American Society of Human Genetics October 22-26 2013

BOSTON

PROGRAM AT-A-GLANCE

Registration and Program Pick-up Hours:

Tuesday: 10:00 am-6:00 pm
Wednesday: 7:00 am-5:00 pm
Thursday: 7:30 am-5:00 pm
Friday: 7:30 am-5:00 pm
Saturday: 7:30 am-10:30 am

THE AMERICAN SOCIETY OF HUMAN GENETICS

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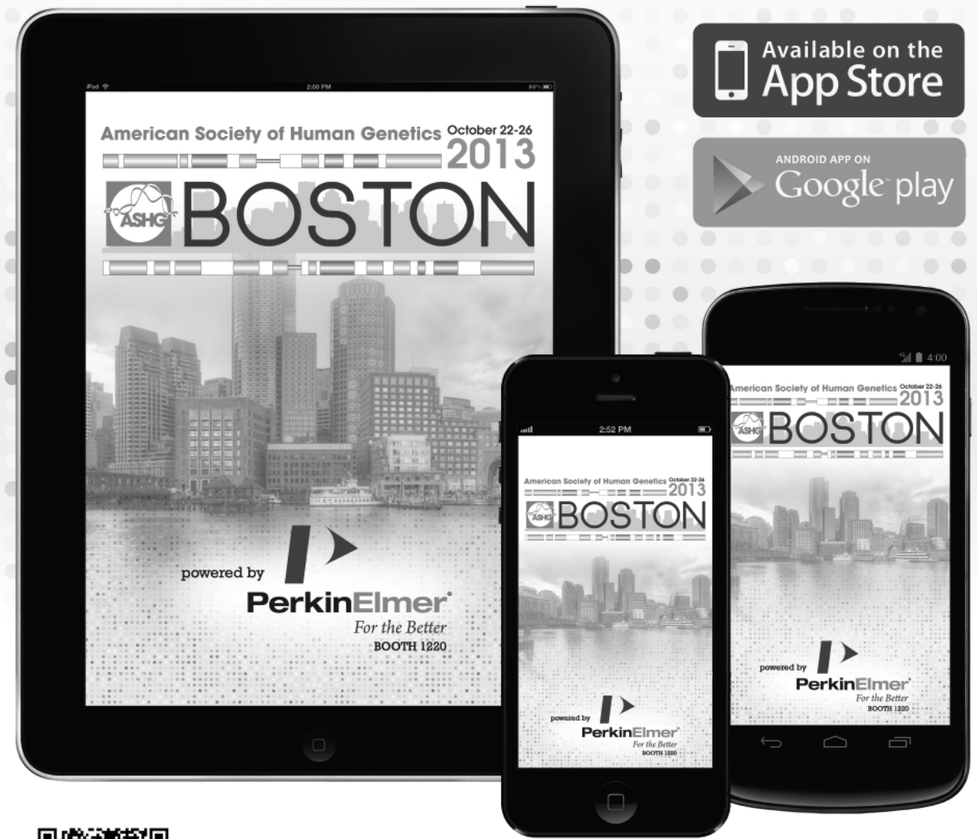

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THE AMERICAN SOCIETY OF HUMAN GENETICS

Schedule of ASHG Scientific Sessions and Events

All events are at the Boston Convention and Exhibition Center (BCEC), unless otherwise indicated.
(*) Asterisk denotes meetings that are by invitation or pre-registration only. Otherwise, attendance may be assumed to be open to all registrants

Tuesday, October 22		
8:00am-3:00pm	ASHG Undergraduate Faculty Genetics Education Workshop*	Room 152, Level 1
8:30am-2:30pm	ASHG High School Workshop for local Boston students and teachers*	Room 052A, Level 0
8:30am-3:00pm	ASHG Board of Directors Meeting*	Room 256, Level 2
10:00am-12:00pm	ASHG Social Issues Committee Meeting*	Room 150, Level 1
10:00am-6:00pm	Scientific Registration Open	North Lobby, Level 1
11:00am-3:00pm	ASHG Program Committee Meeting*	Room 257, Level 2
11:00am-5:00pm	Speaker Presentation/Upload Room Open	Room 203, Level 2
2:00pm-3:30pm	Human Epigenome Atlas and Epigenomic Profile Analysis Using the Genboree Workbench*	Room 104, Level 1
2:00pm-3:30pm	Introduction to Integrative Analysis with GenomeSpace*	Room 102, Level 1
4:30pm-5:00pm	**Session 1:** ASHG Presidential Address: Just Another President's Speech (BUT It's All About You)	Hall B2, Level 0
5:00pm-7:00pm	**Session 2:** Plenary Abstract Presentations	Hall B2, Level 0
7:30pm-9:00pm	The Drama of DNA: Anticipating the Future with WGS*	Room 052, Level 0
7:30pm-9:30pm	#ASHG 2013 Tweet-up	Westin Hotel, M.J. O'Connor's Bar

Wednesday, October 23		
7:00am-5:00pm	Scientific Registration Open	North Lobby, Level 1
7:00am-5:00pm	Speaker Presentation/Upload Room Open	Room 203, Level 2
8:00am-10:00am	**Concurrent Invited Sessions I (3-9):**	
	Session 3: A Renaissance in Gene Therapy: New Tools and Clinical Trials	Room 253, Level 2
	Session 4: DNA Damage Response Network Defects and Cancer Predisposition: Where One Plus One Does Not Equal Two	Room 210, Level 2
	Session 5: Does the Increasingly Blurry Distinction between Research and Clinical Care Create an Obligation to Actively Search for Secondary Findings in Genomic Research or Otherwise Change the Relationship between Researchers and Participants?	Grand Ballroom East, Level 3
	Session 6: Evidence-Based Genetic Counseling for Clinical Genome Sequencing	Grand Ballroom West, Level 3
	Session 7: Functional Interpretation of Genomes Using Biological Networks	Room 205, Level 2
	Session 8: Insights from Large Scale Sequencing	Hall B2, Level 0
	Session 9: Population-Based Animal Models for Discovery of Complex Traits	Room 258, Level 2
10:00am-6:00pm	Exhibits and Posters Open	Exhibit Hall, Level 1
10:00am-6:00pm	ASHG/FASEB Career Resources Open	Exhibit Hall, Level 1
10:30am-12:30pm	**Poster Session I**: Wednesday Poster Authors Present	Exhibit Hall, Level 1
12:30pm-2:00pm	High-Throughput Data Analysis and Visualization with Galaxy*	Room 104AB, Level 1
12:30pm-2:00pm	ASHG Trainee-Mentor Luncheon*	Room 052, Level 0
12:30pm-2:00pm	Navigating Clinical Genomic Resources at NCBI*	Room 102, Level 1

12:30pm-1:15pm	ASHG Trainee Development Program: Beyond the Bench…Preparing for Your Career Transition in the Life Sciences. *Admission on a first-come, first-served basis.*	Exhibit Hall, Level 1
1:15pm-2:00pm	ASHG Trainee Development Program: Academic Job Search: CVs, Letters, Statements, and Start-Ups. *Admission on a first-come, first-served basis.*	Exhibit Hall, Level 1
2:00pm-4:15pm	**Concurrent Platform Session A (10-18):**	
	Session 10: Which Comes First: The Sequence or the Biology?	Hall B2, Level 0
	Session 11: The Shifting Landscape of Genetic Testing: Approaches and Success Stories	Grand Ballroom East, Level 3
	Session 12: Methods in Statistical Genetics	Grand Ballroom West
	Session13: Genetic Variation in Gene Expression	Room 210, Level 2
	Session 14: Cancer Epidemiology: New Loci and Methods	Room 205, Level 2
	Session 15: Psychiatric Disease: GWAS to Genes	Room 253, Level 2
	Session 16: Expanding Knowledge of Mendelian Disorders: Genes, Phenotypes & Treatment	Room 258, Level 2
	Session 17: Structural/Copy Number Variation and Disease	*Westin Hotel,* Grand Ballroom AB
	Session 18: Inborn Errors of Metabolism: From Identification to Treatment	*Westin Hotel,* Grand Ballroom CDE
4:30pm-6:00pm	Celebrate with ASHG at the Welcome Reception	Exhibit Hall, Level 1

Thursday, October 24

7:00am-5:00pm	Speaker Presentation/Upload Room Open	Room 203, Level 2
8:00am-10:15am	**Concurrent Platform Session B (19-27):**	
	Session 19: Hereditary Cancer Syndromes	Hall B2, Level 0
	Session 20: Variants, Variants Everywhere	Grand Ballroom East,
	Session 21: Genetic Epidemiology: Applications and Methods	Grand Ballroom West,
	Session 22: Cardiovascular Genetics: Gene Discovery through GWAS and Sequencing	Room 210, Level 2
	Session 23: From eQTLs to Epigenetics and Beyond	Room 205, Level 2
	Session 24: Neurogenetics: Illuminating Mechanisms	Room 253, Level 2
	Session 25: Genetic Interactions in Complex Traits	Room 258, Level 2
	Session 26: Advances in the Genetics of Skeletal and Morphologic Disorders	*Westin Hotel*, Grand Ballroom AB
	Session 27: Causes and Consequences of Chromosomal Variations	*Westin Hotel,* Grand Ballroom CDE
10:00am-4:30pm	Exhibits and Posters Open	Exhibit Hall, Level 1
10:00am-4:30pm	ASHG/FASEB Career Resources Open	Exhibit Hall, Level 1
10:30am-12:30pm	**Poster Session II**: Thursday Poster Authors Present	Exhibit Hall, Level 1
12:30pm-2:00pm	*AJHG* Editorial Board Meeting*	Room 257, Level 2
12:30pm-2:00pm	ASHG Information & Education Committee Meeting*	Room 101, Level 1
12:30pm-2:00pm	Diagnostic Challenges: Review and Discussion of Unique Cases*	Room 052, Level 0
12:30pm-2:00pm	UCSC Genome Browser Interactive Workshop: Advanced Topics*	Room 104AB, Level 1
12:30pm-1:15pm	ASHG Trainee Development Program: Postdocs: What Should You Be Looking for and How to Find Them. *Admission on a first-come, first-served basis.*	Exhibit Hall Career Resources Theater, Level 1
1:15pm-2:00pm	ASHG Trainee Development Program: Beyond the Bench…Preparing for Your Career Transition in the Life Sciences. *Admission on a first-come, first-served basis.*	Exhibit Hall Career Resources Theater, Level 1
2:00pm-4:15pm	**Concurrent Platform Session C (28-36):**	
	Session 28: Low Frequency Variants for Complex Traits	Hall B2, Level 0
	Session 29: Selection, Demography and Functional Polymorphism	Level 3
	Session 30: Statistical Methods for Family Data	Grand Ballroom West,
	Session 31: Advances and References in Genomic Technology	Room 210, Level 2
	Session 32: Genetic Testing for Neurodevelopmental Disease: Genotype: Phenotype Challenges	Room 205, Level 2
	Session 33: Gene Regulation — At a Multitude of Levels	Room 253, Level 2
	Session 34: Cardiovascular Genetics: Exome Sequencing and Animal Models	Room 258, Level 2
	Session 35: Genomic Medicine: Counseling, Education and Health Services	*Westin Hotel,* Grand Ballroom AB
	Session 36: Biochemical and Clinical Consequences of Mitochondrial Dysfunction	*Westin Hotel,* Grand Ballroom CDE
4:30pm-6:30pm	**Concurrent Invited Session II (37-43):**	
	Session 37: Community Efforts to Decipher the Phenotypic Impact of Genomic Variation	Grand Ballroom West, Level 3
	Session 38: Genetics of Non-communicable Diseases in sub-Saharan Africa	Room 258, Level 2
	Session 39: Guilty by Annotation: The Role of Non-coding Variation in Phenotypic Variation and Disease	Grand Ballroom East, Level 3
	Session 40: Human Genetics of Common Infectious Diseases	Room 210, Level 2
	Session 41: Informed Consent for Whole Genome Sequencing: Experience and Implications for Practice	Hall B2, Level 0
	Session 42: Multimodal Treatment of Lysosomal Storage Diseases as a Portal to Emergent Genetic Therapies	Room 205, Level 2
	Session 43: Nonhuman Primate Genomics: Evolutionary Insights and Relevance to Human Phenotypes	Room 253, Level 2
6:45pm-7:45pm	**Session 44:** ASHG Next: The Future of Genetics and the Future of Your Society.	Room 253, Level 2

Friday, October 25

7:00am-5:00pm	Speaker Presentation/Upload Room Open	Room 203, Level 2
8:00am-10:15am	**Concurrent Platform Session D (45-53):**	
	Session 45: Mo' Data, Mo' Problems?	Hall B2, Level 0
	Session 46: Cancer Genomics	Grand Ballroom East
	Session 47: Demography In and Out of Africa	Grand Ballroom West
	Session 48: Fine-Mapping and Function of Candidate Loci	Room 210, Level 2
	Session 49: New Genes and Disorders	Room 205, Level 2
	Session 50: Neurodegenerative Disease and the Aging Brain	Room 253, Level 2
	Session 51: Epigenetics: From Genomes to Genes	Room 258, Level 2
	Session 52: New Frontiers in Pharmacogenetics	*Westin Hotel,* Grand Ballroom AB
	Session 53: Genomic Approaches for Study of Rare Neurogenetic Disorders	*Westin Hotel,* Grand Ballroom CDE
10:00am-2:30pm	Exhibits and Posters Open	Exhibit Hall, Level 1
10:00am-2:30pm	ASHG/FASEB Career Resources Open	Exhibit Hall, Level 1
10:30am-12:30pm	**Poster Session III**: Friday Poster Authors Present *All authors to remove posters at 2:00 pm*	Exhibit Hall, Level 1
12:30pm-2:00pm	ASHG Program Committee Meeting*	Room 257, Level 2
12:30pm-2:00pm	ASHG Publications Workshop*	Room 052, Level 0
12:30pm-2:00pm	Ensembl Highlights Interactive Workshop: Intermediate/Advanced Users*	Room 104AB, Level 1
12:30pm-2:00pm	Introduction to NGS Visualization with the Integrative Genomics Viewer*	Room 102, Level 1
12:30pm-1:15pm	ASHG Trainee Development Program: Academic Job Search: CVs, Letters, Statements, and Start-Ups	Exhibit Hall Career Resources Theater,

	Admission on a first-come, first-served basis	Level 1
1:15pm-2:00pm	ASHG Trainee Development Program: Postdocs: What Should You be Looking for and How to Find Them *Admission on a first-come, first-served basis.*	Exhibit Hall Career Resources Theater, Level 1
12:30pm-2:00pm	ASHG/FASEB MARC Award Winners Luncheon*	Room 103, Level 1
12:30pm-2:00pm	ASHG/NHGRI Fellows Luncheon*	Room 207, Level 2
2:00pm-4:15pm	**Concurrent Platform Session E (54-62):**	
	Session 54: Hundreds of New GWAS Loci	Hall B2, Level 0
	Session 55: Impact of Bottlenecks and Population Growth on Rare Variation	Grand Ballroom East, Level 3
	Session 56: Haplotypes, Imputation and Interactions	Grand Ballroom West
	Session 57: Autism and Neurodevelopmental Disorders	Room 210, Level 2
	Session 58: Cardiovascular Genetics: Functional Characterization and Clinical Applications	Room 205, Level 2
	Session 59: Prenatal and Reproductive Genetics	Room 253, Level 2
	Session 60: Ethical, Legal, Social and Policy Issues	Room 258, Level 2
	Session 61: Genomics of Developmental Disorders	Westin Hotel, Grand Ballroom AB
	Session 62: Prostate and GI Cancer Susceptibility	Westin Hotel, Grand Ballroom CDE
4:30pm-4:50pm	**Session 63:** ASHG Award for Excellence in Human Genetics Education Presentation	Hall B2, Level 0
4:50pm-5:30pm	**Session 64:** ASHG William Allan Award Presentation	Hall B2, Level 0
5:30pm-6:30pm	**Session 65:** ASHG Membership and Business Meeting	Hall B2, Level 0
6:30pm-8:00pm	ASHG Trainee Networking Session*	East Ballroom Foyer Level 3
7:00pm-9:00pm	ASHG Awards Committee Meeting*	Room 257, Level 2
Saturday, October 26		
8:00am-8:20am	**Session 66:** ASHG Victor A. McKusick Leadership Award Presentation	Hall B2, Level 0
8:20am-8:30am	**Session 67:** C.W. Cotterman Awards Announcement	Hall B2, Level 0
8:30am-8:40am	**Session 68:** ASHG Charles J. Epstein Trainee Awards for Excellence in Human Genetics Research: Announcement of Winners	Hall B2, Level 0
8:40am-9:10am	**Session 69:** ASHG Curt Stern Award Presentation	Hall B2, Level 0
9:30am-11:30am	**Concurrent Invited Session III (70-76):**	
	Session 70: Design, Content and EMR Integration of Clinical Sequencing Reports	Grand Ballroom West, Level 3
	Session 71: More or Less: Copy Number Variation and Human Adaptation	Room 258, Level 2
	Session 72: Somatic Mutations in Human Disease — Piecing Together the Mosaic	Room 210, Level 2
	Session 73: Tandem Repeat-Associated Epigenetic Mechanisms in Neuromuscular Disorders	Room 205, Level 2
	Session 74: Twin Studies: Helping Us Understand and Exploit the Genome (In Honor of Walter Nance's Contributions to Human Genetics on his 80th Birthday)	Hall B2, Level 0
	Session 75: Where Do Risk Variants Act? Interrogating Genomic Studies of Multiple Human Tissues	Room 253, Level 2
	Session 76: Whole Genome Sequencing for Every Baby? Where Diagnostic and Screening Applications Collide	Grand Ballroom East, Level 3
11:45am-1:15pm	**Session 77:** ASHG Distinguished Speakers Invited Symposium: Medical Systems Genomics	Hall B2, Level 0

Tuesday, October 22
4:30 PM–5:00 PM

SESSION 1 – ASHG Presidential Address: Just Another President's Speech (BUT It's All About You)

Hall B2, Level 0 (Lower Level), Convention Center

Presenter: Jeff Murray, ASHG 2013 President, University of Iowa

Although our society turned 65 this year it is far from ready for retirement. Our members have led the way as changes in knowledge, technology, policy and education have challenged our mission over the last seven decades. Genetics has never been more central than it is today to the national and international conversations surrounding science and its application in health, law and teaching. But we must ever focus our commitment to being a part of the solution to the changes that confront us. Having an engaged and active membership who create a strategic vision for our future can ensure that we increase in relevance and utility for ASHG in particular and for advancing the good of society in general.

Tuesday, October 22
5:00 PM–7:00 PM

SESSION 2 – Plenary Abstract Presentations

Hall B2, Level 0 (Lower Level), Convention Center

Moderator: Andrew G. Clark, Cornell Univ.

1/5:00 **Whole exome sequencing of 94 matched brain metastases and paired primary tumors reveals patterns of clonal evolution and selection of driver mutations.** S. L. Carter, P. K. Brastianos, S. Santagata, A. Taylor-Weiner, P. Horowitz, K. Ligon, J. Seaone, E. Martinez-Saez, J. Tabernero, D. Cahill, S. Paek, I. Dunn, B. Johnson, M. Rabin, N.U. Lin, R. Jones, P. Hummelen, A. Stemmer-Rachamimov, D.L. Louis, T.T. Batchelor, J. Baselga, R. Beroukhim, G. Getz, W.C. Hahn.

2/5:20 **Pathogenic de novo SNVs, indels and CNVs in 1,000 children with undiagnosed developmental disorders.** M. Hurles, M. van Kogelenberg, T. Fitzgerald, W. D. Jones, D. King, P. Vijayarangakannan, S. Gerety, K. Morley, S. Gribble, D. Barrett, K. Ambridge, N. Krishnappa, E. Prigmore, D. Rajan, T. Bayzetinova, S. Al-Turki, A. Tivey, S. Clayton, R. Miller, P. Jones, N. Carter, C. Wright, J. Barrett, D. FitzPatrick, H. Firth, DDD Study.

3/5:40 **Chromatin loops and CNVs: The complex spatial organization of the 16p11.2 locus.** M. N. Loviglio, M. Leleu, N. Ghedolf, E. Migliavacca, K. Männik, J. S. Beckmann, S. Jacquemont, J. Rougemont, A. Reymond.

4/6:00 **Fine-mapping GWAS followed by genome editing identifies an essential erythroid enhancer at the HbF-associated *BCL11A* locus.** D. E. Bauer, S. Lessard, S. C. Kamran, J. Xu, Y. Fujiwara, C. Lin, Z. Shao, M. C. Canver, E. C. Smith, L. Pinello, P. J. Sabo, J. Vierstra, R. A. Voit, G. C. Yuan, M. H. Porteus, J. A. Stamatoyannopoulos, G. Lettre, S. H. Orkin.

5/6:20 **Translating dosage compensation to Trisomy 21: A novel approach to Down syndrome.** J. B. Lawrence, J. Jiang, Y. Jing, C. J. Cost, J. Chiang, H. J. Kolpa, A. M. Cotton, D. M. Carone, B. R. Carone, D. A. Shivak, M. Byron, P. D. Gregory, C. J. Brown, F. D. Urnov, L. L. Hall.

6/6:40 **Insights into population history from a high coverage Neandertal genome.** D. Reich, Neandertal Genome Consortium.

Note: The Opening Mixer, traditionally held on Tuesday evening has been moved to Wednesday at 4:30 pm and has been renamed The Welcome Reception.

Wednesday, October 23: Concurrent Invited Session I (3-9)

8:00 AM–10:00 AM

Room 253, Level 2	Room 210, Level 2	Grand Ballroom East, Level 3	Grand Ballroom West, Level 3	Room 205, Level 2	Hall B2, Level 0 (Lower Level)	Room 258, Level 2
SESSION 03 – A Renaissance in Gene Therapy: New Tools and Clinical Trials Co-Moderators: Stephen Kaler, NICHD/NIH; and Luk H. Vandenberghe, Harvard Med. Sch.	**SESSION 04 – DNA Damage Response Network Defects and Cancer Predisposition: Where One Plus One Does Not Equal Two** Co-Moderators: Marc Tischkowitz, Univ. of Cambridge; and William D. Foulkes, McGill Univ.	**SESSION 05 – Does the Increasingly Blurry Distinction between Research and Clinical Care Create an Obligation to Actively Search for Secondary Findings in Genomic Research or Otherwise Change the Relationship between Researchers and Participants?** Co-Moderators: Benjamin E. Berkman, NHGRI/NIH; and Stacy Gray, Dana Farber Cancer Inst.	**SESSION 06 – Evidence-Based Genetic Counseling for Clinical Genome Sequencing** Co-Moderators: Shelin Adam, Univ. of British Columbia; and Myra Roche, Univ. of North Carolina at Chapel Hill	**SESSION 07 – Functional Interpretation of Genomes Using Biological Networks** Moderator: Kasper Lage, Massachusetts Gen. Hosp.	**SESSION 08 – Insights from Large Scale Sequencing** Co-Moderators: Goncalo R. Abecasis, Univ. of Michigan Sch. of Publ. Hlth.; and Gabor Marth, Boston Col.	**SESSION 09 – Population-Based Animal Models for Discovery of Complex Traits** Co-Moderators: John French, NIEHS/NIH, Research Triangle Park; and Kimberly McAllister, NIEHS/NIH, Research Triangle Park
08:00 am Adeno-associated viral vectors for gene therapy: Virus, vector, nanoparticle. **L. H. Vandenberghe.**	08:00 am The molecular crossroads of ataxia telangiectasia, Fanconi anemia and hereditary breast/ovarian cancer. **M. Tischkowitz.**	08:00 am Why researchers do not have a duty to look for incidental findings. **E. W. Clayton.**	08:00 am Evidence-based genomic counseling for 21st century medicine. **M. J. Khoury.**	08:00 am Integrating biological networks and genetics to reverse engineer molecular systems driving diseases. **K. Lage.**	08:00 am Mechanistic and clinical advances from sequencing cancer patients. **N. Rahman.**	08:00 am High-resolution genetic mapping using the mouse collaborative cross and diversity outbred populations. **G. Churchill.**
08:30 am Gene therapy for metabolic CNS disorders. **R. G. Crystal.**	08:30 am Characterizing cancer risks in carriers with mutations in the *FA/BRCA* and *ATM* genes. **A. C. Antoniou.**	08:30 am Appropriately - but narrowly - defining a researcher›s obligations to look for incidental findings. **J. P. Evans.**	08:30 am Measuring patient benefits from genetic counseling and testing interventions: Is patient empowerment a useful outcome? **M. McAllister.**	08:30 am Cell-type specificity of gene networks to understand disease biology. **S. Raychaudhuri.**	08:30 am Accelerating Mendelian genetics. **D. Nickerson.**	08:30 am Genomic characterization of house dust mite-induced allergic airway disease in mice. **S. Kelada.**
09:00 am Brain-directed AAV gene therapy. **S. Kaler.**	09:00 am Ataxia-telangiectasia: From phenotype to biology, and onto treatment strategies. **Y. Shiloh.**	09:00 am Neither duty nor prohibition: Using private ordering to transcend ‹one-size-fits-all› in large-scale sequencing of human beings. **M. N. Meyer.**	09:00 am Can an e-learning platform provide adequate genetic counseling? **P. Birch.**	09:00 am Using networks, functional genomics resources and signals of selection to understand hundreds of complex disease loci. **J. Barrett.**	09:00 am Whole genome mapping, assembly and analysis. **H. Li.**	09:00 am Host genetics and modulation of the gut microbiome as a means to understanding metabolic function. **A. Benson.**
09:30 am Lentiviral-mediated gene therapy for human disease: Extensive genetic engineering of hematopoiesis with therapeutic benefit in metachromatic leukodystrophy patients after lentiviral hematopoietic stem cell gene therapy. **L. Naldini.**	09:30 am Targeting the *FA/BRCA* pathway in cancer therapy. **A. D. D'Andrea.**	09:30 am What we›ve got here is failure to communicate: The growing disconnect between 20th-century research protections and individual agency in an age of whole-genome sequencing. **M. Angrist.**	09:30 am Online genetic education and counseling — Lessons from a direct-access genotyping service. **U. Francke.**	09:30 am Network-based association models for exome-sequencing data. **S. Purcell.**	09:30 am Population genetic insights from 10,000s of human samples. **J. Novembre.**	09:30 am Mapping susceptibility QTLs for gene-environment interactions and environmental toxicity. **J. French.**

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 #ASHG2013** or by posting on the ASHG Facebook page.

Twitter: **@Genetics Society,** #ASHG2013

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

Follow ASHG on Twitter before and during the week of the 2013. 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 and Posters

Wednesday: 10:00 am – 6:00 pm

Thursday: 10:00 am – 4:30 pm

Friday: 10:00 am – 2:30 pm

ASHG Welcome Reception

Exhibit Hall

4:30 pm – 6:00 pm

Wednesday, October 23: Concurrent Platform (abstract-driven) Session A (10-18)

2:00 PM–4:15 PM

	Hall B2, Level 0 (Lower Level)	Grand Ballroom East, Level 3	Grand Ballroom West, Level 3	Room 210, Level 2	Room 205, Level 2	Room 253, Level 2	Room 258, Level 2	Westin Grand Ballroom AB, Concourse Level	Westin Grand Ballroom CDE, Concourse Level
	SESSION 10 – Which Comes First: The Sequence or the Biology? Co-Moderators: Maja Bucan, Univ. of Pennsylvania; and Melissa Wilson Sayres, Univ. of California, Berkeley	**SESSION 11 – The Shifting Landscape of Genetic Testing: Approaches and Success Stories** Co-Moderators: Catherine E. Keegan, Univ. of Michigan; and Stephanie Bielas, Univ. of Michigan	**SESSION 12 – Methods in Statistical Genetics** Co-Moderators: L. Adrienne Cupples, Boston Univ. Sch of Publ. Hlth; and Ingrid B. Borecki, Washington Univ. Sch. of Med.	**SESSION 13 – Genetic Variation in Gene Expression** Co-Moderators: Cisca Wijmenga, Univ. of Groningen; and Barbara Stranger, Univ. of Chicago	**SESSION 14 – Cancer Epidemiology: New Loci and Methods** Co-Moderators: Sharon Savage, NCI/NIH; and Antonis C. Antoniou, Univ. of Cambridge	**SESSION 15 – Psychiatric Disease: GWAS to Genes** Co-Moderators: Brian O'Roak, Univ. of Washington; and Joseph D. Buxbaum, Mount Sinai Sch. of Med.	**SESSION 16 – Expanding Knowledge of Mendelian Disorders: Genes, Phenotypes & Treatment** Co-Moderators: Deb Krakow, UCLA; and Donna M. Martin, Univ. of Michigan	**SESSION 17 – Structural/Copy Number Variation and Disease** Co-Moderators: Steven McCarroll, Harvard Med. Sch.; and David Miller, Boston Children's Hosp.	**SESSION 18 – Inborn Errors of Metabolism: From Identification to Treatment** Co-Moderators: Irini Manoli, NHGRI/NIH; and Manuel Schiff, Univ. of Pittsburgh
2:00	**7** Annotation of pseudogenous gene segments by massively parallel sequencing of rearranged lymphocyte receptor loci. **R. O. Emerson et al.**	**16** The utility of the traditional medical genetics diagnostic evaluation in the context of next-generation sequencing for undiagnosed genetic disorders. **V. Shashi et al.**	**25** Prioritizing sequence variants using statistical evidence: Not all measures are alike. **L. Strug et al.**	**34** Epistasis is widespread in the genetic control of transcription in humans. **J. Powell et al.**	**43** Heritability and familial risk of cancer: An update from the Nordic Twin Registry of Cancer. **L. A. Mucci et al.**	**52** Using brain molecular QTLs to identify novel risk genes shared by multiple psychiatric diseases. **C. Liu et al.**	**61** NIH study, Clinical and Molecular Investigations into Ciliopathies: Findings on Alström syndrome. **J. D. Marshall et al.**	**70** Large-scale parent-child trio sequencing highlights factors influencing spontaneous human mutation. **S. Sunyaev et al.**	**79** A novel treatable disorder of protein glycosylation: Phosphoglucomutase 1 deficiency. **E. Morava et al.**
2:15	**8** High throughput sequence analysis of the TCR repertoire in glioma-associated immune dysregulation. **B. Grinshpun et al.**	**17** The National Institutes of Health Undiagnosed Diseases Program: The first four years. **D. R. Adams et al.**	**26** Mixed model association methods: Advantages and pitfalls. **A. Price et al.**	**35** Low-pass whole-genome sequencing in Europeans identifies 1325 SNPs and indels associated with cis gene expression of which 4% are independent low frequency-large effect associations. **A. R. Wood et al.**	**44** A comprehensive genetic analysis of common cancer risk through the development of the Oncochip. **C. I. Amos et al.**	**53** Functional enrichment analysis on genome-wide epistasis patterns reveals pathway interactions in bipolar disorder. **S. Prabhu et al.**	**62** Genotype/epigenotype/ phenotype correlations define Beckwith-Wiedemann syndromes. **A. Mussa et al.**	**71** Palindromic *GOLGA* core duplicon promotes 15q13.3 microdeletion, inversion polymorphisms, and large-scale primate structural variation. **M. Y. Dennis et al.**	**80** Mutations in *HCFC1* a transcriptional coregulator causes a novel X-linked cobalamin disorder (*cblX*) with a severe neurological phenotype. **T. H. Shaikh et al.**
2:30	**9** Extraction and analysis of clinical traits of multiple sclerosis using electronic medical records. **M. F. Davis et al.**	**18** First year experience of clinical exome sequencing for rare disease diagnosis at UCLA. **H. Lee et al.**	**27** Assessing multivariate gene-metabolome associations using Bayesian reduced rank regression. **P. Marttinen et al.**	**36** Deep whole-genome sequencing in pedigrees to quantify the contribution of private variants to type 2 diabetes and related metabolic traits. **G. Jun et al.**	**45** Detection of large clonal mosaic events in existing genome-wide association study data. **M. Machiela et al.**	**54** Role of the Wnt signaling pathway in bipolar disorder susceptibility: Gene-set analysis of SNP-BMI interaction effects. **M. A. Simonson et al.**	**63** Myhre and LAPS syndromes: Clinical and molecular review of 32 patients. **C. Michot et al.**	**72** Large-scale genotyping of polymorphic inversions in human populations. **S. Villatoro et al.**	**81** Prediction of phenotypes and tetrahydrobiopterin-responsiveness in phenylketonuria using data from the genotypes and locus-specific databases. **S. Wettsten et al.**
2:45	**10** Haplotype of CpG-related SNPs is associated with DNA methylation pattern. **Y. Ma et al.**	**19** Clinical utility of the first one thousand clinical whole exome sequencing tests: Molecular diagnostic rate, changes in medical management, and the impact of incidental findings. **C. Eng et al.**	**28** Characterizing shared pathogenetics from genome-wide association studies via principal component analysis. **A. Keinan et al.**	**37** Inflammatory bowel disease-associated genetic variants preferentially alter gene expression in neutrophil granulocytes. **H. Westra et al.**	**46** Most common 'sporadic' cancers have a substantial germline genetic component. **S. Macgregor et al.**	**55** A novel variant in the HERG3 voltage-gated potassium ion channel gene (*KCHN7*) is associated with bipolar spectrum disorder among the Old Order Amish. **A. R. Benkert et al.**	**64** The phenotype combining Treacher Collins syndrome features with Diamond-Blackfan anemia is a heterogeneous ribosomopathy. **K. Sol-Church et al.**	**73** *SCRIB* and *PUF60* are primary drivers of the multisystemic phenotypes of the 8q23.4 CNV. **C. Golzio et al.**	**82** Three apparent pseudo-deficiency alleles in the *IDUA* gene identified by newborn screening. **L. M. Pollard et al.**
3:00	**11** Comprehensive blood group prediction using whole genome sequencing data from The MedSeq Project. **W. J. Lane et al.**	**20** Discordant karyotype results among non-invasive prenatal screening positive cases. **K. W. Choy et al.**	**29** Power studies for the "extreme versus control" design for exome sequencing studies and identification of a variant of TMC6 as a deleterious modifier for age of onset of chronic *Pseudomonas* infection in children with cystic fibrosis. **M. J. Emond et al.**	**38** Multi-tissue eQTL and pathway analysis of genome-wide genetic association data helps uncover tissue-specific processes of complex disease. **A. V. Segrè et al.**	**47** Meta-analysis of genome-wide association studies in 125,000 women identifies fourteen new breast cancer susceptibility loci. **K. Michailidou et al.**	**56** Exome sequencing in sporadic cases of schizophrenia and functional studies identify 21 putative candidate genes for schizophrenia and establish the *RGS12* gene as a strong candidate. **M. Guipponi et al.**	**65** Weaver syndrome is caused by loss-of-function mutations in *EZH2*. **A. S. A. Cohen et al.**	**74** Functional dissection of the recurrent reciprocal 1q21.1 autism-associated CNV. **N. Katsanis et al.**	**83** Atherosclerosis and glycosaminoglycan metabolism defects: Frequent association of endothelial dysfunction in patients with mucopolysaccharidosis. **S. Yano et al.**
3:15	**12** pVAAST: A new method for family-based rare variant association testing. **C. D. Huff et al.**	**21** Genetic testing for mitochondrial disorders in the next-generation sequencing era: Targeted multiplex PCR gene panel or capture-based WES? **R. Bai et al.**	**30** A novel statistical framework for using 'out of study' control groups in association studies with next-generation data. **A. Derkach et al.**	**39** High-resolution functional analysis of eQTLs in multiple tissues. **X. Wen et al.**	**48** Identification of seven new ovarian cancer susceptibility loci: Risk of ovarian cancer in the general population and in *BRCA1* and *BRCA2* carriers. **K. Kuchenbaecker et al.**	**57** Rare duplications in *RB1CC1* are associated with schizophrenia in large datasets from Europe. **F. Degenhardt et al.**	**66** Novel *ELN* mutations and vascular phenotype in autosomal dominant cutis laxa. **E. Lawrence et al.**	**75** Discovery of genes responsible for neurocognitive disease by large scale integration of sequence and copy number data. **B. P. Coe et al.**	**84** First results of a 6 month, open label, phase I/II clinical trial of intrathecal enzyme replacement therapy and its extension in mucopolysaccharidosis IIIA (Sanfilippo syndrome) patients. **C. Breen et al.**
3:30	**13** Functional genomic confounds of transgenic integration and methods for their delineation. **J. C. Jacobsen et al.**	**22** Comparison of results between karyotyping and microarray testing in postnatal and prenatal specimens: Karyotyping is not dead yet. **D. Warburton et al.**	**31** Using coalescent-based modeling for large-scale fine mapping of complex trait loci using sequencing data in large-scale case-control studies. **Z. Geng et al.**	**40** Network QTLs: A new methodology for multi-tissue eQTL discovery. **B. Iriarte et al.**	**49** Breast cancer risk and survival at the 2q35 locus are mediated through chromatin looping and regulation of *IGFBP5* expression. **M. Ghoussaini et al.**	**58** Variants in *NRG3* associated with delusion have regulatory potential and differentially bind to nuclear proteins. **M. Zeledon et al.**	**67** Mutations in *Lrp5* improve bone properties in a mouse model of osteogenesis imperfecta. **C. M. Jacobsen et al.**	**76** Mind the gap — Exomes and CNVs testing in primary immunodeficiencies. **A. Stray-Pedersen et al.**	**85** Development of AAV8-mediated gene therapy clinical trial for Crigler-Najjar syndrome type I: Optimization of liver-specific expression cassette. **N. Pastore et al.**
3:45	**14** Improved exome prioritization of disease genes through cross species phenotype comparison. **D. Smedley et al.**	**23** Parental studies of 2,248 chromosomal microarray analysis cases: Role of parental studies in facilitating the interpretation of copy number variants. **W. Bi et al.**	**32** Quantifying and partitioning variation due to genetic effects and population stratification using within-family prediction analysis. **J. Yang.**	**41** Identification and characteristics of common genetic variants controlling transcript isoform variation in the Framingham Heart Study. **X. Zhang et al.**	**50** Expression quantitative trait loci analysis in breast cancer tumor and normal adjacent FFPE specimens from the Nurses' Health Study. **A. Quiroz-Zarate et al.**	**59** Testing genetic associations with addiction phenotypes using moderate-depth whole genome sequencing. **S. I. Vrieze et al.**	**68** Teriparatide, the first anabolic agent for treatment of osteogenesis imperfecta improves bone mineral density at the hip and spine: A randomized, blinded, placebo-controlled trial. **S. C. Sreenath Nagamani et al.**	**77** Absence of heterozygosity accompanying complex human genomic rearrangements: Further evidence for replicative mechanisms. **C. M. B. C. Fonseca et al.**	**86** Identification of chemical and pharmacological chaperones to treat Zellweger spectrum patients with the common allele, *PEX1-Gly483Asp*. **N. E. Braverman et al.**
4:00	**15** Phased allele-specific expression analysis in integrated whole exome and mRNA sequencing study in a family with non-random X chromosome inactivation. **S. Szelinger et al.**	**24** Rapid and cost-effective whole exome sequencing for clinical diagnosis and personalized medicine. **D. Muzny et al.**	**33** Meta-imputation: A simple and flexible method to combine multiple reference panels for imputing genetic variants. **P. K. Albers et al.**	**42** Identification of a Sjögren's syndrome-associated variant that influences *OAS1* isoform switching. **H. Li et al.**	**51** Finding genes in animal models of histiocytic sarcoma. **E. A. Ostrander et al.**	**60** A genome-wide association study of alcohol dependence in the Irish affected sib pair study of alcohol dependence. **B. Riley et al.**	**69** Integrin modulating therapies prevent fibrosis and autoimmunity in genetic mouse models of scleroderma. **E. E. Gerber et al.**	**78** Individual gene disruptions from balanced chromosomal rearrangements define novel neurodevelopmental loci and genomic disorders. **H. Brand et al.**	**87** Efficacy of hematopoietic cell therapy in X-linked adrenoleukodystrophy: A multinstitutional study (ALD-101). **G. Raymond et al.**

	Hall B2, Level 0 (Lower Level)	Grand Ballroom East, Level 3	Grand Ballroom West, Level 3	Room 210, Level 2	Room 205, Level 2	Room 253, Level 2	Room 258, Level 2	Westin Grand Ballroom AB, Concourse Level	Westin Grand Ballroom CDE, Concourse Level
	SESSION 19 – Hereditary Cancer Syndromes Co-Moderators: Katherine Nathanson, Univ. of Pennsylvania; and Melissa Southey, Univ. of Melbourne	**SESSION 20 – Variants, Variants Everywhere** Co-Moderators: Gholson Lyon, Cold Spring Harbor Lab.; and Tuuli Lappalainen, Stanford Univ.	**SESSION 21 – Genetic Epidemiology: Applications and Methods** Co-Moderators: Jo Knight, Ctr. for Addiction and Ment. Hlth., Ontario; and Braxton D. Mitchell, Univ. of Maryland Sch. of Med., Baltimore	**SESSION 22 – Cardiovascular Genetics: Gene Discovery through GWAS and Sequencing** Co-Moderators: Paul W. Livermore Auer, Univ. of Wisconsin-Milwaukee; and Leslie Lange, Univ. of North Carolina at Chapel Hill	**SESSION 23 – From eQTLs to Epigenetics and Beyond** Co-Moderators: John M. Greally, Albert Einstein Col. of Med.; and Alexis Battle, Stanford Univ.	**SESSION 24 – Neurogenetics: Illuminating Mechanisms** Co-Moderators: Gary Beecham, Univ. of Miami; and Lindsay Farrer, Boston Univ.	**SESSION 25 – Genetic Interactions in Complex Traits** Co-Moderators: Qibin Qi, Albert Einstein Col. of Med.; and Peter Kraft, Harvard Sch. of Publ. Hlth.	**SESSION 26 – Advances in the Genetics of Skeletal and Morphologic Disorders** Co-Moderators: Charles Venditti, NHGRI/NIH; and Jamie Fitzgerald, Oregon Hlth. & Sci. Univ.	**SESSION 27 – Causes and Consequences of Chromosomal Variations** Co-Moderators: Beverly Emanuel, Univ. of Pennsylvania; and Seema Lalani, Baylor Col. of Med.
8:00	**88** Rare mutations in *RINT1* predispose carriers to early-onset breast cancer. **D. J. Park et al.**	**97** Frequency uniqueness score: Predicting the disease risk of coding variants. **A. C. Alexander et al.**	**106** The effect of CAD/MI SNPs on other vascular domains and the relation with recurrent vascular events. **V. Tragante et al.**	**115** Linkage analysis of hypertriglyceridemia in a single large family identifies 3 novel potentially pathogenic variants. **E. A. Rosenthal et al.**	**124** Interpreting eQTLs by linking enhancers to target genes. **J. Wang et al.**	**133** Unraveling the genetic architecture of multiple sclerosis and the underlying implicated pathways. **N. A. Patsopoulos.**	**142** Daylight exposure may modify the effect of variants at *MTNR1B* and *CRY2* on glucose tolerance: The GLACIER Study. **F. Renström et al.**	**151** Targeted capture and sequencing identifies causative alleles in simplex and multiplex consanguineous Palestinian families with orofacial clefts. **H. Shahin et al.**	**160** Identification of rare genetic variants in high-risk ASD families and their role in a large ASD case/control population. **C. Hensel et al.**
8:15	**89** The BER glycosylase *NEIL1* is a risk gene for familial breast cancer. **M. R. Dufault et al.**	**98** Exome-based linkage mapping and variant prioritization for inherited retinal disorders. **D. C. Koboldt et al.**	**107** Multi-trait meta-analysis of genome-wide association studies of lipid levels and BMI reveals pleiotropy. **V. Lagou et al.**	**116** Rare *APOC3* loss-of-function variants lower plasma triglycerides and protect against clinical coronary heart disease. **J. Crosby et al.**	**125** Genetic architecture of regulatory variation influencing response to human rhinovirus infection. **M. Caliskan et al.**	**134** Autosomal dominant congenital spinal muscular atrophy is caused by mutations in *BICD2*, a golgin and important motor adaptor. **B. Wirth et al.**	**143** *NAT1* in an important genetic effect modifier of tobacco smoke exposure in multiple sclerosis susceptibility in 5,453 individuals. **F. B. S. Briggs et al.**	**152** Dominant mutations in *GRHL3* cause Van der Woude syndrome and disrupt oral periderm development. **M. Peyrard et al.**	**161** Discovery of cryptic chromosomal abnormalities in clinically-referred youth with neuropsychiatric disorders. **V. Pillalamarri et al.**
8:30	**90** More than 25% of breast cancer families with wild-type results from commercial genetic testing of *BRCA1* and *BRCA2* are resolved by BROCA sequencing of all known breast cancer genes. **T. Walsh et al.**	**99** Integrative annotation of variants from 1,092 humans: Application to cancer genomics. **E. Khurana et al.**	**108** Genome-wide association with fasting glucose and insulin in 20,200 African Americans suggests new quantitative trait loci and allelic heterogeneity at known loci: The African American Glucose and Insulin Genetic Epidemiology (AAGILE) Consortium. **J. Meigs et al.**	**117** Novel rare and low frequency coding variants associated with LDL cholesterol levels. **C. Willer et al.**	**126** Genome-wide association of expression response of primary immune cells identifies novel cis and trans loci specific to distinct pathogen responses. **C. Ye et al.**	**135** Rare variants in restless legs syndrome. **E. C. Schulte et al.**	**144** Genome-wide joint meta-analysis for interaction between genetic variants and smoking on waist circumference. **A. E. Justice et al.**	**153** A homeotic maxillary to mandibular transformation in humans resulting from loss of selective ligand affinity of the endothelin receptor type A. **C. Gordon et al.**	**162** Mechanism, prevalence, and more severe neuropathy phenotype of the Charcot-Marie-Tooth, type 1A triplication. **V. Gelowani et al.**
8:45	**91** Nine genes for inherited predisposition to breast cancer among African American women. **O. I. Olopade et al.**	**100** Efficiency of whole exome/genome sequencing for achieving a diagnosis in rare presentations. **M. C. Towne et al.**	**109** Harnessing Web 2.0 social networks for genetic epidemiology studies with millions of people. **Y. Erlich et al.**	**118** Association analysis of C-reactive protein levels in European Americans and African Americans sequenced through the NHLBI Exome-Sequencing Project. **U. M. Schick et al.**	**127** Expression QTL analysis from primary immune cells of a multi-ethnic cohort identifies novel disease-causing regulatory effects. **T. Raj et al.**	**136** Mutations in *PNPLA6* cause a range of neurodegenerative phenotypes. **M. A. Gonzalez et al.**	**145** Interaction between genome-wide variants and physical activity on body mass index: A meta-analysis of 109,924 individuals. **T. O. Kilpeläinen et al.**	**154** Putative gain of function mutations in *FAM111A* result in Kenny-Caffey syndrome and osteocraniostenosis: Identification of an upstream regulator of the PTH axis. **S. Unger et al.**	**163** Whole genome sequencing of two individuals with excessive numbers of de novo CNVs. **P. Liu et al.**
9:00	**92** Germline loss-of-function mutations in 15 different DNA repair genes are present in 22% of 1412 patients with ovarian, peritoneal or fallopian tube cancers not selected for age at diagnosis or family history. **M. I. Harrell et al.**	**101** Computational prediction and in vivo validation of suppressors of human disease mutations. **D. M. Jordan et al.**	**110** Integrated model of multiple types of rare variants and prior information improves the power of detecting risk genes for autism. **X. He et al.**	**119** Genetic association studies illuminate the role of low frequency and rare variation in explaining the variation of blood pressure traits. **A. Manning et al.**	**128** Allele specific expression analysis using transcriptome sequencing in three tissues of a twin cohort reveals large effect of gene-by-gene and gene-by-environment interactions. **A. Buil et al.**	**137** Targeted resequencing of 101 known and candidate epilepsy genes in 600 patients with severe epilepsies identifies recurrently mutated genes. **G. L. Carvill et al.**	**146** Genome-wide analyses highlights gene interaction with processed meat and vegetable intake for colorectal cancer risk. **J. Figueiredo et al.**	**155** Mutations in *PIEZO2* cause Gordon syndrome, Marden-Walker syndrome and distal arthrogryposis type 5. **M. J. McMillin et al.**	**164** Utilization of next-generation sequencing to detect and assign pathogenicity to balanced rearrangements identified by conventional cytogenetics. **U. Aypar et al.**
9:15	**93** The Mainstreaming Cancer Genetics Programme — Integrating genetic testing into routine clinical practice in the United Kingdom. **N. Rahman et al.**	**102** The empowered whole genome cohort: Shareable joint genome interpretation for research and personal insight. **N. M. Pearson et al.**	**111** Tests of aggregate rare variant association applied to a multiethnic sequencing study. **A. D. Ablorh et al.**	**120** Contribution of coding variation to type 2 diabetes-related quantitative traits in 13,000 exomes from multiple ancestries. **X. Sim et al.**	**129** Epigenomic variation between species, tissues, populations and individuals. **A. Kundaje et al.**	**138** A mouse model of Kabuki syndrome demonstrates defective hippocampal neurogenesis rescued with treatment with AR-42, a histone deacetylase inhibitor. **H. T. Bjornsson et al.**	**147** Genome wide gene-environment interaction study identifies a *CYP24A1*-related variant as a modifier of colorectal cancer risk associated with menopausal estrogen plus progesterone therapy. **X. Garcia-Albeniz et al.**	**156** Mutation in the SH2 domain of *PIK3R1* cause SHORT syndrome with partial lipodystrophy. **S. Johansson et al.**	**165** Sequencing of unbalanced translocation junctions reveals mutational mechanisms and gene fusions. **B. Weckselblatt et al.**
9:30	**94** Identification of a second major locus predisposing to an autosomal dominant inherited disorder of multiple schwannomas. **L. Messiaen et al.**	**103** Understanding molecular mechanisms of human disease mutations and coding variants through 3D protein networks. **H. Yu et al.**	**112** Whole-genome sequence based association studies of complex traits: The UK10K project. **N. Timpson.**	**121** A meta-analysis of genome-wide association studies identifies a novel locus associated with thrombin generation potential. **A. Rocañín-Arjó et al.**	**130** Predicting genome-wide DNA methylation using methylation marks, genomic position and DNA regulatory elements. **W. Zhang et al.**	**139** Human iPSC-based models of neuronal ceroid lipofuscinosis capture progressive pre-storage pathology in multiple cellular compartments. **J. F. Staropoli et al.**	**148** Replication of gene-gene interaction models associated with cataracts in the eMERGE Network. **M. A. Hall et al.**	**157** *MAP4* defect underlines centrosomal organization as a central mechanism in growth regulation. **C. T. Thiel et al.**	**166** Identification of a deletion in the *LRP1b* gene associated with megalencephaly in the sudden infant death syndrome. **D. S. Paterson et al.**
9:45	**95** Identification of putative driver mutations in neurofibromatosis type 1-associated plexiform neurofibromas. **A. Pemov et al.**	**104** Evaluation of power of the Illumina HumanOmni5M-4v1 BeadChip to detect risk variants for human complex diseases. **C. Xing et al.**	**113** Genetic variation associated with the susceptibility to herpes zoster in the eMERGE Network. **D. Crosslin et al.**	**122** Genome-wide association analysis of blood pressure traits in nearly 30,000 African ancestry individuals reveals a common set of associated genes in African and non-African populations. **N. Franceschini et al.**	**131** An ENU mutagenesis screen identifies the first mouse mutants of a novel epigenetic modifier, rearranged L-Myc fusion (*Rlf*). **S. K. Harten et al.**	**140** Mutation in *EZR* inhibits the Ras/MAP pathway and causes autosomal recessive intellectual disability. **R. Abou Jamra et al.**	**149** Evidence from multiple genome-wide association studies of a hub of gene-gene interactions affecting human HDL cholesterol levels. **L. Ma et al.**	**158** Best understanding of structural and functional impact of *FGFR3* mutations at the same position (K650N, K650M, K650E) leading to both mild and lethal dwarfism. **D. Komla Ebri et al.**	**167** A comprehensive microarray prenatal study: Efficacy for both copy number and copy neutral changes. **S. Schwartz et al.**
10:00	**96** Somatic structural and rare germline variation in childhood cancers. **D. I. Ritter et al.**	**105** Integrated analysis of protein-coding variation in over 50,000 individuals. **M. Lek et al.**	**114** Whole-genome detection of disease-associated deletions or excess homozygosity in a case-control study of rheumatoid arthritis. **C. C. Wu et al.**	**123** Gene pathway burden test application to cardiovascular disease using whole genome sequencing data. **M. A. A. Almeida et al.**	**132** Zebrafish Mutation Project: Functional genomics of disease. **E. M. Busch-Nentwich et al.**	**141** De novo mutations in the genome organizer *CTCF* cause intellectual disability. **C. Zweier et al.**	**150** Epistasis analysis for quantitative trait with next-generation sequencing data. **F. Zhang et al.**	**159** X-linked osteoporosis and fractures: An unexpected genetic cause. **F. S. van Dijk et al.**	**168** Non-random, locus-specific differences in DNA accessibility are present in homologous metaphase chromosomes. **W. A. Khan et al.**

Thursday, October 24: Concurrent Platform (abstract-driven) Session C (28-36)

2:00 PM–4:15 PM

	Hall B2, Level 0 (Lower Level)	Grand Ballroom East, Level 3	Grand Ballroom West, Level 3	Room 210, Level 2	Room 205, Level 2	Room 253, Level 2	Room 258, Level 2	Westin Grand Ballroom AB, Concourse Level	Westin Grand Ballroom CDE, Concourse Level
	SESSION 28 – Low Frequency Variants for Complex Traits Co-Moderators: Benjamin Voight, Univ. of Pennsylvania; and Ines Barroso, Wellcome Trust Sanger Inst.	**SESSION 29 – Selection, Demography and Functional Polymorphism** Co-Moderators: Xiaoming Liu, Univ. of Texas Hlth. Sci. Ctr. at Houston; and Adam Siepel, Cornell Univ.	**SESSION 30 – Statistical Methods for Family Data** Co-Moderators: Shamil Sunyaev, Brigham and Women's Hosp.; and Nathan L. Tintle, Dordt Col., IA	**SESSION 31 – Advances and References in Genomic Technology** Co-Moderators: Daniel G. MacArthur, Massachusetts Gen. Hosp./Broad Inst.; and Fuli Yu, Baylor Col. of Med.	**SESSION 32 – Genetic Testing for Neurodevelopmental Disease: Genotype: Phenotype Challenges** Co-Moderators: Miriam H. Meisler, Univ. of Michigan; and David R. Adams, NHGRI/NIH	**SESSION 33 – Gene Regulation — At a Multitude of Levels** Co-Moderators: Michael Zwick, Emory Univ.; and Sara Wheelan, Johns Hopkins Univ.	**SESSION 34 – Cardiovascular Genetics: Exome Sequencing and Animal Models** Co-Moderators: Xiaofeng Zhu, Case Western Reserve Univ.; and Bart L. Loeys, Univ. of Antwerp	**SESSION 35 – Genomic Medicine: Counseling, Education and Health Services** Co-Moderators: Wendy Uhlmann, Univ. of Michigan; and Katherine Kim, Lurie Children's Hosp. of Chicago	**SESSION 36 – Biochemical and Clinical Consequences of Mitochondrial Dysfunction** Co-Moderators: Nancy E. Braverman, McGill Univ.; and Devin Oglesbee, Mayo Clin.
2:00	**169** Rare variants contributing to age-related macular degeneration — Results from the International AMD Genomics Consortium. **W. M. Igl.**	**178** A role for host-bacteria interactions in shaping patterns of genetic variation across human populations. **R. Blekhman et al.**	**187** Adjusting family relatedness in data-driven burden test of rare variants. **Q. Zhang et al.**	**196** Statistical model for the joint estimation of mRNA isoforms and individual-specific expression from RNA-seq data. **F. Mordelet et al.**	**205** Clinical experience implementing chromosomal microarray analysis in a clinical psychiatric practice for adults with autism spectrum disorders and related neurodevelopmental disorders. **K. B. Teed et al.**	**214** The evolutionary dynamics of regulatory DNA in the mouse and human genomes. **J. Vierstra et al.**	**223** Identifying multiple causative genes at a single GWAS locus. **M. Flister et al.**	**232** Reasons why patients decline whole genome sequencing in the MedSeq Project. **D. M. Lautenbach et al.**	**241** Phenotype and genotype in 17 patients with succinate-CoA ligase deficiency caused by mutations in *SUCLA2* and *SUCLG1*. **E. Oestergaard et al.**
2:15	**170** Identification of new rare coding variants associated with hemoglobin levels and platelet counts. **P. Livermore Auer et al.**	**179** Using ancient genomes to detect positive selection on the human lineage. **K. Prüfer et al.**	**188** Fast and accurate pedigree-based imputation from sequenced data in a founder population. **O. E. Livne et al.**	**197** Choosing an RNA-seq aligner for QTL and ASE analysis in the Genotype-Tissue Expression Project. **D. S. DeLuca et al.**	**206** New insights into the spectrum of pathogenic variation in epilepsy gained from molecular diagnostic testing of 1600 individuals. **S. Aradhya et al.**	**215** Widespread exonic transcription factor binding directs codon usage and protein evolution. **A. B. Stergachis et al.**	**224** Gene silencing and haploinsufficiency of *Csk* in GWAS locus 15q24 increase blood pressure. **B. Oh et al.**	**233** How do research participants perceive "uncertainty" in genomic sequencing? **B. Biesecker et al.**	**242** Restoration of the mitochondrial citrate transporter by overexpression of *SLC25A1* in primary deficient fibroblasts of patients with combined D-2- and L-2-hydroxyglutaric aciduria. **G. S. Salomons et al.**
2:30	**171** Large-scale whole genome sequencing study for bone mineral density: The UK10K Consortium. **H. Zheng et al.**	**180** Reference sample guided pooled sequencing identifies loss-of-function patterns across human populations. **A. Eran et al.**	**189** Multiple genetic variant association testing by collapsing and kernel methods with pedigree or population structured data. **D. J. Schaid et al.**	**198** mRNA and small RNA sequencing of 465 HapMap cell lines: The feasibility of multicenter RNA-seq studies. **P. A. C. Hoen et al.**	**207** Pathogenic rare copy number variants in community-based schizophrenia suggest a potential role for clinical microarrays. **A. S. Bassett et al.**	**216** Short tandem repeat polymorphisms create an abundant source of expression variability. **M. Gymrek et al.**	**225** A novel genetic basis for systemic vasculitis: Systemic and cutaneous polyarteritis nodosa are caused by recessive mutations in an immune-related gene. **R. Segel et al.**	**234** Factors influencing healthcare utilization in response to personal genetic testing. **S. S. Kalia et al.**	**243** Unprocessed RNA intermediates interfere with mitochondrial translation and cause respiratory chain deficiency. **R. Kopajtich et al.**
2:45	**172** Identification of 6 novel loci associated with amino acid levels using single-variant and gene-based tests. **T. M. Teslovich et al.**	**181** Patterns of IBD sharing inferred from whole genome sequences of 962 European Americans. **C. V. Van Hout et al.**	**190** Evidence for causality of rare variants based on exact sharing probabilities in affected relatives. **I. Ruczinski et al.**	**199** Complete resequencing of extended genomic regions using fosmid targeting and PacBio's single molecule real-time (SMRT®) long-read sequencing technology. **D. E. Geraghty et al.**	**208** Genetic assessment of congenital brain malformations. **U. Hehr et al.**	**217** RNA-DNA sequence differences occur within seconds following RNA exit PolII active sites and are responsive to cellular stress. **V. G. Cheung et al.**	**226** *BMP9* mutations cause a vascular anomaly syndrome with phenotypic overlap with hereditary hemorrhagic telangiectasia. **J. McDonald et al.**	**235** Does personal genome testing drive service utilization in an adult preventive medicine clinic? **N. Hoang et al.**	**244** Mutations in the cytochrome c1 subunit of respiratory chain complex III cause insulin-responsive hyperglycemia and recurrent ketoacidosis. **J. Christodoulou et al.**
3:00	**173** Exome analysis in 65,653 European samples identifies novel low-frequency and common variants for type 2 diabetes. **C. Fuchsberger et al.**	**182** Reconstructing the genetic demography of the United States. **R. Sebro et al.**	**191** A generalized sparse regression model with adjustment of pedigree structure for variant detection from next-generation sequencing data. **S. Cao et al.**	**200** Platinum genomes: A systematic assessment of variant accuracy using a large family pedigree. **M. A. Eberle et al.**	**209** Investigation of *CASK* gene aberrations in 38 patients with severe intellectual disability, microcephaly and disproportionate pontine and cerebellar hypoplasia. **S. Hayashi et al.**	**218** RNA-seq transcriptome profiling uncovers how structural variants influence alternative splicing. **E. Ait Yahya Graison et al.**	**227** Identification of a novel cause of X-linked heterotaxy. **M. Tariq et al.**	**236** Opportunity and cost of clinical whole genome sequencing. **F. Dewey et al.**	**245** Mutations in *FBXL4* cause mitochondrial encephalopathy and a disorder of mitochondrial DNA maintenance. **R. W. Taylor et al.**
3:15	**174** Loss of function mutations in *SLC30A8* protect against type 2 diabetes. **J. Flannick et al.**	**183** Assessing functional potential along the human genome by integrating comparative, population, and functional genomic data. **I. Gronau et al.**	**192** Haplotype phasing across the full spectrum of relatedness. **J. O'Connell et al.**	**201** Sensitive and quantitative measurement of nuclease-mediated genome editing at human endogenous loci using SMRT sequencing. **A. Hendel et al.**	**210** A homozygous *PDE6D* mutation in Joubert syndrome impairs targeting of farnesylated INPP5E protein to the primary cilium. **S. Thomas et al.**	**219** Exploring regulatory and loss-of-function variation in personalized multi-tissue transcriptomes using allele-specific expression. **T. Lappalainen et al.**	**228** Novel and recurrent gain-of-function mutation in *PRKG1* causes thoracic aortic aneurysms and acute aortic dissections. **D. Guo et al.**	**237** Cost-effectiveness analysis of next-generation sequencing in etiologic evaluations for prelingual hearing loss. **M. E. Nunes et al.**	**246** A lipomatosis endophenotype in methylmalonic acidemia: Evidence from patients and mice. **I. Manoli et al.**
3:30	**175** Whole genome sequencing of 2,850 Central-Northern European type 2 diabetes cases and controls reveals insights into functional mechanisms underlying disease pathogenesis. **K. Gaulton et al.**	**184** Genome-wide analysis of cold adaption in indigenous Siberian populations. **A. Cardona et al.**	**193** The theory of genetic interactions and its application to the problem of missing heritability. **A. Young et al.**	**202** Mining genomic feature sets and identifying significant biological relationships with BedTools2. **A. Quinlan et al.**	**211** Assessment of incidental findings in whole exome sequences from the Baylor-Hopkins Center for Mendelian Genomics. **J. Jurgens et al.**	**220** Analysis of the genetic variation and age interplay on gene expression using RNA-seq data from multiple tissues. **A. Viñuela et al.**	**229** Mutations in the *DCHS1* gene cause mitral valve prolapse in humans. **R. Durst et al.**	**238** Documentation of medical decision-making for genetic testing in the health record. **M. T. Scheuner et al.**	**247** Moonlighting in mitochondria: *ACAD9* plays a dual role in energy metabolism. **M. Schiff et al.**
3:45	**176** Exome chip scan of 74,000 subjects of European descent and 18,000 subjects of African descent identify novel genes with functional mutations influencing adiposity traits. **I. B. Borecki.**	**185** Genetic variation is a major source of transcriptional variation in human-induced pluripotent stem cells. **N. Kumasaka et al.**	**194** Tracing individual ancestry in a principal components space. **C. Wang et al.**	**203** Creating a single haplotype human genome assembly. **T. Graves et al.**	**212** Deep sequencing in extended pedigrees reveals a major rare non-synonymous variant influencing the de novo ceramide synthesis pathway. **J. E. Curran et al.**	**221** Transcriptomes of individual cells. **C. Borel et al.**	**230** Robust epistasis between the genes encoding a TGFβ effector and its regulatory microRNA governs modification of cardiovascular phenotypes in TGFβ vasculopathies. **J. Calderon et al.**	**239** Medical genetics and genomics: Parallel revolutions in science and undergraduate medical education. **S. Dasgupta et al.**	**248** Primary ovarian insufficiency is caused by recessive partial loss-of-function mutations in genes for mitochondrial protein homeostasis. **S. B. Pierce et al.**
4:00	**177** Large duplications are associated with increased risk of obesity. **J. S. El-Sayed Moustafa et al.**	**186** Using enhancer activity regulatory motifs to explore evolutionary trajectories and disease mechanisms. **L. D. Ward et al.**	**195** Multiple HLA loci and energy metabolism genes are targeted by recent positive selection in an Ethiopian population. **F. Tekola-Ayele et al.**	**204** A generalized human reference as a graph of genomic variation. **E. Garrison et al.**	**213** From embryonic lethal to no phenotype: What autozygome can teach us about loss of function in the human genome. **F. S. Alkuraya.**	**222** A low-frequency variant in a lincRNA doubles the risk of pneumococcal bacteraemia in Kenyan children. **A. Rautanen et al.**	**231** ERK activation unifies deleterious gene-by-gene and gene-by-environment interactions in Marfan syndrome. **J. J. Doyle et al.**	**240** Genomic medicine in primary care: Views of Ontario family physicians. **J. Carroll et al.**	**249** mtDNA mutations variously impact mtDNA maintenance throughout the human embryo/fetal development. **S. Rondeau et al.**

Thursday, October 24: Concurrent Invited Session II (37-43)

4:30 PM–6:30 PM

Grand Ballroom West, Level 3	Room 258, Level 2	Grand Ballroom East, Level 3	Room 210, Level 2	Hall B2, Level 0 (Lower Level)	Room 205, Level 2	Room 253, Level 2
SESSION 37 – Community Efforts to Decipher the Phenotypic Impact of Genomic Variation Co-Moderators: David H. Ledbetter, Geisinger Hlth. Syst., Danville, PA; and Joyce A. Mitchell, Univ. of Utah	**SESSION 38 – Genetics of Non-communicable Diseases in sub-Saharan Africa** Co-Moderators: Eleftheria Zeggini, Wellcome Trust Sanger Inst., Hinxton, U.K.; and Adebowale Adeyemo, NHGRI/NIH	**SESSION 39 – Guilty by Annotation: The Role of Non-coding Variation in Phenotypic Variation and Disease** Co-Moderators: Stephen B. Montgomery, Stanford Univ.; and Emmanouil T. Dermitzakis, Univ. of Geneva	**SESSION 40 – Human Genetics of Common Infectious Diseases** Co-Moderators: Laurent Abel, INSERM/ Univ. Paris Descartes; and Erwin Schurr, McGill Univ.	**SESSION 41 – Informed Consent for Whole Genome Sequencing: Experience and Implications for Practice** Co-Moderators: Stephanie M. Fullerton, Univ. of Washington Sch. of Med.; and Holly K. Tabor, Seattle Children's Hosp.	**SESSION 42 – Multimodal Treatment of Lysosomal Storage Diseases as a Portal to Emergent Genetic Therapies** Moderator: Ari Zimran, Shaare Zedek Med. Ctr., Jerusalem	**SESSION 43 – Nonhuman Primate Genomics: Evolutionary Insights and Relevance to Human Phenotypes** Co-Moderators: Jeffrey M. Kidd, Univ. of Michigan; and Tomas Marques-Bonet, CSIC-Univ. Pompeu Fabra, Barcelona
04:30 pm Developing approaches to support the community in the evaluation, deposition and curation of genomic variants. **C. L. Martin.**	04:30 pm The African Genome Variation Project. **F. Tekola-Ayele.**	04:30 pm Finding causal regulatory variants with genome and transcriptome sequencing. **E. T. Dermitzakis.**	04:30 pm Immunogenetic variation characterizing exceptional control of HIV. **M. Carrington.**	04:30 pm Research goals and informed consent in clinical genomics research. **J. C. Sapp.**	04:30 pm Enzyme replacement therapy. **G. A. Grabowski.**	04:30 pm Great ape versus human genetic diversity: The Great Ape Genome Project. **T. Marques-Bonet.**
05:00 pm Assessing the evidence for causality of sequence variants: Establishing community standards. **D. MacArthur.**	05:00 pm Developing genomic research in Africa: The case for sickle cell disease. **J. Makani.**	05:00 pm Regulatory genomics and epigenomics of complex disease genetics for fine-mapping and genome-wide integration. **M. Kellis.**	05:00 pm Human genetics and the risk of infectious diseases. **D. B. Goldstein.**	05:00 pm Informed consent and its role in eliciting result return preferences. **W. K. Chung.**	05:00 pm Substrate reduction therapy. **G. Pastores.**	05:00 pm Where ancestry runs deep: Ancient balancing selection in humans. **M. Przeworski.**
05:30 pm Developing standards to represent human phenotypes. **A. Hamosh.**	05:30 pm Discovering podoconiosis susceptibility genes: From molecules to disease control for a neglected tropical disease. **M. Newport.**	05:30 pm Functional analysis of polymorphisms identified using genome-wide association studies. **J. Taipale.**	05:30 pm Human genetics of leprosy: novel insights. **E. Schurr.**	05:30 pm Consent, assent, and WGS studies of children. **I. A. Holm.**	05:30 pm Pharmacologic chaperones. **O. Goker-Alpan.**	05:30 pm Genomic insights into chromosomal evolution in Gibbons. **L. Carbone.**
06:00 pm Using ClinVar as a resource to evaluate genomic variation: A clinical laboratory›s perspective. **S. J. Bale.**	06:00 pm Genomic studies of cardiometabolic traits in sub-Saharan Africa. **M. Sandhu.**	06:00 pm Interpreting loss-of-function variation and complex disease association using RNA-seq and ChIP-seq data. **S. B. Montgomery.**	06:00 pm Human genetic studies on tuberculosis and malaria. **R. Horstmann.**	06:00 pm Informed consent for large-scale clinical mutation testing: Anticipating the future. **R. R. Sharp.**	06:00 pm Future perspectives. **D. Elstein.**	06:00 pm Mapping complex traits in non-human primates. **N. Freimer.**

Thursday, October 24
6:45 PM–7:45 PM

SESSION 44 – ASHG Next: The Future of Genetics and the Future of Your Society

Room 253, Level 2, Convention Center

Moderators:
Jeff Murray, Univ. of Iowa
Cynthia C. Morton, Brigham and Women's Hosp

ASHG is currently engaged in strategic planning that will guide the Society for the next three to five years. Join fellow ASHG members, the elected leadership, and the staff to help chart the future of your professional society. Come prepared to discuss the role of ASHG in the future of human genetics and genetic medicine and to provide your thoughts on the annual meeting, the *American Journal of Human Genetics*, and new services for members.

This session will be an interactive session. Be sure to bring your fully-charged mobile device, with the audience response app downloaded. In advance of the session, go to the iTunes store, Android Market or Blackberry Market to download the ResponseWare app or the QR code provided below. You can also go to www.rwpoll.com to participate. Enler Session ID: ASHG Keypads are available to those without a mobile device. Light refreshments will be available.

Visit the Exhibits and Posters

Wednesday: 10:00 am – 6:00 pm

Thursday: 10:00 am – 4:30 pm

Friday: 10:00 am – 2:30 pm

Friday, October 25: Concurrent Platform (abstract-driven) Session D (45-53)

8:00 AM–10:15 AM

	Hall B2, Level 0 (Lower Level)	Grand Ballroom East, Level 3	Grand Ballroom West, Level 3	Room 210, Level 2	Room 205, Level 2	Room 253, Level 2	Room 258, Level 2	Westin Grand Ballroom AB, Concourse Level	Westin Grand Ballroom CDE, Concourse Level
	SESSION 45 – Mo' Data, Mo' Problems? Co-Moderators: Luke Jostins, Oxford Univ.; and Christopher Brown, Univ. of Pennsylvania	**SESSION 46 – Cancer Genomics** Co-Moderators: Michael Rossi, Emory Univ.; and Jun Z. Li, Univ. of Michigan	**SESSION 47 – Demography In and Out of Africa** Co-Moderators: Jeffrey M. Kidd, Univ. of Michigan; and Philip Awadalla, Univ. of Montreal	**SESSION 48 – Fine-Mapping and Function of Candidate Loci** Co-Moderators: Paivi Pajukanta, UCLA; and Soumya Raychaudhuri, Brigham and Women's Hosp.	**SESSION 49 – New Genes and Disorders** Co-Moderators: Howard P. Levy, Johns Hopkins Univ.; and David Sweetser, Massachusetts Gen. Hosp.	**SESSION 50 – Neurodegenerative Disease and the Aging Brain** Co-Moderators: Jake McCauley, Univ of Miami; and Christine Van Broeckhoven, Univ. of Antwerp	**SESSION 51 – Epigenetics: From Genomes to Genes** Co-Moderators: Erica Davis, Duke Univ.; and Hans Bjornsson, Johns Hopkins Univ. Sch. of Med.	**SESSION 52 – New Frontiers in Pharmacogenetics** Co-Moderators: Joshua Lewis, Univ. of Maryland, Baltimore; and Eileen Dolan, Univ. of Chicago	**SESSION 53 – Genomic Approaches for Study of Rare Neurogenetic Disorders** Co-Moderators: Brunhilde Wirth, Univ. Hosp. Cologne; and Margit Burmeister, Univ. of Michigan
8:00	**250** Selecting likely causal genes, pathways and relevant tissues from genome-wide association studies of complex traits by data-driven expression-prioritized integration. **T. H. Pers et al.**	**259** Network analysis of mutations across cancer types. **M. D. M. Leiserson et al.**	**268** Simultaneous estimation of population size changes and splits times from population level resequencing studies. **M. Forest et al.**	**277** Fine mapping of the MHC in >60,000 samples by the International IBD Genetics Consortium: Identification of multiple predisposing and protective variants that are mostly distinct between Crohn's disease and ulcerative colitis. **P. Goyette.**	**286** A dominant-negative *GFI1B* mutation causes autosomal dominant gray platelet syndrome. **L. Van Laer et al.**	**295** Rare highly penetrant variants of late onset Alzheimers disease. **J. Rehker et al.**	**304** Epigenome-wide association studies in the era of meta-epigenomics. **J. M. Greally et al.**	**313** A genome-wide meta-analysis of the response to inhaled bronchodilators among subjects with chronic obstructive pulmonary disease. **M. Hardin et al.**	**322** A novel mitochondrial *SLC25A* gene causes CMT and optic atrophy. **A. J. Abrams et al.**
8:15	**251** Non-targeted metabolite profiling in large human population-based studies: A new data analysis workflow and metabolome-wide association study of C-reactive protein. **A. Ganna et al.**	**260** The landscape of tumor suppressors in primary tumors. **P. Van Loo et al.**	**269** Inferring complex demographies from PSMC coalescent rate estimates: African substructure and the Out-of-Africa event. **S. Gopalakrishnan et al.**	**278** Mapping the shared and distinct HLA alleles for seropositive and seronegative rheumatoid arthritis. **B. Han et al.**	**287** Identification of disease causing mutations in a new congenital neutrophil defect syndrome. **T. Vilboux et al.**	**296** The identification of high-penetrance variants in late-onset Alzheimer disease by whole exome sequencing in extended families. **M. A. Kohli et al.**	**305** Genome-wide DNA methylation analysis of uniparental disomy cases reveals many novel imprinted loci in the human genome. **R. S. Joshi et al.**	**314** Genome-wide association study of opioid-induced vomiting in the 23andMe cohort. **J. L. Mountain et al.**	**323** Molecular defects in the motor adaptor BICD2 cause proximal spinal muscular atrophy with autosomal-dominant inheritance. **K. Peeters et al.**
8:30	**252** Transcription factor and chromatin features predict genes associated with eQTLs. **D. Y. Wang et al.**	**261** Recurrent somatic mutation altering DNA-binding motif of transcription factor YY1 explains pathogenesis of insulin-producing adenomas. **M. K. Cromer et al.**	**270** Out of Africa, which way? **L. Pagani et al.**	**279** Common genetic variants of autoimmunity confer susceptibility to candididemia. **V. Magadi Gopalaiah et al.**	**288** A functional variant in the *CFI* gene confers a high risk of age-related macular degeneration. **A. den Hollander et al.**	**297** Functional rare genetic variation in Alzheimer's disease: An exome-wide association study in the CHARGE consortium. **J. Jakobsdottir et al.**	**306** Correlation between CpG DNA methylation levels in peripheral $CD4^+$ T cells and brain in aging individuals. **C. M. McCabe et al.**	**315** Transcriptome profiling of human airway smooth muscle cells stimulated with dexamethasone identifies *CRISPLD2* as a regulator of steroid and immune response. **B. E. Himes et al.**	**324** *AMPD2* regulates de novo GTP synthesis and is mutated in a new form of pontocerebellar hypoplasia. **V. Cantagrel et al.**
8:45	**253** Combining regulatory domain and genetic variation information to identify cell types, regulatory elements, and causal genetic variants that influence human disease. **E. Schmidt et al.**	**262** Somatic L1 retrotransposition occurs early during colorectal tumorigenesis. **S. Solyom et al.**	**271** Insights into the genetic architecture of African genomes: The African Genome Variation Project. **I. Tachmazidou.**	**280** PXK and Lupus: Defining novel immunobiology for an SLE risk gene. **S. E. Vaughn et al.**	**289** Recurrent genomic mutation *507delT* in three lipoid proteinosis (Urbach-Wiethe) pedigrees from central Iran. **L. Youssefian et al.**	**298** Whole exome sequencing in early-onset Alzheimer disease families identifies rare variants in multiple Alzheimer-related genes and processes. **B. W. Kunkle et al.**	**307** Genome-wide analysis of Mecp2 dependent DNA methylation and hydroxymethylation at base-resolution in neurons. **K. E. Szulwach et al.**	**316** Potential of integrating human genetics and electronic medical records for drug discovery: The example of *TYK2* and rheumatoid arthritis. **D. Diogo et al.**	**325** Truncating mutations of *MAGEL2* cause autism and Prader-Willi syndrome-like phenotypes. **C. P. Schaaf et al.**
9:00	**254** Genome-wide expression quantitative trait loci: Results from the NHLBI's SABRe CVD Initiative. **R. Joehanes et al.**	**263** Transcriptome sequence analysis of human colorectal cancer samples reveals cancer functional attributes. **H. Ongen et al.**	**272** Genetic evidence for multiple episodes of population mixture in southern and eastern African history. **J. Pickrell et al.**	**281** Allelic heterogeneity of and interactions between polymorphic RET enhancers affecting Hirschsprung disease risk. **S. Chatterjee et al.**	**290** The neuronal endopeptidase *ECEL1* is a frequent cause of autosomal recessive distal arthrogryposis associated with limited knee flexion, ptosis, and limb muscle and tongue atrophy. **K. Dieterich et al.**	**299** Integrated whole transcriptome and DNA methylation analysis identifies new gene network in Alzheimer disease. **C. E. Humphries et al.**	**308** Random replication of the inactive X chromosome. **A. Koren et al.**	**317** Rare variants contribute to bronchodilator drug response in Latino children with asthma. **D. G. Torgerson et al.**	**326** Defective ubiquitination underlies oligogenic cerebellar degeneration and reproductive endocrine axis defects. **M. Kousi et al.**
9:15	**255** A hierarchical multiscale model to infer transcription factor occupancy from chromatin accessibility data. **A. Raj et al.**	**264** Comparative whole genome sequencing to identify candidate somatic driver mutations of Li-Fraumeni syndrome sarcomagenesis in humans and mice. **J. Wong et al.**	**273** Inferring the evolutionary history and the genetic basis of small stature in African Pygmies from whole-genome sequencing data. **M. Sikora et al.**	**282** Identification of human craniofacial, thyroid and heart enhancers at the FOXE1 locus. **A. C. Lidral et al.**	**291** Expanding molecular basis for rasopathies: A new player? **M. Ludwig et al.**	**300** Novel mutations uncovered from exome sequencing of Norwegian families with Parkinson's disease. **M. Lin et al.**	**309** The epigenetic profile of the *SOX9* regulatory region appears Y chromosome dependent. **G. Houge et al.**	**318** Characterization of statin dose-response within electronic medical records. **W. Q. Wei et al.**	**327** Periventricular heterotopia in 6q terminal deletion syndrome: Role of the *C6orf70* gene. **V. Conti et al.**
9:30	**256** Development of a methods-based proficiency test for next-generation sequencing. **N. Aziz et al.**	**265** Comprehensive transcriptome and epigenome sequencing of hypoxic breast cancer reveals non-coding RNAs associated with clinicopathological features. **H. Choudhry et al.**	**274** Reconstructing the population genetic history of the Caribbean. **A. Moreno Estrada et al.**	**283** Irf6 homeostasis is required for neurulation through a direct interaction with Tfap2a. **Y. A. Kousa et al.**	**292** *MC3R* modifies CF lung disease by increasing the level of CFTR. **J. Park et al.**	**301** Gene-environment interaction reveals hidden heritability: Plasma vitamin D concentration and its interaction with vitamin D receptor gene polymorphisms in Parkinson disease. **L. Wang et al.**	**310** Global reduction of 5-hydroxymethylcytosine in a *FMR1* premutation mice model. **B. Yao et al.**	**319** Genome-wide analysis of creatine kinase levels in statin-users. **M. P. Dubé et al.**	**328** A gene implicated in the neurobehavioural abnormalities of Williams-Beuren syndrome, *GTF2IRD1*, encodes a novel epigenetic regulator. **P. Carmona-Mora et al.**
9:45	**257** An integrated nexus of >12,000 genome sequences and analysis tools facilitates novel gene discovery. **J. Reid et al.**	**266** Addressing the complexity of cancer: Integrative genomic and transcriptomic analysis of 775 human cancer cell line reveals novel drivers and regulatory programs. **A. C. Villani et al.**	**275** Selective interference driven by variable recombination impacts mutation load in humans. **J. Hussin et al.**	**284** Deletion of a distant-acting enhancer on Chr16p13.3 causes recessive intractable diarrhea of infancy syndrome (IDIS). **D. Oz-Levi et al.**	**293** Combined exome and whole-genome sequencing identifies mutations in *ARMC4* as a cause of primary ciliary dyskinesia. **A. Onoufriadis et al.**	**302** Genetic variants in longevity gene *KLOTHO* are associated with increased brain volumes in aging. **J. S. Yokoyama et al.**	**311** Coordination of engineered factors with TET1/2 promotes early stage epigenetic modification during somatic cell reprogramming. **Y. Li et al.**	**320** The return of pharmacogenomic variants in the MedSeq Project: Reporting approach and physician response. **J. B. Krier et al.**	**329** Targeted high-throughput sequencing of 220 genes identifies a high proportion of causative mutations in over 80 patients with undiagnosed intellectual disability. **C. Redin et al.**
10:00	**258** Pulling out the 1%: Whole-genome in-solution capture for the targeted enrichment of ancient DNA sequencing libraries. **C. D. Bustamante et al.**	**267** Diagnostic yield of clinical tumor exome sequencing for newly diagnosed pediatric solid tumor patients. **D. W. Parsons et al.**	**276** Human population assembly and error-correction of sequence reads. **Z. Iqbal et al.**	**285** The role of *SIX6* in primary open-angle glaucoma. **M. Ulmer et al.**	**294** Identification of novel molecular defects in chronic intestinal pseudo-obstruction. **G. Romeo et al.**	**303** ENIGMA2: Genome-wide scans of subcortical brain volumes in 16,125 subjects from 28 cohorts. **S. Medland.**	**312** *SMCHD1* mutations in facioscapulohumeral muscular dystrophy type 2. **R. J. L. F. Lemmers et al.**	**321** Genetic evidence improves chances of drug discovery success. **M. R. Nelson et al.**	**330** Genetic analysis and new gene discovery in nemaline myopathy. **V. A. Gupta et al.**

Friday, October 25: Concurrent Platform (abstract-driven) Session E (54-62)

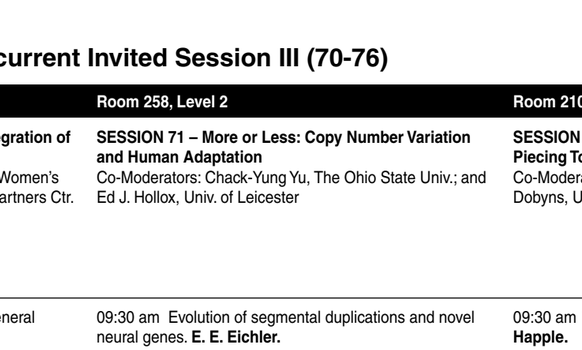
2:00 PM–4:15 PM

	Hall B2, Level 0 (Lower Level)	Grand Ballroom East, Level 3	Grand Ballroom West, Level 3	Room 210, Level 2	Room 205, Level 2	Room 253, Level 2	Room 258, Level 2	Westin Grand Ballroom AB, Concourse Level	Westin Grand Ballroom CDE, Concourse Level
	SESSION 54 – Hundreds of New GWAS Loci Co-Moderators: Ching-Ti Liu, Boston Univ.; and Orli Bahcall, Nature Genetics	**SESSION 55 – Impact of Bottlenecks and Population Growth on Rare Variation** Co-Moderators: Itsik Pe'er, Columbia Univ.; and Shamil Sunyaev, Brigham and Women's Hosp.	**SESSION 56 – Haplotypes, Imputation and Interactions** Co-Moderators: Josee Dupuis, Boston Univ. Sch. of Publ. Hlth; and Michael Nothnagel, Univ. of Cologne	**SESSION 57 – Autism and Neurodevelopmental Disorders** Co-Moderators: Santhosh Girirajan, Pennsylvania State Univ.; and Dimitrios Avramopoulos, Johns Hopkins Univ.	**SESSION 58 – Cardiovascular Genetics: Functional Characterization and Clinical Applications** Co-Moderators: Dan E. Arking, Johns Hopkins Univ. Sch. of Med.; and Myriam Fornage, Univ. of Texas Hlth Sci. Ctr. at Houston	**SESSION 59 – Prenatal and Reproductive Genetics** Co-Moderators: David Chitayat, Mount Sinai Hosp., Toronto; and Mary E. Norton, Stanford Univ.	**SESSION 60 – Ethical, Legal, Social and Policy Issues** Co-Moderators: Dawn Allain, The Ohio State Univ.; and Barbara Biesecker, NHGRI/NIH	**SESSION 61 – Genomics of Developmental Disorders** Co-Moderators: Karen Avraham, Tel Aviv Univ.; and Margarit Urbanek, Northwestern Univ., Chicago	**SESSION 62 – Prostate and GI Cancer Susceptibility** Co-Moderators: Gail Jarvik, Univ. of Washington; and Liesel M. FitzGerald, Cancer Epidemiol. Ctr.
2:00	**331** Large scale meta analysis of 250,000 individuals reveals novel biological pathways involved in adult human height. **T. Esko et al.**	**340** The Ashkenazi Jewish genome. **S. Carmi et al.**	**349** A haplotype map derived from whole genome low-coverage sequencing of over 25,000 individuals. **J. Marchini.**	**358** Utility of a strategic next-generation sequencing approach to genomic diagnosis of patients with neurodevelopmental disorders. **S. Soden et al.**	**367** A homozygous mutation in Smoothened, a member of the Sonic hedgehog (SHH)-GLI pathway is involved in human syndromic atrioventricular septal defect. **W. S. Kerstjens-Frederikse et al.**	**376** Noninvasive fetal trisomy test: A large-scale clinical practice in 78,289 cases. **Y.Gao et al.**	**385** Practical assessment of incidental finding recommendations for use in clinical exome testing. **M. C. Dulik et al.**	**394** Targeted sequencing of GPI anchor synthesis pathway genes identifies a new causal gene of hyperphosphatasia with mental retardation. **P. Krawitz et al.**	**403** Whole exome sequencing of 2126 African American prostate cancer cases and controls from the Multiethnic Cohort. **K. A. Rand et al.**
2:15	**332** Opening the X files: Chromosome X-wide association study reveals new loci for fasting insulin and height and evidence for incomplete dosage compensation. **T. Tukiainen et al.**	**341** Rare variant sharing reveals population histories. **I. Mathieson et al.**	**350** Statistical estimation of haplotype sharing from unphased genotype data. **D. Xifara et al.**	**359** De novo mutations in autism spectrum disorders revealed by whole genome sequencing. **Y. H. Jiang et al.**	**368** Identification of *PRDM16* as a disease gene for left ventricular non-compaction and the efficient generation of a personalized disease model in zebrafish. **A.-K. Arndt et al.**	**377** Comparison of three single-cell whole genome amplification methods for detection of genomic aberrations by array CGH: A step towards noninvasive prenatal diagnosis using intact fetal cells. **A. Breman et al.**	**386** Individual expectations for return of secondary results from exome sequencing. **H. K. Tabor et al.**	**395** 1000 trio exomes: Insights into severe developmental disorders. **M. Van Kogelenberg et al.**	**404** Identification of Y chromosomes associated with risk for prostate cancer. **L. A. Cannon-Albright et al.**
2:30	**333** Expanded and novel loci for A1c levels identified through a trans-ethnic meta-analysis approach in European and African American ancestry samples. **E. Wheeler et al.**	**342** High risk population isolate reveals low frequency variants predisposing to intracranial aneurysms. **M. I. Kurki et al.**	**351** HapFABIA: Identification of very short segments of identity by descent via biclustering. **S. Hochreiter et al.**	**360** Recurrently mutated genes contribute to the risk for developing sporadic autism spectrum disorder. **B. J. O'Roak et al.**	**369** Mutation and copy number variation of *FOXC1* causes cerebral small vessel disease. **C. R. French et al.**	**378** Next-generation sequencing based preimplantation genetic testing of 24-chromosome aneuploidy and monogenic disorders. **N. R. Treff et al.**	**387** The benefits and risks of wanting it all: How parents plan to manage their children's exome sequencing results. **J. Yu et al.**	**396** Atypical Rett Syndrome: Is it really more common in females? **K. Cusmano-Ozog et al.**	**405** Genome-wide scan identifies a novel locus associated with aggressive prostate cancer. **S. I. Berndt et al.**
2:45	**334** Genome-wide association study for serum metabolome reveals 57 associated loci for biomarkers of complex metabolic diseases. **J. Kettunen et al.**	**343** Rare variant association studies: What population genetics models teach us about power and study design. **B. M. Neale et al.**	**352** A new method for genotype calling and phasing for the 1000 Genomes Project leads to improved downstream imputation accuracy. **O. Delaneau et al.**	**361** Identification of biological pathways associated with phenotypically-defined subgroups of autism spectrum disorders. **O. J. Veatch et al.**	**370** Genetic association of common variants with a rare cardiac disease, the Brugada syndrome, in a multi-centric study. **C. Dina et al.**	**379** Genetic normalization of differentiating aneuploid cleavage stage embryos. **P. R. Brezina et al.**	**388** Evaluation of clinical utility of whole genome sequencing: The WGS500 programme. **J. Taylor et al.**	**397** Common molecular networks in Rett, Angelman, Smith-Magenis, Potocki-Lupski, Pitt-Hopkins, and chromosome 2q23.1 deletion syndromes contribute to intellectual disability, seizures, sleep, language, behavior and autism spectrum disorder. **S. V. Mullegama et al.**	**406** Frequent germline mutations in DNA repair genes in familial prostate cancer cases. **D. Leongamornlert et al.**
3:00	**335** Using correlated phenotypes to functionally classify GWAS loci. **N. Eriksson et al.**	**344** Finnish founding bottleneck leads to excess of damaging loss-of-function variants with medically relevant associations. **E. T. Lim et al.**	**353** Identification of genetic epistasis in regulation of gene expression via variance expression quantitative trait loci. **A. Brown et al.**	**362** De novo mutation in the dopamine transporter gene associates dopamine dysfunction with autism spectrum disorder. **N. G. Campbell et al.**	**371** Loss-of-function mutations in *CECR1*, encoding adenosine deaminase 2, cause systemic vasculopathy with fever and early onset strokes. **Q. Zhou et al.**	**380** Maternal age dependent loss of SMC1β transcripts in human oocytes. **V. Jobanputra et al.**	**389** International views on sharing incidental findings from whole genome research. **A. Middleton et al.**	**398** *MBD5* deletion disrupts circadian gene expression and is associated with sleep disturbance in the 2q23.1 deletion syndrome. **S. H. Elsea et al.**	**407** Pleiotropic effect of rare mutation in *HOXB13* on multiple cancers detected in a cohort of >100K individuals via imputation. **J. Witte et al.**
3:15	**336** Meta-analysis of SNP associations with body mass index in >339,000 individuals gives new genetic and biological insights into the underpinnings of obesity. **E. K. Speliotes et al.**	**345** A rare functional variant in *APOC3* is associated with lipid traits and has risen in frequency in distinct population isolates. **E. Zeggini et al.**	**354** Association and replication of SNP-SNP interactions for hundreds of gene expression phenotypes. **A. Fish et al.**	**363** Analysis of synaptic function during neurogenesis and maturation in homogeneous populations of autism-affected GABAergic and glutamatergic neurons. **B. A. DeRosa et al.**	**372** Genetic influence on LpPLA2 activity at baseline as evaluated in the exome chip-enriched GWAS study among ~13600 patients with chronic coronary artery disease in the STABILITY (STabilisation of Atherosclerotic plaque By Initiation of darapLadIb TherapY) trial. **L. Warren et al.**	**381** A clinical algorithm for efficient, high-resolution cytogenomic analysis of uncultured perinatal tissue samples: Study of more than 700 cases. **G. Maire et al.**	**390** No evidence for increase in screening among women given report of moderately higher than average risk for breast cancer from personal genomics services: The PGen Study. **S. W. Gray et al.**	**399** Lysyl-tRNA synthetase (*KARS*) mutations cause autosomal recessive nonsyndromic hearing impairment *DFNB89*. **R. Santos-Cortez et al.**	**408** Detection of large rearrangements in *PMS2*. **D. Mancini-DiNardo et al.**
3:30	**337** Large-scale association analysis identifies novel loci associated with waist-to-hip ratio and suggests underlying biological mechanisms. **D. Shungin et al.**	**346** The impact of recent human demography on deleterious mutation load and the genetic architecture of disease susceptibility. **G. Sella et al.**	**355** Gene-gene interaction analysis for next-generation sequencing. **J. Zhao et al.**	**364** Disruption of the *ASTN2* / *TRIM32* locus at chr9q33.1 in gender modulated risk for autism, ADHD and other neurodevelopmental phenotypes. **K. Tammimies et al.**	**373** Genome-wide association study identifies common and rare genetic variants in caspase-1-related genes that influence IL-18 regulation in patients with acute coronary syndrome. **A. Johansson et al.**	**382** Whole genome oligonucleotide-SNP arrays in prenatal diagnosis: advancement in identification of clinically significant chromosomal abnormalities. **T. Sahoo et al.**	**391** Context is complex: Attitudes to incorporating genomic risk profiling into population screening programs. **S. G. Nicholls et al.**	**400** Multiple de novo variants resulting in combined axial hypotonia with dyskinesia and facial myokymia. **A. Torkamani et al.**	**409** Functional analysis of the chr13q22.1 pancreatic cancer risk locus suggests allele-specific effects on *DIS3* expression. **J. Hoskins et al.**
3:45	**338** Chipping away at the common variant genetics of age related macular degeneration. **L. G. Fritsche.**	**347** Inferring ancient demography using whole-genome sequences from multiple individuals. **M. Steinruecken et al.**	**356** Capturing the geographic and genetic components controlling individual genetic regulation of cardio-metabolic quantitative traits. **Y. Idaghdour et al.**	**365** *TBC1D24*, responsible for early-onset epilepsies associated with intellectual disabilities, plays a role in the formation and maturation of cerebral cortex. **A. Falace et al.**	**374** Prevalence and predictors of pneumothorax in patients with connective tissue disorders enrolled in the GenTAC (National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions) Registry. **J. P. Habashi et al.**	**383** The fetal *FMR1* premutation phenotype: Clues from the amniotic fluid transcriptome. **L. M. Zwemer et al.**	**392** How do citizens balance the benefits and burdens of newborn screening? A choice experiment. **F. A. Miller et al.**	**401** Dominant β-catenin mutations cause intellectual disability with recognizable syndromic features. **T. Kleefstra et al.**	**410** Genome-wide association study of colorectal adenoma in the Nurses' Health Study and the Health Professionals Follow-Up Study. **A. D. Joshi et al.**
4:00	**339** Genetics and biology of rheumatoid arthritis contribute to drug discovery. **Y. Okada et al.**	**348** Inferring human population history and gene flow from multiple genome sequences. **S. Schiffels et al.**	**357** Identification of a set of highly constrained genes from exome sequencing data. **K. E. Samocha et al.**	**366** Transcriptional consequences of 16p11.2 microdeletion/microduplication syndrome in human lymphoblasts and mouse cortex. **I. Blumenthal et al.**	**375** Surprising clinical lessons from targeted next-generation sequencing of thoracic aortic aneurysmal genes. **B. Loeys et al.**	**384** The wide spectrum of alpha and beta-tubulinopathies in foetus : From microlissencephaly to asymmetrical multifocal polymicrogyria. **N. Bahi-Buisson et al.**	**393** Identifying genetic relatives without compromising privacy. **E. Eskin et al.**	**402** Defective initiation of glycosaminoglycan synthesis due to *B3GALT6* mutations causes a pleiotropic Ehlers-Danlos syndrome-like connective tissue disorder. **F. Malfait et al.**	**411** Large numbers of individuals required to classify and define risk for a rare VUS in known cancer risk genes. **B. H. Shirts et al.**

Saturday, October 26: Concurrent Invited Session III (70-76)

9:30 AM–11:30 AM

Grand Ballroom West, Level 3	Room 258, Level 2	Room 210, Level 2	Room 205, Level 2	Hall B2, Level 0 (Lower Level)	Room 253, Level 2	Grand Ballroom East, Level 3
SESSION 70 – Design, Content and EMR Integration of Clinical Sequencing Reports Co-Moderators: Robert C. Green, Brigham and Women's Hosp./Harvard Med. Sch.; and Heidi L. Rehm, Partners Ctr. for Personalized Genet. Med., Cambridge, MA	**SESSION 71 – More or Less: Copy Number Variation and Human Adaptation** Co-Moderators: Chack-Yung Yu, The Ohio State Univ.; and Ed J. Hollox, Univ. of Leicester	**SESSION 72 – Somatic Mutations in Human Disease — Piecing Together the Mosaic** Co-Moderators: Leslie Biesecker, NHGRI/NIH; and William Dobyns, Univ. of Washington	**SESSION 73 – Tandem Repeat-Associated Epigenetic Mechanisms in Neuromuscular Disorders** Co-Moderators: Paul J. Hagerman, Univ. of California, Davis, Sch. of Med.; and Laura Ranum, Col. of Med., Univ. of Florida	**SESSION 74 – Twin Studies: Helping Us Understand and Exploit the Genome (In Honor of Walter Nance's Contributions to Human Genetics on his 80th Birthday)** Co-Moderators: Rita M. Cantor Chiu, David Geffen Sch. of Med. at UCLA; and Cynthia C. Morton, Brigham and Women's Hosp.	**SESSION 75 – Where Do Risk Variants Act? Interrogating Genomic Studies of Multiple Human Tissues** Co-Moderators: Chris Cotsapas, Yale Sch. of Med.; and Kristin Ardlie, Broad Inst. of MIT and Harvard	**SESSION 76 – Whole Genome Sequencing for Every Baby? Where Diagnostic and Screening Applications Collide** Co-Moderators: James O'Leary, Genet. Alliance, Washington, DC; and Natasha Bonhomme, Genet. Alliance, Washington, DC
09:30 am Design and implementation of the General Genome Report. **R. C. Green.**	09:30 am Evolution of segmental duplications and novel neural genes. **E. E. Eichler.**	09:30 am The clinical manifestations of mosaicism. **R. Happle.**	09:30 am Facioscapulohumeral dystrophy: An epigenetic disease with genetic modifiers. **S. Tapscott.**	09:30 am Twin studies in the non-molecular era. **W. Nance.**	09:30 am Leveraging gene expression data to understand cell autonomous effects of inflammatory disease variants. **P. L. De Jager.**	09:30 am Genomic technology and newborn screening — pros and cons. **O. A. Bodamer.**
10:00 am Reporting results not directly related to the indication for testing. **C. Eng.**	10:00 am Human-specific loss of regulatory DNA and the evolution of human-specific traits. **D. M. Kingsley.**	10:00 am Cytogenetics and the historical context of mosaicism. **N. Spinner.**	10:00 am Mechanisms of pathogenesis in fragile X-associated disorders. **P. J. Hagerman.**	10:00 am Twin studies as a tool in genetic epidemiology. **N. Risch.**	10:00 am Cross-tissue meta-analytic approaches result in large gains in regulatory variant identification. **B. Raby.**	10:00 am WGS in newborn screening: What are we screening for? **J. R. Botkin.**
10:30 am Approaches to integrating next-generation sequencing into the electronic health record. **P. Tarczy-Hornoch.**	10:30 am Primate structural genomic variation evolving under positive selection. **C. Lee.**	10:30 am Single gene mutations and mosaic genetic disease. **L. Biesecker.**	10:30 am Repeat associated non-ATG translation in microsatellite expansion disorders: Lessons from SCA8 and myotonic dystrophy. **L. Ranum.**	10:30 am Twin studies as a powerful approach to identifying and understanding molecular pathways that underlie complex traits. **N. Martin.**	10:30 am Unique opportunities for scientific discovery in transcriptome studies across multiple tissues. **N. Cox.**	10:30 am Parental interest in whole genome sequencing of newborns. **A. Goldenberg.**
11:00 am Scaling genomic reporting and clinical decision support. **H. L. Rehm.**	11:00 am Structural variation of beta-defensins: Welcome to the dynamic genome. **E. J. Hollox.**	11:00 am The mosaic landscape of cancer. **E. Mardis.**	11:00 am Development of histone deacetylase inhibitors as therapeutics for Friedreich's ataxia. **J. Gottesfeld.**	11:00 am Twin studies as a valuable approach to omics research. **T. Spector.**	11:00 am Regulatory effect mapping of trait-associated variation identifies causal cell types. **J. Stamatoyannopoulos.**	11:00 am Does the public want to have every baby sequenced at birth? Public participation and expectations of WGS. **Y. Bombard.**



Pattern Change in 2014

Saturday through Wednesday meeting

(not Tuesday through Saturday)

Saturday, October 26

11:45 AM–1:15 PM

SESSION 77 – ASHG Distinguished Speakers Invited Symposium: Medical Systems Genomics

Hall B2, Level 0 (Lower Level), Convention Center

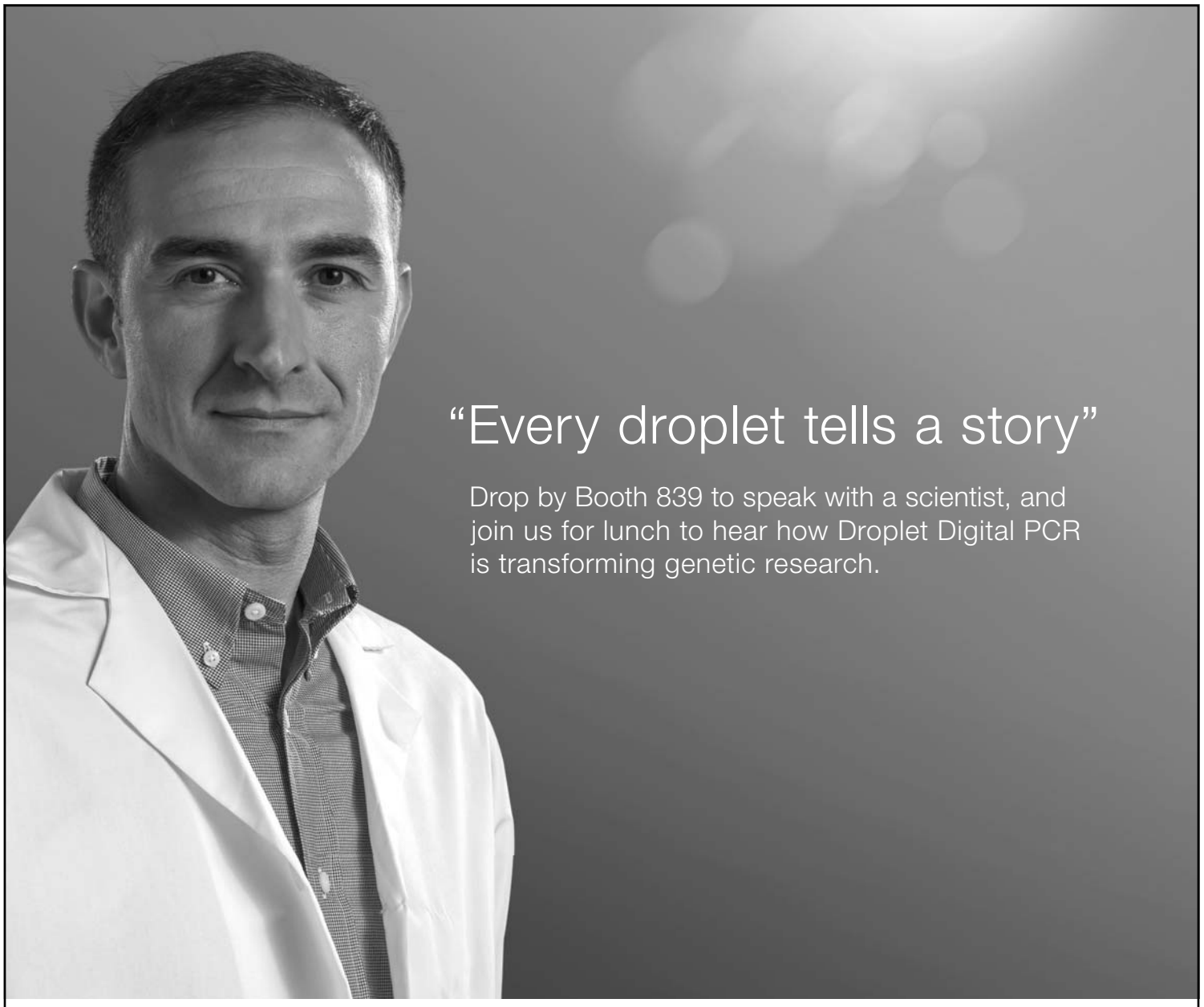
Moderators:
Jeff Murray, Univ. of Iowa
Andrew G. Clark, Cornell Univ.

This symposium will provide the ASHG community with an update from a trio of trailblazing experts on the state of the art of Systems Biology and its applications to medical genetics. Aviv Regev, Marc Vidal, and Garry Nolan will provide varying perspectives on the use of omics data to build predictive models of physiological states of the cell and the organism, including diseased states. These integrative approaches can provide a particularly informative picture of changes in cellular function mediated by cancer, pathogen response, and other stresses. They will challenge us with what the prospects are for methods like these to deliver improved insights regarding disease risk and therapeutic interventions. The speakers will conclude by gazing into the future, providing a vision of where systems medicine will be going in the next decade.

Interactome networks and human disease. M. Vidal. Dana-Farber Cancer Inst., Harvard Univ.

Reconstructing cellular circuits: From individual to single cell variation in immune cells. A. Regev. Broad/MIT/HHMI.

A Definable "Structure" for the Immune System and Cancers at the Single Cell Level. G. Nolan. Stanford School of Medicine.



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Drop by Booth 839 to speak with a scientist, and join us for lunch to hear how Droplet Digital PCR is transforming genetic research.

The QX200 Droplet Digital PCR System
Learn more at **biorad.com/ddpcr**

Attend our luncheon seminar on Thursday, October 24, 12:30–2:00 PM
Room 211, Level 2, Boston Convention and Exhibition Center

Seminar speakers:

"Droplet Digital PCR and Plasma Tumor DNA: Applications in Clinical Oncology"

Ben H. Park, MD, PhD
Associate Professor of Oncology, Breast Cancer Program
Associate Director, Hematology/Oncology Fellowship Training Program
The Sidney Kimmel Comprehensive Cancer Center at Johns Hopkins

"Droplet Digital PCR for the Detection of Copy Numbers in 22q Deletion Syndrome"

Flora Tassone, PhD
Professor in Residence
University of California, Davis
Department of Biochemistry and Molecular Medicine

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

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