



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

YOUR GUIDE TO THE ASHG 2012 MOBILE APP



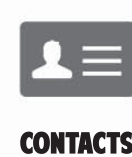

STEP 1:
DOWNLOAD THE APP - ASHG 2012
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STEP 2:
INSTALL AND ACCEPT PERMISSIONS

STEP 3:
BEGIN THE APP EXPERIENCE



DISCOVER ASHG 2012 IN A NEW WAY!



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. <i>AJHG</i> 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. Hallgrímsson, 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-Benkalha, 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.

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
<p>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.</p>	<p>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</p>	<p>SESSION 05 – Gene Regulatory Change: The Engine of Human Evolution? Co-Moderators: James P. Noonan, Yale Univ. Sch. of Med.; and Nadav Ahituv, UCSF</p>	<p>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.</p>	<p>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</p>	<p>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 Françoise Clerget-Darpoux, INSERM U781, Univ. Paris V, France.</p>	<p>SESSION 09 – Surveying Customer Responses to Personal Genetic Services Moderator: J. Scott Roberts, Univ. of Michigan</p>	<p>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.</p>
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

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 prototypic 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: <i>ATR</i> , <i>BAP1</i> , <i>CHEK1</i> , <i>GEN1</i> , <i>KANK4</i> , <i>OBSL1</i> , <i>RAD51B</i> and <i>TP53BP1</i> . 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 <i>TECPR2</i> 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 <i>TGFβ2</i> cause Loews-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 <i>XRCC2</i> 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 <i>ASAH1</i> . 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 <i>HOXB13</i> 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, <i>DUX4</i> 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 <i>MATR3</i> 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 <i>SMAD3</i> 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 <i>OTOGL</i> and <i>SLITRK6</i> are novel genes. M. Tekin et al.	66 Mouse model implicates <i>GNB3</i> 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 (<i>CTNND2</i>): Genetics and function of a novel autism gene. T. Turner et al.	22 Statistics for X-chromosome association. U.zbek et al.	31 Rare allelic forms of <i>PRDM9</i> 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 <i>TCF7L2</i> 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 <i>CHD7</i> mutations. M. Legendre et al.	86 Mutations in <i>OLFML2B</i> 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 <i>PIK3CA</i> 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 <i>FGF21</i> 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.

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<p>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</p>	<p>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.</p>	<p>SESSION 23 – Advancing Gene Therapy to the Clinic: Molecular Medicines Come of Age Moderator: Beverly Davidson, Univ. of Iowa</p>	<p>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</p>	<p>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</p>	<p>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.</p>	<p>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</p>	<p>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</p>
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.



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

Invited Proposals Now Being Accepted:

Deadline, December 5, 2012

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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 <i>DDHD2</i> 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 <i>AUTS2</i> 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 <i>MED12</i> 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 identifies 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 <i>BRAF L597</i> 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 <i>BRCA1</i> -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 <i>BRCA1</i> and <i>BRCA2</i> 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 <i>MBD5</i> 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 <i>MEFV</i> 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 <i>KIAA2022</i> 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 <i>SRCAP</i> . S. M. Nikkel et al.	140 Enhanced detection of low-level mosaic mutations in <i>RB1</i> 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 <i>SLC30A10</i> , 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 <i>DICER1</i> -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 <i>MUTYH</i> 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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<p>SESSION 38 – A Sequencing Jamboree: Exomes to Genomes Co-Moderators: Nancy Cox, Univ. of Chicago; and Orli Bahcall, <i>Nature Genetics</i></p>	<p>SESSION 39 – Admixture and Demography Co-Moderators: Katarzyna Bryc, Harvard Univ.; and John Novembre, UCLA</p>	<p>SESSION 40 – Analysis of Multilocus Systems Co-Moderators: Brendan Keating, Univ. of Pennsylvania; and Laura Almasy, Texas Biomed. Res. Inst., San Antonio</p>	<p>SESSION 41 – Genes Underlying Neurological Disease Co-Moderators: Stylianos Antonarakis, Univ. of Geneva Med. Sch.; and Laurie Ozelius, Mount Sinai Med. Sch.</p>	<p>SESSION 42 – Cancer Genetics III: Common Variants Co-Moderators: Kathleen Cooney, Univ. of Michigan; and Meredith Yeager, NCI/NIH</p>	<p>SESSION 43 – Genetics of Craniofacial and Musculoskeletal Disorders Co-Moderators: Irimi Manoli, NHGRI/NIH; and Siddharth Prakash, Univ. of Texas Hlth. Sci. Ctr. at Houston</p>	<p>SESSION 44 – Tools for Phenotype Analysis Co-Moderators: Donna Maglott, NCI/NIH; and Gregory E. Crawford, Duke Univ.</p>	<p>SESSION 45 – Therapy of Genetic Disorders Co-Moderators: Cynthia J.R. Curry, UCSF; and Brunhilde Wirth, Univ. of Cologne, Germany</p>	<p>SESSION 46 – Pharmacogenetics: From Discovery to Implementation Co-Moderators: Toni Pollin, Univ. of Maryland Sch. of Med.; and Edward Ramos, NHGRI/NIH</p>
<p>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.</p>	<p>178 Differential relatedness of African Americans to populations within West Africa. K. Bryc et al.</p>	<p>187 Gene-based epistasis analysis in genome-wide association studies. L. Ma et al.</p>	<p>196 <i>TMTC4</i>: A novel candidate gene for callosal development. L. Fernandez et al.</p>	<p>205 Identification of 23 novel prostate cancer susceptibility loci using a custom array (the iCOGS) in an international consortium, PRACTICAL. R. Eeles et al.</p>	<p>214 Next-generation sequencing detects mutations in <i>ISPD</i> as a common cause of Walker-Warburg syndrome with defective glycosylation of σ-dystroglycan. T. Roscioli et al.</p>	<p>223 PRIMUS: Pedigree Reconstruction and Identification of the Maximum Unrelated Set. J. Staples et al.</p>	<p>232 Pathogenic exon-trapping by SVA retrotransposon and rescue in Fukuyama muscular dystrophy. M. Taniguchi et al.</p>	<p>241 Pharmacogenomics, ancestry and clinical decision making for global populations. E. Ramos et al.</p>
<p>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.</p>	<p>179 Fine scale population genetic structure of African Americans. E. Y. Durand et al.</p>	<p>188 Building human phenotype networks from shared genetic risk variants. C. Darabos et al.</p>	<p>197 <i>CLK2</i> missense mutation in a family with pontocerebellar hypoplasia type 7. V. R. C. Eggens et al.</p>	<p>206 Large-scale genotyping identifies more than 40 novel breast cancer susceptibility loci. K. Michailidou et al.</p>	<p>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.</p>	<p>224 Pharmacogenoinformatics: Novel approach of in silico drug designing based on genetic variation of <i>MDR1</i> gene involved in statin resistance. A. Munshi et al.</p>	<p>233 Hematopoietic stem cell transplantation for adolescent and adult onset cerebral X-linked adrenoleukodystrophy. T. Matsukawa et al.</p>	<p>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.</p>
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<p>6:00 175 Genome sequencing and analysis in autism spectrum disorder. S. Walker et al.</p>	<p>184 Estimating human population sizes using the coalescent with recombination. S. Sheehan et al.</p>	<p>193 Variants in exons and in transcription factors affect gene expression in trans. A. Kreimer et al.</p>	<p>202 De novo gain of function <i>KCNT1</i> channel mutations cause seizures and developmental delay in malignant migrating partial seizures of infancy. G. Barcia et al.</p>	<p>211 Statistical fine mapping of regions containing melanoma susceptibility genes identified through genome-wide association studies. J. H. Barrett et al.</p>	<p>220 Increased frequency of <i>FBN1</i> variants in adolescent idiopathic scoliosis. J. G. Buchan et al.</p>	<p>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.</p>	<p>238 Beyond cholesterol: Antioxidant treatment for patients with Smith-Lemli-Opitz syndrome. E. Elias et al.</p>	<p>247 Genome-wide association study of vancomycin pharmacokinetics using a de-identified biorepository. S. L. Van Driest et al.</p>
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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, NCB/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
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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 <i>TECTON/C3</i> mutations cause orofacial digital syndrome type IV (Mohr-Majewski). S. Thomas et al.
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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 cilopathy, results from mutation in human <i>Rotatin</i> . 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 <i>ABCA7</i> and <i>GRIN3B</i> , <i>HMHA1</i> and <i>SBNO2</i> , 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 <i>CDKN1C</i> 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 <i>KDM6A</i> 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 <i>LRR6</i> as a novel single-gene cause of primary ciliary dyskinesia. M. Chaki et al.
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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 <i>SLC20A2</i> 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 <i>RET</i> 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 <i>GADD45A</i> . 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 <i>MAP3K1</i> 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 <i>C9ORF72</i> 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 <i>KIAA0649P</i> into the mouse <i>Rb1</i> locus: Modeling the mechanism of imprinted <i>RB1</i> 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 <i>GTF2IRD1</i> . 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 <i>C9orf72</i> 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 <i>BCL9L</i> 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.

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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**



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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.

Saturday, November 10

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 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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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.

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