating the genetic etiology of familial epilepsies using next-generation sequencing. **E. K. Ruzzo et al.**	**212** Combining expression phenotypes with high density imputation to identify melanoma risk genes. **M. H. Law et al.**	**221** Exome sequencing in idiopathic scoliosis reveals rare variants in *VANGL*, a planar cell polarity gene involved in axial development. **S. Sharma et al.**	**230** The Kaiser Permanente/ UCSF Genetic Epidemiology Research Study on Adult Health and Aging: CREX, computerized methodology to identify health conditions using the EMR for GWAS. **S. Sciortino et al.**	**239** Positive effects of short course androgen therapy on the neurodevelopmental outcome in boys with 47, XXY syndrome at 36 and 72 months of age. **C. Samango-Sprouse et al.**	**248** Integrating multiple levels of phenotypic information to map genetic determinants of glucocorticoid sensitivity. **J. Maranville et al.**
6:30	**177** Whole genome sequencing of 2100 individuals in the founder Sardinian population. **C. Sidore et al.**	**186** On the Sardinian ancestry of the Tyrolean Iceman. **M. Sikora et al.**	**195** Incorporating phylogenetic conservation and pedigree information in tests of rare-variant association. **H. Hu et al.**	**204** Autoregulation of the DYT6-gene *THAP1*. **A. Erogullari et al.**	**213** Meta-analysis identifies four new loci for testicular germ cell tumor. **C. C. Chung et al.**	***222** Recessive mutations in *FKBP10*, a PPIase known to cause type XI OI, extend the phenotype to a congenital contracture syndrome (Kuskokwim disease), and cause diminished collagen cross-linking in matrix. **J. Marini et al.**	**231** An informatics approach to analyzing the incidentalome. **M. C. Adams et al.**	**240** A mechanism and treatment strategy for pregnancy-associated aortic dissection in Marfan syndrome. **J. P. Habashi et al.**	***249** Common and rare genetic variation in maturity-onset diabetes of the young genes influence response to interventions for diabetes prevention. **K. Jablonski et al.**

Friday, November 9: Concurrent Platform (abstract-driven) Session D (47-55)

8:00 AM–10:15 AM

	Hall D, Lower Level North	Gateway Ballroom 103, Lower Level South	Room 135, Lower Level North	Room 134, Lower Level North	Gateway Ballroom 104, Lower Level South	Room 124, Lower Level North	Room 132, Lower Level North	Room 130, Lower Level North	Room 123, Lower Level North
	SESSION 47 – Structural and Regulatory Genomic Variation Co-Moderators: Mike Lovett, Washington Univ. in St. Louis; and Greg Elgar, MRC NIMR, London, U.K.	**SESSION 48 – Neuropsychiatric Disorders** Co-Moderators: Dimitrios Avramopoulos, Johns Hopkins Univ.; and Tatiana Foroud, Indiana Univ. Sch. of Med.	**SESSION 49 – Common Variants, Rare Variants, and Everything in-Between** Co-Moderators: Steve Rich, Univ. of Virginia; and Rasika Mathias, Johns Hopkins Univ. Sch. of Med.	**SESSION 50 – Population Genetics Genome-Wide** Co-Moderators: Melissa A. Wilson Sayres, Univ. of California, Berkeley; and Sarah Tishkoff, Univ. of Pennsylvania	**SESSION 51 – Endless Forms Most Beautiful: Variant Discovery in Genomic Data** Co-Moderators: Terry Furey, Univ. of North Carolina at Chapel Hill; and Deanna Church, NCBI/NIH	**SESSION 52 – Clinical Genetics: Complex Mechanisms and Exome-Discovery** Co-Moderators: Michael Gambello, Emory Univ. ; and Antonie D. Kline, Harvey Inst. for Human Genet., Baltimore	**SESSION 53 – From SNP to Function in Complex Traits** Co-Moderators: Praveen Sethupathy, Univ. of North Carolina at Chapel Hill; and Aravinda Chakravarti, McKusick-Nathans Inst. of Genet. Med., Baltimore	**SESSION 54 – Genetic Counseling and Clinical Testing** Co-Moderators: Susan Hahn, Univ. of Miami, Hussman Inst. for Human Genomics; and Andrew Faucett, Geisinger Hlth. Syst., Danville, PA	**SESSION 55 – Mitochondrial Disorders and Ciliopathies** Co-Moderators: Catherine E. Keegan, Univ. of Michigan; and Mitzi Murray, Univ. of Washington
8:00	**250** Germline mosaicism does not explain the maternal age effect on trisomy. **R. Rowsey et al.**	**259** Genetic and functional abnormalities of the melatonin biosynthesis pathway in patients with bipolar disorder. **S. Jamain et al.**	**268** Minimal differences in single nucleotide variation calls between blood- and cell line-derived DNA from the same individuals. **C. M. Schafer et al.**	**277** Direct measure of human somatic base-substitution mutation rate in monozygotic twins. **J. B. Richards et al.**	**286** Dark matter of the diseasome: Annotating personal genomes for gene regulatory disease risk alleles. **G. Bejerano.**	**295** Systematic identification of causal mutations in Mendelian disorders using exome sequence data. **M. Lek et al.**	**304** The type 2 diabetes risk allele of rs11603334 increases *ARAP1* promoter activity and is associated with increased *ARAP1* mRNA in pancreatic islets. **J. R. Kulzer et al.**	**313** Utilization of chromosomal microarrays in pediatrics. **A. H. Seeley et al.**	**322** Combination of modern and traditional techniques identify MCKD1 causal frameshift variants within the *MUC1* VNTR. **A. Kirby et al.**
8:15	**251** Female meiosis II errors prevalence and their impact on human embryo viability. **A. Kuliev et al.**	**260** Massively-parallel sequencing of the brain transcriptome reveals differential expression of novel genes in bipolar disorder. **N. Akula et al.**	**269** The impact of genetic variation on diabetes-related quantitative traits from whole exome sequences: The T2D-GENES Consortium. **H. M. Highland et al.**	**278** Estimating human mutation rate using autozygosity in a founder population. **C. D. Campbell et al.**	**287** Causal mutation discovery using next-generation sequencing data: Development and application of a pipeline to reduce false positive calls and to map regions of shared homozygosity and IBD. **S. Gulsuner et al.**	**296** Exome sequencing of a large cohort of patients with congenital digestive system disorders. **M. Yourshaw et al.**	**305** *NOS1AP* is the major genetic electrocardiographic QT-interval regulator. **A. Kapoor et al.**	**314** Maximizing detection and minimizing noise: The first report of large scale whole exome sequencing data interpretation in a clinical laboratory. **F. Xia et al.**	**323** ARL13B, INPP5E, PDE6D and CEP164 form a functional network involved in Joubert syndrome and nephronophthisis. **S. Seo et al.**
8:30	**252** A population isolate reveals enriched recessive deleterious variants underlying neurodevelopmental traits. **O. Pietiläinen et al.**	**261** Rare and common gain-of-function alleles of the serotonin transporter gene, *SLC6A4*, associated with Tourette disorder. **P. R. Moya et al.**	**270** Whole-exome sequencing in multiplex families identifies novel rare variants in multiple sclerosis. **A. H. Beecham et al.**	**279** The myth of random mating: Evidence of ancestry-related assortative mating across 3 generations in Framingham, MA. **R. Sebro et al.**	**288** A new framework for large-scale genomic variant discovery and validation using pooled sequencing data. **G. del Angel et al.**	**297** Novel defect in kinetochore assembly causes short stature and microcephaly of postnatal onset. **C. Y. Hung et al.**	**306** A regulatory polymorphism in Csk, a Lyp binding partner, associates with systemic lupus erythematosus and affects B cell signaling, maturation and activation. **N. Manjarrez-Orduño et al.**	**315** Efficient detection of causative mutations for rare diseases: Rethinking clinical practice. **H. Lee et al.**	**324** Mainzer-Saldino syndrome is a ciliopathy caused by mutations in the *IFT140* gene. **I. Perrault et al.**
8:45	**253** The role of trans-acting factors on recombination in oocytes with nondisjoined chromosomes 21. **C. D. Middlebrooks et al.**	**262** *GLRB* is the third major gene-of-effect in hyperekplexia or startle disease. **S. K. Chung et al.**	**271** A Mendelian randomization study on vitamin D status and blood pressure: A meta-analysis in up to 89,042 individuals. **K. S. Vimaleswaran et al.**	**280** Combined analysis of loss-of-function variants in protein-coding genes from over 16,000 individuals. **D. G. MacArthur et al.**	**289** Discovery of genomic variants from RNA-sequencing data. **R. Piskol et al.**	**298** Mutations in *PIGO*, a member of the GPI anchor synthesis pathway, cause hyperphosphatasia with mental retardation syndrome. **P. M. Krawitz et al.**	**307** *ITGAM* coding variant, rs1143679 (R77H) that is associated with systemic lupus erythematosus (SLE) susceptibility affects its own expression in monocytes and ligand binding activities in SLE patients. **A. K. Maiti et al.**	**316** Intentions to receive individual results from whole-genome sequencing among participants in the ClinSeq™ study. **B. B. Biesecker et al.**	**325** Mutations in *ALDH1B1*, which encodes a mitochondrial protein belonging to the aldehyde dehydrogenase family, result in hepatic failure and mitochondrial respiratory chain deficiency. **S. Salhi et al.**
9:00	**254** Large-scale function-based enhancer discovery. **D. E. Dickel et al.**	**263** Functional analysis of rare chimeric genes in schizophrenia. **C. Rippey et al.**	**272** APOE modulates the relationship among triglycerides, cholesterol, and CHD through pleiotropy and gene-gene interactions. **T. J. Maxwell et al.**	**281** Abundant selection explains low diversity on human Y chromosomes. **M. Wilson Sayres et al.**	**290** zCall: A rare variant caller for array-based genotyping. **J. I. Goldstein et al.**	**299** The 600 kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. **S. Jacquemont et al.**	**308** Loss-of-function of semaphorins 3C and 3D in Hirschsprung disease. **Q. Jiang et al.**	**317** Changes to control perceptions following disclosure of *APOE*-coronary artery disease associations during genetic susceptibility testing for Alzheimer's disease: Findings from the REVEAL Study. **K. Christensen et al.**	**326** Targeted exome sequencing of 102 patients with clinical evidence of mitochondrial disease. **D. S. Lieber et al.**
9:15	**255** A single enhancer on human chromosome 11 directly controls >1,000 promoters and distal regulatory elements genome-wide. **J. A. Stamatoyannopoulos et al.**	**264** Excess homozygosity in the major histocompatibility complex in schizophrenia. **S. Mukherjee et al.**	**273** Statistical inference of tissue-consistent and tissue-specific eQTLs. **T. Flutre et al.**	**282** The genomic geography of close relatives across Europe. **P. Ralph et al.**	**291** Copy number detection and variant classification in the DDD project. **T. W. Fitzgerald et al.**	**300** Ras/MAPK dysregulation caused by *MEK2* haploinsufficiency: A novel mechanism for a RASopathy phenotype. **M. J. M. Nowaczyk et al.**	**309** Functional assessment of human coding polymorphisms affecting skin pigmentation using zebrafish. **Z. Tsetskhladze et al.**	**318** Decreased prediction ability of common genetic variants on breast cancer risk with age: Possible underlying models and impact on risk prediction. **H. Aschard et al.**	**327** Genetic diagnosis of mitochondrial disorders by whole-exome sequencing. **C. J. Carroll et al.**
9:30	**256** Identification of trait- and disease-relevant genetic polymorphisms in microRNA target sites. **S. Busche et al.**	**265** Significant risk of new mutations for Huntington disease: CAG-size specific risk estimates of intermediate allele repeat instability. **A. Semaka et al.**	**274** Estimates of penetrance for common pathogenic copy number variations. **J. A. Rosenfeld et al.**	**283** Evolutionary history and adaptation inferred from whole-genome sequences of diverse African hunter-gatherers. **J. Lachance et al.**	**292** Removal of mapping biases in sequence-based functional data improves regulatory element identification at heterozygous variants. **M. Buchkovich et al.**	**301** Analysis of ESP5400 exomes for results of clinical utility in genes for conditions tested as part of newborn screening programs and age-related macular degeneration. **H. K. Tabor et al.**	**310** Dosage effects of 169 Chr21 genes on early development events in zebrafish. **S. Edie et al.**	**319** Large-sample size, comprehensive catalog of variants and advanced machine learning technique boost risk prediction for inflammatory bowel disease. **Z. Wei et al.**	**328** Constitutive activation of STIM1 causes tubular aggregate myopathy. **J. Laporte et al.**
9:45	**257** Mapping functional p53 response elements and their variants in human genome. **X. Wang et al.**	**266** Mutations in *AKT3* lead to hemimegalencephaly. **A. Poduri et al.**	**275** Combining Illumina gene expression microarrays from different tissues: Methodological aspects. **K. Heim et al.**	**284** Mapping the human genome's missing pieces using population admixture. **G. Genovese et al.**	**293** SNP discovery in diverse human populations by rapid, very-low-cost next-generation sequencing of reduced representation libraries. **T. F. Cooke et al.**	**302** High congenital malformation rates in a Chornobyl ionizing radiation impacted population isolate in Ukraine. **W. Wertelecki et al.**	**311** Two birds, one stone: Epistasis profiling of many single-nucleotide variants in a human gene. **O. Zill et al.**	**320** A comparison of risk estimates for complex diseases: Navigenics SNP-based testing and family history assessment. **L. Aiyar et al.**	**329** Mutation in *PNPT1* gene, which encodes a mitochondrial polyribonucleotide nucleotidyl-transferase, causes encephalopathy with choreo-athetotic movements. **V. Vedrenne et al.**
10:00	**258** A SNP associated with skin cancer and pigmentation disrupts a melanocyte enhancer in an intron of *IRF4*. **D. U. Gorkin et al.**	**267** De novo somatic mutations in components of the PI3K-AKT3-mTOR pathway cause hemimegalencephaly. **J. Lee et al.**	**276** A DNA variant caller adapted to assess mitochondrial DNA variation in lymphocytes from 1,000 Sardinians. **J. Ding et al.**	**285** When ancestry runs deep: Trans-species polymorphisms in apes. **L. Segurel et al.**	**294** HIBAG -- HLA genotype imputation with attribute bagging. **X. Zheng et al.**	**303** Somatic mosaicism is responsible for congenital melanocytic naevus syndrome, and underpins the associated risk of melanoma. **V. A. Kinsler et al.**	**312** Discovery and replication of pathway-based trans-eQTL associations. **L. Wiley et al.**	**321** Web-based case conferencing: An effective source of cancer genetics training for community-based clinicians. **K. Blazer et al.**	**330** Comprehensive analysis of 101 nuclear genes for molecular diagnosis of mitochondrial disorders. **R. Bai et al.**

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	SESSION 59 – Genome Structure and Variation Co-Moderators: John Moran, Univ. of Michigan Med. Sch.; and Ryan Mills, Univ. of Michigan Med. Sch.	**SESSION 60 – Advances in Neurodegenerative Disease** Co-Moderators: Jeff Vance, Univ. of Miami; and Tricia Thornton-Wells, Vanderbilt Univ.	**SESSION 61 – Missing Heritability, Interactions and Sequencing** Co-Moderators: Dana Crawford, Vanderbilt Univ.; and Alkes Price, Harvard Sch. of Publ. Hlth.	**SESSION 62 – Exome Sequencing Uncovers Etiology of Mendelian Disease** Co-Moderators: Cheryl Maslen, Oregon Hlth. & Sci. Univ.; and Jun Z. Li, Univ of Michigan	**SESSION 63 – Transcriptional Regulation, Variation and Complexity** Co-Moderators: Barbara Stranger, Brigham and Women's Hosp., Harvard Med. Sch; and Ross Hardison, Penn State	**SESSION 64 – Epigenetics** Co-Moderators: Bernd Wollnik, Univ of Cologne, Germany; and Beth A. Sullivan, Duke Univ.	**SESSION 65 – Advances in Ocular Genetics** Co-Moderators: Erica Davis, Duke Univ. Med. Ctr; and Arupa Ganguly, Univ. of Pennsylvania	**SESSION 66 – Cancer Genetics: Somatic Variants** Co-Moderators: Charis Eng, Cleveland Clinic; and Jennelle Hodge, Mayo Clin.	**SESSION 67 – Developmental Insights into Human Malformations** Co-Moderators: Lucy Osborne, Univ. of Toronto, Canada; and Rhona Schreck, Cedars-Sinai Med Ctr.
4:30	**331** A map of human genetic variation: Update from the 1000 Genomes Project. **F. Yu.**	**340** A genome-wide association study for cerebrospinal fluid tau and amyloid beta 42 identify new candidate variants implicated in Alzheimer's disease. **J. S. K. Kauwe et al.**	**349** Empirical and theoretical studies on genetic variance of rare variants for complex traits using whole genome sequencing in the CHARGE Consortium. **C. Zhu et al.**	**358** Loss of function mutations in known human disease genes in 572 exomes. **J. Johnston et al.**	**367** The complete GENCODE human annotation: New insights into the functionality of transcriptional complexity. **J. M. Mudge et al.**	**376** The epitranscriptome reveals novel mechanisms of RNA regulation and spatiotemporal dynamics. **C. E. Mason et al.**	**385** Rare insertion polymorphisms identified by exome sequencing may be associated with age-related macular degeneration. **L. Farrer et al.**	**394** The genomic landscape of childhood pre-B acute lymphoblastic leukemia. **J. Spinella et al.**	**403** *TECTONIC3* mutations cause orofaciodigital syndrome type IV (Mohr-Majewski). **S. Thomas et al.**
4:45	**332** Towards a whole genome map of heritable copy number variation. **S. Aradhya et al.**	**341** Analysis of whole transcriptome specific to the temporal pole of late-onset Alzheimer's disease. **C. E. Humphries et al.**	**350** Leveraging admixture analysis to resolve missing and cross-population heritability in GWAS. **N. Zaitlen et al.**	**359** The problem of multiple plausible molecular diagnoses in next-generation sequencing data: The NIH Undiagnosed Diseases Program experience. **D. Adams et al.**	**368** Genetic analyses in the Genotype-Tissue Expression (GTEx) project. **K. Ardlie et al.**	**377** Epigenome-wide profiling of circulating DNA in colorectal cancer. **R. Cortese et al.**	**386** The role of *SIX6* in primary open-angle glaucoma. **M. Ulmer et al.**	**395** Genomic analysis of serial chronic lymphocytic leukemia samples suggests that epigenetic changes, rather than clonal evolution, drive progression of disease. **E. N. Smith et al.**	**404** Abnormal development of NG2+PDGFRα+ neural progenitor cells causes neonatal hydrocephalus in a ciliopathy mouse model. **C. S. Carter et al.**
5:00	**333** Charting the population-scale landscape of short tandem repeat variation in humans. **M. Gymrek et al.**	**342** Rare variants from high-density exome genotyping in late-onset Alzheimer's disease: Update from Alzheimer's Disease Genetics Consortium. **L.-S. Wang et al.**	**351** Applying a quantitative genetics test of evolutionary neutrality to finger ridge-count, a classical model trait in humans. **S. E. Medland et al.**	**360** Exome sequencing to identify the cause of Mendelian diseases. **J. Lupski et al.**	**369** Characterizing the genetic basis of transcriptome diversity in a large RNA sequencing study. **A. Battle et al.**	**378** Alterations in genomically imprinted miRNA and snoRNA clusters in a mouse model of fetal alcohol spectrum disorders. **B. I. Laufer et al.**	**387** Topical ocular sodium 4-phenylbutyrate rescues glaucoma in a mouse model of primary open angle glaucoma. **G. S. Zode et al.**	**396** Whole-genome sequencing of liver cancers identifies etiological influences on mutation patterns and recurrent mutations in chromatin regulators. **A. Fujimoto et al.**	**405** Malformation of the brain cortex, as the only expression of a ciliopathy, results from mutation in human *Rotatin*. **G. M. S. Mancini et al.**
5:15	**334** Whole-genome sequencing analysis of iPSC lines uncovers lineage-manifested CNVs. **A. E. Urban et al.**	**343** Common variants in *ABCA7* and *GRIN3B, HMHA1* and *SBNO2*, are associated with late-onset Alzheimer's disease in African Americans. **C. Reitz et al.**	**352** Does common variation contribute to the shared genetic basis for schizophrenia and autism? **P. H. Lee et al.**	**361** Domain-specific mutations in *CDKN1C* cause two disorders with opposing phenotypes: The undergrowth disorder IMAGe syndrome or the overgrowth disorder Beckwith-Wiedemann syndrome. **V. Arboleda et al.**	**370** Genetic and molecular basis of RNA-DNA sequence differences in humans. **V. G. Cheung et al.**	**379** *KDM6A* escapes X inactivation and controls expression of reproduction-related homeobox genes in female ES cells and ovary: Deficiency may explain embryonic and ovarian failure in Turner. **C. M. Disteche et al.**	**388** Meta-analysis of GWAS on corneal thickness identifies a total of 27 associated loci, including six risk loci for eye disease keratoconus. **S. Macgregor.**	**397** Breast cancer evolution revealed by deep whole-genome sequencing of early neoplasias and their concurrent carcinomas. **A. Sidow et al.**	**406** Whole exome resequencing identifies mutations in *LRRC6* as a novel single-gene cause of primary ciliary dyskinesia. **M. Chaki et al.**
5:30	**335** SNP markers identify areas with restricted recombination suggesting structural variation across the human genome is widespread. **P. G. Hysi et al.**	**344** Genome-wide association analyses of onset age in late-onset Alzheimer disease demonstrate no strong effect outside of the APOE region. **A. C. Naj et al.**	**353** Ultrafast genome-wide interaction scan on case-control data implicates epistatic calcium channels in bipolar disorder. **S. Prabhu et al.**	**362** SCID newborn screening and exome sequencing identifies ataxia telangiectasia and low T cells early in life. **J. M. Mallott et al.**	**371** Characterizing gene expression variation across seven diverse human populations. **A. R. Martin et al.**	**380** Genome-wide scan of DNA methylation in the aging brain and its relation to Alzheimer's disease. **P. L. De Jager et al.**	**389** Mouse models reveal an essential role for RERE in eye development. **B. Kim et al.**	**398** Intra-tumor genetic heterogeneity in cancer tissues: The key to assessing its significance is the distribution profile of gene variants not just their presence in tumors. **B. Gottlieb et al.**	**407** Temporally and spatially resolved catalogues of in vivo forebrain enhancers. **A. S. Nord et al.**
5:45	**336** Mapping the L1 interactome reveals RISC-associated helicase MOV10 as a potent inhibitor of retrotransposition. **J. Goodier et al.**	**345** Identification by exome analysis of the molecular bases of familial idiopathic basal ganglia calcification not related to *SLC20A2* mutation. **G. Nicolas et al.**	**354** Computational challenges in the analysis of low coverage sequence data in thousands of individuals. **Y. Luo et al.**	**363** Identification of a new melanocyte differentiation gene underlying human autosomal recessive albinism. **K. Grønskov et al.**	**372** Comparative eQTL analyses within and between seven tissue types suggest mechanisms underlying cell type specificity of eQTLs. **B. Engelhardt et al.**	**381** RNA-mediated transcriptional silencing in Friedreich ataxia. **Y. K. Chutake et al.**	**390** Mutations in the nuclear NAD synthesizing enzyme NMNAT1 cause autosomal recessive Leber congenital amaurosis with early-onset severe macular atrophy and optic atrophy. **J. Rozet et al.**	**399** Next -generation sequencing and chromosomal microarray analysis provide novel insight into the genomic landscape of metastatic breast cancer. **M. Li et al.**	**408** SRY regulation of the *RET* gene suggests a potential role of the Y-chromosome gene in sexual dimorphism in Hirschsprung disease. **Y. Li et al.**
6:00	**337** FoSTeS/MMBIR replicative repair mechanisms are error prone: High frequency of nucleotide variation at the breakpoint junctions. **C. M. B. Carvalho et al.**	**346** Mutations in DNAJ cause autosomal dominant Parkinson disease in the Mennonite community. **C. Vilarino-Guell et al.**	**355** Sparse sequencing of 6,000 cases and 6,000 controls from Chinese women for genome-wide association study of major depression. **X. Gan et al.**	**364** Exome sequencing results in 230 patients with severe developmental disorders in the DDD project. **M. van Kogelenberg et al.**	**373** Identification of novel genetic determinants of induced innate immune responses and context specific eQTL in human primary monocytes. **B. P. Fairfax et al.**	**382** P53 regulates 5-hydroxymethylcytosine-mediated epigenetic landscape through *GADD45A*. **Y. Li et al.**	**391** RNA-DNA differences in miRNA transcriptome of retina and retinoblastoma. **A. Ganguly et al.**	**400** The 3D topographic mapping of genetic variations in treatment of naïve advanced ovarian cancer. **E. Cuppen et al.**	**409** *MAP3K1* mutations in 46,XY DGDs alter crosstalk in downstream signal transduction pathways to cause abnormal human gonadal development. **J. Loke et al.**
6:16	**338** Telomere position effect in patients with subtelomeric deletions. **J. Gerfen et al.**	**347** *C9ORF72* repeat expansion is a risk factor for Parkinson disease. **K. Nuytemans et al.**	**356** Deep targeted sequencing of 12 breast cancer loci in 4,700 women across four different ethnicities. **P. Kraft et al.**	**365** Genetic etiology of isolated congenital asplenia. **A. Bolze et al.**	**374** Gene-level and exon-level expression QTL signals in the UK Brain Expression Consortium dataset. **M. E. Weale et al.**	**383** Maps of open chromatin highlight cell type-specific patterns of regulatory sequence variation at hematological trait loci. **C. A. Albers et al.**	**392** Knock-in of human *KIAA0649P* into the mouse *Rb1* locus: Modeling the mechanism of imprinted *RB1* expression in humans. **L. Steenpass et al.**	**401** Transcriptome sequence analysis of human colorectal cancer samples to reveal functional attributes. **H. Ongen et al.**	**410** Soft tissue aspects of the Williams-Beuren syndrome facial phenotype can be attributed to *GTF2IRD1*. **S. J. Palmer et al.**
6:30	**339** De novo CNV formation in mouse embryonic stem cells occurs in the absence of Xrcc4-dependent nonhomologous end joining. **M. F. Arlt et al.**	***348** Age-dependent penetrance of ALS+/-FTD due to *C9orf72* hexanucleotide intronic repeat expansion mutations. **C. Lewis et al.**	**357** Population stratification of human disease-associated SNPs, and their relevance to human disease networks. **S. M. Raj et al.**	**366** Whole genome sequencing in two brothers with heterotaxy reveals *BCL9L* as a novel gene associated with autosomal recessive heterotaxy (HTX6). **C. J. Saunders et al.**	**375** First complete haplotype of the human immunoglobulin heavy chain locus from a single individual and characterization of novel allelic and structural variation. **K. Meltz Steinberg et al.**	**384** Functional epialleles at an endogenous human centromere. **B. A. Sullivan et al.**	**393** Gene therapy provides long-term visual function in a pre-clinical model of retinitis pigmentosa. **K. J. Wert et al.**	**402** Regulatory regions are somatic mutation cold spots in cancer genomes. **S. Sunyaev et al.**	**411** Notch gain of function inhibits chondrocyte differentiation via Rbpj-dependent suppression. **S. Chen et al.**

* Presenting Author Change

Saturday, November 10: Concurrent Invited Session III (73-80)

9:40 AM–11:40 AM

Gateway Ballroom 103, Lower Level South	Hall D, Lower Level North	Gateway Ballroom 104, Lower Level South	Room 134, Lower Level North	Room 132, Lower Level North	Room 135, Lower Level North	Room 124, Lower Level North	
SESSION 73 – Returning Results from Large-Scale Sequencing: Where the Rubber Meets the Road Co-Moderators: Leslie G. Biesecker, NHGRI/NIH; and Robert C. Green, Brigham and Women's Hosp.	**SESSION 74 – Genomic Approaches to Mendelian Disorders** Co-Moderators: Jay Shendure, Univ. of Washington; and David Valle, Johns Hopkins Univ. Sch. of Med. 09:40 am Genomic approaches to Mendelian disorders. **D. Valle.**	**SESSION 75 – Emerging Applications of Identity by Descent Segment Detection** Co-Moderators: Sharon R. Browning, Univ. of Washington; and Brian L. Browning, Univ. of Washington	**SESSION 76 – The Functional Consequences of microRNA Dysregulation in Human Disease** Co-Moderators: Cheryl L. Thompson, Case Western Reserve Univ.; and Ahmad Khalil, Case Western Reserve Univ.	**SESSION 77 – Centralizing the Deposition and Curation of Human Mutations** Co-Moderators: Robert L. Nussbaum, UCSF; and David H. Ledbetter, Geisinger Hlth. Syst., Danville, PA	**SESSION 78 – Stem Cells and Personalized Medicine** Moderator: Stephen H. Tsang, Columbia Univ.	**SESSION 79 – Should Noninvasive Prenatal Diagnosis Augment or Replace Current Prenatal Screening and Diagnosis?** Co-Moderators: Mark E. Nunes, Kaiser Permanente, San Diego; and Mildred K. Cho, Stanford Univ.	**SESSION 80 – Selection Signatures and the Genetics of Autoimmunity and Infectious Diseases** Co-Moderators: Judy H. Cho, Yale Univ.; and Richard A. Spritz, Univ. of Colorado Denver Anschutz Med. Campus
09:40 am ClinSeq: A pilot study of large-scale medical sequencing in research and implications for clinical genomic medicine. **L. G. Biesecker.**	09:55 am FORGE Canada: A nation-wide effort to understand the genomics of childhood disorders. **K. Boycott.**	09:40 am Sharing by descent, phasing, rare variants and population structure. **A. Kong.**	09:40 am OncomiR-1 in cancer and development: A tale of mice and men. **A. Ventura.**	09:40 am Improving the accuracy of variant identification. **D. Church.**	09:40 am Stem cells and personalized medicine in retinal degenerations. **S. H. Tsang.**	09:40 am Lessons from the clinical introduction of noninvasive prenatal diagnosis: How we got here. **A. T. Bombard.**	09:40 am The genetics of autoimmunity. **J. H. Cho.**
10:10 am Expert concordance and discordance for return of incidental findings from whole genome sequencing. **R. C. Green.**	10:10 am Current challenges in exome or genome-based analysis of Mendelian disorders. **J. Shendure.**	10:10 am Length distributions of identity by descent reveal fine-scale demographic history. **I. Pe'er.**	10:10 am microRNA reprogramming in cancer: Mechanisms and consequences. **J. Mendell.**	10:10 am The ISCA Consortium: Standardization and sharing of structural variation data. **C. L. Martin.**	10:10 am Direct reprogramming to generate patient-specific stem cells and neurons. **M. Wernig.**	10:05 am Cell-free fetal DNA in prenatal diagnosis: Where we are going? **D. Bianchi.**	09.55 am The genetics of autoimmunity. **R. A. Spritz.**
10:40 am Using next-generation sequencing for carrier testing for severe childhood recessive diseases. **S. F. Kingsmore.**	10:40 am Lessons from 500 diagnostic exomes. **H. G. Brunner.**	10:40 am Identity by descent within and between pedigrees. **E. A. Thompson.**	10:40 am Exploring circulating miRNAs as blood-based diagnostic biomarkers. **M. Shapero.**	10:40 am Introducing ClinVar. **D. Maglott.**	10:40 am A chemical approach to controlling cell fate. **S. Ding.**	10:30 am Academia and industry in the development of noninvasive prenatal diagnosis. **M. K. Cho.**	10:10 am Selection signatures and mechanisms of host-microbe interactions. **P. Sabeti.**
11:10 am Diagnostic implementation of exome sequencing: Results from 500 patients. **J. Veltman.**	11:10 am Genes, genomes and the future of medicine. **R. Lifton.**	11:00 am Using high resolution identity by descent: From detecting selection to explaining trait variability. **M. Abney.**	11:10 am Circulating microRNAs in obesity and postmenopausal breast cancer. **C. L. Thompson.**	11:10 am Community involvement in centralized mutation curation. **H. L. Rehm.**	11:10 am Patient-specific stem cells and cardiovascular genetics. **B. Conklin.**	10:55 am Ethical and policy implications of early noninvasive prenatal diagnosis. **J. S. King.**	10:40 am Interactions of HLA class I with killer-cell immunoglobulin-like receptors: Influences on human disease. **P. Parham.**
		11:20 am Extending the limits of IBD segment detection with sequence data and new statistical methods. **B. L. Browning.**				11:20 am Discussion. **M. E. Nunes.**	11:10 am Toward a genetic theory of infectious diseases. **J.-L. Casanova.**

During the meeting, attendees are encouraged to post thoughts on exciting scientific or clinical advances heard at a session or workshop and on challenges that the field will face by using **hashtag #ASHG2012** or by posting on the ASHG Facebook page. The posts will be monitored and may be discussed at the Closing Symposium on Saturday.

Twitter: **@Genetics Society,** #ASHG2012

Facebook: **https://www.facebook.com/GeneticsSociety**

Follow ASHG on Twitter before and during the week of the 2012 Meeting to get the latest updates, tips, news, and announcements.

RECORDED PRESENTATIONS AVAILABLE AFTER THE MEETING
Invited Sessions, Award Presentations, and the Plenary Sessions will be recorded and posted on the Web site after the meeting.

These recordings are free to meeting registrants.
Visit the ASHG Web site in early December to view these presentations.

12:00 NOON–1:00 PM

SESSION 81 – Closing Plenary: Human Genetics 2012 and Beyond: Present Progress and Future Frontiers

Hall D, Lower Level North, Moscone Center

Moderator: Joel N. Hirschhorn, 2012 Program Committee Chair
Boston Children's Hosp., Harvard Med. Sch. and Broad Inst.
Presenter: Chris Gunter, HudsonAlpha Inst. for Biotechnol.

Panelists:
Han Brunner, Radboud Univ. Nijmegen
Jay Shendure, Univ. of Washington
Dian Donnai, Univ. of Manchester
Lynn Jorde, Univ. of Utah
Hal Dietz, Johns Hopkins Univ.

An outstanding panel of expert human geneticists with varying perspectives will make will make brief presentations and then participate in a wide-ranging discussion on the most exciting advances and important upcoming challenges in their areas of human genetics.Topics will be driven by questions from the panelists and the audience, but will include many of the following perspectives:

- The importance of education for the public, scientists, and clinicians
- The impact of new technology on human genetics and genomics
- Advances that define biological mechanisms
- Challenges of interpretation of exome and genome sequencing
- Translation of advances into clinical care

During the course of the 62nd Annual Meeting, registrants are encouraged to post their thoughts on scientific or clinical advances they have heard about at the meeting, and on upcoming important challenges in human genetics, via ASHG's social media outlets (Twitter, Facebook).

Twitter: use hashtag #ASHG2012

Facebook: https://www.facebook.com/GeneticsSociety

You can also address your comments through Twitter directly to Chris Gunter, @girlscientist, during the meeting.

Posts by meeting participants will be followed and summarized at the beginning of the session by Chris Gunter, 2012 Program Committee Member.

After the brief presentations by the panelists, there will be opportunities for audience members to ask questions of one or more panel members.

At the conclusion of the session, the moderator will provide a brief summary, and the meeting will be adjourned by the 2012 President, Mary-Claire King

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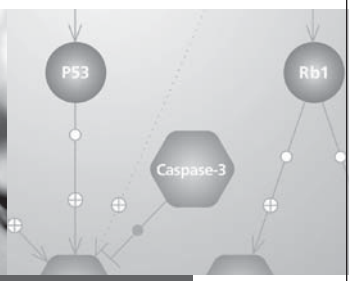

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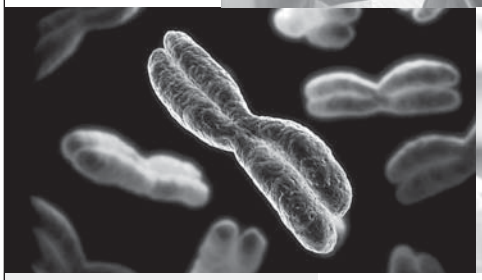

Visit booths #918/920 and #925/927.

Join our luncheon seminars:

Wednesday, Nov. 7, 12:45 – 2:15 PM
Room 302, Esplanade Level (South), Moscone Center
Enabling the Genome Generation #1: population-optimized strategies and genotyping solutions for expanding our understanding of the genetic variations in complex diseases

Thursday, Nov. 8, 12:45 – 2:15 PM
Room 307, Esplanade Level (South), Moscone Center
Enabling the Genome Generation #2: next-generation cytogenetics solution for constitutional and cancer research applications

Box lunch will be provided. First come, first served.