

American Society of Human Genetics October 22-26



2013



BOSTON



The American Society of Human Genetics 63rd Annual Meeting

Program Addendum As of October 15, 2013

Government Shutdown Affects Meeting

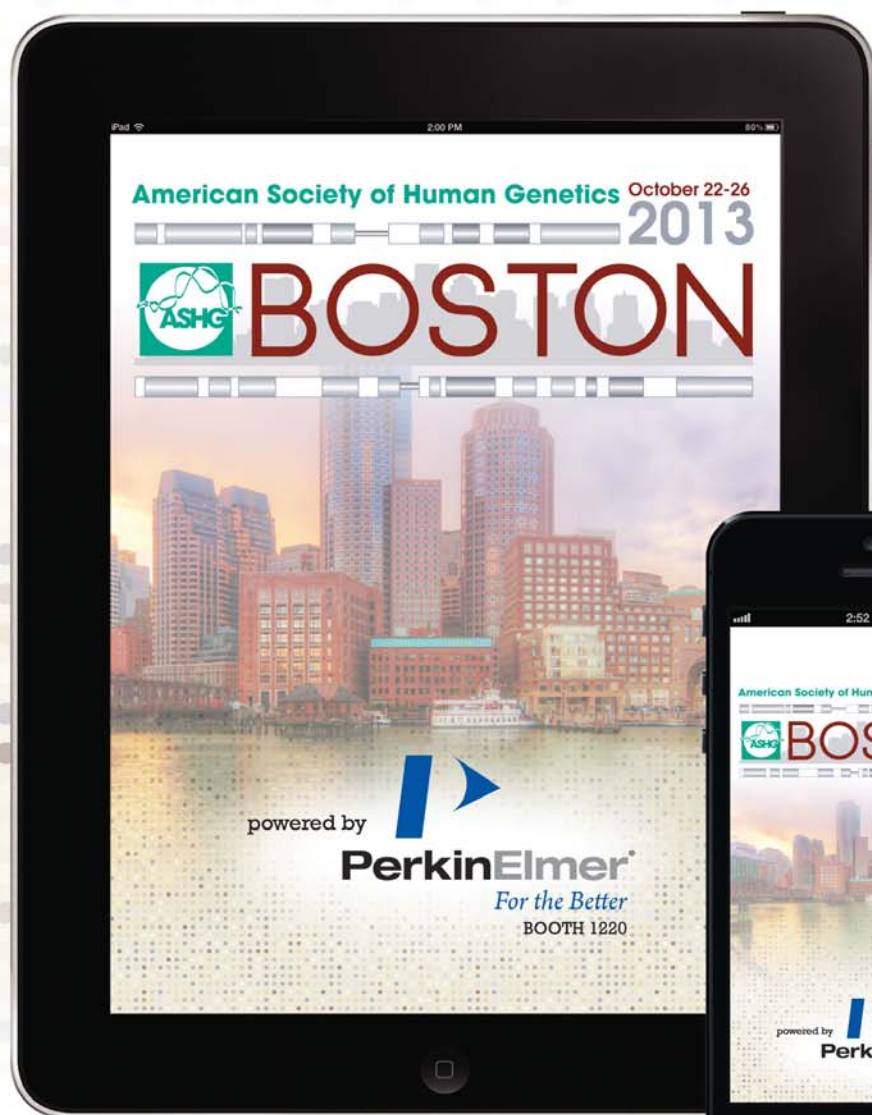
If the government shutdown persists, the impact on our program is as follows:

- **Unable to attend:** 7 invited speakers, 13 speakers in platform sessions, 11 session moderators, and 115 poster presenters.
- **Already Cancelled:** NCBI workshop on navigating clinical genomic resources.
- **Cancelled:** 4 NIH exhibit booths.
- As of October 15, we were able to include any changes or cancellations in the addendum. Changes received after this date will be addressed as follows:
 - At the beginning of each session, moderators will make announcements regarding speaker changes/replacements;
 - Updates will be placed in the mobile app as soon as they are available; and
 - A sign will be placed on poster boards indicating the reason the author is unable to attend.

ASHG Wants to Hear From You: ASHG is seeking feedback, suggestions and ideas for ways that our Society can be of benefit to you, our valued members. Please send comments to society@ashg.org.

Download the ASHG 2013 Mobile App Now!

<http://crowd.cc/ashg2013>



You can also scan this code with a QR Reader on your device.



Reminders/Announcements

Badges must be worn and visible at all times while attending ASHG events in the Boston Convention & Exhibition Center. They will be checked at the entrance to all scientific sessions, poster sessions, and when entering the Exhibit Hall.

Cell Phone/Camera/Video Policy: Cameras and all other recording devices are prohibited from use in all session rooms, on the exhibit floor, and in all poster/oral presentations.

Complimentary Wi-Fi is available at the BCEC. Just click BCEC wireless network under settings on your mobile device.

Complimentary Wi-Fi at the Westin in Scientific Session Rooms: Use the following codes when accessing the Wi-Fi in Ballroom AB and Ballroom CDE. The code changes daily. Wednesday = ashgwed; Thursday = ashgthur; Friday = ashgfri.

Food concessions will be open in the Exhibit Hall Wednesday through Friday, and portable food stands will also be available in several areas throughout the building. The Wicked Food Court, with multiple eateries, is located on Level 1.

Complimentary Dinner Trolley Service: Experience Boston's neighborhood restaurants by taking the complimentary trolley service from the BCEC Wednesday through Friday between the hours of 4:00 pm and 7:00 pm, to Chinatown, South Boston, and the Waterfront. Reservations are recommended. Return service available before 7:00 pm. Stop by the visitors counter in the Northwest Lobby, Level 1 (near the food court) for further details.

Lost and Found Items: Room 254, Level 2.

ASHG Membership Questions: Exhibit Hall at ASHG Central.

Shuttle Buses: BCEC pick-up on Level 1
Tues: 7:00am-10:00am and 1:00pm-8:00pm
Wed-Fri: 6:30am-9:30am and 4:00pm-8:00pm
Sat: 6:30am-9:30am and 11:30am-3:30pm
Route 1: Hilton Financial District; Langham; Intercontinental.
Route 2: Omni; Hyatt Regency,
Route 3: Hilton Logan Airport; Embassy Suites,

Route 4: Doubletree Downtown, Courtyard by Marriott/Tremont; Boston Park Plaza
Telephone Numbers: All offices are located at the Convention Center. Telephone numbers will be operational Tuesday through Saturday.
ASHG Office, Room 254: 617-954-3955
Convention Center Main Number: 617-954-2000
Exhibit Registration, North Lobby: 617-954-3950
First Aid, North Lobby: 617-954-2222
Housing, North Lobby: 617-954-3954
Information, North Lobby: 617-954-3951
Press Office, Room 252: 617-954-3959
Registration Help, North Lobby: 617-954-3952
Restaurant/City Info: 617-954-2566
Security: dial '2222' from any black house phone
Emergency/Police/Fire: dial 2222 from any black house phone

Plenary Authors Corner, ASHG Central	
Meet these plenary authors and ask your questions in a less formal setting at ASHG Central, in the Exhibit Hall. Each session will be moderated by a member of the Program Committee.	
Wednesday, October 23	
1:00-1:30 pm	Maria Nicla Loviglio
1:30-2:00 pm	David Reich
4:30-5:00 pm	Jeanne B. Lawrence
Thursday, October 24	
12:00-12:30 pm	Matthew Hurles
12:30-1:00 pm	Daniel E. Bauer
Friday, October 25	
1:30-2:00 pm	Scott L. Carter

Have you found your match? Visit ASHG Central for your chance to win an iPad mini! The meeting features the theme of great discoveries in genetics. Your name badge has either the name of a person or persons associated with a discovery in genetics OR the name of a discovery in genetics. Your job is to find the appropriate match. For example, if your badge reads "**Hamilton Smiht, Werner Arber, and Dan Nathans**" then you would look for someone with a badge that reads "**restriction enzymes.**" When you find your match, go as a pair to ASHG Central and register to win one of 10 iPad minis. Winners will be announced at the ASHG Business Meeting. At least one person in the matching pair must be present to win.

• WEDNESDAY •

Shuttle Bus Schedule for Wednesday: 6:30am-9:30am and 4:00pm-8:00pm. Buses run every 15-20 minutes. The transportation desk telephone number is 617-939-5782. The number will be operational during the shuttle bus hours. If you need special assistance, such as a wheelchair lift, please contact the transportation desk at 617-939-5782 or the ASHG Management Office at least 24 hours in advance at ashgmeetings@ashg.org.

Reminder: The Welcome Reception, formerly named the opening mixer and traditionally held on Tuesday, will be held on Wednesday, in the Exhibit Hall, to encourage more effective networking and socializing around shared interests. The poster topic categories can be used as convening sites for researchers and clinicians working in a particular discipline

If government authors are unable to travel, announcements will be made at the beginning of each session. For Platform Sessions, the Program Committee will make every effort to upgrade a poster if a replacement author cannot be assigned. If this is not possible, moderators will take a 15 minute break between talks.

Workshops

CANCELLED: NCBI Workshop, Room 102 Wednesday

Ancillary Events

NEW: 6:30 pm - 9:30 pm: 1000 Genomes Data Tutorial, Room 156ABC, BCEC

Platform Sessions

Session 10: Program/Abstract 7: M. Rieder replaces R. Emerson

Session 11: Program/Abstract 20: P. Ankita replaces R. Choy

Session 12: Program/Abstract 25: L. Strug replaces W. Li

Session 12: Program/Abstract 28: D. Chang replaces A. Keinan

Session 12: Program/Abstract 29: M. Emond cancelled. Co-authors cannot attend

Session 14: Program/Abstract 43: J. Kaprio replaces L. Mucci

Session 15: Program/Abstract 60: B. Riley replaces A. Adkins

Session 16: Program/Abstract 66: Z. Urban replaces E. Lawrence

Session 18: Program/Abstract 81: S. Wettsten replaces N. Blau

Posters Cancelled

539W

742W

765W

822 W

894W

951W

1005W

1373W

1434W

1479W

1551W

1754W

1898W*

2081W

2420W

2433W

2570W

2667W

2769W

2808W

3119W

3143W
3167W
3291W

*U.S. Government-employed author, and unable to attend.

Poster Author Changes

581W: D. Cherukuri will present for P.N. Rao
728W: G. Meyer will present for S. Jacobs-Helber
727W: A. Gordon will present for L. Felker
743W: A. Rivera will present for B. Gonzalez
847W: K. Shchetynsky will present for L. Diaz
855 W: D. Crawford will present for R. Villegas
972W: J. Capra will present for D. Kostka
1056W: M. Pino-Yanes will present for M. Acosta-Herrera
1065W: M. Miyake will present for K. Yamashiro
1229W: R. Kim will present for A. Guerin
1232W: U. Hehr will present for S. Herbst
1292W: M. Shohat will present for L. Basel-Vanagaite
1322W: D. Yuan will present for S. Huang
1352W: I. Cendes will present for M. Gonsales
1421W: C. Kao will present for T. Wenger
1431W: P.A.C. t' Hoen will present for Y. Anvar
1473W: T. Park will present for J. Lee
1608W: M. Kamkar will present for A. Hoover
1629W: K. Qaadri will present for C. Olsen
1641W: K. McBride will present for E. Bonachea
1647W: P. McInerney will present for H. Duckworth
1683W: J.H. Park will present for I.H. Lee
1745W: S. Won will present for S. Choi
1892W: M. Salomon will present for P. Marjoram
1934W: C. Gieger will present for E. Albrecht
1944W: L. Carbone will present for M. Konkel
1979W: Y. Espinosa-Parrilla will present for M. Lopez-Valenzuela
2045W: A. Quinto will present for M. Gomez-Ortega

2060W: C. Barbieri will present for A. Duggan
2086W: N. Franceschini will present for S. Cole
2408W: S. Morrison will present for J. Carroll
2413W: S. Dasgupta will present for K. Tuttle
2418W: M. Snyder will present for M Dunkle
2447W: K. Ormond will present for S. Kejriwal
2670W: D. Gelbman will present for L. Wolf
3429W: H. Baris will present for I. Kedar-Barnes
2455W: D. Boeldt will present for C. Bloss
2461W: D. Kaufman and J. Bollinger will present for L. Jamal
2528W: N. Li will present for L. YL Lan
2558W: J. Brunton will present for V. Jayaraman
2606W: M. Nakamura will present for A. Watanabe
2639W: A. Patwardham will present for G. Chandratillake
2643W: J. Rosenfeld will present for C. Jorgez
2703W: A. Verloes will present for A. Lavillaureix
2769W: T. Kisho will present for Y. Narumi
2811W: N. Simpson will present for L. Donohue
2933W: J. Saarela will present for L. Trotta
2951W: S. Ware will present for A. Cast
3002W: G. Stevanin will present for H. Azzedine
3020W: I. Lopes Cendes will present for F. Torres
3044W: J. Marini will present for A. Reich*
3342W: D. Shabsovich is an additional author
3366W: M. Achatz will present for F. Fortes
3407F: G. Matullo will present for A. Russo Jr.

• THURSDAY •

Shuttle Bus Schedule for Thursday: 6:30am-9:30am and 4:00pm-8:00pm. Buses run every 15-20 minutes. The transportation desk telephone number is 617-939-5782. The number will be operational during the shuttle bus hours. If you need special assistance, such as a wheelchair lift, please contact the transportation desk at 617-939-5782 or the ASHG Management Office at least 24 hours in advance at ashgmeetings@ashg.org.

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Platform Sessions

Session 21: Program/Abstract 108: C. Liu replaces J. Meigs

Session 23: Program/Abstract 127: T. Raj replaces B. Stranger

Session 32: Program/Abstract 209: The correct name for one of the co-authors is N. Okamoto.

Session 35: Program/Abstract 240: J. Carroll replaces S. Morrison

Ancillary Events

Cancelled: 1:00 pm: GCAT: An Open and Collaborative Platform for Comparing Genome Analysis Pipelines, Westin Hotel, Douglas Room.

Cancelled: Room 101 at 7:00 am, ACMG Therapeutics Committee Meeting

Posters Cancelled

466T	1899T*
470 T	2100T
576T	2152T
609T*	2198F
651T	2365T
772T*	2385T*
877T	2580T
922T	2683T
946T	2931T*
1000T	3015T
1036T	3051T
1329T	3120T
1359T	3475T
1450T	3496T
1504T	
1555T	
1606T	
1695T	

*U.S. Government-employed author, and unable to attend.

Poster Author Changes

148T: S. Young will present for K. Peterson

S. Young was also added as a co-author

496T: T. Eggermann will present for M. Begemann

630T: C.Czarnecki will present for M. Luo

757T: T.Y. Chang will present for Y.J. Lee

814T: Q.Dai will present for H. Xu

1087T: D. A. Scott will present for T. Beck

1108T: M. Pericak-Vance will present for P.

Whitehead

1218T: Holly Stephens will present for J. Backman

1341T: S. Gandomi will present for S. Tang

1362T: B. Riley will present for E. Loken

1377T: B.Riley will present for D. Brohawn

1528T: D. Abramovitz will present for R. Hariharan

1636T: J. Gerlach will present for G. Geiss

1642T: M. Li and S. Chervitz wil present for H. Lam

1648T: J. Betley and N. Miller will present for K.

Hall

1797T: T. Winkler will present for C. Hoggart
2007T: Y. Espinose-Parrilla will present for I. Balcells
2134T: M. Muenke will present for P. Kruszka*
2273T: G. Cox will present for Y. Xue
2277T: Dr. Nuria Carrillo-Carrasco will present for F. Celeste*
2405T: S.h. Kan will present for M. Vera
2444T: P. Borry will present for S. Janssens
2466T: S. Bale will present for E.V. Haverfield
2538T: J.L. Bluin will present for K. Varvagiannis
2541T: X. Li will present for S. Chen
2601T: J. L Costa will present for A. Justino
2604T: M. Majolini will present for M. Rongioletti

2610T: L. Van Laer will present for A. McInerney-Leo
2613T: X. Chen will present for H. Liu
2850T: J. Tynan will present for R. Tim
3108T: A. Hovnanian will present for S. Duchatelet
3208T: O. Moreno will present for C. Suleima
3242T: M. Melaragno will present for M. Colovati
3252T: G. Johnston will be presenting for D. Weldon
3271T: M. Rotunno will present for A. Goldstein*
3295T: M. Achatz will present for K. Andrade
3322T: A. Chaudhury will present for C.M. Laukaitis
3454T: P. Rosenstiel will present for D. Esser
3487T: Q.Wang will present for Z. Zhao

*U.S. Government-employed author, and unable to attend.

PLAN TO ATTEND

SESSION 44 – ASHG Next: The Future of Genetics and the Future of Your Society Thursday, 6:45 PM–7:45 PM, Room 253, BCEC

Moderators: Jeff Murray, Univ. of Iowa and
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.

You can also go to www.rwpoll.com to participate or you can scan the QR code below. Enter ASHG as the session ID. Key pads are available to those without a mobile device.



• FRIDAY •

Shuttle Bus Schedule for Friday: 6:30am-9:30am and 4:00pm-8:00pm. Buses run every 15-20 minutes. The transportation desk telephone number is 617-939-5782. The number will be operational during the shuttle bus hours. If you need special assistance, such as a wheelchair lift, please contact the transportation desk at 617-939-5782 or the ASHG Management Office at least 24 hours in advance at ashgmeetings@ashg.org.

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Platform Sessions

Session 56: Program/Abstract 356: P. Awadalla replaces Y. Idaghdour

Session 59: Program/Abstract 376: Y. Gao replaces F. Chen

Session 60: Program /Abstract 392: R. Hayeems replaces F. Miller

Session 61: Program/Abstract 395: M. van Kogelenberg, cancelled. Co-authors cannot attend.

Posters Cancelled

441F	2014F
481F	2026F
556F	2083F
607F	2314F
749F	2316F*
971F	2348F
1106F	2478F
1255F	2789F
1372F	2923F
1420F	3049F
1457F	3064F
1478F	3097F
1619F	3503F
1667F*	3506F
1987F	
1888F*	

Poster Author Changes

439F: L. Barcellos will present for A. Baker
477F: R. Sooknanan will present for J. Pease
69F: L. Larizza will present for L. Fontana
788F: T.Y. Chang will present for H. Chi
866F: K. Van Steen will present for F. Van Lishout
914F: N. Franceschini will present for C. Chen
938F: S. Cha will present for Y. Park
1249F: P. Demougin will present for N. Hadziselimovic
1282F: A. Naj will present for L. Wang
1336F: E. Gamsiz will present for E. Morrow
1429F: F. Tsetsos will present for I. Karagiannidis
1496F: D. Leary will present for J. Newcomer
1511F: A. Partch will present for O. Valladares
1517F: S. Pendergrass will present for D. Wolfe
1601F: P. Goldstein will present for H. Heaton
1836F: H.S. Wu will present for K. Cho
1906F: A. Morris will present for C. Grace

1930F: S. Bull will present for Y. Yilmaz
2171F: K. Albers will present for T.E. Galesloot
2270F: T. Donahue will present for C. Samango-Sprouse. T. Donahue was also added as a co-author
2262F: U. Hehr will present for S. M. Herbst
2272F: L. Shihabuddin will present for J. Marshall
2290 F: M. Bailer will present for C. Haller
2344F: S. Kaler will present for K. Patel*
2404F: J. Cohen-Pfeffer will present for K. Mooney
2328F: D. Zand will present for J. Fraser*
2388F: A. Ledger and R. Baek will present for R. Moreland
2479F: R. Hayeems will present for L. Baret
2480F: C. Shuman or D. Chitayat will present for A. Pichini
2482F: J. Youngblom will present for K. Hitch
2483F: J. Schuette will present for L. Kiedrowski

2492F: N.Hoang will present for R. Babul-Hirji
2587F: X. Chen will present for W. Teng
2614F: X. Chen will present for F. Wong
2638F: D. Allingham-Hawkins will present for A. Malhotra
2669F: M.B. Duz will present for E. Kirat
2678F: F. Malfait will present for B. Callewaert
2801F: L. Larizza will present for J. Azzollini

3195F: J.C.P. Rivera will present for J. A. Rojas Martinez
3241F: M. Melaragno will present for S. Bragagnolo
3263F: T. Walsh will present for G. Bernier
3284F: J.Little will present for Z. Montazeri
3458F: A. Dulak will present for D. Greenawalt
3449F: S. Khaliq will present for A. Abid

*U.S. Government-employed author, and unable to attend because of restrictions on travel.

•SATURDAY•

Shuttle Bus Schedule for Saturday: 6:30am-9:30am **and** 11:30am-3:30pm. Buses run every 15-20 minutes. The transportation desk telephone number is 617-939-5782. The number will be operational during the shuttle bus hours.

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Invited Session Changes

If government authors are unable to travel, announcements will be made at the beginning of each session.

• Exhibitors & Exhibit Hall •

Wednesday 10:00 am-6:00 pm
Thursday 10:00 am-4:30 pm
Friday 10:00 am-2:30 pm

The Exhibit Hall entrance is located through the lobby on Level 1. We hope you take some time each day to visit the Exhibit Hall and learn what's new. If you make buying decisions for your lab and have grant funds to allocate, it's one-stop-shopping under one roof! Or gather information to take back to your colleagues. New for 2013, there are 45-minute presentations by exhibitors daily. Check the schedule outside the Theater or the schedule on the Mobile App for more information.

Stop by ASHG Central to recharge your devices, meet the *AJHG* Editors, and get information on ASHG 2014 in San Diego. There will be coffee breaks in the morning, and refreshments during the lunch period. There is something for everyone in the Exhibit Hall!

Government Shutdown Effects on ASHG Exhibit Hall

As of timing of printing (October 15), the federal government remains shut down and as such, the following government exhibiting companies may not be present:

National Center for Biotechnology Information, Booth #755
National Human Genome Research Institute, Booth #840
NIH Therapeutics for Rare and Neglected Diseases, Booth #849

Exhibit Theater Presentation Change

The BioMarin Pharmaceutical session on Thursday, October 24, 11:15 am-12:00 pm title has been changed to:

The Affordable Care Act (ACA) and Insurance Access - Implications for Patients with Rare Genetic Diseases

Please welcome the following additional exhibitors to the ASHG 2013:

Alpaqua Engineering, LLC, Booth #1240

800-690-1620

ostelling@alpaqua.com

<http://www.alpaqua.com>

Alpaqua offers life science research tools including innovative magnet plates for use in bead-based nucleic acid purification. Alpaqua magnet plates are well recognized in the Genomics industry for their performance, reliability, and ease of use. Other products include temperature blocks, SBS tube racks, unique labware stations and Alpiillo® microplate supports.

BioDatomics, Booth #341

877-249-6660

info@biodatomics.com

<http://www.biodatomics.com/>

BioDatomics provides a powerful yet intuitive, open source NGS data analysis platform, BioDT, that helps genomic and biomedical researchers glean insights from their data more quickly. BioDT moves beyond the limitations of existing solutions by providing drag-and-drop workflow creation and

editing; greatly accelerated analysis execution; visualized results; and real-time collaboration.

DeNovix Inc. Booth #1230

302-442-6911

info@denovix.com

<http://www.denovix.com>

DeNovix, Inc. (Wilmington, DE) is an instrumentation company that designs, manufactures and sells laboratory equipment for life science applications. Our products include the DS-11 Spectrophotometer, a stand-alone microvolume spectrophotometer for quantification of nucleic acids and proteins. The DS-11 includes Android based OS, hi-res touchscreen including Wi-Fi, Ethernet and USB connectivity.

Enzymatics, Inc., Booth #456

978- 927-7027

mabraham@enzymatics.com

<http://www.enzymatics.com>

The Focus Foundation, Booth #353

410-798-7934

Info@thefocusfoundation.org

<http://Thefocusfoundation.org>

HughesRiskApps, Booth #452

617-724-0048

kshughes@partners.org

<http://TheBreastCancerSurgeon.org>

HughesRiskApps Identification and management of patients with hereditary syndromes: We use Health Information Technology to facilitate the identification and management of patients with hereditary syndromes before disease occurs. Our suite of software modules is freely available and includes pedigree drawing, risk clinic management and Clinical Decision Support (Risk models and guidelines).

Knome, Inc., Booth #1153

(617) 715-1000

jlee@knome.com

<http://www.knome.com>

Knome is a leading provider of human genome interpretation systems and services. We provide software tools and interpretation services that help researchers and clinicians understand the genetic basis of human disease and drug response. Knome's big data technologies are helping to pave the healthcare industry's transition to molecular-based, precision medicine.

Mawi DNA Technologies, Booth #355

855-DNA-SWAB

info@mawidna.com

<http://www.mawidna.com>

Mawi DNA Technologies is an emerging US biotech company that focuses on noninvasive sample collection device. The patented pending family line of iSWAB are a novel approach for collecting samples from human, animals, and environment. The iSWAB DNA is a simple and effective non-invasive collection device for high recovery human DNA (99%) with low bacterial DNA content (<1%) for for research and clinical testing.

MYcroarray, Booth #331

734-998-0751

info@mycroarray.com

<http://www.mycroarray.com>

MYcroarray is a leading manufacturer of custom molecular probes for genomic applications, including capture baits libraries for targeted sequencing, fluorescent probes libraries, oligonucleotide libraries and oligonucleotide microarrays. We are the only provider of truly error-free oligonucleotides for synthetic biology. We offer microarray hybridization, sequence capture and sequence discovery services.

MEDomics, LLC., Booth #343

626-804-3645

ssommer2013@medomics.com

<http://www.medomics.com>

Renishaw Inc., Booth #454

847-286-9953

usa@renishaw.com

Silicon Valley Biosystems, Inc., Booth #1149

650-265-6909

connect@svbio.com

<http://www.svbio.com>

Bio helps CLIA-certified labs launch and run next-gen sequencing based genetic tests by offering a clinical-grade sequence analysis and reporting platform. The FASTQ-to-results service enables labs to focus on their core competencies in sample processing and expert interpretation, while relieving them of otherwise burdensome software, hardware and personnel investments.

VerdeLabs, Booth #1157

(855) 698-3733

sales@verdelabs.com

VerdeLabs, a leader in next generation sample preparation is focused on improving the quality and consistency of your results. From sample prep to clean up and through the quantification process we are committed to your results. We guarantee our products. If your run fails, we will replace it.

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 to get the latest updates, tips, news, and announcements. Watch the #ASHG2013 live feed by visiting the Boston Commons area in the North Lobby, Level 1.



Invited Sessions and Invited Workshop Proposals: Due by December 6, 2013
Submission Site opens October 22, 2013.
www.ashg.org/2014meeting