National Society of Genetic Counselors

EXPANDING
the Spectrum of Genomic Health

NSGC 31ST ANNUAL EDUCATION CONFERENCE

October 24–27, 2012 | Hynes Convention Center | Boston, Massachusetts

Program Book

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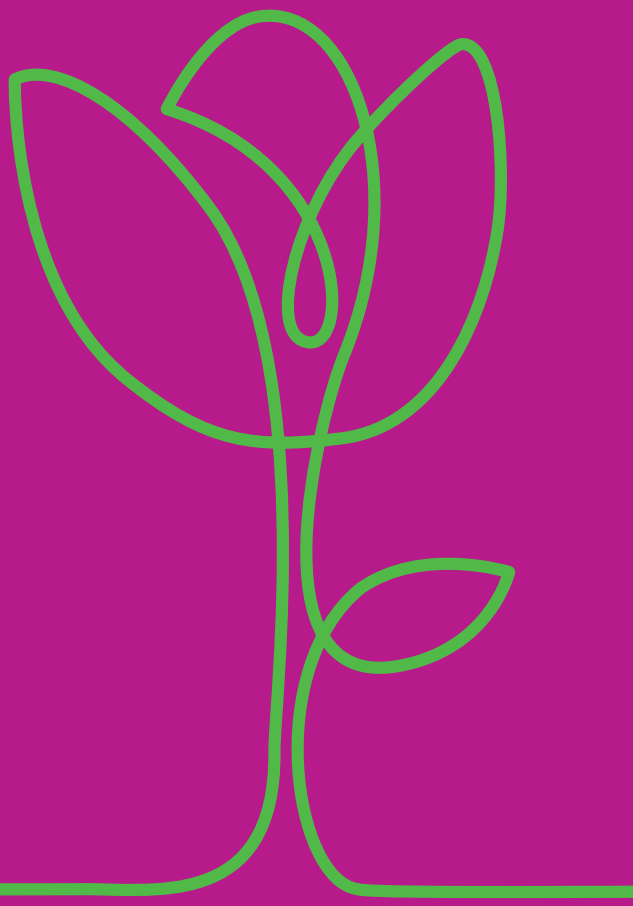

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EXPANDING
the Spectrum of Genomic Health

Welcome to Boston!

On behalf of the NSGC Annual Education Conference (AEC) Planning Subcommittee and the NSGC Board of Directors, thank you for joining us in Boston!

NSGC is expanding the spectrum of genomic health with more than 30 educational sessions designed to support your professional development. Session topics include genetic counseling research, ethical issues in genetic testing, personalized medicine and other topics on the forefront of genetics. Education highlights you will not want to miss include the Clinical Doctorate Plenary Session (page 13), the Dr. Beverly Rollnick Memorial Lecture (page 15) and the NSGC Professional Issues Panel (page 14). Reference pages 11-16 for sessions submitted/sponsored by your NSGC Special Interest Group. Make the most of your AEC schedule and plan to attend education sessions specific to your professional interests.

Expanding your expertise and professional development goes far beyond the valuable education taking place behind the lecture room walls. We encourage you to maximize your AEC experience and take advantage of the Welcome Reception, SIG meetings and other networking opportunities available to you this week. Visit the Exhibitor Suite to see the latest product offerings and services from our industry partners. Catch up with old friends and make new contacts during receptions, program reunions and daily breaks. Attend the NSGC and ABGC Business Meetings to learn more about the latest efforts of your professional organizations. There are so many ways to make this week an incredible experience!

We hope you enjoy your time here in Boston. Take time to learn and connect as we reflect on the advances of the past year and look to the future of genomics in this amazing city!

Claire N. Singletary, MS, CGC
2012 AEC Chair

Quinn Stein, MS, CGC
2012 AEC Vice-Chair

Table of Contents

Download the free 2012 NSGC AEC Mobile App!

This new, easy-to-use mobile app contains program information, the schedule-at-a-glance, educational content, maps and more!

See page 26 for more details.

National Society of Genetic Counselors

Statement of Purpose

The 31st Annual Education Conference (AEC) focuses specifically on the educational needs of genetic counselors. The AEC addresses a wide variety of genetic counseling practice areas and provides the latest information for the genetic counseling profession. Attendees will have access to education designed to support and enhance their current practice. The Exhibitor Suite will provide current information, products and services and the opportunity to talk with exhibitors about new developments in the genetic counseling field. The Pre-conference Symposia will provide in-depth information on specific topics relevant to the field of genetic counseling.

Continuing Education Units

The National Society of Genetic Counselors has been approved as an Authorized Provider by the International Association for Continuing Education and Training (IACET), 1760 Old Meadow Road, Suite 500, McLean, VA 22102. **The National Society of Genetic Counselors is authorized by IACET to offer up to 3.23 Category 1 CEUs or up to 32.25 Category 1 Contact Hours for the 31st Annual Education Conference**. CEUs and Contact hours offered by NSGC will be accepted by The American Board of Genetic Counseling (ABGC) for genetic counselor recertification.

To receive CEUs for a session, attendees must be present for the entire session. The total number of CEUs awarded will be determined by the number of sessions an individual attends. To earn CEUs, attendees MUST:

1. Scan the bar code on the attendee badge upon entering each CEU eligible session.
2. Report and indicate all sessions attended in the online system.
3. Complete an online electronic session evaluation for each session that is attended.

Per IACET requirements, completion of these three steps is mandatory for individuals to receive CEU/Contact Hour credit.

Attendees must retain a copy of this certificate for their records for five years from the date of the course. This form will serve as final Proof of Completion.

IACET requires NSGC to track attendance in sessions for those seeking continuing education credits. To replace the previously used sign-in sheets, there will be a scanner outside of each session room to scan the bar code on your attendee badge upon entering the room. Scanners will be outside of the session rooms for 10 minutes past the start time of the session. If you miss your opportunity to scan in, please see the registration desk to add your name to a session.

IMPORTANT: NSGC will issue a CEU confirmation e-mail following the conference. NSGC will only be able to verify the credits you earned by the portion of the sessions marked as "attended" in the online system.

Evaluation Process

Please assist NSGC in evaluating the AEC sessions and provide valuable input that will help NSGC plan future conferences.

Educational Session Evaluations

Participants are asked to complete online evaluations to provide input regarding individual speakers and educational content. We ask all attendees to complete an online session evaluation for each session attended. **Although individuals requesting CEUs MUST complete evaluations, NSGC would greatly appreciate feedback from all attendees**.

Poster Evaluation

Participants may also complete an electronic Poster Evaluation Form to evaluate the posters displayed. **Those seeking CEUs for viewing posters MUST complete a Poster Evaluation form**.

Concurrent Papers Evaluation

Concurrent papers feature six back-to-back presentations in four different categories running concurrently. The speaker in the first time slot is considered speaker 1, the speaker in the second time slot is considered speaker 2, and so on.

Evaluating various speakers:

If you plan to attend all six abstracts within the same category (no room change), complete just one electronic evaluation by filling in the session code and evaluating speakers 1 thru 6. If you plan to change categories/rooms between abstracts and would like to evaluate each speaker, you may complete up to four evaluations (one for each category, depending how many you attend). For each category, fill in the appropriate session code and then evaluate each speaker based on which time slot they presented in. The speaker in the first time slot is speaker 1, the speaker in the second time slot is speaker 2, and so on.

NOTE: To claim CEUs, you only need to complete one electronic evaluation for one category/session. You will claim the same session as "attended" in the online system.

Overall Conference Evaluation

Following the AEC, you will receive an electronic survey by e-mail requesting feedback about your overall conference experience. Please take a moment to complete this brief survey as your feedback is integral in planning future NSGC events.

Executive Office Information

NSGC Executive Office
401 N. Michigan Ave.
Suite 2200
Chicago, IL 60611 USA

Phone: 312.321.6834
Fax: 312.673.6972
Email: nsgc@nsgc.org
Website: www.nsgc.org

All online electronic forms can be found at https://www.showreg.net/NSGC1210S/CEUCreditlogin.asp.

Registration Hours

Tuesday, October 23	5:00 pm – 8:00 pm
Wednesday, October 24	7:00 am – 8:00 pm
Thursday, October 25	6:30 am – 7:30 pm
Friday, October 26	6:30 am – 7:30 pm
Saturday, October 27	6:30 am – 2:00 pm

Exhibitor Suite Hours

Wednesday, October 24	6:15 pm – 8:15 pm
Thursday, October 25	11:00 am – 2:00 pm
	5:30 pm – 7:30 pm
Friday, October 26	11:30 am – 4:00 pm

Message Center and Job Boards

Bulletin boards with push-pins are available for attendees to leave messages for colleagues or post job opportunities within the genetic counseling field. Advertising is not permitted. Materials posted will be monitored and inappropriate information is subject to removal.

Attendee List Information

Attendee lists are posted on the NSGC website prior to the conference, and an updated list will be posted after the conference. Lists are posted on a board in the Registration Area and are available for making copies (at the attendee's expense) at the Business Center on the Plaza Level near the South Lobby. Attendee lists may not be used for solicitation purposes other than networking. NSGC is not responsible for errors and/or omissions.

Handouts and Presentations

NSGC offers electronic versions of AEC handouts. NSGC does not provide paper copies of AEC session handouts at the conference. All session handouts (if provided by the speaker) are posted on the NSGC Website and will be available following the conference until March 1, 2013. To download handouts go to http://www.nsgc.org/2012AECHandouts/tabid/490/Default.aspx or use this QR Code.

If you are also registered for a Pre-conference Symposium, you will be given a separate link to access these handouts. Handouts will also be available for making copies (at the attendee's expense) in the Business Center.

Business Center Hours of Operation

Monday – Friday	7:00 am – 7:00 pm
Saturday and Sunday	7:00 am – 5:00 pm

Internet Access

Hotel

All NSGC attendees who stay at the Boston Marriott or the Sheraton Boston will receive a discounted rate for internet access in their guest rooms.

Convention Center

The Hynes Convention Center offers free wireless internet service throughout meeting rooms, lobbies and expo halls. Open your internet browser, and look for the "BCEC" or "Hynes Wireless Network." *This service is designed for casual use and is not guaranteed*

Attendees will also have complimentary access to the AEC Internet Pavilion in the Hynes Convention Center throughout the duration of the AEC. The Internet Pavilion is located on the third floor of the convention center.

Sponsored by Pfizer

Sponsored Breakfast, Lunch and Evening Sessions

Sponsored breakfast, lunch and evening sessions are open to the first 300 attendees and are located in the Hynes Convention Center, Room 312. Refer to the session objectives, pages 11-16. Continuing Education Unit approval is noted for eligible sessions.

2012 AEC Online Session Recordings

Maximize your AEC experience! View the sessions you missed in Boston, earn additional CEUs or access the valuable information you gathered on site. Take advantage of the opportunity to purchase the online session recordings — order today!

Session recording packages featuring all Pre-conference Symposia*, or featuring the AEC Plenary and EBS sessions*, are available for purchase. The online recordings will contain synched audio and PowerPoint® presentations for each session. To earn Category 1 CEUs, it is required that you complete and pass a quiz included at the conclusion of each session. Purchase your online recording package in conjunction with your AEC registration for a special discounted rate.**

If you register for the AEC only:

- AEC recordings — $59
- Pre-conference Symposia recordings — $119

If you register for a Pre-conference Symposium only:

- AEC recordings — $119
- Pre-conference Symposia recordings — $59

Best Combo Deal!

If you register for the AEC and Pre-conference Symposia:

- Receive both the Pre-conference Symposia and AEC recordings for the special package price of $99.

Registered attendees are able to order online content both during the AEC and following the conference at an increased rate. Not attending the AEC? Check the NSGC website in January 2013 for additional information and purchase availability.

*** With speaker approval**

**** Discounted package rates only available when purchased in conjunction with a conference registration.**

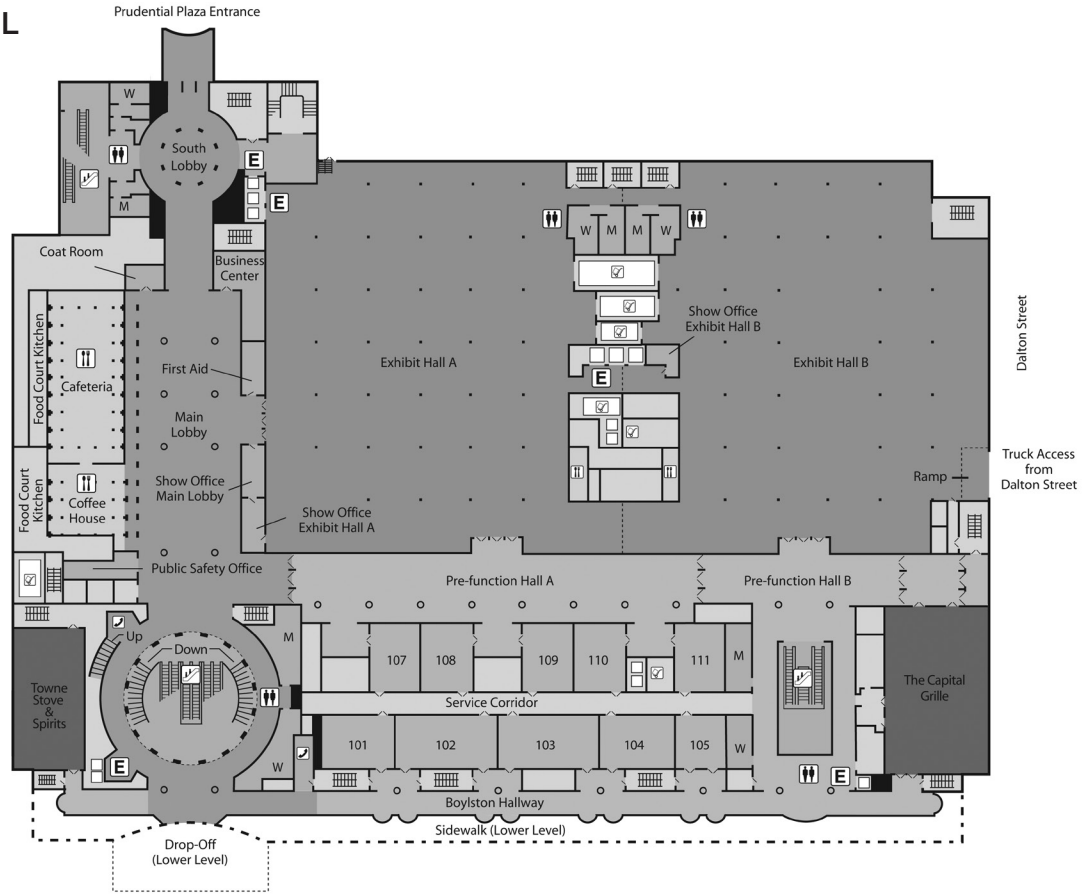
31st Annual Education Conference Schedule-at-a-Glance

Tuesday, October 23						
6:00 pm – 6:30 pm	**Moderator Orientation –** *Room 301*					
Wednesday, October 24						
7:00 am – 7:30 am	**Moderator Orientation –** *Room 301*					
8:00 am – 2:00 pm	**Pre-conference Symposia**					
	101 – Disorders of Sex Development: A Multidisciplinary Approach to Diagnosis, Treatment and Management of Care *Room 201,* See page 11	**102 – Surgical Management for Inherited Cancer Syndromes: Understanding the Options and Effects on Quality of Life** *Room 207,* See page 11	**103 – Measuring Patient Outcomes of Genetic Counseling: Why it's Important, How to Do it, and What Outcomes to Assess** *Room 203*, See page 11	**104 – The Impact of NextGen Sequencing of Cell-Free DNA on Prenatal Genetic Counseling** *Rooms 302/304/306,* See page 11	**105 – Epigenetics: Why DNA Sequence Isn't Everything** *Room 204,* See page 11	**106 – Universal Genetic Counseling Issues Viewed Through a Cardiovascular Lens** *Room 202* See page 11
2:00 pm – 2:30 pm	**Moderator Orientation –** *Room 301*					
2:00 pm – 3:30 pm	**NSGC 2013 Leadership Development Workshop –** *Room 305*					
2:00 pm – 3:30 pm	**Welcome to the AEC –** *Room 207*					
3:30 pm – 3:45 pm	**Opening Remarks –** *Room 302/304/306*					
3:45 pm – 5:15 pm	**Janus Series**					
	Plenary Session 107 – Practical Guidelines for Managing Patients with The 22q11.2 Deletion Syndrome **3:45 pm – 4:15 pm** *Room 302/304/306,* See page 11		**Plenary Session 108 – Complexities of Hereditary Pancreatitis: Clinical Review and Genetic Counseling Implications for Your Practice** **4:15 pm – 4:45 pm** *Room 302/304/306,* See page 11		**Plenary Session 109 – An Overview of Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Current Management and Developing Therapeutic Options** **4:45 pm – 5:15 pm** *Room 302/304/306*, See page 12	
5:30 pm – 5:45 pm	**Beth Fine Kaplan Best Student Abstract Award 110 – Exploring the Relationship Between Diagnoses of Congenital Anomalies or Genetic Conditions and Postpartum Depression: A Retrospective Study –** *Room 302/304/306,* See page 12					
5:45 pm – 6:00 pm	**Best Full Member Abstract Award 111 – Genetic Counseling for Individuals with Serious Mental Illnesses: The First, and Only, Randomized Controlled Trial –** *Room 302/304/306,* See page 12					
6:00 pm – 6:30 pm	**Natalie Weissberger Paul Award and Audrey Heimler Special Project Award –** *Room 302/304/306,* See page 28					
6:15 pm – 8:15 pm	**Exhibitor Suite Open –** *Hall D*					
6:30 pm – 8:00 pm	**Welcome Reception in Exhibitor Suite –** *Hall D*					
Thursday, October 25						
7:00 am – 7:30 am	**Moderator Orientation –** *Room 301*					
7:00 am – 7:45 am	**Sponsored Breakfast Session 201 – Is Noninvasive Prenatal Testing Ready For All Women? The Great Debate, Sponsored by Verinata Health, Inc. –** *Room 312,* See page 12					
8:00 am – 10:00 am	**Educational Breakout Sessions**					
	202 – Pan-ethnic Expanded Carrier Screening *Room 210,* See page 12	**203 – Dissecting the Utility of MSI/IHC on Colon Polyps and Extracolonic Cancers** *Room 302/304/306,* See page 12	**204 – Building a Brain: Understanding and Researching the Genetics of Brain Development and Function** *Room 207,* See page 12	**205 – The Future is Now: The Impact of Next Generation Sequencing as Applied to Whole Exome and Whole Genome Sequencing on the Diagnostic Laboratory and Clinical Practice** *Room 200,* See page 12		
10:15 am – 10:45 am	**NSGC State of the Society Address –** *Room 302/304/306,* See page 30					
10:45 am – 12:15 pm	**Plenary Session 206 – The Clinical Doctorate –** *Room 302/304/306,* See page 13					
11:00 am – 2:00 pm	**Exhibitor Suite Open –** *Hall D*					
12:00 pm – 2:15 pm	**Various Committee and Task Force Meetings**					
	Access and Service Delivery Committee Meeting **12:15 pm – 2:15 pm** *Room 200*	**Communications Committee Meeting** **12:15 pm – 2:15 pm** *Room 204*	**Education Committee Meeting** **12:15 pm – 1:15 pm** *Room 305*	**Genetic Counseling Advanced Degree Task Force** **12:15 pm – 2:15 pm** *Room 303*	**Membership Committee Meeting** **12:15 pm – 2:15 pm** *Room 311*	**Ethics Advisory Group Meeting** **12:30 pm – 2:00 pm** *Room 207*
	Various Special Interest Group Meetings					
	Psychiatric SIG Meeting **12:00 pm – 1:30 pm** *Room 208*	**Health IT SIG Meeting** **12:15 pm – 12:45 pm** *Room 210*				
12:30 pm – 2:00 pm	**Sponsored Lunch Session 207 – Early Experiences in Clinical Whole Exome Sequencing, Sponsored by Baylor College of Medicine –** *Room 312,* See page 13					
2:15 pm – 4:15 pm	**Educational Breakout Sessions**					
	208 – The New Frontier: A Clinical Trial of In Utero Stem Cell Transplantation for Sickle Cell Disease *Room 200,* See page 13	**209 – Beyond Risk Assessment: Novel Application of Whole Genome Technologies to Personalize Oncology Treatment** *Room 302/304/306,* See page 13	**210 – A Hearing Aid for Genetic Counselors: The Full Picture of Hereditary Hearing Loss** *Room 210*, See page 13	**211 – Finding the Teacher Within: Working More Effectively with Patients, Peers and Pupils** *Room 207,* See page 13		
4:30 pm – 5:15 pm	**Plenary Session 212 – Clinical Genetic Resources at NCBI –** *Room 302/304/306,* See page 13					
5:15 pm – 6:00 pm	**Plenary Session 213 – Running with the Trait: Ethical and Societal Implications of Mandatory Testing of Athletes for Sickle Cell Trait –** *Room 302/304/306,* See page 13					
5:30 pm – 7:30 pm	**Exhibitor Suite Open –** *Hall D*					
6:00 pm – 7:30 pm	**Posters with Authors Session 214 –** *Hall D,* See page 21					

Thursday, October 25 (continued)						
7:00 pm – 8:00 pm	**International SIG Meeting –** *Room 311*					
7:30 pm – 9:00 pm	**University of Pittsburgh Reunion –** *Room 200*					
7:30 pm – 9:00 pm	**Sponsored Evening Session 215 – Termination of Pregnancy for Indications of Genetic Disorder and Fetal Abnormality in Advanced Gestations at Boulder Abortion Clinic, Sponsored by Boulder Abortion Clinic, P.C. –** *Room 312,* See page 14					
8:00 pm – 9:00 pm	**Telegenetics SIG Meeting –** *Room 311*					
8:00 pm – 10:00 pm	**Sarah Lawrence College Reception –** *Room 210*					
8:30 pm – 10:00 pm	**Mount Sinai Reunion –** *Room 207*					
9:00 pm – 10:30 pm	**University of Minnesota Reunion –** *Fairfax Room, Sheraton Boston*					
Friday, October 26						
7:00 am – 7:30 am	**Moderator Orientation –** *Room 301*					
7:00 am – 7:45 am	**Past Board Member Breakfast –** *Room 205*					
7:00 am – 7:45 am	**Sponsored Breakfast Session 301 – Non-Invasive Prenatal Testing: A New Era for Fetal Trisomy Detection, Sponsored by Intergrated Genetics –** *Room 312,* See page 14					
8:00 am – 9:00 am	**Plenary Session 302 – Professional Issues Panel,** *Room 302/304/306,* See page 14					
9:00 am – 9:45 am	**Plenary Session 303 – Non-Invasive Prenatal Diagnosis Beyond Trisomy 21,** *Room 302/304/306,* See page 14					
10:00 am – 12:00 pm	**Educational Breakout Sessions**					
	304 – Merging into Clinical Practice: Updates in Non-invasive Prenatal Testing *Room 302/304/306,* See page 1	**305 – Now What am I Supposed to Tell My Fiancé? Psychosocial and Behavioral Outcomes for Unaffected Women with a *BRCA* Mutation** *Room 210,* See page 14	**306 – Genetic Aortopathies: An Explosion of Genetic Knowledge** *Room 200,* See page 14	**307 – Pharmacogenetics: Update on the Field and Roles for Genetic Counselors** *Room 207,* See page 14		
11:30 am – 4:00 pm	**Exhibitor Suite Open –** *Hall D*					
11:45 am – 2:00 pm	**Various Committee Meetings**					
	Public Policy Committee, 11:45 am – 12:45 pm *Room 209*	**Practice Guidelines Committee, 11:45 am – 1:45 pm** *Room 205*	**Perspectives in Genetic Counseling (PGC) Committee, 12:15 pm – 1:15 pm** *Room 208*			
	Various Special Interest Group Meetings					
	Neurogenetics SIG 11:45 am – 12:45 pm *Room 204*	**ART/Infertility SIG 12:15 pm – 1:15 pm** *Room 210*	**Education SIG 12:15 pm – 1:15 pm** *Room 311*	**Cancer SIG 12:15 pm – 2:00 pm** *Room 200*	**Cardiovascular SIG 12:45 pm – 1:45 pm** *Room 204*	**Prenatal SIG 12:45 pm – 1:45 pm** *Room 207*
12:00 pm – 1:00 pm	**Texas Society of Genetic Cousnelors –** *Room 305*					
12:15 pm – 1:45 pm	**Sponsored Lunch Session 308 – Hereditary Cancer Panels: Review of Initial Results and Discussion about Counseling for Next-Generation Tests, Sponsored by Ambry Genetics –** *Room 312,* See page 15					
2:00 pm – 3:30 pm	**Concurrent Papers Sessions 309-312 –** (See session grid on page 18 for room assignments)					
3:45 pm – 4:00 pm	**Passport to Prizes Drawing –** *Hall D*					
4:00 pm – 4:45 pm	**NSGC Annual Business Meeting –** *Room 302/304/306,* See page 30					
4:45 pm – 5:30 pm	**NSGC Leadership Awards –** *Room 302/304/306,* See page 28					
5:30 pm – 7:00 pm	**Dr. Beverly Rollnick Lecture 313 – Part 1: That's Just Crazy Talk: A Play and Discussion; Part 2: Research Evaluation of a Play as a Stigma-Reduction Intervention –** *Room 302/304/306,* See page 15					
7:00 pm – 8:00 pm	**Metabolic/LSD SIG –** *Room 209*					
7:15 pm – 8:45 pm	**Sponsored Evening Session 314 – When More Isn't Better...Genetic Testing from a Legal Perspective, Sponsored by Quest Diagnostics –** *Room 312,* See page 15					
7:30 pm	**Brandeis Reunion –** *Brandeis University*					
Saturday, October 27						
7:00 am – 7:45 am	**Sponsored Breakfast Session 401 – When Bigger is Better: Multi-gene Testing Panels for Genetically Heterogeneous Disorders, Sponsored by GeneDx –** *Room 312,* See page 15					
8:00 am – 9:00 am	**Plenary Session 402 – Late Breaking: Targeted Pharmaceuticals for Neurodevelopmental Disorders –** *Room 302/304/306,* See page 15					
9:00 am – 9:30 am	**Incoming Presidential Address –** *Room 302/304/306,* See page 30					
9:30 am – 10:30 am	**Plenary Session 403 – Jane Engelberg Memorial Fellowship (JEMF) Presentation –** *Room 302/304/306,* See page 15					
10:30 am – 12:30 pm	**Educational Breakout Sessions**					
	404 – Counseling Difficulties and Ethical Concerns in an Uncertain Prenatal Diagnosis *Room 200,* See page 15	**405 – Familial Pancreatic Cancer: From Evaluation to Endoscopy** *Room 210,* See page 15	**406 – The New Landscape of Genetic Testing: How to Approach Testing Minors for Adult-onset Conditions in the Era of Large-scale Genomic Testing** *Room 302/304/306,* See page 16	**407 – Hints From the Experts: Job Interviewing Skills** *Room 207,* See page 16		
12:30 pm – 2:00 pm	**Sponsored Lunch Session 408 – Using Cost Effectiveness Modeling to Demonstrate the Value of Genetic Services, Sponsored by Myriad Genetic Laboratories, Inc. –** *Room 312,* See page 16					
12:30 pm – 2:00 pm	**ABGC Annual Business Meeting –** *Room 302/304/306,* See page 30					
12:30 pm – 2:15 pm	**Various Group Meetings**					
	Pediatric and Clinical Genetics SIG 12:30 pm – 1:30 pm *Room 305*	**Research SIG 12:30 pm – 1:30 pm** *Room 309*	**Student/New Member SIG 12:45 pm – 1:45 pm** *Room 200*	**Industry SIG 12:45 pm – 2:00 pm** *Room 208*	**Personalized Medicine SIG 12:45 pm – 2:15 pm** *Room 207*	
2:00 pm – 3:30 pm	**Concurrent Papers Sessions 409-412 –** (See session grid on page 19 for room assignments)					

Hynes Convention Center Floor Plan

PLAZA LEVEL



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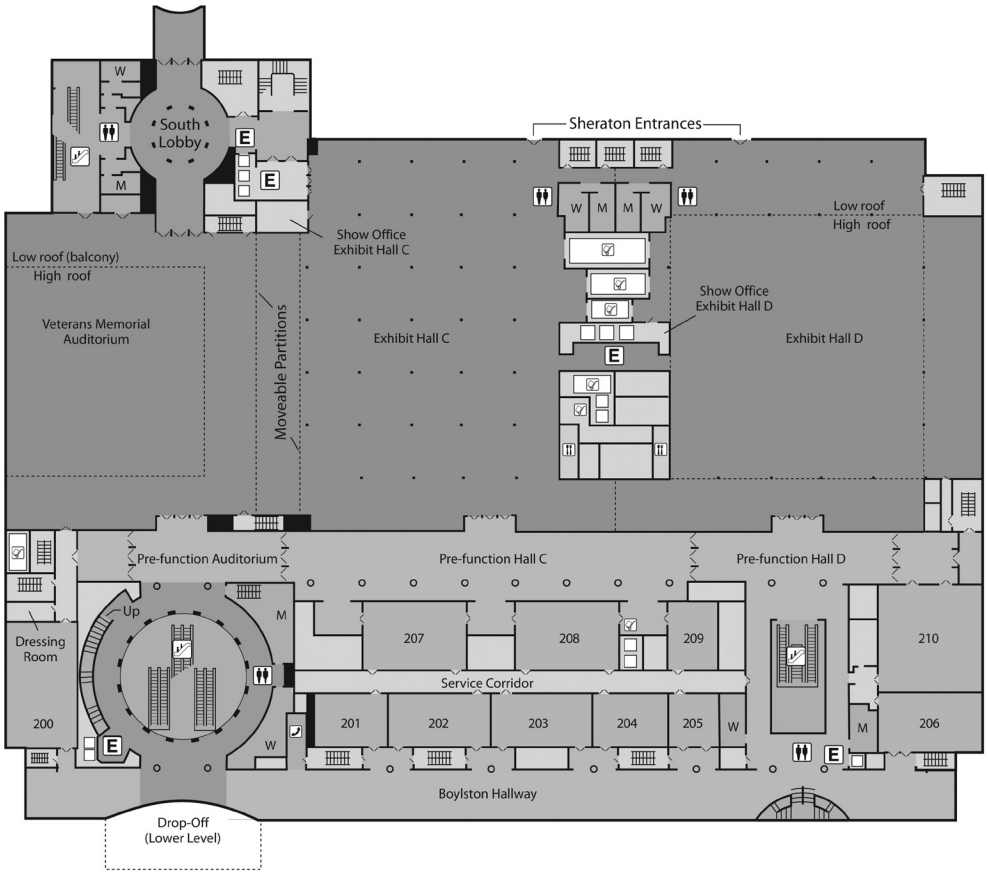

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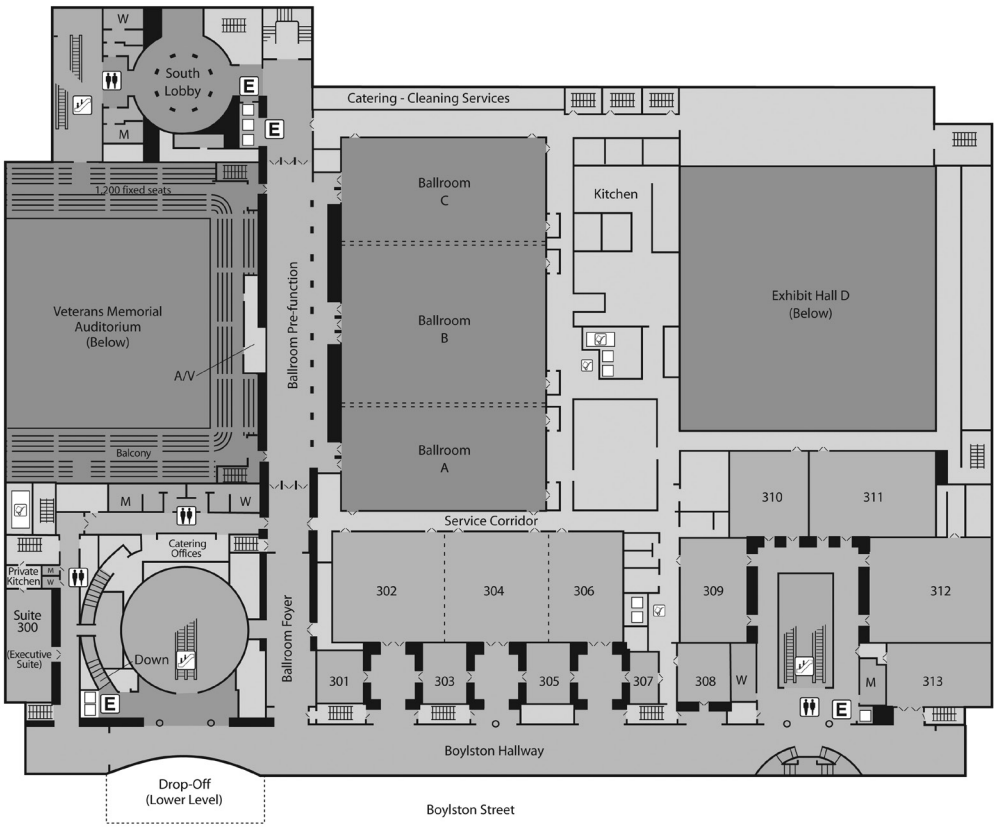

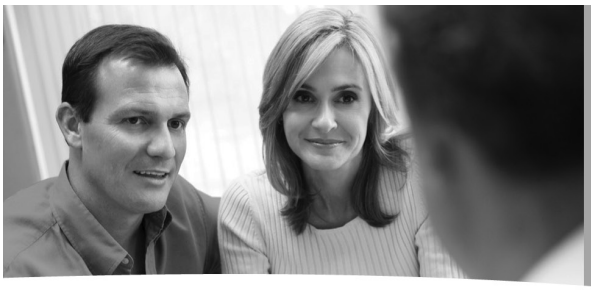

MaterniT21™ PLUS

A noninvasive prenatal laboratory-developed test that analyzes the relative amount of 21, 18, 13 and Y chromosomal material.

Clear

Developed by Sequenom CMM, the technology and data behind the MaterniT21 PLUS test are extensively studied and well published.

Convenient

A noninvasive laboratory-developed test that can be used in the 1st or 2nd trimester, as early as 10 weeks' gestation.

- Commercial experience with thousands of tests
- Genetic counselors available to consult about our testing services
- Customer service and support are available to answer your questions

Compelling

The MaterniT21 PLUS test detects fetal chromosomal 21, 18, 13 aneuploidies, and the presence of the Y chromosome, in singletons, twins and higher order multiple pregnancies.

Learn More

For more information, or to have a Business Development Manager contact you, please call us toll-free (within the US) at

877.821.7266

Visit www.SequenomCMM.com

Visit our booth #100 at NSGC

1. DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. Palomaki GE, Kloza EM, Lambert-Messerlian GM, Haddow JE, Neveux LM, Ehrich M, van den Boom D, Bombard AT, Deciu C, Grody WW, Nelson SF, Canick JA. *Genet Med*. 2011 Nov;13(11):913-920.
2. DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13, as well as Down syndrome: an international collaborative study.Palomaki GE, Deciu C, Kloza EM, Lambert-Messerlian GM, Haddow JE, Neveux LM, Ehrich M, van den Boom D, Bombard AT, Grody WW, Nelson SF, Canick JA. *Genet Med*. 2012 Mar;14(3):296-305.
3. Canick JA, Kloza EM, Lambert-Messerlian GM, Haddow JE, Ehrich M, van den Boom D, Bombard AT, Deciu C, Palomaki GE. DNA sequencing of maternal plasma to identify Down syndrome and other trisomies in multiple gestations. [published online ahead of print May 14, 2012] *Prenat Diagn*. doi: 10.1002/pd.3892.

SEQUENOM® CMM®
CENTER FOR MOLECULAR MEDICINE

Clarity for cancer.

Hereditary cancer testing from Ambry Genetics delivers real results. Our panels for hereditary breast, ovarian, colon and other cancers are helping clinicians—and their patients—discover answers to help guide important management decisions. To learn more stop by the Ambry booth (#109) at NSGC or visit ambrygen.com

DON'T MISS AMBRY'S SPONSORED LUNCH AT NSGC

Hereditary Cancer Panels: Review of Initial Results and Discussion About Counseling for Next-Generation Tests

Friday, October 26, 2012

12:15 – 1:45 pm

Ambry Genetics™

Wednesday, October 24

Pre-conference Symposia

8:00 am – 2:00 pm

101 Disorders of Sex Development: A Multidisciplinary Approach to Diagnosis, Treatment and Management of Care

0.50 CEU

1. V. Reid Sutton, MD, Department of Molecular & Human Genetics Baylor College of Medicine & Texas Children's Hospital; 2. Elise G. Austin, MS, GC, Department of Molecular & Human Genetics Baylor College of Medicine & Texas Children's Hospital; 3. Ingrid A. Holm, MD, MPH, Divisions of Genetics and Endocrinology Children's Hospital Boston; 4. David A. Diamond, MD, Children's Hospital Boston Gender Management Service

- Define disorders of sex development (DSD) and review strategies to sensitively counsel individuals with DSD.
- Examine diagnostic complications and treatment options of DSD; illustrate the importance of a multidisciplinary, patient-centered approach to care.
- Identify ethical and psychosocial issues associated with gender assignment.

Submitted/Sponsored by the Pediatric SIG

102 Surgical Management for Inherited Cancer Syndromes: Understanding the Options and Effects on Quality of Life

0.50 CEU

1. Michael A. Howard, MD, NorthShore University Health System; 2. Sarah Y. Boostrom, MD, The Mayo Clinic; 3. Jack Basil, MD, TriHealth - Tristate Gynecologic Oncology; 4. Sam S. Yoon, MD, Massachusetts General Hospital Cancer Center; 5. Pardeep Kaurah, MSc, BC Cancer Agency

- Review prophylactic surgical options (including reconstructive techniques) for common inherited cancer predisposition syndromes (e.g. HBOC, Lynch, FAP, HDGC, etc.), including technical aspects of each surgery.
- Describe clinical outcomes and risk reduction achieved through prophylactic surgery.
- Examine quality of life outcome data for the various surgeries as it pertains to high risk populations.

Submitted/Sponsored by the Cancer SIG

103 Measuring Patient Outcomes of Genetic Counseling: Why It's Important, How to Do it, and What Outcomes to Assess

0.50 CEU

1. Jehannine C. Austin, CGC/CCGC, PhD, University of British Columbia; 2. Barbara B. Biesecker, MS, CGC, PhD, NHGRI; 3. Gillian Hooker, PhD, ScM, CGC, NHGRI; 4. Catriona Hippman, MSc, CGC, BC Women's Hospital & Health Centre

- Discuss the importance of randomized controlled trials in relation to rigorously evaluating the effects of genetic counseling on client outcomes.
- Identify gaps in the existing literature relating to the client outcomes of genetic counseling in different contexts.
- Develop ideas for how to evaluate client outcomes of genetic counseling in different contexts.

Submitted by the Research SIG and JEMF Advisory Group
Sponsored by the JEMF Advisory Group

104 The Impact of NextGen Sequencing of Cell-Free DNA on Prenatal Genetic Counseling

0.50 CEU

1. Edward Kloza, MS, CGC, Division of Medical Screening and Special Testing Department of Pathology and Laboratory Medicine, Women & Infants' Hospital; 2. Glenn Palomaki, PhD, Division of Medical Screening and Special Testing Department of Pathology and Laboratory Medicine, Women & Infants' Hospital; 3. Margaret Piper, PhD, MPH, Blue Cross Blue Shield Association Technology Evaluation Center; 4. Stephanie Dukhovny, MD, Brigham and Women's Hospital; 5. Brian Skotko, MD, Massachusetts General Hospital - Division of Genetics; 6. Diana Bianchi, MD, Tufts University School of Medicine

- Identify the challenges posed to third party payers, primary prenatal care providers, and maternal-fetal medicine specialists by the expansion of cfDNA technology.
- Determine whether next-gen prenatal testing influences genetic counselors' perceptions of individuals with Down syndrome and other trisomies.
- Evaluate the potential role of next-gen technology in prenatal testing for sex aneuploidies, single gene disorders and pregnancy complications.

Submitted/Sponsored by the Prenatal SIG

105 Epigenetics: Why DNA Sequence Isn't Everything

0.50 CEU

1. J. Cecelia Bellcross, PhD, MS, CGC, Emory University School of Medicine; 2. Bradford Coffee, PhD, Emory University School of Medicine; 3. Rosanna Weksberg, MD, PhD, The Hospital for Sick Children; 4. Emily Edelman, MS, CGC, NCHPEG; 5. Kate Reed, MPH, ScM, CGC, NCHPEG; 6. Angela Ting, PhD, Genomic Medicine Institute (NE50); 7. Scott Weissman, MS, CGC, NorthShore University Health System; 8. Robert A.Waterland, PhD, Depts. of Pediatrics and Molecular & Human Genetics at Baylor College of Medicine

- Describe the epigenome's role in human development and disease.
- Recognize the testing techniques available for epigenetic assessment in research and clinical practice.
- Apply knowledge of epigenetics to patient care for risk assessment, diagnosis and management.

Submitted by the Personalized Medicine SIG and Public Health SIG
Sponsored by the Personalized Medicine SIG

106 Universal Genetic Counseling Issues Viewed Through a Cardiovascular Lens

0.50 CEU

1. Christina M. Rigelsky, MS, CGC, Cleveland Clinic; 2. Ana Morales, MS, CGC, The Ohio State University; 3. Peggy Crawford, PhD, University of Cincinnati, Department of Neurology; 4. Sonia Suter, MS, JD, George Washington University; 5. Brittan Sutphin, Sudden Arrhythmia Death Syndromes Foundation; 6. Heather MacLeod, MS, CGC, Informed Medical Decisions, Inc.

- Identify best practices to communicate difficult information to children and overcome family communication barriers.
- Assess a family history of heart attack in the pedigree and learn appropriate follow-up questions to help determine different etiologies of this common pedigree finding.
- Recognize important issues for genetic counselors to consider with respect to postmortem genetic testing.

Submitted/Sponsored by the Cardiovascular SIG

Janus Series

3:45 pm – 4:15 pm

107 Practical Guidelines for Managing Patients With The 22q11.2 Deletion Syndrome

0.05 CEU

1. Donna M. McDonald-McGinn, MS, CGC, The Children's Hospital of Philadelphia

- Recognize the current best scope of practice specific to The 22q11.2 Deletion Syndrome, including all aspects of genetic counseling.
- Incorporate the published Practical Guidelines for Managing Patients With The 22q11.2 Deletion Syndrome into everyday best practice and understand how these apply to individuals with atypical nested deletions and 22q11.2 duplications.
- Recognize the importance of experienced coordinated team care for individuals with The 22q11.2 Deletion Syndrome and define the purpose and outcomes to date of The International 22q11.2 Deletion Syndrome Consortium.

4:15 pm – 4:45 pm

108 Complexities of Hereditary Pancreatitis: Clinical Review and Genetic Counseling Implications for Your Practice

0.05 CEU

1. Sheila Solomon, MS, CGC, University of Pittsburgh

- Provide a comprehensive clinical and genetic review of hereditary pancreatitis.
- Evaluate the multi-factorial data associated with a clinical diagnosis of pancreatitis in patients and their at-risk relatives.
- Explain the complexities of genetic testing on predicting phenotype and recurrence risk along with the associated psychosocial/genetic counseling issues.

Submitted/Sponsored by the Cancer SIG

Wednesday, October 24 (Continued)

4:45 pm – 5:15 pm

109 An Overview of Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Current Management and Developing Therapeutic Options

0.05 CEU

1. Catherine Walsh Vockley, MS, CGC, Children's Hospital of Pittsburgh

- Distinguish among the three forms of VLCAD deficiency, including presentations and current recommended management.
- Explain the various experimental therapeutic options for patients and have resources for referral of patients to study sites.
- Recognize the value of early identification of patients with rare disorders in order to develop long term follow up programs like the Inborn Errors of Metabolism Collaborative, to clarify natural history and contribute to development of new therapies and management practices.

Submitted/Sponsored by the Metabolism/Lysosomal Storage Disorders SIG

Best Abstract Awards

5:30 pm – 5:45 pm

Beth Fine Kaplan Best Student Abstract Award

110 Exploring the Relationship Between Diagnoses of Congenital Anomalies or Genetic Conditions and Postpartum Depression: A Retrospective Study

0.025 CEU

1. Meagan F. Wiesenhart, MS, The University of Alabama at Birmingham

- Examine the association of a woman having an infant with a congenital anomaly and/or specific genetic diagnosis and experiencing postpartum depression.
- Consider utilization of the Edinburgh Postnatal Depression Scale as a method of screening for postpartum depression in genetic counseling sessions.
- Recognize areas of genetic counseling practice that may include postpartum depression anticipatory guidance and/or screening as well as genetic counselors' suitability for these roles.

5:45 pm – 6:00 pm

Best Full Member Abstract Award

111 Genetic Counseling for Individuals with Serious Mental Illnesses: The First, and Only, Randomized Controlled Trial

0.025 CEU

1. Catriona Hippman, MSc, CGC, BC Women's Hospital & Health Centre

- Identify key components of genetic counseling for psychiatric illnesses and recognize the similarities and differences between genetic counseling for psychiatric illnesses as compared to other illnesses.
- Evaluate evidence from the first RCT of psychiatric genetic counseling, including the impact of genetic counseling on the outcome measures: risk perception, knowledge, internalized stigma, and perceived control over illness.
- Recognize the benefit of leveraging research results to enhance clinical practice and expand service delivery.

Thursday, October 25

Sponsored Breakfast Session

7:00 am – 7:45 am

201 Is Noninvasive Prenatal Testing Ready For All Women? The Great Debate

0.05 CEU

1. Christin Coffeen, MS, CGC; 2. Patricia Devers, MS, CGC; 3. Amy Swanson, MS, CGC

- Recognize similarities/differences between the major NIPT studies published to date.
- List reasons why NIPT should currently be offered to all pregnant women.
- Identify reasons why NIPT should not necessarily be offered to all pregnant women at this time.

Sponsored by Verinata Health, Inc.

Educational Breakout Sessions

8:00 am – 10:00 am

202 Pan-ethnic Expanded Carrier Screening

0.20 CEU

1. Shivani Nazareth, MS, CGC, Counsyl; 2. Gabriel A. Lazarin, MS, CGC, Counsyl; 3. Jason Flanagan, MS, CGC, Sanford Women's Health; 4. Krista Moyer, MS, CGC, San Francisco Perinatal Associates

- Describe the experience of pan-ethnic, expanded carrier screening in the preconception and prenatal setting.
- Review current allele frequency data on testing a nationwide, multi-ethnic population for many recessive diseases.
- Discuss established criteria and public health precedents in reference to pan-ethnic carrier screening.

Submitted/Sponsored by the Prenatal SIG

203 Dissecting the Utility of MSI/IHC on Colon Polyps and Extracolonic Cancers

0.20 CEU

1. Elena Stoffel, MD, MPH, University of Michigan Health System; 2. Russell Broaddus, MD, PhD, The University of Texas: MD Anderson; 3. Sarah Kerr, MD, Mayo Clinic; 4. Maegan Roberts, MS, CGC, Mayo Clinic

- Determine the positive or negative predictive value and clinical significance of MSI/IHC testing on endometrial and ovarian tumors, sebaceous neoplasms and colorectal polyps.
- Discuss the utility of MSI/IHC testing on gynecologic tumors based on histologic subtype.
- Identify key points for selecting a tumor for MSI/IHC testing.

Submitted/Sponsored by the Cancer SIG

204 Building a Brain: Understanding and Researching the Genetics of Brain Development and Function

0.20 CEU

1. A. James Barkovich, MD, University of California San Francisco; 2. Christopher Walsh, MD, PhD, Children's Hospital Boston; 3. Annapurna Poduri, MD, MPH, Boston Children's Hospital ; 4. Bernard Chang, MD, Harvard Medical School/ Beth Israel Deaconess Medical Center

- Define normal brain formation, the basis of structural abnormalities seen on brain imaging and the spectrum of normal.
- Review the genetic basis of various congenital brain anomalies and seizure conditions.
- Summarize the status, successes and pitfalls in using traditional gene analysis and whole genome and exome technologies in diagnosing and researching brain-based anomalies.

205 The Future is Now: The Impact of Next Generation Sequencing as Applied to Whole Exome and Whole Genome Sequencing on the Diagnostic Laboratory and Clinical Practice

0.20 CEU

1. Monica Giovanni, MS, CGC, Brigham and Women's Hospital; 2. Sherri Bale, PhD, FACMG, GeneDx; 3. Michael Murray, MD, Brigham and Women's Hospital

- Review next generation sequencing and CLIA-approved whole exome testing to provide a basic understanding of the technology and the testing laboratory's processes for analyzing and filtering genetic variants.
- Describe how genome-level data can be used to identify actionable variants and the impact on patients' test results.
- Discuss how physicians and genetic counselors approach and apply whole exome/genome sequencing tests in the clinic including pre and post-test considerations.

Submitted/Sponsored by the Personalized Medicine SIG

Plenary Session

10:45 am – 12:15 pm

206 The Clinical Doctorate

0.15 CEU

1. Ann Walker, MA, LCGC, Past President ABGC and NSGC, University of California-Irvine; 2. Brenda Finucane, MS, CGC, President, NSGC; 3. Catherine Reiser, MS, CGC, President, Association of Genetic Counseling Program Directors, University of Wisconsin-Madison; 4. Robin Grubs, PhD, CGC, Past President ABGC, University of Pittsburgh

- Distinguish between the PhD, the entry level CD and the Advanced Practice CD as distinct academic pursuits.
- List a potential benefit and a potential disadvantage of a Clinical Doctorate for Genetic Counselors from each of the following perspectives: professional, educational and accreditation/credentialing.
- Identify and prioritize factors that influence opinions about the possible development of a Clinical Doctorate.

Submitted by the Association of Genetic Counseling Program Directors

Sponsored Lunch Session

12:30 pm – 2:00 pm

207 Early Experiences in Clinical Whole Exome Sequencing

0.10 CEU

1. Alicia Braxton, MS, CGC; 2. Elise Austin, MS, CGC; 3. Gayle Patel, MS, CGC; 4. Sandra Darilek, MS, CGC

- Discuss the history, methodology, strengths, and limitations of whole-exome sequencing (WES).
- Review the ideal candidates for WES and points to consider in the consent process for testing.
- Illustrate results a provider will receive from WES.

Sponsored by Baylor College of Medicine

Educational Breakout Sessions

2:15 pm – 4:15 pm

208 The New Frontier: A Clinical Trial of In Utero Stem Cell Transplantation for Sickle Cell Disease

0.20 CEU

1. Kwaku Ohene-Frempong, MD, The Children's Hospital of Philadelphia; 2. Stefanie Kasperski, MS, CGC, The Children's Hospital of Philadelphia; 3. Alan Flake, MD, FACS, FAAP, The Children's Hospital of Philadelphia; 4. Moriah Eberhard, MS, CGC, The Children's Hospital of Philadelphia

- Describe the rationale and experimental support for IUHCT for the treatment of genetic disease.
- Compare and contrast potential risks and benefits in utero stem cell transplantation for sickle cell disease to postnatal bone marrow transplantation.
- Incorporate knowledge gained into prenatal genetic counseling practice.

209 Beyond Risk Assessment: Novel Application of Whole Genome Technologies to Personalize Oncology Treatment

0.20 CEU

1. Victoria Raymond, MS, CGC, University of Michigan; 2. Scott Kim, PhD, MD, Department of Psychiatry and Center for Bioethics and Social Sciences in Medicine, University of Michigan; 3. Jessica Everett, MS, CGC, University of Michigan

- Describe the current and future approaches to genome-based personalized oncology treatment.
- Recognize the oncologic, genomic and psychosocial factors important for care collaborations in personalized oncology treatment.
- Identify unique roles for genetic counselors in multidisciplinary personalized oncology treatment programs.

Submitted/Sponsored by the Personalized Medicine SIG and Cancer SIG

210 A Hearing Aid for Genetic Counselors: The Full Picture of Hereditary Hearing Loss

0.20 CEU

1. Margaret Kenna, MD, MPH, Children's Hospital Boston Department of Otolaryngology and Communication Enhancement; 2. Heidi Rehm, PhD, FACMG, Partners Healthcare Center for Personalized Genetic Medicine; 3. Katherine A. Lafferty, MS, CGC, PCPGM Laboratory for Molecular Medicine; 4. Mark Dunning, The Coalition of Usher Syndrome Research

- Summarize the current clinical and genetic aspects of hearing loss and outline the future directions of this field.
- Communicate the many components, both clinical and technical, that are involved in determining the etiology of hearing loss including, recognizing the benefits and limitations to genetic testing, testing strategies and subsequent impacts on patient care.
- Illustrate the psychosocial implications of hearing loss on a patient and their family and describe available support and resources using examples from an experienced counselor in the specialty of hearing loss and a parent advocate story.

211 Finding the Teacher Within: Working More Effectively with Patients, Peers and Pupils

1. Janet Mulliner, EdD, New York University; 2. Leslie Cohen, MS, CGC, University Hospitals Case Medical Center - Center for Human Genetics; 3. Kelly East, MS, CGC, Hudson Alpha Institute for Biotechnology

- Identify key concepts of adult learning theory, including experiential learning, collaborative learning and the power of stories in learning.
- Apply the concepts of adult learning theory to the day-to-day interaction of genetic counselors with patients, healthcare professionals and students.
- Recognize the needs of the NSGC membership for additional resources on educational topics, as well as identify resources that are currently available.

Submitted/Sponsored by the Education SIG

Plenary Session

4:30 pm – 5:15 pm

212 Clinical Genetic Resources at NCBI

0.075 CEU

1. Brandi L. Kattman, MS, CGC, NCBI/NLM/NIH; 2. Adriana J. Malheiro, MS, NCBI/NLM/NIH

- Discover how to become a power user of the GTR with MyNCBI and other navigation tips.
- Demonstrate how to find a test by a variant, and identify the variant's clinical significance.
- List at least four databases at NCBI of use to genetics professionals.

5:15 pm – 6:00 pm

213 Running with the Trait: Ethical and Societal Implications of Mandatory Testing of Athletes for Sickle Cell Trait

0.075 CEU

1. Charmaine Royal, PhD, MS, Duke University, Duke Institute for Genome Sciences & Policy

- Describe the historical context of sickle cell screening and need for a socially responsible athletic screening program.
- Identify areas of knowledge among athletes and athletic personnel that need to be enhanced to ensure informed consent and ultimate safety of the student-athlete.
- Demonstrate ways genetic counselors can use their skills to meet the needs of such screening programs in their area.

Submitted by the Diversity Subcommittee

Thursday, October 25 (Continued)

Sponsored Evening Session

7:30 pm – 9:00 pm

215 Termination of Pregnancy for Indications of Genetic Disorder and Fetal Abnormality in Advanced Gestations at Boulder Abortion Clinic
0.10 CEU

1. Warren M. Hern, MD, MPH, PhD

- Identify the relevance of these services to genetic counselors.
- Identify purpose, basic principles and components of clinical practice including grief support.
- Identify the basic operative procedures and clinical results of this care.

Sponsored by Boulder Abortion Clinic, P.C.

Friday, October 26

Sponsored Breakfast Session

7:00 am – 7:45 am

301 Non-invasive Prenatal Testing: A New Era for Fetal Trisomy Detection
0.05 CEU

1. Tom Musci, MD; 2. Vivian Weinblatt, MS, CGC

- Describe current prenatal aneuploidy screening and barriers to patient acceptance.
- Compare and contrast the available technical approaches to analyzing cell free DNA in maternal blood.
- List two reasons why the percentage of cell free fetal DNA in maternal blood is an important feature of overall non-invasive prenatal testing technology.
- Relate the overall sensitivity and specificity of cell free DNA technology in detecting common fetal trisomy when counseling prenatal clients about non-invasive prenatal testing (NIPT) options.

Sponsored by Integrated Genetics

Plenary Sessions

8:00 am – 9:00 am

302 Professional Issues Panel
0.10 CEU

1. John Richardson, Government Relations Director, NSGC; 2. Joy Larsen Haidle, MS, CGC, Humphrey Cancer Center; 3. Wendy Uhlmann, MS, CGC, Division of Molecular Medicine and Genetics, Department of Internal Medicine, University of Michigan

- Describe the current status of NSGC's federal advocacy efforts to obtain reimbursement for services performed by genetic counselors.
- Review the progress of NSGC's state licensure efforts.
- Identify strategies NSGC is using in approaching payers to cover and promote genetic counseling services and progress on these efforts.
- Outline a leadership role for genetic counselors in promoting an interdisciplinary framework for integrating new genomic tests into patient care.

9:00 am – 9:45 am

303 Non-Invasive Prenatal Diagnosis Beyond Trisomy 21
0.075 CEU

1. Brynn Levy, MSc, PhD, FACMG, CGC, Columbia University Medical Center

- Compare and contrast technologies for NIPD detection of chromosome abnormalities (past and present).
- Describe the complexities involved in isolating the fetal genome from maternal blood.
- Discuss the scope of clinical disorders detectable and implications for clinical practice.

Submitted/Sponsored by the Prenatal SIG

Educational Breakout Sessions

10:00 am – 12:00 pm

304 Merging into Clinical Practice: Updates in Non-invasive Prenatal Testing
0.20 CEU

1. Lee Shulman, MD, FACOG, FACMG, Department of Obstetrics & Gynecology, Feinberg School of Medicine, Northwestern University; 2. Christin Coffeen, MS, CGC, Verinata Health, Inc; 3. Annette Batey, MS, Ariosa Diagnostics; 4. Melissa Savage, MS, CGC, Natera; 5. Jennifer Hoskovec, MS, CGC, University of Texas Houston Medical School

- Compare and contrast published data on various NIPD models.
- Identify the benefits and limitations of non-invasive testing for aneuploidy as well as possible future applications of non-invasive technology.
- Discover attitudes of providers and patients regarding non-invasive testing and how they are applying testing in their clinical practice.

Submitted/Sponsored by the Prenatal SIG

305 Now What am I Supposed to Tell My Fiancé? Psychosocial and Behavioral Outcomes for Unaffected Women with a *BRCA* Mutation
0.20 CEU

1. Lindsey M. Hoskins, PhD, National Cancer Institute; 2. Andrea Patenaude, PhD, Dana-Farber Cancer Institute; 3. Adam Buchanan, MS, MPH, CGC, Duke University Medical Center; 4. Bright Pink Panel

- Review current literature on psychosocial outcomes and medical decision-making in unaffected women with a *BRCA* mutation.
- Report preliminary data on adherence to current risk management recommendations among unaffected women with a *BRCA* mutation.
- Report first-person narratives of ways in which having a *BRCA* mutation has influenced young unaffected women's medical and psychosocial decision-making.

Submitted/Sponsored by the Cancer SIG

306 Genetic Aortopathies: An Explosion of Genetic Knowledge
0.20 CEU

1. Christina M. Rigelsky, MS, CGC, Cleveland Clinic; 2. Patricia Arscott, MS, CGC, University of Michigan; 3. Dianna Milewicz, MD, PhD, University of Texas Medical School at Houston

- Identify the genes associated with aortic disease and appreciate the spectrum of clinical findings associated with genetic aortic disease.
- Recognize important aspects of imaging evaluations that help define genetic arterial disease.
- Describe the role of new technologies in gene discovery for aortic diseases.

Submitted/Sponsored by the Cardiovascular SIG

307 Pharmacogenetics: Update on the Field and Roles for Genetic Counselors
0.20 CEU

1. Howard McLeod, PharmD, University of North Carolina; 2. Susanne Haga, PhD, Duke Institute for Genome Sciences & Policy; 3. Amy C. Sturm, MS, CGC, The Ohio State University Medical Center; 4. Kevin Sweet, MS, CGC, The Ohio State University Medical Center

- Gather an understanding of the current status of the field of pharmacogenetics including basic principles as well as the clinical potential and use particularly in the areas of cardiology, oncology, neurology and psychiatry.
- Recognize ethical and policy issues related to the delivery of pharmacogenetics including interpretation and communication of results as well as data storage and management.
- Explore current and potential professional roles for genetic counselors in the establishment and support of pharmacogenetic programs.

Submitted/Sponsored by the Personalized Medicine SIG

Friday, October 26 (Continued)

Sponsored Lunch Session

12:15 pm – 1:45 pm

308 Hereditary Cancer Panels: Review of Initial Results and Discussion about Counseling for Next-Generation Tests

1. Steven Keiles, MS, CGC, Ambry Genetics; 2. Aaron Stuenkel, MS, CGC, Ambry Genetics; 3. Anu Chittenden, MS, LGC, Dana Farber Cancer Institute; 4. Scott Weissman, MS, CGC, NorthShore University HealthSystem; 5. Kory Jasperson, MS, CGC, Huntsman Cancer Institute; 6. Heather Hampel, MS, CGC, The Ohio State University; 7. Megan Myers, MS, CGC, UCSF Cancer Risk Program

- Review of preliminary cancer panel data: mutation and variant rates, insurance coverage and ordering clinician demographics.
- Describe clinicians' experiences ordering and counseling for hereditary cancer panels, regardless of testing laboratory.
- Evaluate the utility and appropriateness of a next-generation panel testing approach in various clinical scenarios.
- Discuss the current and future roles of next-generation panels in hereditary cancer testing.

Sponsored by Ambry Genetics

5:30 pm – 7:00 pm

Dr. Beverly Rollnick Memorial Lecture

313 Part 1: That's Just Crazy Talk: A Play and Discussion

0.15 CEU

1. Victoria Maxwell, BFA/BPP, Crazy for Life Co.

- Illustrate symptoms of mania, depression, psychosis and anxiety.
- Describe various experiences and consequences of self-stigma and external stigma.
- Improve skills of empathy and insight into mental illness, treatment and recovery.
- Explore the effectiveness of humor as a recovery tool.

313 Part 2: Research Evaluation of a Play as a Stigma-Reduction Intervention

1. Sagar V. Parikh, MD, FRCPC, University Health Network Director of Continuing Mental Health Education, University of Toronto

- Clarify the role of drama as a tool for stigma reduction.
- Identify key measures of stigma measurement in mental health.
- Demonstrate successful stigma reduction through use of a play.

Sponsored Evening Session

7:15 pm – 8:45 pm

314 When More Isn't Better…Genetic Testing from a Legal Perspective

1. Phil Duffy, Esq.

1. Recognize the importance of genetic screening.
2. Recognize the challenges of over-utilization.
3. Recognize the ideal mix of clinically appropriate and standard of care.

Sponsored by Quest Diagnostics

Saturday, October 27

Sponsored Breakfast Session

7:00 am – 7:45 am

401 When Bigger is Better: Multi-gene Testing Panels for Genetically Heterogeneous Disorders

0.05 CEU

1. Chris Lauricella, MS, CGC; 2. Elizabeth Butler, MS, CGC

- Recognize the benefits and limitations of multi-gene testing panels for genetically heterogeneous disorders.
- Identify patients who can benefit from genetic testing using multi-gene panels for severe combined immunodeficiency (SCID) and epilepsy.

Sponsored by GeneDx

Plenary Sessions

8:00 am – 9:00 am

Late-breaking Plenary Session

402 Targeted Pharmaceuticals for Neurodevelopmental Disorders

0.10 CEU

1. Paul P. Wang, MD, Seaside Therapeutics; 2. Kira A. Dies, ScM, CGC, Boston Children's Hospital

- Describe the "targeted pharmacotherapy" research approach for neurodevelopmental disorders.
- Identify potential drug targets for the treatment of Fragile X Syndrome.
- Define the manifestations of Tuberous Sclerosis Complex that Rapamycin may affect.

9:30 am – 10:30 am

403 Jane Engelberg Memorial Fellowship (JEMF) Presentation

0.10 CEU

1. Kevin Sweet, MS, CGC, The Ohio State University Medical Center; 2. Dawn C. Allain, MS, CGC, The Ohio State University Medical Center; 3. Kate P. Shane-Carson, MS, CGC, The Ohio State University Medical Center, Clinical Cancer Genetics; 4. Sara Fitzgerald-Butt, MS, CGC, The Research Institute at Nationwide Children's Hospital

- Describe the purpose and mission of the Jane Engelberg Memorial Fellowship.
- Outline new resources and CEU opportunities for genetic counselors through JEMF and NSGC to enhance their teaching and counseling skills.
- Discover the findings of original research and provide discussion on the value of genetic counselors in the genetic testing process for Hereditary Breast Ovarian Cancer.

Submitted by the JEMF Advisory Group

Educational Breakout Sessions

10:30 am – 12:30 pm

404 Counseling Difficulties and Ethical Concerns in an Uncertain Prenatal Diagnosis

0.20 CEU

1. Jamie Chance, MS, CGC, Regional Perinatal Center, Odessa Regional Medical Center; 2. Curtis R. Coughlin II, MS, MBe, University of Colorado; Children's Hospital of Colorado; 3. Holly H. Zimmerman, MS, CGC, University of Mississippi Medical Center; 4. Martha Dudek, MS, CGC, Vanderbilt University; 5. Rebecca Carter, MS, CGC, University of Texas Health Science Center

- Describe and review counseling techniques and management options in pregnancies that have a likely, but not an absolutely, lethal diagnosis.
- Illustrate how to present the option of comfort care: tips, do's and don'ts.
- Discuss the controversy and ethical implications in management of a pregnancy with a lethal or a potentially lethal diagnosis.

Submitted/Sponsored by the Prenatal SIG and Ethics Advisory Group

405 Familial Pancreatic Cancer: From Evaluation to Endoscopy

0.20 CEU

1. Jennifer Axilbund, MS, CGC, The Johns Hopkins University; 2. Alison Klein, PhD, The Johns Hopkins University; 3. Michael Goggins, MD, The Johns Hopkins University

- Describe the currently known inherited cancer syndromes that predispose to pancreatic cancer, both adenocarcinoma and neuroendocrine, and the clinical utility of genetic testing for each.
- Recognize ongoing new gene discovery efforts and how to use those discoveries in patient care.
- Identify the benefits and limitations of currently available pancreatic screening methods, and how to manage individuals and families with hereditary pancreatic cancer.

Submitted/Sponsored by the Cancer SIG

Saturday, October 27 (Continued)

406 The New Landscape of Genetic Testing: How to Approach Testing Minors for Adult-onset Conditions in the Era of Large-scale Genomic Testing

0.20 CEU

1. Laura Hercher, MS, CGC, Sarah Lawrence College; 2. Barbara A. Bernhardt, MS, CGC, University of Pennsylvania; 3. Regan Veith, MS, CGC, Children's Hospital of Wisconsin; 4. Amy White, MS, CGC, Children's Hospital of Wisconsin; 5. Benjamin Berkman, JD, MPH, National Human Genome Research Institute, National Institutes of Health; 6. Flavia M. Facio, MS, CGC, National Human Genome Research Institute, National Institutes of Health

- Outline the application of whole genome and exome sequencing technologies in the pediatric setting and describe the current challenges in obtaining informed consent and returning results.
- Report the early experiences of genetic counselors working in this novel setting and outline the complexities of providing genetic counseling for whole genome and exome sequencing through the use of case examples.
- Describe broad ethical guidance and recommendations on how to approach issues related to informed consent and return of results in the pediatric population, specifically in cases where minors are found to carry variants that are known or suspected to cause or predispose to adult-onset conditions.

Submitted by the Public Policy Committee and Ethics Advisory Group

407 Hints from the Experts: Job Interviewing Skills

0.20 CEU

1. Mary Jarvis Ahrens, MS, CGC, University of Minnesota Medical Center, Fairview; 2. Elizabeth Balkite, MS, Sarah Lawrence College; 3. Debra Collins, MS, CGC, KUMC; 4. Elizabeth Gettig, MS, CGC, University of Pittsburgh; 5. Teresa Kruisselbrink, MS,CGC, Mayo Medical Center; 6. Bonnie S. LeRoy, MS, CGC, University of Minnesota; 7. Caroline Lieber, MS, CGC, Sarah Lawrence College; 8.Maureen E. Smith, MS, CGC, Center for Genetic Medicine; 9. Wendy Uhlmann, MS, CGC, Division of Molecular Medicine and Genetics, Department of Internal Medicine University of Michigan; 10. LuAnn Weik, MS, CGC, Children's Hospital of Wisconsin; 11. Vivian Weinblatt, MS, CGC, Integrated Genetics

- Recognize challenging interview questions and learn how to answer them appropriately during a job interview.
- Determine the critical factors for evaluation of applicants by genetic counselors in a position to hire them.
- Review and teach genetic counseling students effective communication skills that the presenters have learned in their jobs and in interviewing genetic counselors for positions at their institutions.

Sponsored Lunch Session

12:30 pm – 2:00 pm

408 Using Cost Effectiveness Modeling to Demonstrate the Value of Genetic Services

0.10 CEU

1. David Stenehjem, PharmD

- Outline the definitions and terms used in cost effectiveness modeling studies and health economics and why they are valuable measures of healthcare outcomes.
- Define how cost effectiveness modeling studies are done, how the models work and how they should be interpreted in the context of a practicing healthcare provider.
- Discuss recent cost effectiveness modeling studies in genetics, highlighting how they have been informative to testing guidelines, medical policy and payer coverage.

Sponsored by Myriad Genetic Laboratories, Inc.

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Ariosa™ DIAGNOSTICS

Friday, October 26

	309 – Education Papers 1. Describe approaches and issues in the education of genetic counseling students. 2. Discuss genetics education of lay and professional groups.	**310 – Pediatrics Papers** 1. Discuss the latest developments in genetic counseling for pediatric patients and their families. 2. Discuss genetic conditions seen in the pediatric genetics clinic.	**311 – Cancer Papers** 1. Identify the latest developments in evaluation and testing for inherited cancer predispositions. 2. Discuss issues that are specific and unique to individuals with an inherited cancer predisposition.	**312 – Genetic Testing Papers** 1. Discuss latest developments in the field of diagnostic testing and test interpretation. 2. Review potential testing opportunities in the genetics clinic and issues related to genetic testing.
	Room 207	***Room 200***	***Room 302/304/306***	***Room 210***
2:00 pm – 2:15 pm	Genetic Education Materials for School Success (GEMSS): A Guide for the Classroom for Children with Genetic Conditions *C. Giummo*	Deletion Mutations Bordering Exon 44 of the DMD Gene: The CINRG Experience *N. Mans*	Implementation of Institutional MSI and IHC Testing for Diagnosis of Lynch Syndrome: The Cleveland Clinic Experience *B. Leach*	Consumer Preferences for the Reporting of Genetic Variants of Uncertain Significance *N. Smith*
2:15 pm – 2:30 pm	The Impact of Supervision Training on Genetic Counselors' Supervisory Identity Development *K. Lewis*	Mosaicism Due to Reversion: A Report of a Rare 20pter-p13 Deletion *M. Martin*	Universal Lynch Syndrome Screening Result Notification *H. Hampel*	Role of Numeracy and Genetic Literacy in the Desire to Receive Genetic Risk Information *I. Yurkiewicz*
2:30 pm – 2:45 pm	The Pregnancy and Health Profile Pilot Project: Evaluating the Patient Satisfaction and Usability of a Novel Prenatal Family History and Genetic Screening Tool *E. Edelman*	Gaucher Disease Type 3 Due to Chromosome 1 Uniparental Disomy *C. Brasington*	Age of Onset and Predictors of Medullary Thyroid Cancer in Patients with Lower Risk *RET* Proto-Oncogene Mutations *T. Rich*	Deaf Genetic Testing and Psychological Well-Being *C. Palmer*
2:45 pm – 3:00 pm	The New Face of Genetics: A Multimedia Educational Tool *A.Fan* *J. Martineau** *(*Presenter)*	Integration of Genetic Services into Pediatric Patient Care: A National Survey of Pediatric Primary Care Physicians *W. Uhlmann*	Turcot Syndrome: A Case Report of a 15-Year-Old with Diffuse Colon Polyposis and Medulloblastoma *J. Fulbright*	Interpreting and Delivering Microarray Results of Variants of Unknown Significance: Genetic Counselors' Perspectives *V. Mehta*
3:00 pm – 3:15 pm	Evaluation of Knowledge Regarding Diagnostic Strategies for Genetic Diseases in Select Residents *S. Penney*	Evidence for Maternal Mosaicism in *OPHN1* *A. Schreiber*	The Clinical Presentation of an Adult Patient with Biallelic Mutations in *PMS2* *L. Mar*	Accuracy of the BRCAPro Risk Assessment Model in Males Presenting to MD Anderson Cancer Center for *BRCA1* and *BRCA2* Testing *C. Garby*
3:15 pm – 3:30 pm	Development of a Clinical Supervisor Support Program *C. Atzinger*	Associations Between Parents' Psychosocial Outcomes and Their Perception of Providers' Communication After Newborn Screening Identifies Genetic Carrier Status for Cystic Fibrosis or Sickle Cell Hemoglobinopathy *A. La Pean*	Comprehension and Family Communication of Hereditary Cancer Risk Among African Americans *T. Morgan*	Assessment of Individuals with *BRCA1* and *BRCA2* Large Genomic Rearrangements in High-Risk Breast Cancer and Ovarian Cancer Families *A. G. Arnold*

Saturday, October 27

	409 – Counseling/ Psychosocial Issues Papers 1. Describe common psychosocial and counseling techniques in genetic counseling sessions. 2. Discuss patient concerns and quality of life.	**410 – Pre- and Perinatal Papers** 1. Discuss the latest developments in prenatal testing and prenatal genetic counseling. 2. Describe the attitudes and perceptions of patients and providers in the prenatal clinic.	**411 – Adult Papers** 1. Discuss genetic conditions seen in adult patients. 2. Review genetic issues related to common complex diseases.	**412 – Professional Issues Papers** 1. Recognize the professional and personal experiences of genetic counselors. 2. Identify potential future opportunities for the field of genetic counseling.
	Room 200	***Room 302/304/306***	***Room 210***	***Room 207***
2:00 pm – 2:15 pm	The Role Of Family Functioning in Promoting Adaptation Among Siblings of Individuals with Duchenne Muscular Dystrophy (DMD) *B. Pappa*	Value of Non-invasive Prenatal Testing in the Third Trimester *W. DiNonno*	Disclosure of Whole Genome Sequencing Findings: Perspectives of Individuals Affected with and Predisposed to Genetic Disease *K. Bontempo*	Diagnostic Yield of a 180K Array Based Comparative Genomic Hybridization in a Pediatric Population *J. Kaylor*
2:15 pm – 2:30 pm	Importance of Confirmation for Positive Non-invasive Prenatal Testing Results *V. Cherepakho*	Utility of SNP Microarray for Chromosomal Abnormality Analysis in POC by Trimester *H. Taylor*	Utilizing Illness Representations to Improve Family Communication Within A Population At-Risk for Cardiomyopathy *B. Batte*	Creating Professional Development Opportunities in Research for Practicing Genetic Counselors *M. Myers*
2:30 pm – 2:45 pm	Retrospective Study of Little People (LP) Pregnancy: Experiences, Opinions and Utilization of Genetic Counseling Services *C. Blout*	Exploring the Perceptions and the Role of Genetic Counselors in the Emerging Field of Perinatal Palliative Care *M. Dudek*	Comparison of Informed Consent Preferences for Multiplex Genetic Carrier Screening Among a Diverse Population *A. Reeves*	A Strategy to Improve the Quality and Economics of Laboratory Send-outs at Seattle Children's Hospital: The Role of the Laboratory Genetic Counselor *J. Conta*
2:45 pm – 3:00 pm	Parental Attitudes Regarding Newborn Screening for Duchenne Muscular Dystrophy *K. Buser*	Non-vascular Ehlers-Danlos Syndrome and Pregnancy: What are the Risks? *K. Sondergaard*	Phenotypic and Neurocognitive Characterization of Adults with 17p13.3 Microduplication Syndrome *S. Goldstein*	Training Genetic Counselors for the Variety of Roles Available within the Field *E. Heckaman*
3:00 pm – 3:15 pm	Motivations and Characteristics of Families who Adopt Children with Special Needs: A Qualitative Study *B. Gallinger*	Long Term Follow-Up of Morbidity and Quality of Life Associated with Isolated Gastroschisis *R. Mostafavi*	A Collaborative Approach to Integrating Genetics into Healthcare *A. Trepanier*	Expanding the Genetic Counseling Workforce: Program Directors Views on Increasing the Number of Graduates and Factors Influencing the Growth of Graduate Programs *V. Pan*
3:15 pm – 3:30 pm	Genetic Testing in Potential *BRCA1/2* Carriers Aged 18 to 25 Years: Decision-making and Impact *A. Hershner*	Prenatal Testing for Noonan Spectrum Disorders in Fetuses with Increased NT and Cystic Hygroma *E. Kramer*	Sickle Cell Trait Testing at NCAA Division 1 Universities: Sports Physicians and Athletic Trainers' Perspectives *N. Brown*	The Long and Short of Genetic Counseling Summary Letters: A Case-Control Study *J. Roggenbuck*

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Posters with Authors

Objective:

Describe the most recent research, techniques and approaches in the field of genetic counseling.

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1 Cholesteryl Ester Storage Disease (CESD): An Under-Recognized Disease of Lysosomal Acid Lipase Deficiency with Liver Dysfunction/Failure and Hyperlipidemia
D. L. Bernstein

2 Liver Disease and Chronic Liver Failure have Complete Penetrance in Cholesteryl Ester Storage Disease, a Form of Lysosomal Acid Lipase Deficiency
D. L. Bernstein

3 Left Ventricular Hypertrophy Aids in Identification of Patients with Aneurysm Osteoarthritis Syndrome Caused by *SMAD3* Mutations
D. S. Clements

4 The Iceberg and the Helix: Investigating the Role of Genetic Testing and Genetic Counselors in the Diagnosis and Management of Celiac Disease
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5 Factors Influencing Clinician Satisfaction with *APOE* Genotype Disclosure Counseling Sessions: The REVEAL Study
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6 Prevalence of Premature Ovarian Failure in Women with Tuberous Sclerosis Complex
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7 Differences in Management Recommendations for Adults with Cognitive Disability Based on Genetic Diagnosis Status
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8 Developing and Validating the Family Members of Individuals with Mental Illness Internalized Stigma (FaMIL-IS) Scale: A New Genetic Counseling Outcome Measure
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9 Interest in Genetic Testing for Complex Diseases: Age-Related Macular Degeneration (AMD) and the Elderly Perspective
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10 Huntington Disease-Ten Years After Testing
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11 Referral Patterns and Utilization of Clinical Genetic Testing for Patients with Inherited Cardiac Disease
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13 Evaluating the Current Genetic Testing Practices around Huntington Disease
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14 Evaluation of Mutations Causative of Inherited Cardiovascular Disease
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15 Implications for the Prevention of Type 2 Diabetes: Investigating the Relationship Between Religiosity, Genetic Explanations for Diabetes and Control Over Diabetes Risk
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16 A Clinical Tool for Classification of Frontotemporal Degeneration Pedigrees
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NSGC Awards and Fellowships

The Jane Engelberg Memorial Fellowship

Response to Genetic Counseling: Understanding the Experience and Needs of Individuals Receiving a Prenatal Diagnosis of Down Syndrome

Katie Sheets, MS, CGC and
Blythe Crissman, MS, CGC

The Audrey Heimler Special Projects Award

Heidi Cope, MS, CGC

NSGC Leadership Awards

Natalie Weissberger Paul National Leadership Award
Joan H. Marks, MS

New Leader
Samantha M. Baxter, MS, CGC
Catriona L. Hippman, MSc, CGC

Strategic Leader
Scott M. Weissman, MS, CGC

Outstanding Volunteer
Joy Larsen Haidle, MS, CGC
Leigha Senter-Jamieson, MS, CGC

Best Abstract Awards

Best Full Member Abstract Award
Genetic Counseling for Individuals with Serious Mental Illnesses: The First, and Only, Randomized Controlled Trial

Catriona L. Hippman, MSc, CGC

Beth Fine Kaplan Student Abstract Award
Exploring the Relationship Between Diagnoses of Congenital Anomalies or Genetic Conditions and Postpartum Depression: A Retrospective Study

Meagan Wiesenhart, MS

AEC Planning Subcommittee

NSGC expresses its gratitude to these volunteers for their hard work and dedication.

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Networking Activities and Business Meetings

Welcome Reception

Wednesday, October 24 • 6:30 pm – 8:00 pm • *Hall D*

Make new contacts and greet your friends as you preview the vendors and their services in the Exhibitor Suite. Join your colleagues for this special kickoff to the AEC. Light hors d'oeuvres and cash bar will be available.

State of the Society Address

Thursday, October 25 • 10:15 am – 10:45 am • *Room 302/304/306*

Join President Brenda Finucane, MS, CGC, as she provides an overview of NSGC activities and accomplishments over the past year. Brenda will review NSGC's advocacy efforts, strategic initiatives and provide highlights from 2012.

NSGC Annual Business Meeting

Friday, October 26 • 4:00 pm – 4:45 pm • *Room 302/304/306*

Incoming Presidential Address

Saturday, October 27 • 9:00 am – 9:30 am • *Room 302/304/306*

Hear NSGC President Elect Rebecca Nagy, MS, CGC, as she introduces herself to NSGC members and outlines her vision for NSGC in 2013.

ABGC Annual Business Meeting

Saturday, October 27 • 12:30 pm – 2:00 pm • *Room 302/304/306*

Breaks and Lunches

There are many opportunities to meet and mingle with your colleagues throughout the conference. Make note of these scheduled breaks to network with your peers:

Thursday, October 25

10:00 am – 10:15 am	*Level 3 Foyer*
4:15 pm – 4:30 pm	*Level 2 Foyer*

Friday, October 26

9:45 am – 10:00 am	*Level 3 Foyer*
3:30 pm – 4:00 pm	*Hall D*

Saturday, October 27

10:30 am – 10:45 am	*Level 2 Foyer*

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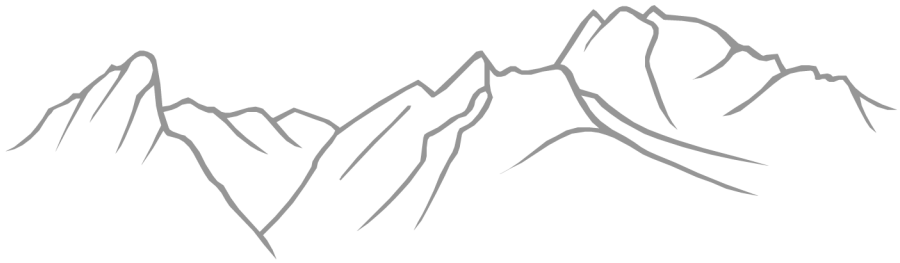

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Fellow, American College of Preventive Medicine
Assistant Clinical Professor, Department of Obstetrics & Gynecology
University of Colorado Health Sciences Center

Author

Abortion Practice
Philadelphia: J.B. Lippincott, 1984
Boulder: Alpenglo Graphics, 1990
(soft cover edition)

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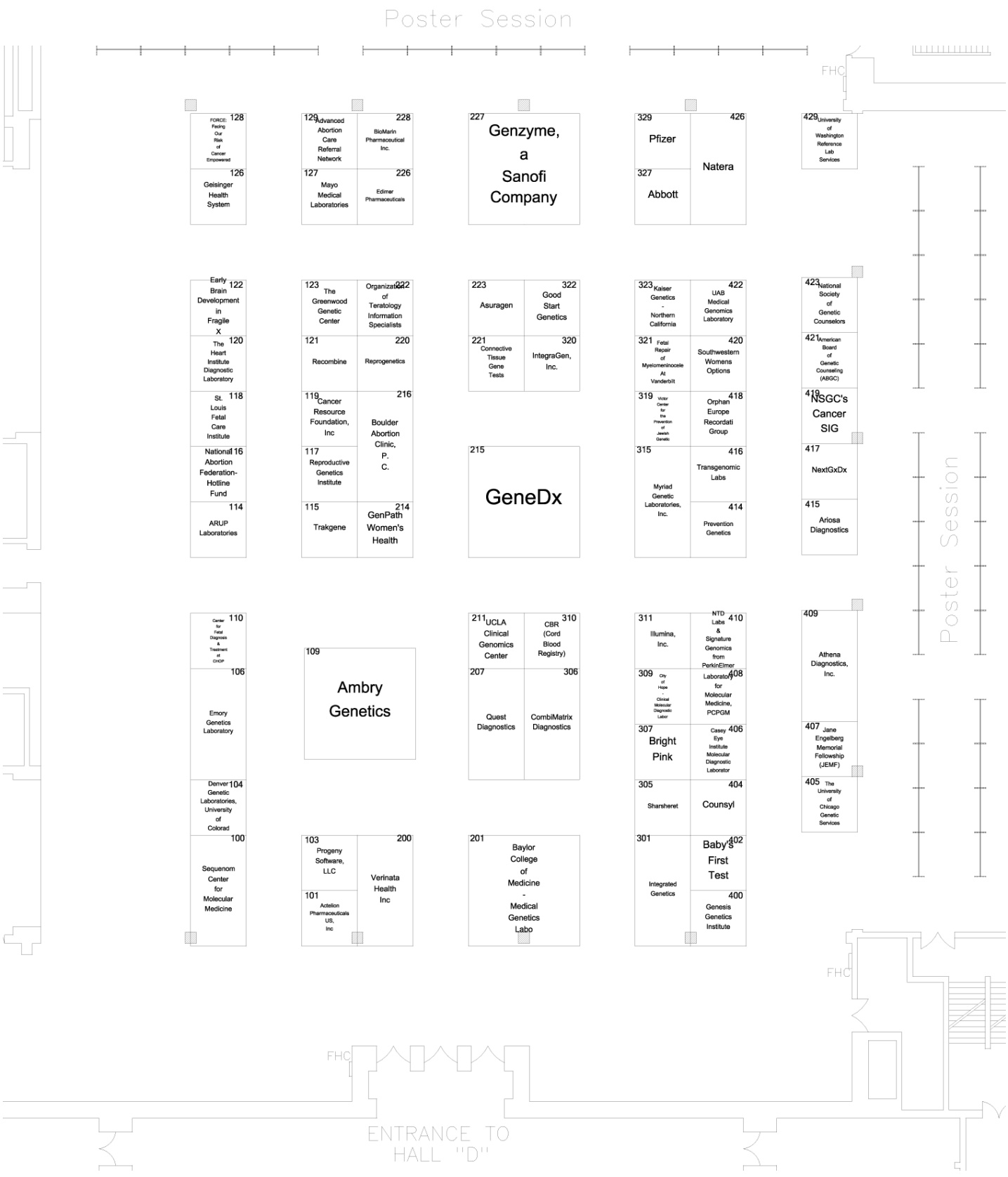

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Booth#: 327

Phone: 847.937.6100
www.abbott.com

Abbott is a global, broad-based healthcare company devoted to discovering new medicines, new technologies and new ways to manage health. Our products span the continuum of care, from nutritional products to medical devices and pharmaceutical therapies. Our comprehensive product line encircles life itself — addressing important health needs for all ages.

Actelion Pharmaceuticals US, Inc
Booth#: 101

Phone: 650.624.6900
www.actelion.com

Actelion Pharmaceuticals Ltd is a biopharmaceutical company headquartered in Allschwil/Basel, Switzerland, that focuses on the discovery, development and commercialization of innovative treatments to serve high unmet medical needs.

Advanced Abortion Care Referral Service
Booth#: 129

Phone: 856.667.5910
Email: jmoore@cherryhillwomenscenter.com
www.cherryhillwomenscenter.com

Providing safe and compassionate referrals for later abortion care for women and their families.

Ambry Genetics*
Booth#: 109

Phone: 949.900.5500
www.ambrygen.com

Ambry Genetics, an innovator in genetic testing and leader in next generation sequencing technology, offers a comprehensive menu of diagnostic tests for more than 300 genes, plus whole exome sequencing. Ambry's insurance pre-verification, easy ordering and enhanced reporting let you focus on the patient instead of the paperwork.

American Board of Genetic Counseling, Inc. (ABGC)
Booth #: 421

Phone: 913.895.4617
Email: info@abgc.net
www.abgc.net

The American Board of Genetic Counseling establishes standards of competence through accreditation of graduate training programs and certification and recertification of genetic counselors to advance the profession and protect the public.

Ariosa Diagnostics
Booth#: 415

Phone: 855.9.ARIOSA (855.927.4672)
Email: clientservices@ariosadx.com
www.ariosadx.com

Ariosa Diagnostics, Inc. is committed to providing safe, highly accurate and affordable tests for maternal and fetal health. Ariosa's Harmony Prenatal Test offers reliable detection of the common trisomies associated with Down syndrome and other conditions.

ARUP Laboratories
Booth#: 114

Phone: 800.522.2787
Email: clientservices@aruplab.com
www.arup.com/genetics

A nonprofit enterprise of the University of Utah, ARUP Laboratories is a leading national reference laboratory offering esoteric molecular assays and a comprehensive genetics test menu, including biochemical, cytogenetic and molecular genetic testing.

Asuragen, Inc.
Booth#: 223

Phone: 512.681.5200
Email: clinicallabsupport@asuragen.com
www.asuragen.com

Asuragen Clinical Laboratory offers a suite of Fragile X testing methodologies including Xpansion Interpreter™, a breakthrough molecular diagnostic test that utilizes novel PCR technologies to detect stabilizing AGG sequences within the FMR1 gene in order to refine the risk of expansion in future generations.

Athena Diagnostics, Inc.
Booth#: 409

Phone: 800.394.4493
Email: info@athenadiagnostics.com
www.athenadiagnostics.com

Athena Diagnostics, a leader in specialized diagnostic testing, provides the most comprehensive test menu for neurological, endocrine and renal conditions through more than 350 diagnostic tests.

Baby's First Test
Booth#: 402

Phone: 202.966.5557
Email: info@babysfirsttest.org
www.babysfirsttest.org

Baby's First Test is the nation's newborn screening education center for families and providers. This site provides information on all of the conditions screened for, details screening procedures and has information on state panels.

Baylor College of Medicine, Medical Genetics Laboratories*
Booth#: 201

Phone: 713.798.6555
Email: genetictest@bcm.edu
www.bcmgeneticlabs.org

Baylor College of Medicine, Medical Genetics Laboratories offer a broad range of diagnostic genetics tests including DNA diagnostics, sequencing, cytogenetics, FISH diagnostics, cancer cytogenetics, chromosomal microarray analysis, whole exome sequencing, biochemical genetics, and Mitochondrial DNA analysis. Please visit our booth for more information!

BioMarin Pharmaceutical Inc.
Booth#: 228

Phone: 415.506.6700
Email: kward@bmrn.com
www.bmrn.com

BioMarin develops and commercializes innovative biopharmaceuticals for serious diseases and medical conditions. Approved products include the first and only enzyme replacement therapies for MPS I and MPS VI and the first and only FDA-approved medication for PKU. Visit www.BMRN.com to learn more.

Boulder Abortion Clinic, P.C.*
Booth#: 216

Phone: 303.447.1361 / 800.535.1287
Email: bachern@msn.com
www.drhern.com

Our purpose is to provide the safest possible abortion care and termination of pregnancies for fetal anomalies or medical indications. We provide this care in a confidential, comfortable, humane, and dignified outpatient setting.

Bright Pink, NFP
Booth#: 307

Phone: 312.787.4412
Email: brightpink@bebrightpink.org
http://www.bebrightpink.org

Bright Pink is a national non-profit organization that provides education and support to young women who are at high risk for breast and ovarian cancer. We arm young women with knowledge, options and a great attitude, and offer companionship and empathy during their journey. We empower them to take control of their breast and ovarian health and, in turn, grant them the freedom and peace of mind to live beautiful and fulfilling lives.

Cancer Resource Foundation, Inc.
Booth#: 119

Phone: 508.630.2242
Email: info@cancer1source.org
www.cancer1source.org

The Cancer Resource Foundation's (CRF) mission is to advocate and provide support for the prevention, early detection, diagnosis, treatment and survivorship of cancer through patient assistance, education and research.

Casey Eye Institute Molecular Diagnostic Laboratory
Booth#: 406

Phone: 503.494.5838
Email: chiangj@ohsu.edu
www.ohsucasey.com/diagnostics

The Casey Eye Institute Molecular Diagnostics Laboratory is a CLIA certified laboratory providing comprehensive molecular testing for genetic diseases involving the eye. Our mission as a laboratory is to identify the causative genetic mutation(s) for evey person in a timely and cost-effective manner.

CBR ® (Cord Blood Registry®)
Booth#: 310

Phone: 800.588.0258
Email: providers@cordblood.com
www.cordblood.com

CBR® (Cord Blood Registry ®) is the largest, most experienced newborn stem cell bank in the world. CBR has banked stem cells for more than 400,000 children, and has released more cord blood for treatment than any other family bank.

Center for Fetal Diagnosis & Treatment at CHOP
Booth#: 110

Phone: 800.IN-UTERO (468.8376)
Email: fetalsurgery@email.chop.edu
www.fetalsurgery.chop.edu

For pregnancies complicated by birth defects, the Center is one of the most experienced programs in the world, providing the complete spectrum of care — expert prenatal diagnosis, fetal therapy including fetal repair of spina bifida, delivery and postnatal treatment.

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City of Hope — Clinical Molecular Diagnostic Laboratory
Booth#: 309

Phone: 888.826.4362
Email: mdl@coh.org
mdl.cityofhope.org

The City of Hope Molecular Diagnostic Laboratory (CMDL) specializes in clinical genetic testing services for cancer predisposition, coagulopathies, connective tissue disorders, muscular dystrophies, neuropsychiatric disorders and pharmacogenetics. For more up-to-date information, please visit http://mdl.cityofhope.org.

CombiMatrix Diagnostics
Booth#: 306

Phone: 800.710.0624
Email: info@cmdiagnostics.com
www.combimatrix.com

CombiMatrix Diagnostics, an industry-leader in genomic testing, offers clinicians a broad menu of innovative microarray tests. We provide state-of-the-art genetic testing services in the areas of Prenatal & Reproductive Health (including microarray, FISH & chromosome analysis); Pediatrics/Postnatal; and Hematology-Oncology.

Connective Tissue Gene Tests
Booth#: 221

Phone: 484.244.2900
Email: leena.ala-kokko@ctgt.net
www.ctgt.net

CTGT is committed to providing the broadest range of molecular diagnostics for inherited connective tissue disorders – over 180 tests and growing rapidly. CTGT has high test sensitivity, fast service, expert advice and superior customer service.

Counsyl
Booth#: 404

Phone: 888.COUNSYL (888.268.6795)
Email: info@counsyl.com
www.counsyl.com

Counsyl has developed an affordable and universal pre-pregnancy test which every parent can take to increase her chances of having a healthy child. The test is covered by insurance and prescribed by thousands of physicians internationally.

Denver Genetic Laboratories, University of Colorado
Booth#: 104

Phone: 303.724.3826 (Biochemical); 303.724.3801 (DNA)
Email: michael.woontner@ucdenver.edu (Biochemical); elaine.spector@ucdenver.edu (DNA)
www.denvergenetics.org

Denver Genetic Laboratories includes the DNA Diagnostic Laboratory (Dr. Elaine Spector) and Biochemical Genetics Lab (Dr. Stephen Goodman), University of Colorado. Our molecular, biochemical, mitochondrial, clinical and cytogenetics expertise is backed by ongoing academic research.

Early Brain Development in Children with Fragile X
Booth #122

Phone: 800.793.5715 or 888.845.6786
Email: ibisnetwork@gmail.com
www.ibisnetwork.org/fragilex

A longitudinal MRI study focusing on brain development in infants with Fragile X. Looking for participants 0-24 months with a Fragile X diagnosis. Scans and assessments are conducted at 6, 12 and 24 months of age.

Edimer Pharmaceuticals
Booth#: 226

Phone: 617.758.4300
Email: Tessa@edimerpharma.com
www.edimerpharma.com

Edimer Pharmaceuticals is developing EDI200, a treatment for X-linked Hypohidrotic Ectodermal Dysplasia. Edimer has an active IND for EDI200; clinical studies will begin Q4 2012. EDI200 has Orphan Drug designation in the US and Europe.

Emory Genetics Laboratory
Booth#: 106

Phone: 404.778.8499
Email: kellianne.martin@emory.edu
www.geneticslab.emory.edu

Emory Genetics Laboratory (EGL) is a worldwide leader in rare disease clinical genetic testing. EGL's biochemical, cytogenetic, and molecular laboratories perform integrated and comprehensive testing including whole exome sequencing, prenatal microarrays and metabolic disorder testing.

Fetal Repair of Myelomeninocele At Vanderbilt
Booth#: 321

Phone: 877.875.3737
Email: mary.dabrowiak@vanderbilt.edu
http://www.childrenshospital.vanderbilt.org/fetalcenter

Vanderbilt University Medical Center is the only centrally located facility in the U.S. currently offering prenatal treatment for myelomeningocele. We are unique in our focus on the care of both mom and unborn baby.

FORCE: Facing Our Risk of Cancer Empowered
Booth#: 128

Phone: 866.288.RISK (7475)
Email: info@facingourrisk.org
www.facingourrisk.org

FORCE is a national nonprofit dedicated to fighting hereditary breast and ovarian cancer. With 50 outreach groups throughout the US, FORCE programs include education, support, advocacy and research specific to BRCA mutations and hereditary cancers.

Geisinger Health System
Booth#: 126

Phone: 570.214.9275/800.845.7112
Email: jheid1@geisinger.edu
www. geisinger.org

Geisinger serves 3 million people in Northeastern and Central Pennsylvania, and has been nationally recognized for research, innovation and quality care. An electronic health record connects multiple hospitals, community practice sites and 900 physicians.

GeneDx*
Booth#: 215

Phone: 301.519.2100
www.genedx.com

GeneDx tests for more than 350 rare Mendelian disorders using DNA sequencing and deletion/duplication analysis of the associated gene(s), and offers oligonucleotide microarray-based testing and next-generation sequencing based panels for cardiac, mitochondrial and neurodevelopmental disorders.

Genesis Genetics Institute
Booth#: 400

Phone: 313.259.5507
Email: dorothy@rbdcreative.com
www.genesisgenetics.org

Genesis Genetics Institute is the pioneer of pre-implantation testing of embryos for inherited genetic abnormalities. Founded by world renowned scientist Dr. Mark Hughes MD, PhD., one of the original scientists working on the National Institutes of Health's Human Genome Project, Dr. Hughes' team was largely responsible for discovering pre-implantation genetic diagnosis (PGD) as a clinical practice and performed the first successful cases in the world.

GenPath Women's Health
Booth#: 214

Phone: 800.633.4522
www.genpathdiagnostics.com

GenPath Women's Health, a division of BioReference Laboratories, Inc. and sister division of GeneDx, specializes in diagnostic needs for MFM and ObGyn, including prenatal/maternal risk assessment, carrier testing, prenatal diagnosis, pregnancy thrombophilia and infectious diseases.

Genzyme, a Sanofi Company
Booth#: 227

Phone: 617.252.7600
www.genzyme.com

Genzyme discovers and delivers transformative therapies for patients with rare and special unmet medical needs, providing hope where there was none before. Visit www.genzyme.com.

Good Start Genetics, Inc.
Booth#: 322

Phone: 617.714.0800
www.gsgenetics.com

Good Start Genetics® is a molecular diagnostics company improving the standard of care for routine genetic carrier screening. By harnessing best-in-class technologies, GoodStart Select ™ delivers relevant results through simple testing and reporting solutions tailored to each clinician and patient and supported by customer care.

Illumina, Inc.*
Booth#: 311

Phone: 858.202.4566
Email: info@illumina.com
www.illumina.com

Illumina's technologies are providing the foundation for understanding disease at the molecular level. Our genotyping and sequencing technologies are fueling the emerging field of consumer genomics and are beginning to be applied to create innovations in molecular diagnostics.

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IntegraGen, Inc.
Booth#: 320

Phone: 877.992.7475
Email: larry.yost@integragen.com
www.arisktest.com

IntegraGen is a biotechnology company dedicated to molecular biomarker discovery. The Company has recently launched the ARISk™ Test, a genetic screening tool for the early identification of children at risk for autism spectrum disorder.

Integrated Genetics*
Booth#: 301

Phone: 800.848.4436
www.integratedgenetics.com

Integrated Genetics is a leading provider of reproductive genetic testing services with an expansive menu of complex tests in prenatal and postnatal genetic testing. For more information, visit www.integratedgenetics.com.

Jane Engelberg Memorial Fellowship (JEMF)
Booth#: 407

Email: kevin.sweet@osumc.edu
www.osumc.edu

The prestigious Jane Engelberg Memorial Fellowship is awarded by the NSGC in memory of Jane Engelberg, MS to promote the professional development of individual counselors and to improve our practice by providing support for scholarly investigation.

Kaiser Genetics - Northern California
Booth#: 323

Email: Cindy.E.Soliday@kp.org
www.genetics.kp.org

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Laboratory for Molecular Medicine, PCPGM
Booth#: 408

Phone: 617.768.8500
Email: lmm@partners.org
http://pcpgm.partners.org/lmm

The Harvard-affiliated Laboratory for Molecular Medicine is a CLIA-certified molecular diagnostic laboratory within the Partners Healthcare Center for Personalized Genetic Medicine. Our major areas of focus are cardiovascular diseases and syndromes, cancer and hearing loss.

Mayo Medical Laboratories
Booth#: 127

Phone: 800.533.1710
Email: mml@mayo.edu
www.mayomedicallaboratories.com

Mayo Medical Laboratories is a global reference laboratory operating within Mayo Clinic's Department of Laboratory Medicine and Pathology. Mayo Medical Laboratories' personal approach to patient care extends into every aspect of our business.

Myriad Genetic Laboratories, Inc.*
Booth#: 315

Phone: 800.469.7423
Email: cscommentis@myriad.com
www.myriadpro.com

Myriad Genetics is a leading molecular diagnostic company dedicated to making a difference in patient's lives through the discovery and commercialization of transformative tests to assess a person's risk of developing disease, guide treatment decisions and assess risk of disease progression and recurrence.

Natera*
Booth#: 426

Phone: 650.249.9090x357
Email: ksong@natera.com
www.natera.com

Natera provides reproductive genetic tests using bioinformatics in combination with SNP microarrays. Tests include preimplantation genetic diagnosis (PGD) for IVF, rapid turnaround molecular karyotyping of products of conception, non-invasive prenatal paternity testing, and non-invasive prenatal diagnosis.

National Abortion Federation – Hotline Fund
Booth#: 116

Phone: 800.772.9100
Email: chhebert@prochoice.org
www.prochoice.org

The NAF Hotline is the only toll-free source of information about abortion and referrals to providers of quality care in the U.S. and Canada. We provide callers unbiased, factual information in English, Spanish and French.

National Society of Genetic Counselors (NSGC)
Booth#: 423

Phone: 312.321.6834
Email: nsgc@nsgc.org
www.nsgc.org

The National Society of Genetic Counselors (NSGC) advances the roles of genetic counselors in healthcare by fostering education, research, and public policy to ensure the availability of quality genetic services. Visit the booth for membership services information, to view products and more.

NextGxDx
Booth#: 417

Phone: 615.236.4569
Email: info@nextgxdx.com
www.nextgxdx.com

Wed-based marketplace that offers one-stop shopping for genetic testing. NextGxDx provides genetic counselors with tools to search, compare, order and receive test results online.

NSGC Cancer SIG
Booth#: 419

Members of the NSGC Cancer SIG will be available during breaks to answer your questions about SIG projects and how you can get involved. Please stop by to view and receive samples of materials that have been developed by the SIG recently.

NTD Labs & Signature Genomics from PerkinElmer
Booth#: 410

Phone: 888.NTD.LABS or 877.SigChip
Email: ntdlabs@perkinelmer.com;
info@ www.ntdlabs.com
www.signaturegenomics.com

NTD Labs offers the most accurate first trimester aneuploidy screening. Signature Genomics offers a range of prenatal diagnostic testing to suit every patient case. PerkinElmer Labs give providers critical information needed to ensure timely management of the pregnancy.

Organization of Teratology Information Specialists
Booth#: 222

Phone: 877.311.8972
www.otispregnancy.org

Healthcare providers and their patients can utilize OTIS as a resource for accurate and current information about the possible effects of medications and other environmental exposures during pregnancy and lactation.

Orphan Europe Recordati Group
Booth#: 418

Phone: 949.637.5199
Email: us-west@orphan-europe.com
www.orphan-europe.com

At Orphan Europe, part of the Recordati Group, we are fully committed to improving the lives of people suffering from rare diseases through effective and safe treatments. Our products treat rare diseases and rare diseases only. In cooperation with the scientific community and patient organizations, we want to assure that everything is done from our side to make life as easy as possible for these patients.

Pfizer*
Booth#: 329

Phone: 212.733.2323
www.pfizer.com

At Pfizer, we apply science and our global resources to improve health and well-being at every stage of life. Every day, Pfizer colleagues work across developed and emerging markets to advance wellness, prevention, treatments and cures that challenge the most feared diseases of our time.

Prevention Genetics LLC
Booth#: 414

Phone: 715.387.0484
Email: s.samuels@preventiongenetics.com
www.preventiongenetics.com

Prevention Genetics offers over 500 clinical DNA sequencing tests and DNA banking services. We provide fast turnaround times, outstanding personalized service and the most reasonable prices in the industry. We are CLIA accredited. Please visit www.preventiongenetics.com.

Progeny Software, LLC
Booth#: 103

Phone: 800.progeny (776.4369)
Email: info@progenygenetics.com
www.progenygenetics.com

Progeny Software provides customizable software to manage pedigrees, genotypes and samples for genetic counselors, clinicians and scientists worldwide. Our NEW web-based Family History Questionnaire captures patient data online or via tablet, automatically drawing the pedigree.

Quest Diagnostics*
Booth#: 207

Phone: 866.MYQUEST
www.questdiagnostics.com

Quest Diagnostics, the world's leading provider of diagnostic testing, information and services, offers a comprehensive test menu including prenatal and genetics, toxicology and immunology, endocrinology and oncology.

Exhibitor Index

Recombine
Booth#: 121

Phone: 855.OUR.GENES
Email: info@recombine.com
www.recombine.com

Recombine is a clinical genetic testing company. We provide a complete and integrated service covering everything from sample collection to genetic counseling. We simplify the genetic testing process so you can focus on caring for your patients.

Reproductive Genetics Institute
Booth#: 117

Phone: 773.472.4900
Email: rgiworld@gmail.com
www.reproductivegenetics.com

RGI is a world-renowned provider of preimplantation testing (PGD) with expertise spanning three decades. PGD testing is available through RGI for nearly any single-gene condition, chromosome rearrangements or aneuploidy testing via PCR, FISH or aCGH.

Reprogenetics
Booth#: 220

Phone: 973.436.5003
Email: jfischer@reprogenetics.com
www.reprogenetics.com

Reprogenetics is a full-service preimplantation genetic diagnosis (PGD) laboratory offering 24 chormosome aCGH for aneuplooidy, translocations and inversions, PGD for single gene disorders and HLA matching, a CGH for POC testing, and FISH on sperm.

Sequenom Center for Molecular Medicine*
Booth#: 100

Phone: 877.821.7266
Email: info@sequenomcmm.com
www.sequenomCMM.com

Sequenom CMM® is a wholly owned subsidiary of Sequenom, Inc., and a CLIA-certified, CAP accredited molecular diagnostics laboratory dedicated to the development and commercialization of laboratory-developed tests for prenatal and ophthalmic conditions. Utilizing innovative proprietary technologies, Sequenom CMM provides test results that can be used by ordering clinicians in managing patient care. New for 2012 is the MaterniT21™ PLUS laboratory-developed test, a noninvasive test that analyzes the relative amount of chromosomes 21, 18 and 13 from a maternal blood sample. Other laboratory-developed tests include SensiGene® Cystic Fibrosis Carrier Screening, SensiGene® Fetal RHD Genotyping and RetnaGene™ AMD.

Sharsheret
Booth#: 305

Phone: 866.474.2774
Email: jthompson@sharsheret.org
www.sharsheret.org

Sharsheret is a national not-for-profit organization supporting young women and their families, of all Jewish backgrounds, facing breast cancer. Our mission is to offer a community of support to women diagnosed with breast cancer or at increased genetic risk, by fostering culturally-relevant individualized connections with networks of peers, health professionals and related resources.

Southwestern Women's Options
Booth#: 420

Phone: 505.242.7512
Email: boyd02@covad.net
www.curtisboydmd.com

Curtis Boyd, MD-owned clinics provide a full range of medical and surgical abortion services. The Albequerque office specializes in third trimester abortion care and offers a unique Fetal Indications Program geared to the special needs of the patient and her family.

St. Louis Fetal Care Institute
Booth#: 118

Phone: 314.268.4037
Email: fetalcare@ssmhc.com
www.stlouisfetalcare.com

The St. Louis Fetal Care Institute is a comprehensive diagnostic and therapeutic program that specializes in treating congenital problems and structural abnormalities in babies both in the womb and after birth.

The Greenwood Genetic Center
Booth#: 123

Phone: 800.473.9411
Email: kking@ggc.org
www.ggc.org

The Greenwood Genetic Center is a nonprofit institute organized to provide clinical genetic services, diagnostic laboratory testing, educational resources, and research in the field of medical genetics. Our laboratory offers biochemical, cytogenetic and molecular testing.

The Heart Institute Diagnostic Laboratory
Booth#: 120

Phone: 513.803.1751
Email: heartdx@cchmc.org
www.cincinnatichildrens.org/heartdx

The Heart Institute Diagnostic Lab at Cincinnati Children's Hospital Medical Center specializes in sequencing of genes associated with cardiovascular disease and molecular analysis of viruses known to cause myocarditis.

The University of Chicago Genetic Services Laboratory
Booth#: 405

Phone: 773.834.0555
Email: ucgslabs@genetics.uchicago.edu
www.dnatesting.uchicago.edu

Our laboratory is committed to high quality genetic diagnostics and translational research toward the development of tests for orphan diseases. Our focus is neurodevelopmental disorders. Some of our services include genetic testing for brain malformation syndromes, microcephaly, epileptic encephalopathy and Cornelia de Lange syndrome.

Trakgene
Booth#: 115

Email: rcarbone@kintrak.com
www.kintrak.com

Kintrak is a clinical genetics information management system. KinTrak has been developed by clinicians and researchers for clinicians and researchers. It provides a repository for all patient demographic data, surveillance, investigations, process management, pre-natal management, pedigree drawing, and risk analysis.

Transgenomic Labs
Booth#: 416

Phone: 877.274.9432
Email: Geneticstestinfo@transgenomic.com
www.transgenomiclabs.com

Transgenomic Clinical Laboratories advances personalized medicine using proprietary technologies and services. We provide molecular diagnostics for cardiology, neurology, mitochondrial disorders, and oncology.

UAB Medical Genomics Laboratory
Booth#: 422

Phone: 205.934.5562
Email: medgenomics@uab.edu
www.genetics.uab.edu

The UAB Medical Genomics Laboratory (MGL) is a CAP-certified, non-profit clinical laboratory, offering comprehensive testing for both common and rare genetic disorders, while specializing in the neurofibromatoses: NF1, Legius syndrome, NF2, segmental NF, and schwannomatosis.

UCLA Clinical Genomics Center
Booth#: 211

Phone: 310.825.7099
Email: scwebb@mednet.ucla.edu
www.pathology.ucla.edu/genomics

The comprehensive services of the UCLA Clinical Genomics Center include CLIA-certified clinical exome sequencing along with professional interpretation and genetic counseling. Additional areas of diagnostic expertise include Sanger sequencing, FISH, and chromosomal microarray analysis.

University of Washington Reference Lab Services
Booth#: 429

Phone: 206.465.6717
Email: kdestro@u.washington.edu
http://depts.washington.edu/labweb

University of Washington Reference Lab offers genetics testing. BROCA - Breast and Ovarian Cancer Risk Panel: Uses next-generation sequencing to detect mutations in 40 genes associated with cancer risk. Coloseq™ - Lynch and Polyposis Panel: Uses next-generation sequencing to detect mutations in multiple genes associated with Lynch and polyposis syndromes.

Verinata Health Inc.*
Booth#: 200

Phone: 650.603.5200
Email: info@verinata.com
www.verinata.com

Verinata Health, Inc. (Verinata), a privately-held company, is driven by a sole and extraordinary purpose — maternal and fetal health. To learn more about Verinata and the verifi™ prenatal test, visit www.verinata.com.

Victor Center for the Prevention of Jewish Genetic Diseases
Booth#: 319

Phone: 877.401.1093
Email: info@victorcenter.edu
www.victorcenters.org

Our mission is to ensure ongoing access to comprehensive genetic education, screening and counseling services. This is accomplished through Jewish community education programs and screening programs.

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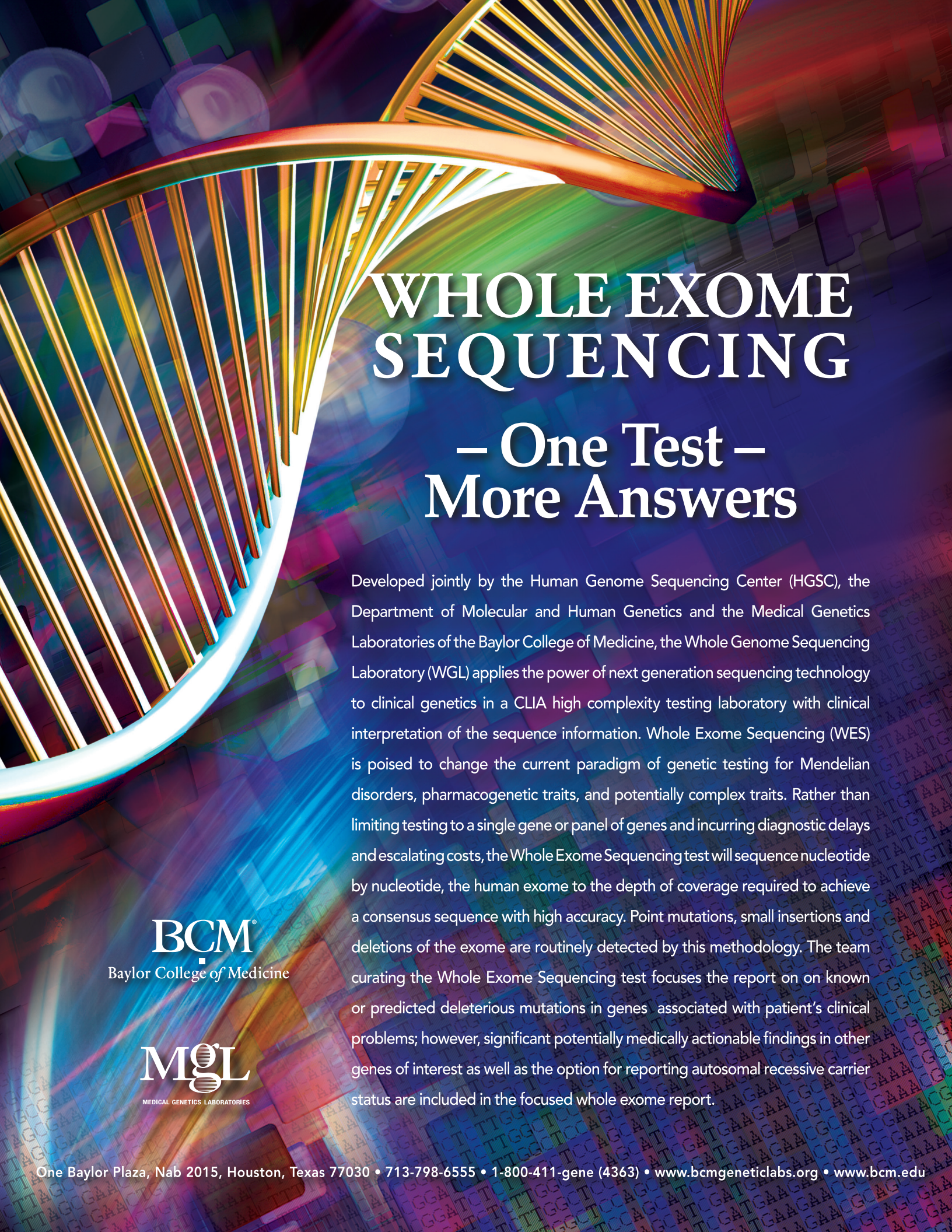

WHOLE EXOME SEQUENCING

– One Test – More Answers

Developed jointly by the Human Genome Sequencing Center (HGSC), the Department of Molecular and Human Genetics and the Medical Genetics Laboratories of the Baylor College of Medicine, the Whole Genome Sequencing Laboratory (WGL) applies the power of next generation sequencing technology to clinical genetics in a CLIA high complexity testing laboratory with clinical interpretation of the sequence information. Whole Exome Sequencing (WES) is poised to change the current paradigm of genetic testing for Mendelian disorders, pharmacogenetic traits, and potentially complex traits. Rather than limiting testing to a single gene or panel of genes and incurring diagnostic delays and escalating costs, the Whole Exome Sequencing test will sequence nucleotide by nucleotide, the human exome to the depth of coverage required to achieve a consensus sequence with high accuracy. Point mutations, small insertions and deletions of the exome are routinely detected by this methodology. The team curating the Whole Exome Sequencing test focuses the report on on known or predicted deleterious mutations in genes associated with patient's clinical problems; however, significant potentially medically actionable findings in other genes of interest as well as the option for reporting autosomal recessive carrier status are included in the focused whole exome report.

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National Society of Genetic Counselors

401 N. Michigan Ave.
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Chicago, IL 60611 USA
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