

WELCOME TO NEW ORLEANS

On behalf of the National Society of Genetic Counselors (NSGC) Annual Education Conference (AEC) Planning Subcommittee and the NSGC Board of Directors, thank you for joining us!

NSGC is building a stronger future for genomic health with more than 32 educational sessions designed to support your professional development. Sessions will cover a variety of topics at the forefront of genomics such as genetic test utilization, the clinical and psychosocial complexities of obtaining a diagnosis, and informed consent for genomic sequencing. Educational highlights you do not want to miss include the pre-conference symposium *"Practicing with Change: Best Practices in Prenatal Screening and Diagnosis"* (page 11), the Dr. Beverly Rollnick Memorial Lecture (page 14) and the NSGC Professional Issues Panel (page 14). Reference pages 11-16 for sessions submitted/sponsored by your NSGC Special Interest Group (SIG). Maximize your AEC experience by building your schedule to include education sessions specific to your professional interests.

Valuable experiences await you outside of lecture room walls as well. Take advantage of the Welcome Reception, SIG meetings and the new AEConnect area to network with 1,800 of your peers. Visit the Exhibitor Suite to see the latest product offerings and services within the profession. Catch up with old friends and make new connections during receptions, program reunions and daily breaks. Attend the State of the Society address, the NSGC Business Meeting and the new SIG Fair to learn more about the latest efforts of your professional organization. Experience all of the incredible activities this week has to offer!

We hope you enjoy your time here in New Orleans, absorbing content on the latest innovations and developments in the profession of genetic counseling, all while enjoying the rich culture of this amazing city!



Katherine Dunn, MS, CGC
2014 AEC Chair



Lori A.H. Erby, ScM, PhD, CGC
2014 AEC Vice-Chair

Table of Contents

About the AEC	4
General Information	5
AEC Schedule-At-A-Glance	6
Reunion Information	8
AEC Session Objectives	11
Concurrent Papers	18
Posters	21
NSGC Sponsors	26
NSGC Awards, Fellowships and AEC Planning Subcommittee	27
Networking Activities and Business Meetings	28
AEC Exhibitor Directory	29
AEC Exhibitor Suite Map	30
AEC Exhibitor Index	32
Vendor Solutions Presentations	36

Download the Official NSGC 2014 AEC Mobile App



NSGC is delivering everything AEC directly to your fingertips via the 2014 NSGC AEC mobile app. Download the app to gain early access to session descriptions, speakers and scheduling. Use

the interactive maps to navigate the show floor with ease, search the exhibitor directory and stay in-the-know with show alerts.

With your smartphone or tablet, search for "NSGC 2014 AEC" in your app store or direct your mobile browser to http://m.core-apps.com/nsgc_ec2014.

Engage with NSGC and fellow genetic counselors at the AEC on social media using hashtag #NSGC2014.

About the 33rd Annual Education Conference

Statement of Purpose

The 33rd 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 gain important information to support and enhance their current practice at sessions such as: *Lesbian, Gay, Bisexual, Transgender, Queer/Questioning (LGBTQ) Clients in Genetic Counseling: Awkward Questions? Complex Answers? Let's Start the Discussion* and *Tumor Genomic Testing: Technology, Clinical Implications and the Role of the Genetic Counselor*. The Exhibitor Suite will provide current information and the opportunity to talk with exhibitors about new developments in genetics. The pre-conference symposia will provide in-depth information on specific topics relevant to the field of genetic counseling.

Continuing Education

NSGC has been approved to offer up to 3.25 CEUs or 32.5 Contact Hours at the Annual Education Conference. CEUs earned through these activities will be accepted by ABGC as Category 1 CEUs for purposes of certification and recertification. Individuals must be certified at the time of participation in the activity in order for it to count towards recertification.

Earn up to:	CEUs	Contact Hours
Pre-conference Symposia	0.5	5.0
AEC General Sessions	2.1	21.0
Sponsored Meal Sessions	0.65	6.5
Total	3.25	32.5

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 that are marked as "attended" and have been evaluated in the online system.

Evaluation Process

Please assist NSGC in evaluating the AEC sessions; your input will help us plan future conferences.

Educational Session Evaluations

Participants are asked to complete online evaluations to provide input regarding individual speakers and educational content. For each educational session, please evaluate each speaker based on the time slot for the presentation. The speaker in the first time slot is speaker "1," the speaker in the second time slot is speaker "2," and so on. We ask all attendees to complete an online session evaluation for each session attended. **Although individuals claiming CEUs MUST complete evaluations, NSGC greatly appreciates feedback from all attendees.**

Poster Evaluation

Participants are asked to complete an electronic poster evaluation for attending the Posters with Authors sessions (215 and 309). **Those seeking CEUs for viewing posters MUST complete a poster evaluation.**

Concurrent Papers Evaluation

The Concurrent Papers sessions (302 – 305 and 410 – 413) feature six back-to-back presentations in four different categories, which run concurrently. The speaker in the first 15 minute time slot is considered speaker "a", the speaker in the second 15 minute time slot is considered speaker "b", and so on. For example, the second speaker in session 302 will be listed as 302b. If you plan to attend all six abstracts within the same category (no room change), complete six evaluations for that session number (example: complete evaluations 302a, 302b, 302c, 302d, 302e and 302f). If you plan to change categories/rooms between abstracts, complete the evaluation that corresponds with the session number and speaker order. **In order to receive the full 1.5 Contact Hours, you must attend and evaluate six speakers.** If you attend and evaluate five or fewer speakers, your CEUs will be credited proportionally.

All online electronic evaluation forms can be found at:
<http://www.showreg.net/2014AEEvaluationSite>

A link to these forms can also be found at www.nsgc.org/2014aec or in the AEC mobile app.

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.

2014 AEC Online Session Recordings

Maximize your AEC experience – listen to the sessions you missed in New Orleans, 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 recordings can be purchased and added while on-site as well!

Session recording packages featuring all pre-conference symposia* or featuring the AEC plenary and educational breakout sessions* are available for purchase. The online recordings will contain synced audio and PowerPoint® presentations for each session. You will be required to complete and pass a quiz included at the conclusion of each session and submit an evaluation to earn Category 1 CEUs for participating in the online course recordings. Purchase your online recording package in conjunction with your AEC registration for a special discounted rate.** The recordings will be released in early 2015 and will be available for two years after the release date.

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 low package price of \$99.

Registered attendees will be able to order online content during the AEC at these special rates and following the conference at an increased rate. Not attending the AEC? Check the NSGC website in January 2015 for additional information and purchase availability. Visit www.nsgc.org/2014AEC to add session recordings to your registration.

* With speaker approval

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

General Information

Registration Hours

Ernest N. Morial Convention Center

Tuesday, September 16	5:00 pm – 8:00 pm
Wednesday, September 17	7:00 am – 8:00 pm
Thursday, September 18	6:30 am – 8:00 pm
Friday, September 19	7:00 am – 7:00 pm
Saturday, September 20	7:00 am – 2:00 pm

Exhibitor Suite Hours

Ernest N. Morial Convention Center, Hall B

Wednesday, September 17	6:30 pm – 8:00 pm
Thursday, September 18	11:00 pm – 8:00 pm 5:00 pm – 7:30 pm
Friday, September 19	11:30 am – 3:00 pm 2:45 pm (<i>Passport to Prizes drawing</i>)

Message Center and Job Boards

Bulletin boards with push-pins are available in the AEConnect section of the Exhibitor Suite for attendees to leave messages for colleagues or to post job opportunities within the genetic counseling field. Advertising is not permitted. Material posted will be monitored and inappropriate information is subject to removal at NSGC's discretion.

Attendee List Information

Attendee lists were posted on the NSGC website prior to the conference and an updated list will be posted after the conference. Lists are available at the registration desk and are available to copy (at the attendee's expense) at the FedEx Office Print and Ship Center across the street from the Convention Center. Attendee lists are provided solely for networking and may not be used for solicitation purposes. NSGC is not responsible for errors and/or omissions.

Handouts and Presentations

NSGC offers electronic versions of AEC session handouts if provided by AEC speakers. A copy of the handouts will be available for reproduction (at the attendee's expense) at the FedEx Office Print and Ship Center across the street from the Convention Center. All session handouts (if provided by the speaker) are posted on the NSGC website and will be available following the conference until March 1, 2015. To download handouts go to <http://www.nsgc.org/2014AECHandouts>.

If you are also registered for a pre-conference symposium, you will be given a separate link to access your handouts. Handouts will also be available to copy (at the attendee's expense) at the FedEx Office Print and Ship Center across the street from the Convention Center.

Business Center: Hours of Operation

The FedEx Office Print and Ship Center is located across the street from the Ernest N. Morial Convention Center at 901 Convention Center Blvd. #100 and is open during the following hours:

Monday – Friday	7:30 am – 9:00 pm
Saturday	8:00 am – 6:00 pm
Sunday	12:00 pm – 6:00 pm

The UPS Store/Business Center is located in Lobby F of the Ernest N. Morial Convention Center and is open during the following hours:

Monday – Tuesday	9:00 am – 5:30 pm
Wednesday – Friday	8:00 am – 6:00 pm

Internet Access

NSGC attendees staying at the Hilton New Orleans Riverside will receive complimentary internet access in their guest rooms.

NSGC will also have wireless internet available in all meeting space and common areas at the Ernest N. Morial Convention Center.

NSGC gratefully acknowledges our wireless internet sponsor at the Convention Center:



Sponsored Sessions

Sponsored Meal Sessions are available for pre-registration. If you pre-registered to attend a session, a ticket will be printed with your badge at registration. In order to be admitted to each session, please bring your conference badge and the ticket that pertains to that session. We encourage you to arrive early for each session to allow all attendees time to be seated. If you did not pre-register for a session, but are still interested in attending, you are welcome to join the waiting line outside the room. We cannot guarantee you will be able to attend the session, but if all pre-registered attendees are seated and we still have room, we will be happy to accommodate you.

Please note that not all attendees will receive food and beverage with their admittance. Please check your ticket to see if the sponsor will provide food or if you are encouraged to bring your own.

Executive Office Information

NSGC Executive Office
330 N. Wabash Avenue
Suite 2000
Chicago, IL 60611 USA

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

Executive Director
Meghan Carey
mcarey@nsgc.org

Schedule-At-A-Glance

Wednesday, September 17							
8:00 AM - 2:00 PM	<div>CEU</div> Pre-conference Symposia						
	101 - Genetic Counseling Training: How to Start, Expand or Revitalize a Program <i>Room 211/212/213</i>	102 - Tumor Genomic Testing: Technology, Clinical Implications and the Role of the Genetic Counselor <i>Room 208/209/210</i>	103 - Laboratories and Industry: Depth and Variety of Genetic Counseling Career Opportunities <i>Room 206/207</i>	104 - Saving the Data: A Writer's Workshop <i>Room 215/216</i>	105 - Career Trajectories in Genetic Counseling <i>Room 217/218/219</i>	106 - Practicing with Change: Best Practices in Prenatal Screening and Diagnosis <i>Room 220/221/222</i>	
2:00 PM - 2:30 PM	New! NSGC SIG Fair (see page 28 for more information) - <i>Room R05</i>						
2:00 PM - 3:15 PM	Welcome to the AEC: How to Make the Most of the Conference and NSGC (see page 28 for more information) - <i>Room R02/R03/R04</i>						
3:30 PM - 3:45 PM	AEC Opening Remarks - <i>The Great Hall AD</i>						
3:45 PM - 5:15 PM	<div>CEU</div> Janus Series						
	107 - Fanconi Anemia: Breaking Apart the Complex Cancer and Genetic Counseling Issues <i>The Great Hall AD</i>		108 - Eye Can See Clearer Now: Genetic Testing and Genetic Counseling for Retinitis Pigmentosa <i>The Great Hall AD</i>		109 - Advances in the Field of Bleeding Disorders <i>The Great Hall AD</i>		
5:15 PM - 5:30 PM	<div>CEU</div> 110 - Beth Fine Kaplan Best Student Abstract Award - <i>The Great Hall AD</i>						
5:30 PM - 5:45 PM	<div>CEU</div> 111 - Best Full Member Abstract Award - <i>The Great Hall AD</i>						
5:45 PM - 6:05 PM	Natalie Weissberger Paul National Achievement Award - <i>The Great Hall AD</i>						
6:05 PM - 6:30 PM	Audrey Heimler Special Project Award - <i>The Great Hall AD</i>						
6:30 PM - 8:00 PM	Welcome Reception in the Exhibitor Suite - <i>Hall B</i>						
7:30 PM - 11:00 PM	Various Program Reunions (See page 8 of the program book for more information)						
	Canadian Programs 7:30 pm - 9:30 pm <i>Café Soulé</i>	University of Arkansas 7:30 pm <i>Location TBA</i>	University of Cincinnati (Prior RSVP required) 8:30 pm <i>Hilton Garden Inn</i>	The University of Michigan 8:30 pm - 11:00 pm <i>The Rusty Nail</i>	Wayne State University 8:30 pm <i>The Rusty Nail</i>		
8:30 PM	Journal of Genetic Counseling Editorial Board Meeting - <i>Room R02</i>						
Thursday, September 18							
7:00 AM - 7:45 AM	<div>CEU</div> 201 - Use of Chromosomal SNP Array in Today's Clinical Practice - Sponsored by Integrated Genetics - <i>The Great Hall BC</i>						
7:00 AM - 8:00 AM	AEC Breakfast - <i>Great Hall Prefunction Space</i>						
7:00 AM - 7:45 AM	Various NSGC Ancillary Meetings						
	NSGC Leadership Orientation <i>Room R03</i>		Research SIG <i>Room R02</i>		NSGC Leadership Development Program (Incoming Committee Vice-Chairs and Incoming Board Members) <i>Room 214</i>		
8:00 AM - 9:30 AM	<div>CEU</div> Educational Breakout Sessions						
	202 - Adoption of Children with Genetic Disorders: Essential Knowledge for Genetic Counselors <i>Room 211/212/213</i>	203 - Non-Invasive Prenatal Screening: Data, Marketing and Women's Choices <i>Room 208/209/210</i>	204 - The Evolution of Hereditary Cancer Susceptibility Genetic Testing: The Clinical Utility of Integrating Whole Exome and Genome Sequencing into the Practice of Cancer Genetic Counselors <i>Room 206/207</i>	205 - Psychiatric Genetic Counseling in the Era of Direct to Consumer Genetic Testing <i>Room 217/218/219</i>	206 - Lesbian, Gay, Bisexual, Transgender, Queer/Questioning (LGBTQ) Clients in Genetic Counseling: Awkward Questions? Complex Answers? Let's Start the Discussion <i>Room 220/221/222</i>		
9:30 AM - 9:45 AM	AEC Break - <i>Great Hall Prefunction Space</i>						
9:45 AM - 10:45 AM	<div>CEU</div> 207 - National Efforts Towards Standardizing Variant Interpretation - <i>The Great Hall AD</i>						
10:45 AM - 11:30 AM	NSGC State of the Society Address - <i>The Great Hall AD</i>						
11:30 AM - 2:00 PM	Exhibitor Suite Open - <i>Hall B</i>						
11:30 AM - 12:00 PM	American Board of Genetic Counseling (ABGC) Business Meeting - <i>The Great Hall AD</i>						
12:00PM - 12:30 PM	Accreditation Council for Genetic Counseling (ACGC) Presentation - <i>The Great Hall AD</i>						
12:30 PM - 2:00 PM	<div>CEU</div> 208 - Next Generation Panel Testing for Genetically Heterogeneous Cancer and Mitochondrial Disorders - Sponsored by Baylor College of Medicine - <i>The Great Hall BC</i>						
12:30 PM - 2:00 PM	Various NSGC Committee Meetings						
	Access and Service Delivery 12:30 pm - 1:30 pm <i>Room R01</i>	AEC Subcommittee 12:30 pm - 1:15 pm <i>Room R04</i>	Education 1:15 pm - 2:00 pm <i>Room R04</i>	Ethics Advisory Group 12:30 pm - 1:30 pm <i>Room R02</i>	Membership 12:30 pm - 1:30 pm <i>Room R03</i>	Practice Guidelines 1:00 pm - 2:00 pm <i>Room 214</i>	Public Policy 12:30 pm - 1:30 pm <i>Room 214</i>
2:00 PM - 3:30 PM	<div>CEU</div> Educational Breakout Sessions						
	209 - Psychiatric Genetics for the Pediatric Counselor <i>Room 217/218/219</i>	210 - Sex Chromosome Aneuploidies: A Multidisciplinary Perspective on Counseling and Current Treatment Recommendations <i>Room 220/221/222</i>		211 - Hereditary Cancer Communication with Underserved Patients <i>Room 208/209/210</i>	212 - How to Review a Manuscript for a Journal: A Practical Workshop Aimed at Professional Development for Genetic Counselors <i>Room 211/212/213</i>	213 - Myotonic Muscular Dystrophy: Global Impact! <i>Room 206/207</i>	
3:30 PM - 3:45 PM	AEC Break - <i>Great Hall Prefunction Space</i>						
3:45 PM - 4:45 PM	<div>CEU</div> 214 - Dr. Beverly Rollnick Memorial Lecture - Far From the Tree: Parents, Children and the Search for Identity - <i>The Great Hall AD</i>						
4:45 PM - 5:30 PM	NSGC Leadership Awards - <i>The Great Hall AD</i>						
5:00 PM - 7:30 PM	Exhibitor Suite Open - <i>Hall B</i> (see page for 36 for vendor-sponsored presentation information)						
5:30 PM - 6:30 PM	Andrew Solomon (Rollnick Speaker) Book Signing - <i>Hall B</i>						
6:00 PM - 10:00 PM	Various Program Reunions (see page 8 of the program book for more information)						
	Brandeis University 7:30 pm - 9:00 pm <i>Café Soule Paris Room</i>	Mount Sinai 6:00 pm - 8:00 pm <i>The Wine Room at Café Adelaide</i>	Stanford University 7:30 pm <i>W.I.N.O</i>	University of Colorado 7:45 pm <i>Warehouse Grille</i>	University of Maryland 7:00 pm - 10:00 pm <i>The Rusty Nail</i>	University of South Carolina 7:30 pm -10:00 pm <i>Barcadia</i>	University of Texas 7:30 pm <i>ACME Oyster House</i>
6:30 PM - 7:30 PM	<div>CEU</div> 215 - Posters with Authors: Even Numbered Posters - <i>Hall B</i>						
7:45 PM - 9:15 PM	<div>CEU</div> 216 - NIPT: To Expand or Not to Expand? That is the Question - Sponsored by Illumina - <i>The Great Hall BC</i>						

Friday, September 19						
7:00 AM - 7:45 AM	<div>CEU</div> 301 - Application of an Enhancement Exome in Diagnosis of Rare Genetic Diseases - Sponsored by Personalis - <i>The Great Hall BC</i>					
7:00 AM - 8:00 AM	AEC Breakfast - <i>Great Hall Prefunction Space</i>					
7:00 AM - 7:45 AM	Various NSGC Ancillary and SIG Meetings					
	Cancer SIG - <i>Room R04</i>			NSGC Past Board Member Breakfast - <i>Room R03</i>		
8:00 AM - 9:30 AM	<div>CEU</div> Concurrent Papers					
	302 - Cancer <i>Room 208/209/210</i>	303 - GC Professional Roles <i>Room 211/212/213</i>	304 - Genetic Testing I: New Technology <i>Room 206/207</i>	305 - Prenatal <i>Room 220/221/222</i>		
9:30 AM - 9:45 AM	AEC Break - <i>Great Hall Prefunction Space</i>					
9:45 AM - 10:45 AM	<div>CEU</div> 306 - Informed Consent for Genomic Sequencing: Experience and Recommendations for Clinical and Research Settings - <i>The Great Hall AD</i>					
10:45 AM - 11:30 AM	<div>CEU</div> 307 - Professional Issues Panel: State Licensure and Advocacy - <i>The Great Hall AD</i>					
11:30 AM - 12:30 PM	NSGC Business Meeting - <i>The Great Hall AD</i>					
11:30 AM - 3:00 PM	Exhibitor Suite Open - <i>Hall B</i> (see page 36 for Vendor-Sponsored presentation information)					
12:30 PM - 2:00 PM	<div>CEU</div> 308 - Unmasking he Genetic Diagnosis: Updates to Whole Exome Sequencing and Inherited Cancer Teting - Sponsored by GeneDx - <i>The Great Hall BC</i>					
12:30 PM - 2:00 PM	Various NSGC Ancillary and SIG Meetings					
	ART/Infertility SIG 12:30 pm - 2:00 pm <i>Room R03</i>	Cardiovascular SIG 12:30 pm - 2:00 pm <i>Room R05</i>	Industry SIG 12:30 pm - 2:00 pm <i>Room R02</i>	Personalized Medicine SIG 12:45 pm - 1:45 pm <i>Room R01</i>	Psychiatric SIG 12:30 pm - 2:00 pm <i>Room 214</i>	Student/New Member SIG 1:00 pm - 2:00 pm <i>Room R04</i>
2:00 PM - 3:00 PM	<div>CEU</div> 309 - Posters with Authors: Odd Numbered Posters - <i>Hall B</i>					
2:45 PM	Passport to Prizes Drawing in Exhibitor Suite - <i>Hall B</i>					
3:00 PM - 3:15 PM	AEC Break - <i>Great Hall Prefunction Space</i>					
3:15 PM - 4:45 PM	<div>CEU</div> Educational Breakout Sessions					
	310 - The Clinical and Psychosocial Complexities of Obtaining a Diagnosis for Rare Genetic Disorders: Navigating the Diagnostic Odyssey <i>Room 211/212/213</i>	311 - ART Matters: Clinical Considerations for the Non-ART Genetic Counselor <i>Room 206/207</i>	312 - Inside the Pediatric Cancer Genetics Clinic <i>Room 208/209/210</i>	313 - Institutional Genetic Test Utilization: Developing Programs that Benefit the Healthcare System and our Profession <i>Room 217/218/219</i>	314 - A Next Generation Approach to Hypertrophic Cardiomyopathy <i>Room 220/221/222</i>	
5:00 PM - 6:00 PM	<div>CEU</div> 315 - Shared Decision Making in Genetic Counseling - <i>The Great Hall AD</i>					
6:00 PM - 7:00 PM	<div>CEU</div> 316 - Jane Engelberg Memorial Fellowship Presentation - <i>The Great Hall AD</i>					
7:00 PM	NSGC SIG Meeting					
	Industry SIG - <i>Room R03</i>					
7:00 PM - 10:00 PM	Various Program Reunions (see page 8 of the program book for more information)					
	Arcadia University 8:00 pm <i>Location TBD</i>	Boston University 7:30 pm <i>Domenica</i>	California State University and UC Berkley 8:30 pm <i>Location TBD</i>	Northwestern University 8:00 pm - 11:00 pm <i>The Maison</i>	Sarah Lawrence College 7:00 pm <i>Ernest. N. Morial Convention Center, R06</i>	University of Alabama at Birmingham 7:00 pm <i>Court of Two Sisters Restaurant</i>
					University of Minnesota 8:30 pm <i>The Crazy Lobster</i>	University of Oklahoma 7:00 pm <i>Le Bayou Restaurant</i>
7:15 PM - 8:45 PM	<div>CEU</div> 317 - Termination of Pregnancy for Indications of Genetic Disorder in Advanced Gestations - Sponsored by Boulder Abortion Clinic - <i>The Great Hall BC</i>					
Saturday, September 20						
7:00 AM - 7:45 AM	<div>CEU</div> 401 - Noninvasive Prenatal Testing for Microdeletions: One Year Later - Sponsored by Sequenom Laboratories - <i>The Great Hall BC</i>					
7:00 AM - 8:00 AM	AEC Breakfast - <i>Great Hall Prefunction Space</i>					
8:00 AM - 9:30 AM	<div>CEU</div> Educational Breakout Sessions					
	402 - Congenital Disorders of Glycosylation: Clinical and Genetic Variability <i>Room 211/212/213</i>	403 - The Down Syndrome Consensus Statement Five Years Later: Making Progress and Evaluating the Impact of Non-Invasive Prenatal Screening/Testing on a Fragile Compromise between the Disability and Genetics Communities <i>Room 208/209/210</i>		404 -The Non-Cancerous Female Breast <i>Room 220/221/222</i>	405 - Crossing the Generation Gap: Engaging Millenial Learners <i>Room 206/207</i>	406 - Double-Edged Sword: The Impact of Mass Media on Genetic Counseling <i>Room 217/218/219</i>
9:30 AM - 9:45 AM	AEC Break - <i>Great Hall Prefunction Space</i>					
9:45 AM - 11:00 AM	<div>CEU</div> 407 - Bringing ELSI Issues to Life: The Drama of DNA - <i>The Great Hall AD</i>					
11:00 AM - 11:30 AM	Incoming Presidential Address - <i>The Great Hall AD</i>					
11:30 AM - 12:30 PM	<div>CEU</div> 408 - Late-Breaking Session - Cell-Free Nucleic Acids: To Prenatal and Beyond - <i>The Great Hall AD</i>					
12:30 PM - 2:00 PM	<div>CEU</div> 409 - Genetic Testing Beyond the Individual: Finding Answers for Families - Sponsored by Invitae Corporation - <i>The Great Hall BC</i>					
12:30 PM - 2:00 PM	NSGC SIG Meeting					
	Pediatric SIG - <i>Room R02</i>					
2:00 PM - 3:30 PM	<div>CEU</div> Concurrent Papers					
	410 - Access and Service Delivery <i>Room 211/212/213</i>	411 - Education/ELSI <i>Room 206/207</i>		412 - Genetic Testing II: Implementation <i>Room 220/221/222</i>	413 - Pediatrics <i>Room 217/218/219</i>	

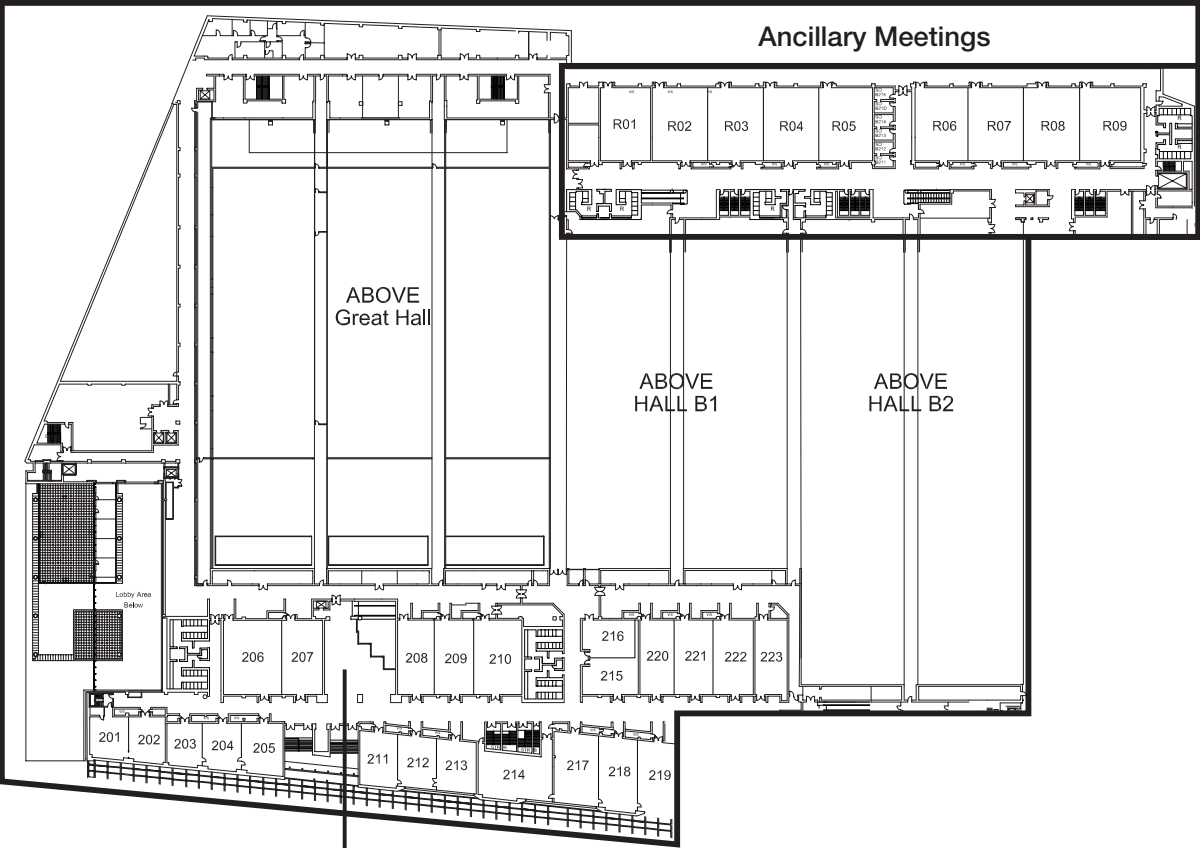
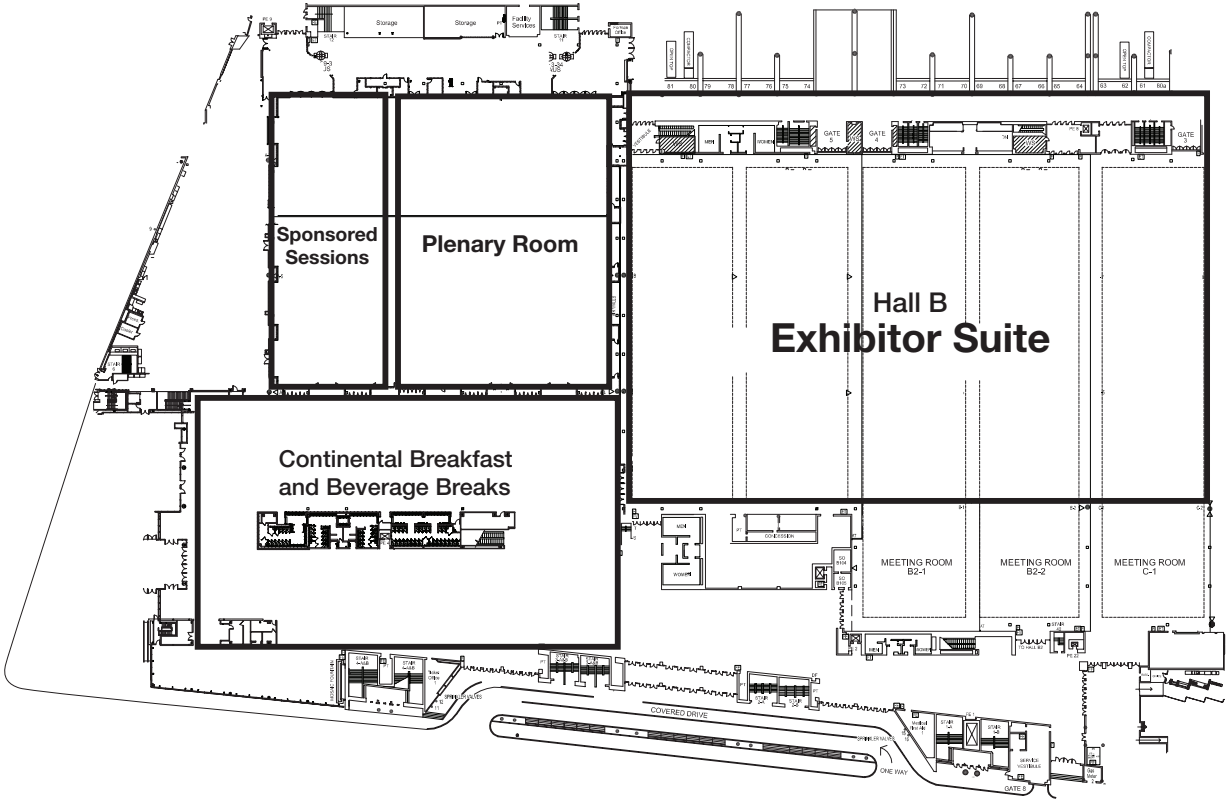
Reunion Information

Wednesday, September 17		
8:30 pm	University of Cincinnati <i>Prior RSVP Required</i>	Hilton Garden Inn 1001 South Peters Street New Orleans, LA
7:30 pm - 9:30 pm	Canadian Programs (McGill University, University of British Columbia, Université de Montréal, University of Toronto)	Café Soulé 720 Saint Louis Street New Orleans, LA 70116
7:30 pm	University of Arkansas for Medical Science	To Be Determined
8:30pm	Wayne State University	The Rusty Nail 1100 Constance Street New Orleans, LA 70130
8:30 pm – 11:00 pm	University of Michigan	The Rusty Nail 1100 Constance Street New Orleans, LA 70130
Thursday, September 18		
6:00 pm - 8:00 pm	Mount Sinai	The Wine Room at Cafe Adelaide 300 Poydras St New Orleans, LA 70130
TBA	University of Wisconsin - Madison	Barcadia 601 Tchoupitoulas Street New Orleans, LA 70130
7:00 pm - 10:00 pm	University of Maryland	The Rusty Nail 1100 Constance Street New Orleans, LA
7:30 pm - 9:00 pm	Brandeis University	Cafe Soule Paris Room 720 Saint Louis Street New Orleans, LA 504.304.4636
7:45 pm	University of Colorado	Warehouse Grille 869 Magazine Street New Orleans, LA 504.322.2188
7:30 pm	Stanford University	W.I.N.O. 610 Tchoupitoulas St. New Orleans, LA 70130
7:30 pm	University of Texas	ACME Oyster House 724 Iberville Street New Orleans, LA 504.522.5973
7:30 pm - 10:00 pm	University of South Carolina	Barcadia 601 Tchoupitoulas Street New Orleans, LA 504.335.1740

Friday, September 19		
7:00 pm	Sarah Lawrence College	Ernest N. Morial Convention Center Room R07 900 Convention Center Blvd, New Orleans, LA 70130
7:00 pm	University of Alabama at Birmingham <i>Prior RSVP Required</i>	Court of Two Sisters Restaurant 613 Royal Street New Orleans, LA
7:30 pm	Boston University	Domenica 123 Baronne Street New Orleans, LA 504.648.6020
8:00 pm	Arcadia University	To Be Determined
8:00 pm - 11:00 pm	Northwestern University	The Maison 508 Frenchman Street New Orleans, LA 70116
8:30 pm	California State University-Stanislaus and UC Berkeley alumnae	To Be Determined
8:30 pm	University of Minnesota Genetic Counseling Program	The Crazy Lobster 500 Port of New Orleans Place Suite 83 New Orleans, LA 504.569.3380
7:00 pm	University of Oklahoma Health Sciences Center	Le Bayou Restaurant 208 Bourbon Street New Orleans, LA 504.525.4755

Please visit the AEC Message Center board for additional reunion location and contact information.

Ernest N. Morial Convention Center Floor Plan



Pre-conference Symposia
and Education Breakout Sessions



Clear ANSWERS to Questions that Matter

Accuracy for All

Harmony is Validated in Women of All Ages¹⁻¹⁰

- Harmony Prenatal Test is clinically validated for use in all pregnant women, regardless of age or risk, to assess the risk of trisomy 21, 18, and 13

Harmony is the most broadly studied cell-free DNA-based maternal blood test

In blinded studies of over 22,000 pregnant women age 18 to 50 for trisomy 21:¹⁻⁷

- False positive rate was less than 0.1%
- Detection was greater than 99%

harmonytest.com



1. Sparks AB et al., Am J Obstet Gynecol. 2012 Apr;206(4):319.e1-9.
2. Ashoor G et al., Am J Obstet Gynecol. 2012 Apr;206(4):322.e1-5.
3. Norton M et al., Am J Obstet Gynecol. 2012 Aug;207(2):137.e1-8.
4. Nicolaides KH et al., Am J Obstet Gynecol. 2012 Nov;207(5):374.e1-6.
5. Ashoor G et al., Ultrasound Obstet Gynecol. 2013 Jan;41(1):21-5.

6. Verweij EJ et al., Prenat Diagn. 2013 Oct;33(10):996-1001.
7. Data on file at Ariosa Diagnostics, Inc.
8. Hooks J et al., Prenat Diagn. 2014 May;34(5):496-9.
9. Nicolaides K et al., Fetal Diagn Ther. 2014;35(1):1-6.
10. Gil M et al., Fetal Diagn Ther. 2014;35:204-11.

AEC Session Objectives

Wednesday, September 17

Pre-conference Symposia

8:00 am – 2:00 pm

101 Genetic Counseling Training: How to Start, Expand or Revitalize a Program

0.50 CEU

1: Quinn Philip Stein, MS, CGC, Sanford Health; 2: Catherine A. Reiser, MS, CGC, University of Wisconsin; 3: MaryAnn Campion, MS, CGC, Boston University School of Medicine; 4: Robin L. Bennett, MS, CGC, ScD Hon, University of Washington Medical Center; 5: Noelle R. Danylichuk, MS, CGC, University of Arkansas for the Medical Sciences; 6: Dee Quinn, MS, CGC, University of Arizona - OTIS; 7: Ian Wallace, MS, CGC, Pullman Regional Hospital

- Describe the steps required to start a new genetic counseling training program and the essential elements needed to achieve ACGC accreditation.
- Describe the purpose of using an external consultant and/or an AGCPD mentor for program planning.
- Recognize the factors involved with expanding and/or revitalizing an existing training program.

102 Tumor Genomic Testing: Technology, Clinical Implications and the Role of the Genetic Counselor

0.50 CEU

1: Scott Kopetz, MD, PhD, UT MD Anderson Cancer Center; 2: Steven T. Lott, PhD, Thermo Fisher Scientific; 3: Marilyn M. Li, MD, Baylor College of Medicine Medical Genetics Laboratories; 4: Molly S. Daniels, MS, CGC, The UT MD Anderson Cancer Center; 5: Elizabeth Varga, MS, LGC, Nationwide Children's Hospital; 6: Jessica N. Everett, MS, CGC, University of Michigan - Cancer Genetics Clinic; 7: Megan Frone, MS, CGC, UT Southwestern Medical Center - Simmons Cancer Center; 8: Emily Edelman, MS, CGC, The Jackson Laboratory; 9: Shannon Kieran, MS, CGC, Life Technologies, Inc; 10: Lisa Madlensky, PhD, CGC, University of California San Diego; 11: Cecelia Bellcross, PhD, MS, CGC, Emory University School of Medicine

- Compare available tumor genomic testing technologies and approaches and assess their relative strengths and limitations in clinical and research settings.
- Describe roles for the genetic counselor in the tumor genomic testing process including program setup, results reporting, addressing legal and ethical issues, coordinating appropriate follow-up testing and providing education to both patients and providers.
- Investigate challenging genetic counseling issues related to potential germline mutations found during tumor genomic testing via case examples and panel discussion.

Submitted/Sponsored by: Cancer SIG and Personalized Medicine SIG

103 Laboratories and Industry: Depth and Variety of Genetic Counseling Career Opportunities

0.50 CEU

1: Elyse Mitchell, MS, CGC, Mayo Medical Laboratories; 2: Jessie Conta, MS, CGC, Seattle Children's Hospital; 3: Katrina Kotzer, MS, CGC, Mayo Clinic; 4: Gabriel Lazarin, MS, CGC, Counsyl; 5: Amy Swanson, MS, CGC, Illumina; 6: Erin Riggs, MS, CGC, International Collaboration for Clinical Genomics (ICCG)/Geisinger Health System; 7: Christina Zaleski, MS, CGC, Prevention Genetics

- Describe emerging areas of practice for genetic counselors employed by laboratories and other non-clinical fields, including roles in pharmacogenomic testing, in hospital laboratories facilitating test utilization and management, as field specialists supporting client education and marketing efforts, and as international collaborators in areas such as data curation and translation to clinical utility.
- Assess how the depth and variety of laboratory genetic counseling roles align with the NSGC Scope of Practice, how counseling skills are applied, and how test utilization and management are optimized through examination of daily activities, inception and development of positions, and ethical issues encountered.
- Identify future directions, career development and growth opportunities for laboratory genetic counselors through the use of alternative service delivery models such as the design of web interfaces, telephone consultations to care for and educate patients regarding test results or appropriate test utilization for unique circumstances and new ways in which laboratories are utilizing genetic counselors.

Submitted by: Industry SIG

104 Saving the Data: A Writer's Workshop

0.50 CEU

1: Jehannine Austin, PhD, CGC, CCGC, University of British Columbia; 2: Gillian W. Hooker, PhD, ScM, CGC, NextGxDx

- Appreciate the importance of dissemination of genetic counseling research.
- Develop plans for the successful publication of genetic counseling research.
- Compose manuscripts for submission to peer-reviewed journals.

Submitted by: Research SIG and Editorial Board of the Journal of Genetic Counseling

105 Career Trajectories in Genetic Counseling

0.50 CEU

1: Lesley Bainbridge, BSR(PT), MEd, PhD, The University of British Columbia; 2: Angela M. Trepanier, MS, CGC, Wayne State University; 3: Lori A.H. Erby, ScM, PhD, CGC, National Human Genome Research Institute/The Johns Hopkins Bloomberg School of Public Health; 4: Catriona Hippman, MSc, CGC, Women's Health Research Institute; 5: Mary E. Freivogel, MS, CGC, Invision Sally Jobe; 6: Dayna-Lynn Dymianiw, CCGC, Xenon Pharmaceuticals Inc.; 7: Claire Davis, MS, CGC, Mount Sinai School of Medicine; 8: Bonnie Jeanne Baty, MS, University of Utah Sciences Center

- Describe a framework for developing career trajectories that has been used in other professions.
- List specific examples of career trajectories in genetic counseling and the factors/critical incidents that led individual genetic counselors down these trajectories.
- Apply the framework and case examples to develop a personal career trajectory and identify novel trajectories for the genetic counseling profession.

Submitted by: Committee on Advanced Training for Certified Genetic Counselors

106 Practicing with Change: Best Practices in Prenatal Screening and Diagnosis

0.50 CEU

1: Raynah M. Lobo, MS, CGC, Quest Diagnostics, Nichols Institute; 2: Joseph R. Biggio, Jr, MD, University of Alabama at Birmingham; 3: Elizabeth A. Kramer Dugan, MS, CGC, GeneDx; 4: Melanie Mahtani, PhD, CellScape Corporation; 5: Cori Feist, MS, CGC, Oregon Health Sciences University; 6: Megan Allyse, PhD, Duke University; 7: Anthony R. Gregg, MD, FACOG, FACMG, University of Florida College of Medicine; 8: Ignatia Van den Veyner, MD, Baylor College of Medicine; 9: Lisa Demers, MS, CGC, Dartmouth-Hitchcock Nashua

- Describe the value and role of maternal serum screen and ultrasound markers for a variety of genetic syndromes and purposes in pregnancy. Understand the strengths and weaknesses of the various screening methods.
- Appreciate both the recommended uses and limitations of cell-free DNA screening (NIPT). Identify future uses for NIPT and the role of NIPT in unusual circumstances.
- Evaluate the risks and benefits of chromosome microarray vs. karyotype for invasive testing. Understand, and become comfortable with, best practices for counseling VOUS, variable penetrance and incidental findings.
- Review society recommendations and best practices for all prenatal testing methods. Using case studies, explore the nuances of new prenatal technologies, and identify the most powerful ways to educate you and your colleagues.

Submitted by: Prenatal Counseling/Ultrasound Anomalies SIG

Janus Series

3:45 pm – 5:15 pm

107 Fanconi Anemia: Breaking Apart the Complex Cancer and Genetic Counseling Issues

0.05 CEU

1: Heather Zierhut, PhD, MS, University of Minnesota

- Review the types of Fanconi Anemia (FA) and how they are genetically and medically diverse.
- Assess the current and potential future obstacles of genetic counseling for FA carriers.
- Illustrate the reproductive risks and options for parents of children with FA.

AEC Session Objectives *(continued)*

Wednesday, September 17 *(continued)*

108 Eye Can See Clearer Now: Genetic Testing and Genetic Counseling for Retinitis Pigmentosa

0.05 CEU

1: Kari Haag Branham, MS, CGC, University of Michigan Kellogg Eye Center

- Describe different aspects of the genetic complexity associated with Retinitis Pigmentosa (RP).
- Effectively interpret pedigrees from families affected with RP.
- Recognize challenges in genetic testing for RP.

109 Advances in the Field of Bleeding Disorders

0.05 CEU

1: Elizabeth Varga, MS, LGC, Nationwide Children's Hospital

- Illustrate the clinical utility of genotyping in the setting of bleeding disorders.
- Discuss the latest research related to treatment and management of bleeding disorders, particularly as related to females.
- Identify roles for the genetic counselor in the setting of the hemostasis and thrombosis.

Best Abstract Awards

5:15 pm – 5:45 pm

Beth Fine Kaplan Best Student Abstract Award

110 Genetic Counselors as Choice Architects: Some Considerations for Presenting Genetic Testing Decisions in a Complex Choice Environment

1: Marci Barr, NHGRI/Johns Hopkins University

- Summarize existing knowledge about how the structure of a choice problem may impact judgment and decision making.
- Formulate a framework for structuring genetic testing choice tasks from the perspective of genetic counselors as choice architects.
- Identify areas of research need and methodologies for building an evidence base for how to design genetic testing choices.

Best Full Member Abstract Award

111 Analysis of Billing and Reimbursement of Genetic Counseling Services in a Single Institution in a State Requiring Licensure

1: Jennifer Leonhard, MS, Sanford Health

- Describe problems associated with limiting reimbursement of genetic counseling services.
- Recognize factors that can influence reimbursement rates for genetic counseling services.
- Identify future research avenues to better characterize reimbursement rates for genetic counseling services.

Thursday, September 18

Sponsored Breakfast Session

7:00 am – 7:45 am

201 Use of Chromosomal SNP Array in Today's Clinical Practice

0.05 CEU

1: Stuart Schwartz, PhD, Laboratory Corporation of America; 2: Romela Pasion, MS, CGC, Laboratory Corporation of America

- Explain SNP technology to patients.
- Counsel homozygous (HMZ) regions detected in SNP arrays and its utilization in detection of AR disorders.

Sponsored by: Integrated Genetics

Educational Breakout Sessions

8:00 am – 9:30 am

202 Adoption of Children with Genetic Disorders: Essential Knowledge for Genetic Counselors

0.15 CEU

1: Carrie Lynn Blout, MS, CGC, Johns Hopkins McKusick - Nathans Institute of Genetic Medicine; 2: Martha Osborne, RainbowKids Adoption Advocacy; 3: Susan Dibs, MD, Johns Hopkins University; 4: Colleen Gioffreda, BA, Greenberg Center for Skeletal Dysplasias, Johns Hopkins Hospital

- Describe the legal aspects of domestic and international adoption for children with special needs.
- Underline what is included in the medical work-up of internationally and domestically adopted children.
- Recognize available adoption organizations and resources available for children with genetic disorders.

Submitted by: Public Policy Committee and Pediatric and Clinical Genetics SIG

203 Non-Invasive Prenatal Screening: Data, Marketing and Women's Choices

0.15 CEU

1: Katie Stoll, MS, CGC, Group Health Cooperative; 2: George Estreich, MFA, Oregon State University; 3: Beth Daley, New England Center for Investigative Reporting

- Identify and analyze persuasive strategies in non-invasive prenatal screening (NIPS) advertising.
- Compare NIPS advertising with the nondirective goals of genetic counseling.
- Give examples of how NIPS advertising has affected individual reproductive decisions.

204 The Evolution of Hereditary Cancer Susceptibility Genetic Testing: The Clinical Utility of Integrating Whole Exome and Genome Sequencing into the Practice of Cancer Genetic Counselors

0.15 CEU

1: Laura Amendola, MS, CGC, University of Washington; 2: Sarah Scollon, MS, CGC, Baylor College of Medicine/Texas Children's Hospital; 3: Elaine Hiller, MS, CGC, Dana-Farber Cancer Institute

- Assess the clinical utility of the current and possible future testing modalities that may be implemented in the clinical hereditary cancer genetics setting.
- Recognize possible next steps when incidental findings are received from whole exome or genome germline sequencing in a research or clinical setting.
- Predict ethical issues that may arise with the implementation of this technology in the clinical care of hereditary cancer patients, as well as patients receiving genetic counseling that incorporates these tests for other indications.

Submitted/Sponsored by: Cancer SIG

205 Psychiatric Genetic Counseling in the Era of Direct to Consumer Genetic Testing

0.15 CEU

1: Jehannine Austin, PhD, CGC, CCGC, University of British Columbia; 2: Catriona Hippman, MSc, CGC, Women's Health Research Institute; 3: Edith Kolozsi, MS, CGC, University of Ottawa Heart Institute; 4: Hannah White, MS, CSU Stanislaus

- Describe some of the practical and psychosocial issues that can emerge in the context of direct to consumer genetic testing for psychiatric disorders.
- Explain the etiology of mental illness in a manner suitable for patients/family members, and how to integrate direct to consumer genetic testing results into this explanation.
- Describe of the psychosocial issues associated with genetic counseling for mental illness, and with receiving direct to consumer genetic testing results.

Submitted by: Psychiatric Disorders SIG

206 Lesbian, Gay, Bisexual, Transgender, Queer/Questioning (LGBTQ) Clients in Genetic Counseling: Awkward Questions? Complex Answers? Let's Start the Discussion

0.15 CEU

1: Robin L. Bennett, MS, CGC, ScD Hon, University of Washington Medical Center; 2: June A. Peters, MS, CGC, National Cancer Institute /Epidemiology; 3: Luba Djurdjinovic, MS, Ferre Institute-Genetics Programs; 4: Robert Pilarski, MS, CGC, Ohio State University; 5: Bradley Rolf, MS, CGC, Genetic Medicine Clinic, University of Washington Medical Center; 6: Susan Silber, JD, Silber, Perlman, Sigman & Tiley, P.A.

- Produce a consensus pedigree documenting appropriate genetic information, with and without representation of LGBTQ issues.
- List at least three issues that a LGBTQ client or couple may encounter in your genetic counseling practice setting.
- Identify at least one practice change to make to your genetic counseling practice setting that would make it more client-centered to LGBTQ issues.

Plenary Sessions

9:45 am – 10:45 am

207 National Efforts Towards Standardizing Variant Interpretation

0.10 CEU

1: W. Andrew Faucett, MS, LCGC, Geisinger Health System; 2: Kelly E. Ormond, MS, LCGC, Stanford University

- Describe the current NIH efforts to collect genotype and phenotype information from clinical laboratories.
- Utilize the ClinVar resource to gather information about clinically relevant variants.
- List at least three ways that genetic counselors can participate in and improve variant interpretation.

Sponsored Lunch Session

12:30 pm – 2:00 pm

208 Next Generation Panel Testing for Genetically Heterogeneous Cancer and Mitochondrial Disorders

0.10 CEU

1: Eric S. Schmitt, PhD, MS, Baylor Medical Genetics Laboratories; 2: Alicia A. Braxton, MS, Baylor Medical Genetics Laboratories

- List the advantages of next generation sequencing (NGS) panels relative to whole exome sequencing or whole genome sequencing.
- List the advantages of NGS panels vs Sanger sequencing.
- Use clinical and genetic information to guide test selection.

Sponsored by: Baylor College of Medicine

Educational Breakout Sessions

2:00 pm – 3:30 pm

209 Psychiatric Genetics for the Pediatric Counselor

0.15 CEU

1: Brenda Finucane, MS, LGC, Geisinger Health System, Autism and Developmental Medicine Institute; 2: Emily Morris, MSc, CCGC, University of British Columbia; 3: Anne S. Bassett, MD, FRCPC, University of Toronto

- Familiarize the pediatric genetic counselor with the concept of a continuum of developmental brain dysfunction that may give rise to psychiatric features in genetic conditions.
- Learn how to appropriately discuss the risk of psychiatric features with parents in a pediatric genetic counseling setting.
- Outline how psychiatric features of genetic conditions are managed by psychiatrists.

Submitted by: Psychiatric Disorders SIG and Pediatric and Clinical Genetics SIG

210 Sex Chromosome Aneuploidies: A Multidisciplinary Perspective on Counseling and Current Treatment Recommendations

0.15 CEU

1: Nicole Tartaglia, MD, Children's Hospital Colorado, University of Colorado School of Medicine; 2: Pravin Rao, MD, The Johns Hopkins University School of Medicine; 3: Susan Howell, MS, CGC, MBA, The eXtraordinary Kids Clinic and Fragile X Clinic, Children's Hospital Colorado

- Describe the features associated with sex chromosome aneuploidies (SCAs) from developmental, endocrinological, fertility and psychological perspectives.
- Summarize medical management recommendations across the lifespan.
- Discuss genetic counseling issues and identify strategies and resources to provide effective prenatal and postnatal genetic counseling.

Submitted/Sponsored by: Prenatal Counseling/Ultrasound Anomalies SIG

211 Hereditary Cancer Communication with Underserved Patients

0.15 CEU

1: Galen Joseph, PhD, University of California, San Francisco; 2: Robin Tropp Lee, MS, LCGC, UCSF, Cancer Risk Program

- Appreciate the experience of genetic counseling clients through the research of a medical anthropologist who has done extensive observations, interviews and focus groups aimed at better understanding the strengths and limitations of Hereditary Breast and Ovarian Cancer (HBOC) in the public hospital setting.
- Identify some key factors that present barriers for underserved patients in the HBOC setting.
- Identify strategies aimed at improving communication with clients of color, low income and/or low health literacy.

212 How to Review a Manuscript for a Journal: A Practical Workshop Aimed at Professional Development for Genetic Counselors

0.15 CEU

1: Christina Palmer, PhD, CGC, UCLA; 2: Pat McCarthy-Veach, PhD, MA, University of Minnesota; 3: John M. Quillin, PhD, MPH, MS, CGC, Virginia Commonwealth University; 4: Ian MacFarlane, PhD, Austin College; 5: Jehannine C. Austin, PhD, CGC, CCGC, University of British Columbia

- Understand the personal and professional benefits of acting as a peer reviewer for manuscripts that have been submitted for potential publication in journals.
- Appreciate how to practically approach the task of reviewing a manuscript, including how to identify issues and how to structure your written review.
- Discuss the ethical considerations associated with reviewing a manuscript for a journal, including how reviewers should/should not use the knowledge they gain from reviewing the work.

Submitted by: Research SIG and Editorial Board of the Journal of Genetic Counseling

AEC Session Objectives *(continued)*

Thursday, September 18 *(continued)*

213 Myotonic Muscular Dystrophy - Global Impact!

0.15 CEU

1: Carly Siskind, MS, CGC, Stanford Hospital and Clinic; 2: William Groh, MD, MPH, Indiana University School of Medicine; 3: Chad Heatwole, MD, University of Rochester Medical Center

- Summarize the main clinical findings in myotonic dystrophy and the genetic cause and complexities.
- Describe the most common heart phenotype and treatments, and how it could impact the genetic counseling encounter.
- Identify three features of central nervous system dysfunction in myotonic muscular dystrophy.

Submitted by: Cardiovascular Genetics SIG, Neurogenetics SIG and Psychiatric Disorders SIG

Sponsored by: Cardiovascular Genetics SIG and Neurogenetics SIG

Dr. Beverly Rollnick Memorial Lecture

3:45 pm – 4:45 pm

214 Far From the Tree: Parents, Children and the Search for Identity

0.10 CEU

1: Andrew Solomon, Author, *Far From the Tree: Parents, Children and the Search for Identity*

- Learn how differences unite us.
- Examine the tension between ideas of illness and identity as used to describe the same conditions.
- Reflect on how people forge meaning out of experiences of difficulty.

Sponsored by: The Dr. Beverly Rollnick Memorial Fund

Sponsored Evening Session

7:45 pm – 9:15 pm

216 NIPT: To Expand or Not to Expand? That is the Question.

0.10 CEU

1: Patricia Taneja, MS, LCGC, Illumina; 2: Holly Snyder, MS, CGC, Illumina; 3: Patricia Devers, MS, CGC, Illumina

- Cite most recent non-invasive prenatal testing (NIPT) performance data and clinical experience from NIPT CLIA laboratories.
- List clinical considerations (risks and benefits) in NIPT test menu expansion.
- Describe the general process NIPT clinical laboratories may use for test menu expansion.

Sponsored by: Illumina

Friday, September 19

Sponsored Breakfast Session

7:00 am – 7:45 am

301 Application of an Enhanced Exome in Diagnosis of Rare Genetic Diseases

0.05 CEU

1: Sarah Garcia, PhD, MS, CGC, Personalis; 2: Gemma Chandratillake, MPhil, PhD, MS, LCGC, Personalis

- Describe the makeup and features of an augmented exome with genome-wide structural variant (SV) detection.
- Show examples where an augmented exome with genome-wide (SV) detection enabled diagnoses that would have been missed with a standard exome.
- Outline the decision making process of using panels vs. an augmented exome with genome-wide SV detection for suspected genetic syndromes.

Sponsored by: Personalis, Inc.

Plenary Session

9:45 am – 10:45 am

306 Informed Consent for Genomic Sequencing: Experience and Recommendations for Clinical and Research Settings

0.10 CEU

1: Barbara Bernhardt, MS, Hospital of the University of Pennsylvania; 2: Sarah Scollon, MS, CGC, Baylor College of Medicine/Texas Children's Hospital; 3: Denise Lautenbach, MS, CGC, Brigham and Women's Hospital

- Compare the similarities and differences between obtaining informed consent for genomic sequencing vs. traditional targeted genetic tests and genomic sequencing in the clinical vs. research settings.
- Discuss strategies for addressing common concerns, questions and misconceptions from patients and families considering genomic sequencing.
- Describe recommendations for obtaining informed consent for genomic sequencing in clinical and research contexts.

10:45 am – 11:30 am

307 NSGC Professional Issues Panel: State Licensure and Advocacy

1: John Richardson, Director of Government Relations, National Society of Genetic Counselors; 2: Dawn Cardeiro, MS, CGC, Chairperson, National Society of Genetic Counselors Licensure Subcommittee

- Review the current status of NSGC's federal advocacy efforts to obtain reimbursement for services performed by genetic counselors.
- Report on the progress of NSGC's state licensure efforts.
- Describe the current climate for licensure and practice and address recent issues that are relevant to the genetic counseling profession.
- Educate and identify strategies of how to overcome licensure barriers.

Sponsored Lunch Session

12:30 pm – 2:00 pm

308 Unmasking the Genetic Diagnosis: Updates to Whole Exome Sequencing and Inherited Cancer Testing

0.10 CEU

1: Kristen Vogel Postula, MS, CGC, GeneDx; 2: Jackie Tahiliani, MS, CGC, GeneDx; 3: Erica Vaccari, MS, GeneDx

- Describe the inherited cancer genetic testing offerings and whole exome sequencing (WES) provided by GeneDx and discuss testing strategies.
- Review variant classification methods and variant testing program.
- Examine the clinical utility and genetic counseling considerations of inherited cancer next generation sequencing (NGS) gene panels, WES and XomeDxSlice.

Sponsored by: GeneDx

Educational Breakout Sessions

3:15 pm – 4:45 pm

310 The Clinical and Psychosocial Complexities of Obtaining a Diagnosis for Rare Genetic Disorders: Navigating the Diagnostic Odyssey

0.15 CEU

1: Elizabeth Chao, MD, FACMG, University of California, Irvine; 2: Stephanie Gandomi, MS, CGC, LGC, Ambry Genetics; 3: Kelly Gonzalez, MS, CGC, LGC, Ambry Genetics; 4: Amy Clugston, Syndromes Without A Name (SWAN USA); 5: Jonathon Rodis, BS, MBA, Massachusetts Chapter of the Marfan Foundation; 6: Nicole Boice, Global Genes/RARE Project

- Evaluate and assess the benefits and limitations of new technologies and their impact on patient care as it applies to the genetic counseling practice.
- Appreciate, from the patient and family perspective, the journey to find a diagnosis and the subtle and obvious life changes that emerge after receiving a long-awaited diagnosis.
- Describe the complexities of obtaining a clinical diagnosis in the context of a rare disease and the implications of these complexities for the genetic counseling process.

311 ART Matters: Clinical Considerations for the Non-ART Genetic Counselor

0.15 CEU

1: Lauren Isley, LCGC, California Cryobank; 2: Elizabeth Herr Cameron, MS, CGC, Genesis Genetics; 3: Andria G. Besser, BEd, MS, CGC, Bonei Olam

- Define general concepts and clinical applications of assisted reproductive technology (ART), including recent advancements in the field.
- Illustrate the application of ART knowledge across all areas of clinical care and its importance to genetic counselors practicing in prenatal, pediatric and cancer settings, as well as specialty clinics.
- Examine the psychosocial issues faced by ART patients and associated ethical considerations.

Submitted/Sponsored by Assisted Reproductive Technology and Infertility SIG

312 Inside the Pediatric Cancer Genetics Clinic

0.15 CEU

1: Kami Wolfe Schneider, MS, CGC, University of Colorado Denver; 2: Joyce Tannenbaum Turner, MS, CGC, Children's National Medical Center; 3: Martha Thomas, MS, CGC, Martha Jefferson Hospital; 4: Sarah Scollon, MS, CGC, Baylor College of Medicine/Texas Children's Hospital; 5: Amanda Knoth, MS, CGC, Myriad Genetic Laboratories

- Recognize the features of a medical and family history that are suggestive of pediatric cancer predisposition syndromes.
- Identify the health professionals who are essential players in the care of individuals with pediatric cancer predisposition syndromes.
- Gain an appreciation for the avenues genetic counselors have taken in the establishment of a pediatric cancer genetics clinic.

Submitted/Sponsored by: Cancer SIG

313 Institutional Genetic Test Utilization: Developing Programs that Benefit the Healthcare System and our Profession

0.15 CEU

1: Asheley Supik, MS, CGC, Riverside Health System; 2: Darci Sternen, MS, CGC, Seattle Children's Hospital; 3: Jacquelyn Riley, MS, CGC, Cleveland Clinic; 4: Julie Kaylor, MS, CGC, Arkansas Children's Hospital; 5: Fallon Brewer, MS, CGC, University of Alabama at Birmingham; 6: Lee Zellmer, MS, CGC, Children's Mercy Hospital

- Describe the development and implementation of test utilization review at various institutions, including the growing evidence of the value of the genetic counselor in this role.
- Review the inner workings of different test utilization programs, from how a test is flagged through tracking data on test alterations, considering how institutional differences may dictate which methods will be most successful.
- Discuss the cost savings and improvements in patient care that result from the test utilization review and the benefits of test utilization programs to institutions and the overall health care system.

Submitted by: Access and Service Delivery Committee and Industry SIG

314 A Next Generation Approach to Hypertrophic Cardiomyopathy

0.15 CEU

1: Carolyn Ho, MD, Harvard Medical School and Brigham and Women's Hospital; 2: Stephanie Ware, MD, PhD, FACMG, Indiana University School of Medicine; 3: Samantha Baxter, MS, CGC, GeneInsight and Partners Healthcare

- Review our current understanding of the natural history, genetic etiology and current management of hypertrophic cardiomyopathy (HCM).
- Outline recent advancements in genetic testing, diagnosis and treatment of HCM.
- Summarize cardiomyopathy gene panel technologies and discuss strategies for interpretation of results.

Submitted/Sponsored by: Cardiovascular Genetics SIG

Plenary Session

5:00 pm – 6:00 pm

315 Shared Decision Making in Genetic Counseling

0.10 CEU

1: Barbara Biesecker, PhD, MS, NHGRI/NIH; 2: Sarah Kobrin, PhD, MPH, National Cancer Institute, NIH; 3: Amy Turriff, ScM, NEI/NIH; 4: Julie Sapp, ScM, NHGRI/NIH

- Familiarize genetic counselors with shared decision making and evidence of its effectiveness.
- Describe the critical components of shared decision making and relate them to the process of facilitating informed decisions in genetic counseling.
- Illustrate how a shared decision making model may be used in genetic counseling, highlighting use of new skills.
- Propose key research questions to assess the value of a shared decision making model in genetic counseling.

Submitted by: Research SIG

6:00 pm – 7:00 pm

316 Jane Engelberg Memorial Fellowship Presentation

0.10 CEU

1: Kelly Ormond, MS, CGC, JEMF Chair; 2: 2014 JEMF Awardee - Flavia Malheiro Facio, MS, CGC; 3: 2013 JEMF Awardee - Blythe Crissman, MS, CGC, Duke University; 4: 2013 JEMF Awardee - Kathryn Berrier Sheets, MS, CGC, Duke University Medical Division of Medical Genetics

- Describe the purpose and mission of the Jane Engelberg Memorial Fellowship.
- Provide an overview of the newly awarded 2014 research project.
- Share preliminary results of the Duke Down Syndrome (DS) Prenatal Diagnosis study and the role of the genetic counselor in the prenatal diagnosis experience.
- Describe patients' experience participating in research following a prenatal diagnosis of DS.

Sponsored Evening Session

7:15 pm – 8:45 pm

317 Termination of Pregnancy for Indications of Genetic Disorder in Advanced Gestations

0.10 CEU

1: Warren M. Hern, MD, MPH, PhD, Boulder Abortion Clinic

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

Sponsored by: Boulder Abortion Clinic

Saturday, September 20

Sponsored Breakfast Session

7:00 am – 7:45 am

401 Noninvasive Prenatal Testing for Microdeletions: One Year Later

0.05 CEU

1: Nicole Teed, MS, CGC, Sequenom Laboratories; 2: Heather Marin, MS, LCGC, Center for Prenatal Diagnosis; 3: Lauren Korty, MS LCGC, University of California San Diego Health System

- Describe the performance of non-invasive prenatal testing (NIPT) for microdeletions in clinical laboratory experience.
- Explain the benefits and limitations of prenatal screening for microdeletions.
- Illustrate the application of NIPT for microdeletions in clinical prenatal practice.

Sponsored by: Sequenom Laboratories

Educational Breakout Sessions

AEC Session Objectives *(continued)*

Saturday, September 20 *(continued)*

8:00 am – 9:30 am

402 Congenital Disorders of Glycosylation: Clinical and Genetic Variability

0.15 CEU

1: Eva Morava-Kozicz, MD, PhD, Tulane University Hayward Genetics Center; 2: Claire Teigen, CGC, GeneDx

- Describe the clinical variability observed in patients with congenital disorders of glycosylation (CDGs).
- Evaluate tests and diagnostic strategies for identifying CDGs based upon clinical presentations.
- Counsel patients and family members regarding treatment options, recurrence risks and recommendations for testing of additional family members based upon test results.

403 The Down Syndrome Consensus Statement Five Years Later: Making Progress and Evaluating the Impact of Non-Invasive Prenatal Screening/Testing on a Fragile Compromise between the Disability and Genetics Communities

0.15 CEU

1: Stephanie Meredith, MA, University of Kentucky; 2: Angela M. Trepanier, MS, CGC, Wayne State University; 3: Campbell K. Brasington, MS, CGC, Carolinas Medical Center; 4: Kathryn Berrier Sheets, MS, CGC, Duke University Medical Center Division of Medical Genetics; 5: Richard Ferrante, PhD, Center for Disability Resources, USC Columbia; 6: David Hoppe, Bipartisan Policy Center; 7: Janice G. Edwards, MS, CGC, University of South Carolina; 8: Judith Benkendorf, MS, CGC, American College of Medical Genetics and Genomics; 9: Nancy Rose, MD, University of Utah Health Sciences

- Understand the historical misperceptions between the medical/genetics and Down Syndrome (DS) communities and the unprecedented work at the DS Consensus Group meeting to bridge the gaps and establish common ground.
- Identify how the proposed areas of collaboration have been addressed through the development and dissemination of patient education materials, professional guidelines and research, and how those outcomes might be replicated for other conditions.
- Identify challenges and stressors that persist beyond the DS Consensus Group conversations, such as non-invasive prenatal testing (NIPT), community fractures and funding disparities, and how a working group of experts can tackle those issues.

404 The Non-Cancerous Female Breast

0.15 CEU

1: Adam Cohen, MD, MS, Huntsman Cancer Institute, University of Utah

- Describe the stages of development of the breast and label the parts using correct nomenclature.
- Recognize normal breast variants and those associated with an increased risk for breast cancer.
- Incorporate DCIS into genetic risk analysis using currently available data.

405 Crossing the Generation Gap: Engaging Millennial Learners

0.15 CEU

1: Caroline Lieber, MS, CGC, Sarah Lawrence College; 2: Anne Elizabeth Greb, MS, CGC, The Joan H. Marks Graduate Program in Human Genetics, Sarah Lawrence College; 3: Bernard R. Robin, PhD, University of Houston; 4: Paula Gregory, PhD, LSU Health Science Center-New Orleans; 5: Lori Dean, MS, CGC, University of Arkansas for the Medical Sciences

- Explore and evaluate learning characteristics of the millennial generation.
- Define and examine new technologies that can be employed in the education of millennial learners that can enhance the teaching experience.
- Demonstrate the incorporation of these technologies in genetic counselor and medical student training.

406 Double-Edged Sword: The Impact of Mass Media on Genetic Counseling

0.15 CEU

1: Jill M. Fischer, MS, CGC, Reprogenetics LLC; 2: Laura Hercher, MA, MS, CGC, Sarah Lawrence College; 3: Christine Colón, MS, LCGC, MotherToBaby Studies, Conducted by OTIS; 4: Rebecca Nagy, MS, CGC, The Ohio State University Wexner Medical Center - James Cancer Center; 5: Sara Riordan, MS, LCGC, Life Technologies

- Examine experiences and current information available about impact the media has on the public's perception of genetics and genomics.
- Discuss how the media affects the uptake and delivery of genetic counseling services.
- Outline possible strategies for working with patients and other providers in these situations.

Plenary Sessions

9:45 am – 11:00 am

407 Bringing ELSI Issues to Life: The Drama of DNA

0.125 CEU

1: Lynn Bush, PhD, MS, MA, Columbia University Medical Center; 2: Karen H. Rothenberg, JD, MPA, NHGRI, NIH, University of Maryland School of Law; 3: Wendy Uhlmann MS, CGC, University of Michigan; 4: Barbara Biesecker, PhD, MS, NHGRI/NIH; 5: W. Andrew Faucett, MS, CGC, Geisinger Health System; 6: Steve Keiles, MS, CGC, Ambry Genetics; 7: Rebecca Nagy, MS, CGC, The Ohio State University Wexner Medical Center - James Cancer Center; 8: Cate Walsh Vockley, MS, CGC, Children's Hospital of Pittsburgh

- Develop a deeper understanding of the ethical, psychosocial and policy implications of genomic research and medicine through this creative educational approach for the genetic professional and students who are attendees.
- Provide a creative pedagogical approach for the genetic counselor and other healthcare professionals to use themselves in their own teaching of genetic counseling students, medical students, residents and fellows.
- Enhance discourse and synthesize some key "teachable moments" for further analysis to be shared with colleagues and students well beyond the conference.

11:30 am – 12:30 pm

Late-Breaking Session

408 Cell-Free Nucleic Acids: To Prenatal Screening and Beyond

0.10 CEU

1: Megan Hall, PhD, ISMPP CMPP, Jazz Pharmaceuticals

- Explore utilization of cell free nucleic acids in settings other than prenatal.
- Discuss potential of tumor cell free DNA evaluation in cancer setting.
- Discuss potential for cell free DNA as marker for infectious agents, transplant rejection, and neurologic injury/disease.

Sponsored Lunch Session

12:30 pm – 2:00 pm

409 Genetic Testing Beyond the Individual: Finding Answers for Families

0.10 CEU

1: Linda Robinson, MS, CGC, University of Texas, Southwestern Medical Center, Simmons Cancer Center; 2: Colleen Caleshu, MSc, CGC, LGC, Stanford University School of Medicine; 3: Randy Scott, PhD, Invitae Corporation

- Develop strategies for identifying and counseling patients regarding multi-gene hereditary cancers and cardiology panels.
- Describe the unique counseling issues associated with communication of multi-gene panel results to family members.
- Demonstrate the value and utility of genetic testing and counseling to ensure patient understanding and facilitate communication of risk to relatives.
- Describe Invitae's clinical offering and commitment to family.

Sponsored by: Invitae Corporation



BOULDER ABORTION CLINIC

Specializing in Late Abortion for Fetal Disorders
Quality Care for Women Since 1973

Warren M. Hern, MD, MPH, PhD

Director

American Board of Preventive Medicine
Fellow, American College of Preventive Medicine
Associate 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)

- Outpatient abortion over 30 menstrual weeks for selected patients with documented fetal anomaly, fetal demise, or medical indications
- Routine preoperative ultrasound evaluation for all patients
- Routine preoperative use of laminaria for maximum safety
- Individual counseling and support
- Highly experienced and dedicated professional staff
- Tradition of research and teaching in abortion services
- Founding institutional member, National Abortion Federation
- Bilingual staff and physician: fluent in Spanish, Portuguese, and Italian
- Recent research publications on request
- Consultations on request

1130 Alpine Avenue, Boulder, CO 80304
Tel: (303) 447-1361 • (800) 535-1287 • Fax: (303) 447-0020
www.drhern.com

Concurrent Papers

Friday, September 19				
	302 - Cancer 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.	303 – GC Professional Roles 1. Discuss the professional and personal experiences of genetic counselors. 2. Identify potential future opportunities for the field of genetic counseling.	304 – Genetic Testing I: New Technology 1. Discuss latest developments in the field of diagnostic testing and test interpretation. 2. Describe the impact of Next-Gen Sequencing on patient diagnosis and counseling.	305 – Prenatal 1. Discuss the latest developments in prenatal testing and prenatal genetic counseling. 2. Describe the attitudes and experiences of patients and providers in the prenatal clinic.
	Room 208/209/210 This paper track is sponsored by:  Ambry Genetics™	Room 211/212/213	Room 206/207 This paper track is sponsored by:  BIOBASE <small>BIOLOGICAL DATABASES</small>	Room 220/221/222
8:00 am – 8:15 am	Reasons Patients Do Not Pursue BRCA Genetic Testing following Genetic Counseling <i>V. Raymond</i>	Genetic Counseling Licensure: An Oral History <i>K. Valverde</i>	Maximizing the Effectiveness of Exome Testing: A Retrospective Comparison of Diagnostic Yield in Singletons versus Multiple Family Members in Over 1500 Cases Submitted for Whole Exome Sequencing <i>H. Hanson Pierce</i>	Current Practices in Prenatal Genetic Counseling Regarding a Diagnosis of Down Syndrome <i>E. Moe</i>
8:15 am – 8:30 am	Large Scale Changes in Cancer Genetic Testing with Variable Integration of Expanded Gene Panels <i>G. Hooker</i>	The Genetic Counseling Assistant: Is Our Profession Ready for Multiple Career Levels? <i>L. Robinson</i>	Revised Diagnosis Through Exome Sequencing of an Infant with Congenital Cataracts Expands Phenotypic Spectrum of COL4A1-Associated Disorders <i>G. Chandratillake</i>	Continuing a Pregnancy Following Prenatal Diagnosis of a Lethal Fetal Defect is Associated with Improved Psychological Outcome <i>H. Cope</i>
8:30 am – 8:45 am	The Use of Diagnostic Exome Sequencing in the Identification of a Molecular Diagnosis in Cases Presenting with Cancer Phenotypes <i>C. Espenschied</i>	Career Advancement in Genetic Counseling: Perceived Opportunities and Barriers among Practicing Genetic Counselors <i>N. Poullard</i>	Complexities of Genetic Counseling for Variants of Unknown Significance on Whole Exome Sequencing <i>M. Harr</i>	Termination of Pregnancy for Aneuploidy: Are Things Changing? <i>K. Morris</i>
8:45 am – 9:00 am	Cancers Associated with <i>BRCA1</i> and <i>BRCA2</i> Mutations Other than Breast and Ovarian <i>J. Mersch</i>	Development, Experience and Expression of Meaning in Genetic Counselors' Lives: An Exploratory Analysis <i>P. McCarthy-Veach</i>	Patient Perceptions of Whole Genome Sequencing Results and Non-Actionable Findings <i>L. Jamal</i>	"I Kind of Just Went Along With It." An Exploration of the Experiences and Needs of Partners of Women Receiving Uncertain Prenatal Microarray Results <i>K. Stumm</i>
9:00 am – 9:15 am	Germline BRCA Mutations in an Unselected Cohort of Patients with Pancreatic Adenocarcinoma <i>S. Holter</i>	The Expanding Role of Genetic Counselors in Industry-Based Employment and Emerging Professional Issues: The Intersection of Innovation and Conflicts of Interest <i>S. Gandomi</i>	Attitudes of Genetic Counselors Toward Return of Incidental Results from Exome and Whole Genome Sequencing <i>M. J. Bamshad</i>	Evaluation of the Quality and Literacy of Commercial Non-Invasive Prenatal Test Websites <i>M. Bell</i>
9:15 am – 9:30 am	Physician Experiences and Understanding of a Genomic Sequencing Project for Oncology Patients <i>C. Weipert</i>	Emerging Genetic Counselor Roles within the Biotechnology and Pharmaceutical Industries: As Industry Interest Grows in Rare Genetic Disorders, How Are Genetic Counselors Joining the Discussion? <i>T. Field</i>	Consumers Report Lowered Confidence in their Genetics Knowledge following Personal Genomic Testing: Findings from the PGGen Study <i>D. A. Carere</i>	Clinical Experience of Trisomy 16 and 22, and Microdeletion Detection by Noninvasive Prenatal Testing <i>N. Dharajiya</i>

Saturday, September 20				
	410 – Access and Service Delivery 1. Recognize novel approaches in the delivery of genetic counseling services. 2. Describe issues in billing and reimbursement for genetic counseling services.	411 – Education/ELSI 1. Describe approaches and issues of genetic counseling education. 2. Discuss ELSI implications of providing genetic services.	412 – Genetic Testing II: Implementation 1. Explore emerging data on genetic test detection rates and issues in results interpretation. 2. Identify potential testing opportunities in the genetics clinic.	413 - Pediatrics 1. Discuss the latest developments in genetic counseling for pediatric patients and their families. 2. Describe the attitudes and experiences of patients and providers in pediatric clinics.
	Room 211/212/213	Room 206/207	Room 220/221/222	Room 217/218/219
2:00 pm – 2:15 pm	Development of a Preauthorization and Predetermination Process to Improve Access to Hereditary Cancer Risk Assessment Services and Subsequent Review of Reimbursement of CPT 96040 for Services Rendered <i>J. Polk</i>	Moving Beyond Likert Scales: Competency-Based Milestones and Implications for Genetic Counseling Education <i>C. Guy</i>	Diagnostic Yield of Genetic Evaluation and Testing at the Children's Hospital Colorado Autism Specialty Genetics Clinic <i>B. Miller</i>	A Follow-Up Study to the Possible Role of Pediatric Genetic Counselors in the International Adoption Process of Children with Special Needs <i>E. White</i>
2:15 pm – 2:30 pm	Perinatal Palliative Care and Bereavement: Establishing A Compassionate, Multidisciplinary Program for Families Facing a Life-Limiting Prenatal Diagnosis <i>S. Chadwick</i>	Assessment of the Readability of Genetic Counseling Patient Letters <i>E. Brown</i>	The Challenge of Comprehensive and Consistent Sequence Variant Interpretation across Clinical Laboratories <i>M. Pepin</i>	The Relationship Between Birth Weight and the Variability of Medical Complications in Patients with Cystic Fibrosis <i>R. Nelson</i>
2:30 pm – 2:45 pm	A Comparison of Telephone and In Person Genetic Counseling from the Genetic Counselor's Perspective <i>K. Burgess</i>	Video Patient Encounter for Genetic Counsellor Skill Development <i>J. Scott</i>	Yield of Pathogenic/Expected Pathogenic Variants in Young Women with Breast Cancer Undergoing Hereditary Cancer Panel Testing <i>L. Andolina</i>	Parents Report Impaired Health-Related Quality of Life in Children with Methylmalonic Acidemia with Improvements in Development and Health following Liver Transplantation <i>K. Splinter</i>
2:45 pm – 3:00 pm	Show Me the Money: Half A Million Downstream Billing Generated From Three Genetic Counselors' Visits over Nine Months <i>M. Dudek</i>	Genetic Counseling Student Experiences of Mental Health Concerns <i>A. Cantor</i>	Comparison of Mutation Detection in Cancer Specific versus Pan-Cancer Approaches to an At-Risk Population <i>P. Kaushik</i>	Family History of Epilepsy and Central Nervous System Comorbidities as a Prognostic Indicator for Respective Surgery Outcomes Among Pediatric Epilepsy Patients <i>K. Qualman</i>
3:00 pm – 3:15 pm	Resource Stewardship: The Children's Hospital Colorado Laboratory Experience to Improve the Value of Expensive Genetic Reference Laboratory Tests with Active Utilization Management <i>M. Lovell</i>	Balancing Cost Management and Ethics: A Proposed Process for Inpatient Genetic Testing Appraisal for Atypical Cases <i>M. Smith</i>	Performance of Single-Nucleotide Polymorphism-Based Non-Invasive Prenatal Testing in Low-Risk Women <i>M. Stosic</i>	Chinese Parents' Perception of Autism Spectrum Disorders: An Exploration of the Influence of Culture <i>J. Chen</i>
3:15 pm – 3:30 pm	Family History as a Predictor of Mutation Positive Status in Hypertrophic Cardiomyopathy: Refining a Genotype Risk Algorithm <i>L. Hipp</i>	Informed Consent in Pediatric Genome-Wide Sequencing: Content of Informed Consent Sessions and Parental Understanding <i>S. Wiley</i> Withdrawn	Performance of Noninvasive Prenatal Aneuploidy Testing at Different Fetal Fractions <i>R. McCullough</i>	Left Out in the Cold: Barriers to Clinical Trial Participation and the Impact on Parents of Sons with Duchenne Muscular Dystrophy <i>L. Murray</i>

Join us for a Product Theater

“Myriad myRisk™ Hereditary Cancer Panel: Clinical Data Supporting a Pan-Cancer Panel Approach to Hereditary Cancer Testing”

with **Jennifer Saam, MS, CGC, PhD**

Thursday, September 18th

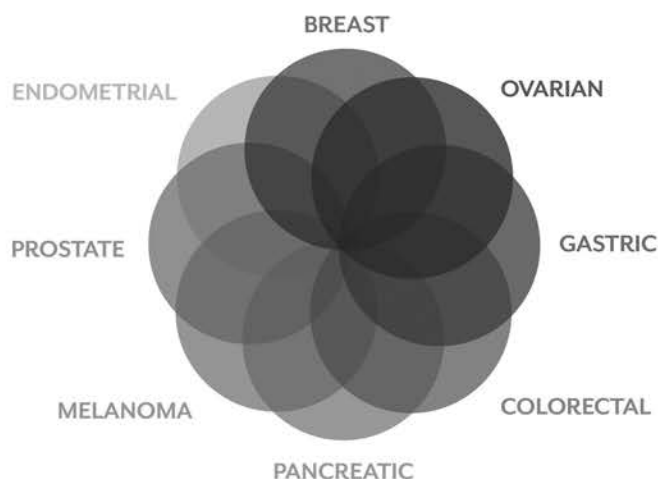
12:15 - 12:45 PM

Exhibitor Suite, Hall B



The Advancement of Hereditary Cancer Testing

Emerging Data Demonstrates the Benefits of a Pan-Cancer Panel Over Disease-Specific Testing



	Myriad myRisk	Breast Panel	Colon Panel
Breast	●	●	
Ovarian	●	●	●
Gastric	●		●
Colorectal	●		●
Pancreatic	●	●	●
Melanoma	●		
Prostate	●	●	
Endometrial	●		●

Myriad, the Myriad logo, Myriad myRisk, the Myriad myRisk logo, BRACAnalysis, the BRACAnalysis logo, COLARIS, and the COLARIS logo are either trademarks or registered trademarks of Myriad Genetics, Inc. in the United States and other jurisdictions.

©2014, Myriad Genetic Laboratories, Inc.

Posters with Authors

Objective:

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

215 Even Numbered Posters

Thursday, September 18

6:30 pm – 7:30 pm

0.10 CEU

309 Odd Numbered Posters

Friday, September 19

2:00 pm – 3:00 pm

0.10 CEU

Access and Service Delivery

- 1 Access to Genetic Counseling and BRCA Testing among a Population-Based Sample of Black Women with Early-Onset Breast Cancer
D. Bonner
- 2 Clinical Practices of Neurologists Related to Predictive Testing of Presymptomatic Patients At Risk for Huntingtons Disease
I. Bradley
- 3 A Survey of Genetic Counselors regarding the Impact of Recent State Legislation Restricting Access to Abortion on Patients and Practice
C. Cooney
- 4 The Impact of *Association for Molecular Pathology v. Myriad Genetics, Inc.* on Cancer Genetic Counseling Practice
V. Costello
- 5 A Descriptive Study of Current Hereditary Breast Cancer Knowledge and Clinical Practices among Florida Providers
D. Cragun
- 6 The Misdiagnosis of a Rare Disease: The Journey to a Hermansky-Pudlak Syndrome Diagnosis
L. Giannetti
- 7 Genetic Testing without Genetic Counselors: Exploring the BRCA Testing Experiences of Patients with Breast Cancer
M. Hayes
- 8 One Family, One Counselor: Continuity of Genetics Care in a Fetal Health Center
J. Kussmann
- 9 Ethical Patient Care through Financial Means: Requiring Genetic Counseling Code for Billing Prenatal Genetic Testing
M. Leach
- 10 The Implementation of a Multidisciplinary Care Clinic for Von Hippel-Lindau Disease at the University of Alabama at Birmingham
A. Mariano
- 11 Cost Should Not Be a Barrier to Genetic Testing in Patients with Paragangliomas and Pheochromocytomas
S. Merrill
- 12 Developing a Specialty Clinic for Patients with *PTEN* Hamartoma Tumor Syndrome: Benefits and Barriers
J. Mester
- 13 Impact of Genetic Counseling in the Cardiac Intensive Care Unit for Infants with Isolated Congenital Heart Defects
R. Palmquist
- 14 Service Delivery Model and Experiences in a Genetics Clinic for an Underserved Population
J. Profato

- 15 Centering Pregnancy: An Untapped Delivery Care Model for Genetic Counseling
P. Robbins Furman
- 16 Population Screening for Hereditary Cancer: Does it Really Work for Everyone?
L. Robinson
- 17 Jewish Genetic Disease Carrier Screening in Atlanta: Success of Marketing and Outreach Campaigns
Y. Shao
- 18 High Frequency of Genetic Services for Patients with Inborn Errors of Metabolism
Q. Stein
- 19 Genetic Testing Practices of Physicians for Primary Immunodeficiency Diseases: Are There Unmet Patient Needs?
S. Waltermann
- 20 Receipt of Cancer Genetics Services among Young Breast Cancer Survivors in Georgia
R. Webster

Adult

- 21 Assessment of the Clinical Presentation of Females Heterozygous for Fabry Disease: A Comparison of Classic and Later-Onset Forms
A. Conner
- 22 De Novo Mutation Rate in the *RYR2* Gene: Implications for Genetic Counseling
K. Davis
- 23 The Use of Social Media and the Impact of Support on the Well-Being of Adult Cystic Fibrosis Patients
M. Faust
- 24 A Role for Preventative Genetics? The Impact of *FTO* Results on Intention to Lose Weight
E. Gordon
- 25 Examining Differences in Symptoms in Individuals with Hypermobile Ehlers-Danlos Syndrome in Relation to Puberty
K. Heraty
- 26 A Novel Homozygous Variant in *RRM2B* in Two Siblings with Mitochondrial DNA Depletion Syndrome
W. Mu
- 27 The Complexity Continues: Identification of Pathogenic Sarcomeric Mutations in Families with a Clinical Diagnosis of Arrhythmogenic Right Ventricular Cardiomyopathy
B. Murray
- 28 Disclosure of Psychiatric Manifestations of 22q11.2 Deletion Syndrome: A Retrospective Chart Review Documenting Clinical Practice of Medical Geneticists
S. Talcott Baughman

Cancer

- 29 How Do We Counsel on Somatic Tumor Testing Reports? Red Herring or the Real Deal?
M. Dreon
- 30 Cancer Multi-Gene Panel Testing: Clinical Experience and Impact on Medical Management
M. Gabree
- 31 Risk Stratification of Women at Intermediate or High Risk of Breast Cancer: Developing a Consensus Framework for Screening and Prevention
J. Gagnon
Withdrawn
- 32 Atypical Phenotypes of Familial Adenomatous Polyposis and MYH-Association Polyposis Patients Ascertained through Multi-Gene Hereditary Cancer Panels
J. Guiltinan
- 33 Identification of Lynch Syndrome Families with Female Reproductive Tract Cancers in the Era of Next Generation Sequencing
C. Ikard
- 34 I Wish I Had Known This! Impact of Age on Life Choices and Testing Satisfaction for *BRCA1/2* Mutation Carriers Who Underwent Genetic Testing by Age 25
S. King
- 35 Returning Hereditary Cancer Panel Results to Patients is Clinically Feasible and Appreciated by the Patients
K. Kingham
- 36 *APC* Mutations in Children with Hepatoblastoma: Evidence for Genetic Evaluation for Familial Adenomatous Polyposis as Standard of Care
S. Knapke
- 37 Features of Hereditary Breast and Ovarian Cancer in a Lynch Syndrome Cohort Ascertained through Multi-Gene Panel Testing
H. LaDuca
- 38 The Angelina Jolie Effect: Assessing the Impact of a Celebrity's Story on Cancer Genetic Counseling
M. MacCuaig
- 39 Hereditary Breast and Ovarian Cancer: Implementation of Genetic Counseling within the High Risk Program of the Breast Center at Clinica Alemana of Santiago, Chile
S. Margarit
- 40 Hereditary Cancer Panels: Clinical Utilization, Testing Strategies and Genetic Counselors' Knowledge of Technology
D. McKenna
- 41 A Year of Unexpected Results: How the New Panels and a Non-Geneticist Diagnosed Three Patients with Hereditary Cancer Syndromes that a Genetic Counselor Would Have Missed
S. Morrill-Cornelius
- 42 Cancer Genetics Knowledge in Orthodox Jewish Women with and without a Family History of Cancer in the Greater Detroit Area
T. Paling
- 43 Do Personal or Family History of Renal Cell Carcinoma Predict the Likelihood of an Inherited Cancer Syndrome? Preliminary Results from a Multi Gene Hereditary Renal Cancer Test
L. Panos

- 44 Before It's Too Late: Broad Hereditary Cancer Panel Testing at the End of Life
M. Rabideau
- 45 Detection of Pathogenic Mutations in Moderate Penetrance Breast Cancer Genes Significantly Increases the Number of Patients Identified as Candidates for Increased Screening
E. Rosenthal
- 46 Multiple Lessons Learned from a Single Cancer Genetics Referral: Unusual Presentation of Monoallelic Mismatch Repair Deficiency
K. Schneider
- 47 Hereditary Breast Cancer Testing: When Results Are Not Straightforward
L. Servais
- 48 Newly-Described Genes on the GeneDx Comprehensive Cancer Panel: Pathogenic/Likely Pathogenic Variants Detected in Patients Satisfying National Comprehensive Cancer Network Testing Guidelines
S. Solomon
- 49 Examining Gastrointestinal Stromal Tumor Patients' Understanding of Tumor Mutation Analysis and Personalized Medicine
S. Stictevers
- 50 Unexpected *RAD51C* and *RAD51D* Findings in Breast Cancer Only Families
A. Stuenkel
- 51 Patients with Multiple Pathogenic Mutations Detected by Multi-Gene Panel Testing in a Lynch Syndrome Cohort
P. Summerour
- 52 *NF1* Mutations Detected on Multi-Gene Cancer Panel Testing in Proband with Atypical Phenotypes
P. Summerour
- 53 Usability of a Breast Cancer Risk Assessment Tool in a General Mammographic Screening Population: Utilization and Implications for Future Practice
M. Truelson
- 54 Predictors of Therapeutic and Prophylactic Mastectomy in Breast Cancer Patients
F. Tubito
- 55 Low Risk of *TP53* and *CDH1* Secondary Findings on Inherited Cancer Panels
K. J. Vogel

Counseling

- 56 "All in the Family:" Barriers and Motivators to the Use of Family History Questionnaires
S. Armel
- 57 Parents' Dreams for Their Young Adults with Down Syndrome: What Resources are Needed to Achieve Them?
J. Baker
- 58 Exploring Patient Reactions about Genetic Testing for Treatment Response to Bariatric Surgery
L. Balay
- 59 Genetic Counseling Clients' Views on Religious and Spiritual Assessment in Genetic Counseling
A. Bartenbaker

- 60 The Psychosocial Implications of Hereditary Diffuse Gastric Cancer
M. Beaston-Casey
- 61 An Investigation into the Factors that Influence Parental Decision to Disclose Carrier Status to Daughters in Families with Hemophilia
K. Bisordi
- 62 *SCN5A*: A Complex Channelopathy Gene with Counseling Challenges
D. Clements
- 63 Exploring Communication about Type 2 Diabetes and Perceptions of Risk Reduction Methods in Unaffected First Degree Relatives of an Affected Individual
S. Fernandes
- 64 Assessing the Impact of BRCA Testing Decisions on Breast Cancer Worry, Decision Regret and Cancer Risk Management
J. Frank
- 65 Parental Gender Differences: Perception of a Child Diagnosed with a Craniofacial Difference and Effects on Child Adjustment
J. Harris
- 66 Communication of Psychiatric Risk in 22q11.2 Deletion Syndrome
S. Hart
- 67 Factors Influencing Decisions to Undergo Preimplantation Genetic Diagnosis for Fanconi Anemia and Long-Term Interpersonal Outcomes of Those Decisions: A Qualitative Investigation of Parents' Experiences
K. Haude
- 68 Confirmed Versus Suspected: The Social Significance of a Genetic or Non-Genetic Diagnosis of Mitochondrial Disease
E. Krieg
- 69 Who Should I Bring? A Qualitative Investigation of Genetic Counselors' Perspectives on the Role of the Support Person in Cancer Genetic Counseling Sessions
B. LeRoy
- 70 An Evaluation of Genetic Counseling Effectivity as Perceived by Parents with Surviving Children of Trisomy 13, Trisomy 18, and Mosaic Trisomy 16
T. Lewis
- 71 The Role of Uncertainty in Coping Efficacy: The Experience of Parents of Children with Undiagnosed Medical Conditions
E. Macnamara
- 72 Duchenne Muscular Dystrophy: A Survey of Families' Perspectives on Carrier Testing and Communication within the Family
B. Mellicker
- 73 The Impact of Culture and Ethnicity on the Counseling Process: Perspectives of Genetic Counselors from Minority Ethnic Groups
B. Morris
- 74 How Does Family Communication about Cancer Work? Exploration of a Mediational Model
J. Quillin
- 75 Support Desired by Women following Termination of Pregnancy for a Fetal Anomaly
A. Ramdaney

- 76 Parent Reflections on the Diagnostic Odyssey
A. Richardson
- 77 Teens with Glycogen Storage Disease Types I and III: Planning to Take Responsibility
H. Rocha
- 78 "Who is the Deciding Factor?" Analysis of Parental Perspectives Regarding the Discontinuation of Elaprase in Children with MPS II
E. Schindewolf
- 79 Adaptation to Living with a *BRCA1/2* Mutation in Carriers and Their Partners
R. Shapira
- 80 Perceptions of Latinas on the Traditional Prenatal Genetic Counseling Model
S. Thompson
- 81 Exploring Fathers' Roles and Experiences with Dissemination of Sexual Health Information to Their Children with Down Syndrome
L. Torrey
- 82 Predictive Testing for Huntington's Disease: An Exploration of the Partner's Role in Decision-Making
S. Towner

Education/ELSI

- 83 Genetic Counselors' Knowledge and Perspectives of Cord Blood Banking and Stem Cell Therapies
S. Brummitt
- 84 An Analysis of Online Education Methods for an At Home Genetic Carrier Screening Service
J. Denton
- 85 The Impact of Increased Education on Career Interest in the Genetic Counseling Field among High School Students
J. Dix
- 86 Topics of Discussion in Families with Youth with Special Health Care Needs during Health Care Transition
C. Grabarits
- 87 The Student Voice: Learner-Centered Changes in a Molecular Genetics Laboratory Rotation to Increase Student Satisfaction and Knowledge Integration
C. Guy
- 88 The Conceptual and Practical Evolution of Education to Obtain Clinical Readiness in Genetic Counseling
S. Hassed
- 89 MedGen: A Portal for Medical Genetics Information
B. Kattman
- 90 Mapping and Evaluation of a Genetic Counseling Training Program Curriculum
T. Lepard Tassin
- 91 Twitter Activity Before and After Association for Molecular Pathology v. Myriad Genetics, Inc. Supreme Court Decision Using NodeXL
A. Lewis
- 92 The Impact of Rosa's Law on Describing Persons with Intellectual Disability
A. Lutter

- 93 Professional, Ethical and Legal Issues of Genetic Testing and Personal Insurance
I. Ngueng Feze
- 94 Implementation of Crisis Intervention Training in Genetic Counseling Training Programs
R. Reese
- 95 Disability Experiences and Perspectives regarding Reproductive Decisions, Parenting and the Utility of Genetic Services: A Qualitative Study
C. Roadhouse
- 96 Knowledge of Sickle Cell Disease in Ghana
D. Schlegel
- 97 Hereditary Breast and Ovarian Cancer Education Needs of Georgia Primary Care Residency Training Programs
E. Schmitt
- 98 Patient and Family Outreach for Marfan Syndrome and Related Disorders: Findings from a Multidisciplinary Education Day
A. Shikany
- 99 Group Prenatal Genetic Counseling Facilitates Patient Decision-Making
E. Sturm
- 100 "It Really Permeates Your Consciousness:" Examining the Impact of Genetic Counseling Training on Students' Prenatal Risk Perception and Reproductive Decision Making
I. Thompson
- 101 Parental Satisfaction and Teacher Perspectives on Inclusive Education of Students with Asperger Syndrome: An Educational Tool
H. Warren
- 102 Form Follows Function: Development of a Model for Clinical Supervision Practice in Genetic Counseling
C. Wherley
- 103 Traditional Textbook Photographs Negatively Impact Student Perception of Individuals with Visible Genetic Conditions
N. Wood
- GC Professional Roles**
- 104 The Importance of Suicide Screening in the Genetic Counseling Setting
C. Anderson
- 105 Current Practices and Perceptions of Ophthalmic Genetic Counselors
S. Chen
- 106 Clinical Exome Sequencing Test Development: Roles for Laboratory Genetic Counselors
E. H. Denenberg
- 107 U.S. Preventive Task Force and Genetic Counseling for BRCA Mutations: Tools to Help your Primary Care Colleagues
M. Doerr
- 108 Incorporating Computer-Aided Facial Analysis Software into Genetic Counseling Practice
D. Gelbman
- 109 The Relationship between Burnout and Occupational Stress in Genetic Counselors
B. Johnstone
- 110 Behind Laboratory Doors: Lab Genetic Counselors' Experiences, Professional Identity and Unique Ethical and Professional Challenges
C. Koellner
- 111 Genetics Laboratory Directors' Perspectives on the Role of Genetic Counselors in Acquired Mutation Testing: Current and Expanded Opportunities
C. Lewis
- 112 Assessing the Practices of Genetic Counselors regarding Head Circumference Measurement in Hereditary Cancer Assessment
A. Matchette
- 113 A Review of the Current State of Clinical Pharmacogenomic Testing: An Examination of Existing and Potential Roles for Genetic Counselors and Pharmacists
A. McKittrick
- 114 Role of a Genetic Counselor in the Next Generation of Oncology Clinical Trials
A. Varma
- Genetic Testing I**
- 115 Meanings Parents Attribute to an Answer from Whole Exome Sequencing Research
B. Blosser
- 116 Disomic Placentas, Trisomic Babies: Reverse Mosaicism and Implications for Noninvasive Prenatal Testing
T. Boomer
- 117 Multigene Panel Testing vs. Whole Exome Sequencing: What is the Best Testing Approach for Patients with Epilepsy and Neurodevelopmental Disorders?
M. Bradbury
- 118 Genomes, Exomes and Targeted Disease Panels: Understanding their Relative Strengths and Weakness When Providing Genetic Counseling
C. Campbell
- 119 A Negative Result on Exome Sequencing: What a Genetic Counselor Should Know
G. Chandratillake
- 120 Exploring Parental Perspectives on the Return of Genomic Results for Children Enrolled in a Pediatric Genetic Biorepository
P. Connors
- 121 The Importance of Clinical Indications in the Analysis and Interpretation of Next Generation Sequencing Gene Panel Data
G. Douglas
- 122 Identification of Recurrent De Novo Alterations in the Clinically Novel *PURA* Gene through Diagnostic Exome Sequencing
D. El-Khechen
- 123 Homocystinuria Diagnosed by Whole Exome Sequencing in Siblings from an Isolated Central American Village
C. Farkas
- 124 Use of an Enhanced Exome with Genome-Wide Structural Variant Detection for the Diagnosis of Mendelian Disease
S. Garcia
- 125 Prenatal Screening and Testing in the Non-Invasive Era: A New Paradigm in Patient Desire?
A. Hanson
- 126 Preferences of Undergraduate Students Towards the Reporting of Incidental Findings in Whole Genome and Whole Exome Sequencing
C. Hong
- 127 Trends in a Rapidly Expanding Genetic Testing Marketplace
G. Hooker
- 128 Whole Genome Sequencing of De Novo Balanced Chromosome Rearrangements Empowers Prenatal Genetic Counseling
T. Kammin
- 129 Assessing Factors Affecting the Decision Regarding Noninvasive Prenatal Testing after Genetic Counseling
K. Kobara
- 130 Pharmacogenomics: An Analysis of the Baseline Knowledge of ClinSeq® Participants
J. Kohler
- 131 Assessing Patient Comprehension Using a Modified Consent Document in a Pharmacogenomics Study
S. Lahiri
- 132 Medical EmExome: Making "Whole" Exome Sequencing Truly Whole!
A. Narravula
- 133 Germline *STAT2* Variant Implicated through Clinical Whole Exome Testing in Patient with Familial Non-Medullary Thyroid Cancer
J. Pickard Brzosowicz
- 134 Diagnostic Exome Sequencing for Patients with a Family History of Consanguinity: Over 40% of Positive Results Do Not Follow an Autosomal Recessive Pattern
Z. Powis
- 135 Associations between Life Stage and Preferences regarding Return of Results from Whole Genome Sequencing: An Online Survey Study
E. Quinn
- 136 Interpretation and Clinical Implications of Significant Variants Detected via Whole Genome Sequencing in a Healthy Adult Cohort
E. Ramos
- 137 Parental Attitudes Toward Whole-Genome Newborn Screening
S. Rego
- 138 A Trio-Based Whole Genome Sequencing Longitudinal Cohort Study: Experiences and Genetic Counselor Roles
S. Ruppert
- 139 250,000 Cases and Counting: The High Volume Clinical Experience with Noninvasive Prenatal Testing
J. Saldivar
- 140 Homozygous Mutations in Newly Described Gene, *CNTNAP1*, Detected through Whole Exome Sequencing Provide Diagnosis for Siblings
A. Shealy
- 141 Genotype-Phenotype Correlation with Exome Sequencing
E. Smith

- 142 Reporting Incidental Findings in Clinical Whole Exome Sequencing: Incorporation of the 2013 American College of Medical Genetics Recommendations into Current Practices of Genetic Counseling
L. Smith
- 143 Importance of Clinical Data in the Interpretation of a Novel *EHMT1* (Kleefstra Syndrome) Variant Identified by Whole Genome Sequencing
B. Smith-Packard
- 144 Motivation and Consumer Willingness for Self-Pay Noninvasive Prenatal Testing at the Medcan Clinic in Canada
E. Stephenson
- 145 Significance of Reporting Candidate Genes with a Potential Relationship to a Disease Phenotype Identified by Whole Exome Sequencing
J. Tahilliani
- 146 Clinical Experience Reporting Trisomy 16 and 22 on Noninvasive Prenatal Testing: Test Performance and Implications for Genetic Counseling
N. Teed
- 147 Opt-Out Rates for Receiving Secondary Findings from Whole Exome Sequencing: The Experience of a Clinical Laboratory
A. Telegrafi
- 148 Successful Utilization of Enhanced Exome Sequencing to Identify the Genetic Cause of Retinal Disorders in a Case Series
J. Tirsch
- 149 Parents' Perspectives: Child's Whole Exome Sequencing Research Results of Uncertain Significance
G. Tran
- 150 Application of Risk-Score Analysis to Low-Coverage Whole Genome Sequencing Data for the Noninvasive Detection of Trisomy 21 and Trisomy 18
J. Tynan
- 151 Understanding Next Generation Sequencing Results: An Update on Elements of Variant Interpretation and Classification
K. Vikstrom
- 152 Detection of Maternal 22q Deletions by Noninvasive Prenatal Testing
J. Wardrop
- 153 Secondary *PTEN* Mutation Identified on Whole Exome Sequencing
C. Williams
- 154 Psychological Impact of Receiving Secondary Findings from Whole Exome Sequencing
J. Wynn
- 155 Perceived Obligations by Researchers in Genetics to Return Individual Research Results and Incidental Findings to Participants
C. Young
- 158 Motivations for Presymptomatic Genetic Testing in *C9orf72* Related Frontotemporal Degeneration and Amyotrophic Lateral Sclerosis Families
C. Chambers
- 159 Products of Conception Testing by SNP Microarray Analysis on Monozygotic Twin Gestations Reveals Discrepant Chromosome Results and the Scope of Aneuploidy in Mosaic Pregnancies
D. Clark
- 160 Exploring Genetic Counselors' Perspectives on the Clinical and Personal Utility of Genetic Testing
L. Coon
Withdrawn
- 161 Hereditary Pancreatic Cancer Multi-Gene Test: Preliminary Results
E. Dalton
- 162 Expanded Carrier Screening in the Ashkenazi Jewish Population
L. Elkhoury
- 163 200 Days of Multi-Gene Testing: The Clinical Experience
A. Forman
- 164 "The Sooner the Better:" Genetic Testing following Ovarian Cancer Diagnosis
E. Fox
- 165 Male Rett Syndrome Resulting from an *MECP2* Missense Variant
J. Hartley
- 166 Novel Variant Identified in *CYP7B1* Using Next-Generation Sequencing
C. Hartshorne
- 167 Factors Affecting Decision Making of Social Workers about Preadoption Genetic Testing
D. Julian
- 168 Going Beyond The Guidelines: A Call for Expanded Jewish Carrier Screening Based on an Analysis of 500 Clinical Samples Screened via an Expanded Carrier Screening Platform
G. Kellogg
- 169 A Single Institution's Experience with the myRisk Panel: Exploring the Rates of Incidental Findings and Variants of Uncertain Significance
B. Leach
- 170 A Case of Prenatal Detection of Duplication (21)(q22.2q22.13): Use of Multiple Testing Methodologies to Clarify Apparently Discordant Results
K. Levandoski
- 171 A Comparison of Traditional and Panel Testing for Hereditary Breast and Ovarian Cancer in over 900 Patients
S. Lincoln
- 172 One Disease, Multiple Solutions: A Family Case Study of Preconception and Prenatal Testing for Duchenne Muscular Dystrophy and Exploration of Reproductive Testing Options
M. Maisenbacher
- 173 ClinVar: The Respository for Interpretations of Clinically-Relevant Variations
A. Malheiro
- 174 Meckel-Gruber Syndrome in a Fetus Found to Be Compound Heterozygous for Mutations in *TMEM231* and Heterozygous for a Mutation in *MKS6*
K. Marchand
- 175 Understanding Patient Motivation and Factors that Influence Decisions on Prenatal Testing
L. Masso
- 176 Genetic Analysis of 113 Usher Syndrome Cases Using a Nine Gene Next Generation Sequencing Panel and a Targeted Array Comparative Genomic Hybridization
M. Meltzer
- 177 The Impact of Fragile X Newborn Screening Results on Reproductive Choices and Surveillance for Fragile X-Associated Disorders
P. Miranda
- 178 Genotype Does Not Always Clarify Phenotype: Diagnosis Doesn't Stop with DNA
K. Morphy
- 179 Oocyte Donor Genetic Screening Practices
E. Moyle
- 180 A Novel Genotype-Phenotype Correlation of a Patient with a 13q13 Deletion: A Case Report
M. Nardini
- 181 Exploring Reasons for Genetic Testing Not Pursued in Patients Referred for Genetic Risk Evaluation
J. Osborne
- 182 Confirming Prenatal FISH Results: A Rare Prenatal Diagnosis of Duplication (21)(q22.13q22.2)
K. Patek
- 183 Quality of Care Surrounding Genetic Tests Ordered by Non-Geneticists
J. Peredo
- 184 Chromosomal Microarray in Prenatal Diagnosis: The Wayne State University Experience
K. Rauch
- 185 Family History or Carrier Screening? How Patients are Identified to Be Candidates for Preimplantation Genetic Diagnosis for Three Common Conditions: Cystic Fibrosis, Spinal Muscular Atrophy and Fragile X Syndrome
E. Repass
- 186 Detection of Uniparental Disomy by SNP Microarray
B. Rush
- 187 Next-Generation Sequencing Reveals a Novel Duplication in *BRCA2*
V. Semenysty
- 188 Scientific Explanation: When the "False Positive" Noninvasive Prenatal Test Result is Not False
C. Settler
- 189 A Case of Hypertrophic Cardiomyopathy: The First with Likely-Significant Alterations in Both *TPM1* and *TMEM43*
K. Spoonamore
- 190 Attitudes and Motivations towards Genetic Testing among College-Level Athletics
A. Squire
- 191 From Last to First: Repositioning Genetic Testing in the Diagnostic Process
C. Stanley

Genetic Testing II

- 156 Determining the Clinical Significance of *BRCA1* and *BRCA2* Intronic and Exonic Splicing Variants
K. Bowles
- 157 The Impact of Testing Minors for Alpha1-Antitrypsin Deficiency from the Perspective of Adults Tested as Children
A. Brown

- 192 Rethinking the Family Cancer History Questionnaire in the Era of Next Generation Sequencing Panels: Are We Asking the Right Questions?
K. Vikstrom
- 193 Evaluation of Laboratory Perspectives on Multiplex Genetic Testing for Hereditary Cancer Susceptibility by Next-Generation Sequencing
J. Stoll
- 194 International Data Sharing Efforts: Lessons from Council on Governmental Relations
S. White
- 195 Multi-Gene Testing for Paragangliomas and Pheochromocytomas: Diagnostic Yield and Phenotypic Spectrum
S. Witherington
- 196 Utilization of Next Generation Panel Genetic Testing Diagnoses a Patient with Atypical Colon Polyp Presentation
C. Williams
- Pediatrics**
- 197 Biochemical and Clinical Findings of Patients with the Common Variant (c.625G>A) in *ACADS* Identified through Minnesota Newborn Screening
C. Alexander
- 198 Hemophilia B Acquired from Liver Transplantation: A Case Report and Literature Review
K. Bergstrom
- 199 Development of Newborn Screening Connect (NBS Connect): A Self-Reported Patient Registry and Its Role in Improvement of Care for Inherited Metabolic Disorders
A. Devarajan
- 200 Pediatric Neurometabolic Disorders: Medical Needs, Service Use and Disease Impact on the Family
J. Diaz
- 201 If the Microarray Diagnosis Doesn't Quite Fit, You Must Not Quit: A Case of 16p11.2 Deletion
H. Dubbs
- 202 Perceptions of Severity of Children's Bleeding Disorders: Impact on Parental Quality of Life and Reproductive Decisions
E. Holt
- 203 Reference Growth Curves for Children with Classic Galactosemia
J. Howell
- 204 Discontinuing Enzyme Replacement Therapy in Patients with Lysosomal Storage Diseases Due to Significant Clinical Decline
A. Kim
- 205 Low Adiposity of Cystic Fibrosis Mice Due to Small Adipocyte Size
J. Klavarian
- 206 Carrying Cri du Chat: A Case Report of 46, XX, del (5) (p15.32)mat
K. Lammers
- 207 Case Series: Eliminating Barriers to Genetic Testing in the Duchenne/Becker Muscular Dystrophy Community
A. Martin
- 208 When Café Au Lait Spots Do Not Equal *NF1*: Constitutional Mismatch Repair Deficiency as an Important Differential Diagnosis for *NF1* Stigmata and Pediatric Cancer
R. McGee
- 209 Chromosome Microarray Results and Implications on Genetic Counseling: A Case Report Involving a 22q11.2 Distal Deletion
R. Mostafavi
- 210 A Multidisciplinary Approach to Individualized Management of Cornelia de Lange Syndrome at The Children's Hospital of Philadelphia: The Parents' Perspective
S. Noon
- 211 Insulin Misregulation Underlies Circadian and Cognitive Deficits in a *Drosophila* Fragile X Model
B. Schoenfeld
- 212 Further Evidence of a Milder Phenotype Associated with the V80F and D132A Mutations in *EXOSC3*
A. Schreiber
- 213 Comparing Face Perception Skill among Individuals with Williams Syndrome, Autism Spectrum Disorder, Prader-Willi Syndrome, Mild Intellectual Disability and Typical Development
K. Schultz
- 214 Prevalence of Genetic Conditions and Development Delays in Emergency Department Visits for Poison and Foreign Body Ingestions
E. Sengstock
- 215 Disclosure of a Fragile X Syndrome Diagnosis to Symptomatic Females: A Qualitative Study of the Parental Approach
L. Stobie
- 216 *FBN1* Mutation Identified in Critically Ill Neonate with a Diagnosis of Marfan Syndrome
J. Tarpinian
- 217 Death of a Child to Tay-Sachs and Other Progressive Neurological Disorders: Long-Term Impact on Parents' Emotional and Personal Lives
E. Williams
- 218 Expanding the Phenotype of Mitochondrial Disease: What Every Genetic Counselor Needs to Know
S. Wong
- Prenatal**
- 219 The Down Syndrome Prenatal Diagnosis Study: Understanding the Informational and Emotional Needs of Individuals with a Prenatal Diagnosis of Down Syndrome
K. Berrier Sheets
- 220 Prenatal Indicators of Sotos Syndrome
S. Burnett
- 221 Attitudes of Prenatal Genetic Counselors regarding Pan Ethnic Carrier Screening and Development of a Pre-Counseling Informational Mobile Webpage
K. Curd
- 222 Not the Answer We Expected: Detection of Fetal 47,XXX via Non-Invasive Prenatal Testing in the Setting of Increased Nuchal Translucency
M. Discenza
- 223 An Evaluation of Referral Indications for Prenatal Genetic Counseling Over a 15-Year Period
D. Durand
- 224 Walking the Edge with Controversial Use of Preimplantation Genetic Diagnosis (PGD): Opinions and Attitudes of Genetic Counselors
K. Everton
- 225 Recurrent Bilateral Renal Agenesis: Expanding the Spectrum of Fraser Syndrome
M. Ferguson
- 226 Termination Rates and Influencing Variables for Pregnancies Affected by Aneuploidy
K. Garfield
- 227 The Paternal Age Effect: A Preliminary Study of Current Challenges for Prenatal Care
A. Gunter
- 228 Conception through Gamete Donation: Challenges in Genetic Testing and Counseling of Gamete Donor Recipients
L. Isley
- 229 Genetic Evaluation of Products of Conception and Deceased Fetuses after Miscarriage and in Cases with Abnormal Ultrasound Findings: Experiences of a Diagnostic Laboratory
A. Janze
- 230 Prenatal Diagnosis of Pallister Hall Syndrome: A Case Report
S. Kasperski
- 231 What is the Chance for Women with a History of Depression to Develop Psychosis in the Postpartum Period? Preliminary Data to Inform Prenatal Genetic Counseling Practice
C. Mighton
- 232 Making the Choice to Continue: How Noninvasive Prenatal Screening is Utilized in Southeastern United States
J. Nichols
- 233 The Declining Rate of Amniocentesis Procedures at One Center
L. Rhee-Morris
- 234 Challenges in Expanded Carrier Screening: Determining Accurate Detection Rates and Residual Carrier Risks
S. Rodriguez
- 235 Clinical Description of a Newborn Male with a Prenatal Diagnosis of 46,XY,del(16)(q12.1q21)dn
B. Tucker
- 236 Parent Perspectives on Support Received from Physicians and Genetic Counselors following a Decision to Continue a Pregnancy with a Prenatal Diagnosis of Trisomy 13/18
S. Wallace
- 237 Potential Biological Explanations for No Results for Sex Chromosome Aneuploidy Assessment Using Directed Cell-Free DNA Analysis: A Summary of Three Cases
K. White

Thank You to Our Sponsors

NSGC Year-Round Premier Sponsors

Gold



Silver



33rd Annual Education Conference Sponsors

Gold



Silver



Bronze



NSGC Awards and Fellowships

The Audrey Heimler Special Projects Award

Development of a Website to Facilitate the Recruitment of African Americans and Latinos into the Field of Genetic Counseling
Kara Anstett, BSc and Sharon Chen, BA

Genetic Counseling Assistants: An Integral Piece of the Evolving Genetic Counseling Service Delivery Model
Beth Crawford, MS, CGC, Linda Robinson, MS, CGC and Sara Pirzadeh-Miller, MS, CGC

The Jane Engelberg Memorial Fellowship

How Does Family History Influence Psychosocial Adaptation in Individuals with Inherited Cardiomyopathies and Their At-Risk Family Members?
Cynthia A. James, ScM, PhD, CGC

NSGC Leadership Awards

Natalie Weissberger Paul National Achievement Award
Barbara Bowles Biesecker, PhD, MS

International Leader
Alexandra J. Obregon-Tito, PhD, MSc, MA, CGC

New Leader
Melanie Walker Hardy, MS, MS, CGC

Strategic Leader
Monica Marvin, MS, CGC

Outstanding Volunteer
Kami Wolfe Schneider, MS, CGC

Outstanding Volunteer
Heather Zierhut, PhD, MS, CGC

Best Abstract Awards

Best Full Member Abstract Award
Analysis of Billing and Reimbursement of Genetic Counseling Services in a Single Institution in a State Requiring Licensure
Jennifer Leonhard, MS

Beth Fine Kaplan Student Abstract Award
Genetic Counselors as Choice Architects: Some Considerations for Presenting Genetic Testing Decisions in a Complex Choice Environment
Marci Barr

Cultural Competency Scholarship
Michale Kieke, PhD
Alexander Ing

AEC Planning Subcommittee

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

Chair
Katherine R. Dunn, MS, LCGC

Vice Chair
Lori A.H. Erby, ScM, PhD, CGC

Subcommittee Members
Fallon Brewer, MS, CGC
Jason Flanagan, MS, CGC
Stephanie Gandomi, MS, CGC, LGC
McKinsey Goodenberger, MS, CGC
Lauren Hache, MS, CGC
Andrea Harbison, MS, CGC
Kelly Jackson, MS, CGC
Kate Lamvik Loranger, MS, CGC
Kirsty McWalter, MS, CGC
Ana Morales, MS, CGC

Abstract Chair
Emily Edelman, MS, CGC

Abstract Vice Chair
Beverly Yashar, MS, CGC, PhD

Abstract Workgroup
Sharon Aufox, MS, CGC
Melanie Baxter, ScM, CGC
Barbara Biesecker, PhD, MS, CGC
Meg Bradbury, MS, CGC
Kari Haag Branham, MS, CGC
Carrie Castonguay, MS, CGC
Kathleen Collins, MS, CGC
Jennifer Czerwinski, MS, CGC
Martha Dudek, MS, LCGC
Kristi K. Fitzgerald, MS, CGC
Sara M. Fitzgerald-Butt, MS, CGC, LGC
Stephanie Gandomi, MS, CGC, LGC
Tracey Grant, MS
Catriona Hippman, MSc, CGC
Jodie Ingles, GDGC, PhD
Cindy James, PhD, ScM, CGC
Emily James, MS, CGC
Yelena Kemel, MS, ScM, CGC
Katie Krepkovich, MS, CGC
Andrea Kwan, MS, CGC
Dawn Laney, MS, CGC, CCRC
Erin Miller, MS, CGC
Sarah Noblin, MS, CGC
Victoria Raymond, MS, CGC, CCRP
Thereasa Rich, MS, CGC
Christina Rigelsky, MS, CGC
Taylor Sale, MS, MEd, LCGC
Carly Siskind, MS, CGC
Christine Spaeth, MS, CGC
Christopher Tan, MS, CGC
Karen Wain, MS, CGC
Susan Walther, MS, CGC
Elisabeth Wood, MS, CGC
Heather Zierhut, PhD, MS

Education Committee Chair
Renee Chard, MSc, CGC

Education Committee Vice-Chair
Heather MacLeod, MS, CGC

Education Committee Board Liaison
Carrie L. Atzinger, MS, CGC

Networking Activities and Business Meetings

New! - NSGC SIG Fair

Wednesday, September 17

2:00 pm – 2:30 pm • Room R05

NSGC Members: Attend the new SIG Fair to meet with SIG leaders and learn more about current projects and how you can become involved.

First-Time Attendees

Welcome to the AEC: How to Make the Most of the Conference and NSGC

Wednesday, September 17

2:00 pm – 2:45 pm • Room R02/R03/R04

Are you a first-time AEC attendee? Make your way to this event to network with other new attendees and learn about the different types of educational sessions available at the AEC.

Welcome to the AEC SIG Fair

Wednesday, September 17

2:45 pm – 3:15 pm • Room R05

First-time AEC attendees and new NSGC members: Meet with SIG leaders at this event devoted specifically to fostering relationships between SIGs and new NSGC members.

Welcome Reception

Wednesday, September 17

6:30 pm – 8:00 pm • Exhibit Hall B

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, September 18

10:45 am – 11:30 am • The Great Hall AD

Join President Jennifer Malone Hoskovec, MS, CGC, as she provides an overview of NSGC activities and accomplishments over the past year; reviews NSGC's advocacy efforts and strategic initiatives; and provides highlights from 2014.

ABGC Annual Business Meeting

Thursday, September 18

11:30 am – 12:00 pm • The Great Hall AD

ACGC Presentation

Thursday, September 18

12:00 pm – 12:30 pm • The Great Hall AD

NSGC Annual Business Meeting

Friday, September 19

11:30 am – 12:30 pm • The Great Hall AD

Incoming Presidential Address

Saturday, September 20

11:00 am – 11:30 am • The Great Hall AD

Hear NSGC President Elect Joy Larsen Haidle, MS, CGC, as she introduces herself to NSGC members and outlines her vision for NSGC in 2015.

Meals and Breaks

Breakfast and Breaks

Continental breakfast will be served Thursday through Saturday outside of The Great Hall from 7:00 am – 8:00 am.

Concessions will be located in the Exhibitor Suite on Thursday and Friday. Concessions will also be available in the convention center main foyer on Wednesday and Saturday. Refreshment breaks will be located in the AEConnect area (Exhibitor Suite, Hall B) on Thursday from 9:30 am – 9:45 am and on Friday from 3:30 pm – 3:45 pm.

All refreshment breaks are sponsored by

Baylor
College of
Medicine

Thursday, September 18

9:30 am – 9:45 am

3:30 pm – 3:45 pm

Friday, September 19

9:30 am – 9:45 am

3:00 pm – 3:15 pm

Saturday, September 20

9:30 am – 9:45 am

Network

Synergy

Gather

Connect

Network

Share

Grow

Collaborate

Network

Socialize

Gain

Convene

Network

Synergy

Gain

Collective

Network

Succeed

Gather


Collaborate

Network

Share

Grow

Connect



AEConnect

Located in the Exhibitor Suite
Open Wednesday - Friday During Exhibit Hours

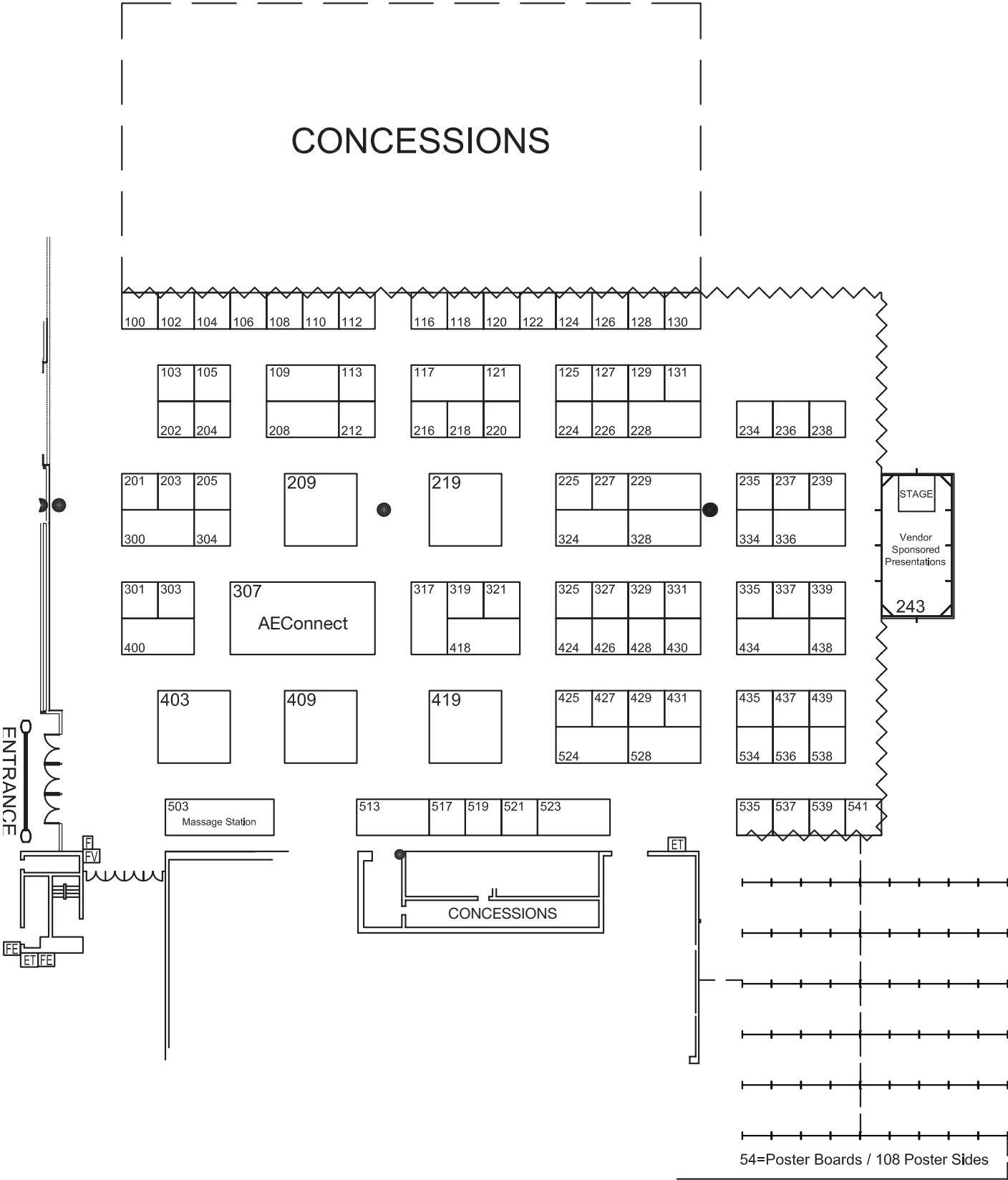
AEConnect will be the premier place at the Annual Education Conference to network with your professional community. While in the Exhibitor Suite, stop by to check out available job postings, learn more about our social media efforts, connect with exhibiting companies one-on-one, and meet up with old colleagues or friends. We invite you to visit AEConnect during Exhibitor Suite hours Wednesday through Friday.

Exhibitor Directory by Company Name

Exhibitor	Booth
Ambry Genetics	409, 541
American Board of Genetic Counseling (ABGC)	321
American Thrombosis & Hemostasis Network (ATHN)	118
Ariosa Diagnostics	523
ARUP Laboratories	331
Association of Public Health Laboratories	234
Asuragen, Inc.	425
Basser Research Center for BRCA	429
Baylor College of Medicine Medical Genetics Laboratory	403
BIOBASE	303
BioMarin Pharmaceutical Inc.	339
Boulder Abortion Clinic	304
Bright Pink	521
Cancer Treatment Center of America	128
Casey Molecular Diagnostics Laboratory	437
Cedar River Clinics	537
Center for Jewish Genetics	439
City of Hope - Clinical Molecular Diagnostic Laboratory	216
Claritas Genomics	212
Clinical Genome Resource (ClinGen)	329
CombiMatrix	224
Connective Tissue Gene Tests	424
Cord Blood Registry	325
Counsyl Inc	434
Courtagen Diagnostics Laboratory	125
Denver Genetics Laboratories at Children's Hospital Colorado	106
Edimer Pharmaceuticals	337
Emory Genetics Laboratory	418
FORCE: Facing Our Risk of Cancer Empowered	426
Fulgent Diagnostics	220
Geisinger Health System	534
Gene By Gene	328
GeneDx	219
GeneTests	121
Genetic Alliance	203
Genome Magazine	112
Genomic Healthcare Innovations	129
GenPath Women's Health	228
Genzyme, a Sanofi Company	535
Greenwood Genetic Center	201
Heart Institute Diagnostic Company	110
Human Genetics Laboratory UNMC	120

Exhibitor	Booth
Hyperion Therapeutics	113
Illumina, Inc.	400
Improve Labs	117
Integrated Genetics	528
Invitae Corporation	419
Kaiser Permanente - Northern California	519
Laboratory for Molecular Medicine, Partners Personalized Medicine	327
Mauli Ola Foundation	539
Mayo Medical Laboratories	235
MotherToBaby Conducted by the Org. of Teratology Information Specialists	127
Mount Sinai Genetic Testing Laboratory	335
Myriad Genetic Laboratories, Inc.	524
Natera, Inc.	209, 229
National Society of Genetic Counselors (NSGC)	317
NextGxDx, Inc.	225
NSGC Cancer SIG	319
NSGC Prenatal SIG	108
Omicia	438
Pathway Genomics	336
PerkinElmer Labs	324
Personalis, Inc.	205
Pfizer, Inc.	116
PreventionGenetics	208
Proband by The Children's Hospital of Philadelphia	226
Progeny Software, LLC	435
Quest Diagnostics	300
Recombine	218
Recordati Rare Diseases	103
Reproductive Genetic Innovations (RGI)	227
Reprogenetics	431
Seattle Children's Hospital	202
Sequenom Laboratories	513
Sharsheret	122
Sinai Surgical Center	428
Southwestern Women's Options	517
St. Louis Fetal Care Institute	237
Transgenomic, Inc.	334
UAB Medical Genomics Laboratory	427
UCLA Clinical Genomics Center	536
University of Chicago Genetic Services	430
University of Washington Reference Lab Services	200

Exhibitor Suite Map





Prenatal Genetic Counselors

2nd & 3rd trimester abortions
Fetal genetic & anatomic anomalies

Accredited surgical center
Affiliated with Cedars-Sinai Medical Center

Sinai Surgical Center

Booth 428 Exhibit Hall

Meet the providers

Beverly Hills, California
(310) 247-0553

Exhibitor Index

Ambry Genetics **Booth # 409, 541**

Phone: 949.900.5500 | Fax: 949.900.5501
info@ambrygen.com
www.ambrygen.com

Ambry Genetics is a College of American Pathologists (CAP)-accredited and Clinical Laboratory Improvement Amendments (CLIA)-certified commercial clinical laboratory with headquarters in Orange County, California. Since its founding in 1999, it has become a leader in providing genetic services focused on clinical diagnostics and genomic services, particularly in sequencing and array services. Ambry has established a reputation for unparalleled service and for over a decade has been at the forefront of applying new technologies to the clinical molecular diagnostics market and to the advancement of disease research.

American Board of Genetic Counseling (ABGC)

Booth # 321
Phone: 913.895.4617
Fax: 913.895.4652
info@abgc.net
www.abgc.net

The American Board of Genetic Counseling (ABGC) is the credentialing organization for the genetic counseling profession in the United States and Canada. The ABGC certifies and recertifies qualified genetic counseling professionals.

American Thrombosis & Hemostasis Network (ATHN)

Booth # 118
Phone: 800.360.2846 | Fax: 847.572.0967
info@athn.org
www.athn.org

The American Thrombosis and Hemostasis Network (ATHN) is a non-profit organization committed to advancing and improving care for individuals affected by bleeding and thrombotic disorders. ATHN manages a national database of patient health data that can be used to improve care and support vital research.

Ariosa Diagnostics **Booth # 523**

Phone: 925.854.6246 | Fax: 408.229.7510
ClientServices@ariosadx.com
www.ariosadx.com

Ariosa Diagnostics, Inc. is a molecular diagnostics company committed to innovating together to improve patient care. Our flagship product, the Harmony™ Prenatal Test, is a safe, highly accurate and affordable prenatal test for maternal and fetal health. Led by an experienced team, we use our proprietary technology to perform a directed analysis of cell-free DNA in blood.

ARUP Laboratories **Booth # 331**

Phone: 801.583.2787
deanna.lemke@aruplab.com
www.aruplab.com

A nonprofit enterprise of the University of Utah, ARUP Laboratories is a leading national reference laboratory offering more than 3,000 tests and test combinations, ranging from routine screening tests to esoteric molecular and genetic assays.

Association of Public Health Laboratories **Booth # 234**

Phone: 240.485.2745 | Fax: 240.485.2700
info@aphl.org
www.aphl.org/aphlprograms

APHL represents state, county and city government laboratories that analyze disease agents and other health threats, delivering answers for effective public health response. These laboratories perform 97% of the newborn screening and genetics testing in the US.

Asuragen, Inc. **Booth # 425**

Phone: 512.681.5200 | Fax: 512.681.5201
www.asuragen.com

Asuragen offers innovative PCR-based approaches for fragile X testing, including AmpliDeX® PCR and mPCR assays to determine CGG size and methylation status, and Xpansion Interpreter®, to determine AGG interruption status and refine the risk of expansion from mother to child.

Basser Research Center for BRCA **Booth # 429**

Phone: 215.662.2748 | Fax: 215.349.5314
basserinfo@uphs.upenn.edu
www.basser.org

The Basser Research Center for BRCA of the University of Pennsylvania aims to deliver cutting edge research in basic and clinical sciences to advance the care of individuals who carry BRCA mutations.

Baylor College of Medicine Medical Genetics Laboratory **Booth # 403**

Phone: 713.798.6555 | Fax: 713.798.2787
GeneticTest@bcm.edu
www.bcmgeneticlabs.org

Baylor College of Medicine's Medical Genetics Laboratories offer a broad range of diagnostic genetic tests including DNA diagnostics, sequencing, cytogenetics, FISH diagnostics, cancer cytogenetics, chromosomal microarray analysis, whole exome sequencing, biochemical genetics, and Mitochondrial DNA analysis. Additionally we have a full range of testing for Autism Spectrum Disorders. Please visit our booth for more information.

BIOBASE **Booth # 303**

Phone: 978.922.1643
info@biobase-international.com
www.biobase-international.com

BIOBASE is a leading provider of manually-curated databases for molecular diagnostics that offer well-structured data, assembled by qualified experts and organized in an easily searchable manner that enables clinical interpretation of data arising from NGS efforts.

BioMarin Pharmaceutical Inc. **Booth # 339**

Phone: 415. 506.6700
IR@BMRN.com
www.BMRN.com

BioMarin develops and commercializes innovative biopharmaceuticals for serious diseases and medical conditions. Approved products include the first and only medications for PKU and LEMS, and the first and only enzyme replacement therapies for MPS I, MPS VI and Morquio A syndrome. Visit www.BMRN.com to learn more.

Boulder Abortion Clinic **Booth # 304**

Phone: 303.447.1361 | Fax: 303.447.0020
bachern@msn.com
www.drhern.com

At Boulder Abortion Clinic, Dr. Hern offers the safest possible abortion care and termination of pregnancies for fetal anomalies or other medical indications. These specialized services are provided in a confidential and comfortable outpatient setting.

Bright Pink **Booth # 521**

Phone: 312.787.4412
Fax: 312.787.4414
BrightPink@BeBrightPink.org
www.brightpink.org

Bright Pink is the only national non-profit organization focusing on the risk reduction and early detection of breast and ovarian cancer in young women while providing support for high-risk individuals.

Cancer Treatment Center of America **Booth # 128**

Phone: 623.207.3058
Bobby.Farrell@ctca-hope.com
www.azjobs.cancercenter.com

We deliver state-of-the-art, high quality care through an integrative model where a team of experts puts patients at the center of their own care. Every day, you will help patients win the fight against cancer.

Casey Molecular Diagnostics Laboratory **Booth # 437**

Phone: 503.494.5838
Fax: 503.494.6261
ceidiagnostics@ohsu.edu
www.ohsucasey.com/diagnostics

The Casey Molecular Diagnostics Laboratory is a CLIA certified laboratory located in Portland, Oregon providing comprehensive molecular testing for genetic diseases involving the eye.

Cedar River Clinics **Booth # 537**

Phone: 800.572.4223 | Fax: 425.255.0262
MonaW@CedarRiverClinics.org
www.CedarRiverClinics.org

Compassionate abortion care, to 26 weeks elective, with personal sedation plans. Our Fetal Indication Services include a dedicated clinical care advocate and private waiting rooms. We can assist clients with lodging, transportation, and funding resources.

Center for Jewish Genetics **Booth # 439**

Phone: 312.357.4718
jewishgeneticsctr@juf.org
www.jewishgenetics.org

The Center for Jewish Genetics is an educational resource for hereditary cancers and Jewish genetic disorders. Working closely with community members and support organizations, the Center aims to inform and empower individuals so they can plan for a healthy future.

City of Hope - Clinical Molecular Diagnostic Laboratory **Booth # 216**

Phone: 888.826.4362 | Fax: 626.301.8142
cmdl@coh.org
cmdl.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://cmdl.cityofhope.org>

Claritas Genomics **Booth # 212**

Phone: 617.553.5800 | Fax: 617.553.5842
clientservices@claritasgenomics.com
www.claritasgenomics.com

Claritas Genomics is a pediatric genetic testing company that combines the power of DNA sequencing technology with the clinical expertise of the world's best pediatric specialists to inform and improve patient care.

Clinical Genome Resource (ClinGen) **Booth # 329**

clingen@clinicalgenome.org
www.clinicalgenome.org

The Clinical Genome Resource (ClinGen) is an NIH-funded program dedicated to harnessing genetic and genomic data from research and clinical settings for the purpose of identifying clinically relevant variants.

CombiMatrix

Booth # 224

Phone: 949.753.0624 | Fax: 949.753.1504
info@combimatrix.com
www.combimatrix.com

CombiMatrix provides valuable molecular diagnostic solutions and comprehensive clinical support to promote the highest quality of care - specializing in cytogenomic miscarriage analysis, prenatal and pediatric healthcare.

Connective Tissue Gene Tests

Booth # 424

Phone: 484.244.2900 | Fax: 484.244.2904
inquiries@ctgt.net
www.ctgt.net

CTGT is committed to providing the broadest range of molecular diagnostics for inherited connective tissue disorders – over 300 tests and growing rapidly. CTGT has high test sensitivity, fast service, expert advice and superior customer service.

Cord Blood Registry

Booth # 325

Phone: 888.932.6568
providersupport@cordbloodregistry.com
www.cordblood.com

At Cord Blood Registry, our mission is to enable breakthrough medical treatments for more families by significantly advancing the real-life clinical applications of newborn stem cells. For more, visit cordblood.com or call 888-CORDBLOOD.

Counsyl Inc.

Booth # 434

Phone: 888.268.6795
support@counsyl.com
www.counsyl.com

Counsyl strives to give men and women access to vital information. We offer affordable high-quality screening, automated results delivery and genetic counseling so you can focus on patient care.

Courtagen Diagnostics Laboratory

Booth # 125

Phone: 877.395.7608 | Fax: 617.892.7192
genomics@courtagen.com
www.courtagen.com

Courtagen is a diagnostic sequencing and molecular information company that converts NGS sequencing data into actionable clinical information for neurological and metabolic disorders such as mitochondrial disease, epilepsy, and intellectual disability, including autism spectrum disorders.

Denver Genetic Laboratories at Children's Hospital Colorado

Booth # 106

Phone: 720.777.0500 | Fax: 720.777.7886
Elaine.Spector@childrenscolorado.org
www.denvergenetics.org

Denver Genetic Laboratories aims to provide Complete Genetic Solutions™ for genetic disorders and some cancers, to contribute to a better tomorrow for patients, families and healthcare providers.

Edimer Pharmaceuticals

Booth # 337

Phone: 617.758.4300 | Fax: 866.334.4240
info@edimerpharma.com
www.edimerpharma.com

Edimer Pharmaceuticals is a biopharmaceutical company dedicated to improving the lives of future generations living with XLHED. Edimer is developing EDI200, a potential treatment for future generations living with XLHED.

Emory Genetics Laboratory

Booth # 418

Phone: 404.778.8499 | Fax: 404.778.8559
egl.marketing@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.

FORCE: Facing Our Risk of Cancer Empowered

Booth # 426

Phone: 866.288.7475 | Fax: 954.827.2200
info@facingourrisk.org
www.facingourrisk.org

FORCE is a national nonprofit dedicated to fighting hereditary breast and ovarian cancer (HBOC). With over 50 outreach groups throughout the U.S., FORCE provides support, education, awareness, advocacy and research on behalf of anyone affected by HBOC.

Fulgent Diagnostics

Booth # 220

Phone: 626.350.0537 | Fax: 626.454.1667
info@fulgentdiagnostics.com
www.fulgentdiagnostics.com

Fulgent Diagnostic Lab, a cancer-focused CLIA lab, currently offers various clinical molecular genetic testing services. These tests are primarily used for cancer risk assessment, early detection of cancer regression, monitoring of disease progression, patient stratification, targeted therapy, treatment monitoring, and companion diagnostics.

Geisinger Health System

Booth # 534

Phone: 570.214.6117 | Fax: 570.271.6988
jhaid1@geisinger.edu
www.geisinger.org

Geisinger serves nearly 3 million people and is nationally recognized for research, innovative practices, quality and integrated healthcare delivery. We have an institutional commitment to research and a focus on personalized medicine.

Gene By Gene

Booth # 328

Phone: 713.474.2401 | Fax: 713.230.8999
sales@genebygene.com
www.genebygene.com

Gene By Gene is a leader in genetic testing. With a state-of-the-art laboratory and industry leading bioinformatics, we offer cutting-edge solutions in genetic testing across a variety of industry applications, including pre-conception carrier screening.

GeneDx

Booth # 219

Phone: 301.519.2100 | Fax: 301.519.2892
GeneDx@GeneDx.com
www.GeneDx.com

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

GeneTests

Booth # 121

Phone: 888.729.1204 | Fax: 201.212.6457
GeneTests@GeneTests.org
www.GeneTests.org

GeneTests is an online medical genetics information resource with capability to search by test, disorder, or gene. GeneTests searches retrieve links to GeneReviews™ chapters, other online resources and genetic testing information. Visit www.GeneTests.org.

Genetic Alliance

Booth # 203

Phone: 202.966.5557 | Fax: 202.966.8553
info@geneticalliance.org
www.genesinlife.org

Genetic Alliance, a nonprofit founded in 1986, engages individuals, families, and communities to transform health. Explore our extensive suite of technology platforms, educational resources, and advocacy tools and help educate and empower your patients.

Genome Magazine

Booth # 112

Phone: 972.905.2920
editor@genomemag.com
www.genomemag.com

Genome magazine explores the world of personalized medicine and the genomic revolution that makes it possible, empowering readers to make informed health decisions that will help them live better, longer.

Genomic Healthcare Innovations

Booth # 129

Phone: 414.955.2550
Fax: 414.955.6516
jnorthup@mcw.edu
www.genomichi.com

Genomic Healthcare Innovations is a genomic diagnostics company that uses results from clinical genome and exome sequencing to guide the care of patients through the CLIA-certified clinical sequencing laboratory at the Medical College of Wisconsin.

GenPath Women's Health

Booth # 228

Phone: 800.633.4522 | Fax: 201.791.3046
Info@GenPath.com
www.GenPath.com

GenPath Women's Health, a division of BioReference Laboratories, Inc., and a sister division of GeneDx, offers an extensive prenatal genetic portfolio from preconception to post-menopause, including prenatal/maternal risk assessment, carrier testing, prenatal diagnosis, pregnancy thrombophilia and infectious diseases which will improve the quality of care through better detection and diagnosis of gynecological and obstetric conditions.

Genzyme, a Sanofi Company

Booth # 535

Phone: 617.252.7500 | Fax: 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.

Greenwood Genetic Center

Booth # 201

Phone: 800.473.9411
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.

Heart Institute Diagnostic Laboratory

Booth # 110

Phone: 513.803.1751 | Fax: 513.803.1748
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 lab draws upon the expert analysis and consultation services of the Heart Institute's cardiologists, cardiac-focused geneticists and dedicated genetic counselors.

Human Genetics Laboratory UNMC Booth # 120

Phone: 402.559.5070 | Fax: 402.559.7248
humangenetics@unmc.edu
www.unmc.edu/geneticslab

Human Genetics Laboratory (UNMC) is a full-service clinical cytogenetic and molecular genetic laboratory specializing in both constitutional (prenatal and postnatal) and cancer diagnostics for over 40 years. Our comprehensive test menu includes conventional cytogenetics, FISH, microarray, next generation sequencing (autism/ID/MA, Rett/Angelman syndromes, cardiomyopathy, connective tissue disorders, craniosynostosis, Noonan syndrome/RASopathy disorders, and osteogenesis imperfecta), and intragenic deletion/duplication analysis.

Hyperion Therapeutics Booth # 113

Phone: 650.745.7802
www.hyperiontx.com

Hyperion Therapeutics, Inc. is a commercial-stage biopharmaceutical company committed to advancing science and developing treatments for orphan and hepatic diseases.

Illumina, Inc. Booth # 400

Phone: 858.202.4500 | Fax: 858.202.4766
info@verifitest.com
www.verifitest.com

Illumina is a leading developer, manufacturer, and marketer of life science tools and integrated systems for the analysis of genetic variation and function.

Improve Labs Booth # 117

Phone: 617.819.4727
info@improvelabs.com
www.improvelabs.com

Improve Labs offers a modular clinical data platform that seamlessly integrates with EMRs and streamlines existing workflows to give you more time with patients. A reliable, comprehensive, and intuitive interface unites pedigree drawing, questionnaires, risk assessment, requisitioning and clinical decision support. www.powerlineage.com.

Integrated Genetics Booth # 528

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 and the largest commercial genetic counseling network in the laboratory industry.

Invitae Corporation Booth # 419

Phone: 415.374.7782 | Fax: 415.520.9486
clinical@invitae.com
www.invitae.com

Invitae, a genetic information company, is aggregating the world's genetic tests into a single service with better quality, faster turnaround time and a lower price than most single-gene diagnostic tests today. Please visit www.invitae.com.

Kaiser Permanente - Northern California Booth # 519

www.genetics.kp.org

PRACTICE WHAT YOU BELIEVE, PRACTICE AT KAISER PERMANENTE! Kaiser Genetics is the employer of choice for over 70 genetic counselors in Northern California. Stop by our booth to learn about our unique brand of care and our rewarding positions!

Laboratory for Molecular Medicine, Partners Personalized Medicine Booth # 327

Phone: 617.768.8500
Fax: 617.768.8513
lmm@partners.org
www.partners.org/personalizedmedicine/lmm

The Laboratory for Molecular Medicine, a CLIA-certified molecular diagnostic laboratory within Partners HealthCare Personalized Medicine, translates genetic discoveries into clinical tests. Testing areas include disease-targeted panels, clinical genome and exome sequencing with interpretation services provided by experts.

Mauli Ola Foundation Booth # 539

Phone: 949.900.5583 | Fax: 949.900.5501
www.mauliola.org

The Maui Ola Foundation began as a group of surfers who banded together to introduce surfing as a natural treatment to people with cystic fibrosis. Since 2007, Maui Ola has taken nearly 1,300 CF patients surfing at nearly 100 Surf Experience Days and has now expanded it's reach with hospital visits and other activities that touch the lives of kids with cancer and a variety of other health challenges. In 2010, MOF was awarded The Agent of Change Award by SURFER Magazine for its positive contributions and example to the surfing community.

Mayo Medical Laboratories Booth # 235

Phone: 800.533.1710 | Fax: 507.284.0947
mml@mayo.edu
mayomedicallaboratories.com

Mayo Medical Laboratories provides comprehensive testing and unparalleled expertise in laboratory genetics. Over 35 board certified geneticists and genetic counselors at Mayo Clinic assist in appropriate test selection and interpretation of results.

MotherToBaby Pregnancy Studies conducted by the Organization of Teratology Information Specialists Booth # 127

Phone: 877.311.8972 | Fax: 858.246.1710
otisresearch@ucsd.edu
pregnancystudies.org

MotherToBaby, a non-profit service of the Organization of Teratology Information Specialists (OTIS), provides information about environmental exposures during pregnancy and lactation. MotherToBaby conducts research studies evaluating the safety of medications and vaccinations used during pregnancy.

Mount Sinai Genetic Testing Laboratory Booth # 335

Phone: 212.241.7518 | Fax: 212.241.0139
www.icsm.mssm.edu/geneticstesting

MGTL offers comprehensive molecular, cytogenetic and biochemical testing in our CLIA-certified, NY-state approved and CAP-accredited facility. Our laboratory directors, genetic counselors, account managers, and client service representatives provide superior service and state-of-the art testing.

Myriad Genetic Laboratories, Inc. Booth # 524

Phone: 800.469.7423
cscomments@myriad.com
www.myriad.com

Myriad Genetics is a leading molecular diagnostic company dedicated to making a difference in patients' 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, Inc. Booth # 209, 229

Phone: 650.249.9090
ccruz@natera.com
www.natera.com

Natera is a genetic testing company that specializes in analyzing microscopic quantities of DNA for reproductive health to help families conceive and deliver. Natera provides a host of preconception and prenatal genetic testing services, including the Panorama™ non-invasive prenatal test, genetic carrier screening, preimplantation genetic diagnosis (PGD), miscarriage testing and paternity testing.

National Society of Genetic Counselors (NSGC) Booth # 317

Phone: 312.321.6834 | Fax: 312.673.6972
nsgc@nsgc.org
www.nsgc.org

The National Society of Genetic Counselors 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, product information and more.

NextGxDx, Inc. Booth # 225

Phone: 615.861.2634 | Fax: 615.422.0857
info@nextgxdx.com
www.nextgxdx.com

NextGxDx is dedicated to improving the genetic test ordering process for the genetic counselor community. GeneSource, NextGxDx's genetic testing database, is the most comprehensive, current and easy-to-use tool available to search and compare genetic tests.

NSGC Cancer SIG Booth # 319

Phone: 312.321.6834
nsgc@nsgc.org
www.nsgc.org/CancerSIG/tabid/248/Default.aspx

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 developed by the SIG recently.

NSGC Prenatal SIG Booth # 108

Phone: 312.321.6834
nsgc@nsgc.org
www.nsgc.org/PrenatalSIG

The mission of the Prenatal Special Interest Group is to advocate for genetic counseling as an integral part of preconception and prenatal care by serving as an expert resource for NSGC and the board of directors, providing resources for prenatal genetic counselors, and promoting research and education within reproductive genetics.

Omicia Booth # 438

Phone: 510.595.0800 | Fax: 510.588.4523
info@omicia.com
www.omicia.com

Omicia is advancing genomics-based personalized medicine. We are developing a comprehensive platform of genome interpretation software, methodologies, and clinical services to find the medical significance of genetic variations found in individuals' DNA sequences.

Pathway Genomics Booth # 336

Phone: 877.505.7374 | Fax: 858.450.6604
clientservices@pathway.com
www.pathway.com

Pathway Genomics is a clinical diagnostics company that specializes in hereditary cancers, pharmacogenomics and lifestyle genomics. We accept both saliva and blood samples. All our tests have a TAT of 3 weeks or less.

PerkinElmer Labs

Booth # 324

Phone: 800.762.4000
www.perkinelmer.com

PerkinElmer Labs, along with our partner lab Good Start Genetics, offers a variety of prenatal and carrier screening test options to suit every patient case. We provide one source to support your genetic testing needs.

Personalis, Inc.

Booth # 205

Phone: 650.752.1300 | Fax: 650.752.1301
info@personalis.com
www.personalis.com

Personalis® is a clinical testing laboratory that gives clinicians the most accurate and comprehensive sequencing and interpretation solutions available. Designed to enhance diagnostic yield, the ACE Clinical Exome™ Test combines gene finishing and structural variant analysis to find answers for your patients.

Pfizer, Inc.

Booth # 116

Phone: 212.733.2323
www.pfizer.com

At Pfizer, we apply science and our global resources to bring therapies to people that extend and significantly improve their lives.

PreventionGenetics

Booth # 208

Phone: 715.387.0484 | Fax: 715.207.6602
clinicaldnatesting@preventiongenetics.com
www.preventiongenetics.com

PreventionGenetics is a leader in providing comprehensive clinical DNA testing offering NextGen Sequencing, Sanger sequencing and deletion/duplication testing via array CGH for over 1000 genes.

Proband by The Children's Hospital of Philadelphia

Booth # 226

Phone: 267.426.7522 | Fax: 215.590.5245
millerjm1@email.chop.edu
www.probandapp.com

Proband is an iPad application designed to replace paper for drawing pedigrees during family history interviews. Built from the ground up for the iPad, Proband uses intuitive gestures to make creating pedigrees fast and efficient.

Progeny Software, LLC

Booth # 435

Phone: 800.776.4369 | Fax: 888.584.1210
info@progenygenetics.com
www.progenygenetics.com

Progeny offers family history and genetic pedigree software for family-based clinical data management. Capture family history electronically before the clinic visit and identify at-risk patients via our customizable online questionnaire.

Quest Diagnostics

Booth # 300

Phone: 866.697.8378
www.questdiagnostics.com

Quest Diagnostics, the world's leading provider of diagnostic testing, information and services, offers a comprehensive Genetics Testing menu including Prenatal and Neonatal, Oncology, Neurology, and Endocrinology. We empower health with diagnostic insights. Visit QuestDiagnostics.com/Genetics.

Recombine

Booth # 218

Phone: 855.687.4363 | Fax: 212.214.0377
info@recombine.com
www.recombine.com

Recombine was founded by experts in fertility and reproductive genetics. We offer CarrierMap, a Comprehensive Carrier Screen for > 200 genetic conditions, and FertilityMap, a Complete Fertility Genetic Assessment for infertility. From sample collection to genetic counseling, we manage the entire genetic testing process. It is Genetic Testing. Simplified.

Recordati Rare Diseases

Booth # 103

Phone: 908.236.0888
dutch.c@recordati.com
www.recordatirarediseases.com

Recordati Rare Diseases (RRD) Inc is a member of the Recordati Group, which consists of Recordati S.p.A and Orphan Europe. RRD's mission is to partner with patients, healthcare providers, advocacy and industry to make products available to treat rare and severe diseases.

Reproductive Genetic Innovations (RGI)

Booth # 227

Phone: 847.400.1515 | Fax: 847.400.1516
info@rgipgd.com
www.rgipgd.com

Reproductive Genetic Innovations (RGI) is a world-renowned provider of Preimplantation Genetic Diagnosis (PGD). With experience spanning three decades, RGI is a leader in PGD technology and offers testing for nearly any single gene disorder, as well as for chromosomal rearrangements and aneuploidy by PCR, FISH, and 24-chromosome aCGH.

Reprogenetics

Booth # 431

Phone: 973.436.5003 | Fax: 973.710.4238
www.reprogenetics.com

Reprogenetics is a full-service preimplantation genetic diagnosis (PGD) laboratory offering 24 chromosome aCGH for aneuploidy, translocations and inversions, PGD for single gene disorders and HLA matching via standard methods or karyomapping, aCGH for POC testing and FISH on sperm.

Seattle Children's Hospital

Booth # 202

Phone: 206.987.3361 | Fax: 206.987.3840
plugs@seattlechildrens.org
www.seattlechildrenslab.org

PLUGS is a Utilization Management service that helps hospital laboratories and practitioners decrease costs and errors associated with unnecessary laboratory testing, which will ultimately save you money! Visit our booth at NSGC.

Sequenom Laboratories

Booth # 513

Phone: 877.821.7266 | Fax: 858.202.9205
info@sequenom.com
www.laboratories.sequenom.com

Sequenom Laboratories, a molecular diagnostics laboratory dedicated to improving patient care, commercialized the first noninvasive prenatal test for pregnant woman at increased risk. Through a routine blood draw, MaterniT21™ PLUS laboratory-developed test analyzes and reports clinically relevant fetal chromosomal abnormalities—including the core trisomies 21, 18, and 13, as well as trisomies 16 and 22; fetal sex aneuploidies; and select microdeletions.

Sharsheret

Booth # 122

Phone: 866.474.2774
Fax: 201.837.5025
info@sharsheret.org
www.sharsheret.org

Sharsheret is a national not-for-profit organization supporting young Jewish women and families facing breast cancer and ovarian cancer – those who are at high risk, those who are diagnosed, and survivors.

Sinai Surgical Center

Booth # 428

Phone: 310.247.0553
Fax: 310.289.1694
SinaiSurgical@aol.com
www.ProChoiceMedical.com

Second and third trimester abortions for fetal indications. Accredited surgical center; hospital affiliated. Josepha Seletz MD, board certified Ob Gyn. Rob MacLennan, CRNA. Los Angeles, California.

Southwestern Women's Options

Booth # 517

Phone: 505.242.7512
Fax: 505.242.0540
boyd02@covad.net
www.southwesternwomens.com

Curtis Boyd, MD owned clinics provide a full range of abortion services. The Albuquerque office specializes in third trimester abortion care offering a unique Fetal Indications Program geared to the special needs of the patient.

St. Louis Fetal Care Institute

Booth # 237

Phone: 314.268.4037
Fax: 314.678.4499
katie_francis@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.

Transgenomic, Inc.

Booth # 334

Phone: 877.274.9432 | Fax: 203.907.2615
info@transgenomic.com
labs.transgenomic.com

Transgenomic provides proprietary technology and molecular genetics expertise for fully integrated molecular diagnostic solution through our three integrated divisions — Biomarker Identification, Genetic Assays and Platforms, and Patient Testing.

UAB Medical Genomics Laboratory

Booth # 427

Phone: 205.934.5562
Fax: 205.996.2929
medgenomics@uabmc.edu
www.genetics.uab.edu/medgenomics

The Medical Genomics Laboratory (MGL) is a CAP-certified not-for-profit clinical laboratory at the University of Alabama at Birmingham, offering comprehensive testing for common and rare genetic disorders. The MGL specializes in testing for all forms of the neurofibromatosis, including NF1, Legius syndrome, NF2, segmental NF and schwannomatosis.

UCLA Clinical Genomics Center Booth # 536

Phone: 310.825.7099
Fax: 310.267.2685
scwebb@mednet.ucla.edu
www.pathology.ucla.edu/genomics

The UCLA Clinical Genomics Center offers testing for hereditary disorders and cancer diagnosis/management, and genetic counseling. Our own CLIA-certified CAP-accredited labs provide exome sequencing with expert interpretation by our Genomic Data Board, custom Sanger sequencing, chromosomal microarray and more.

University of Chicago Genetic Services Booth # 430

Phone: 773.834.0555
Fax: 773.702.9130
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 neurodevelopmental disorders. Some of our services include genetic testing for brain malformation syndromes, microcephaly, epilepsy, ataxia, congenital muscle diseases and Cornelia de Lange syndrome.

University of Washington Reference Lab Services Booth # 200

Phone: 1.800.713.5198
commserve@uw.edu
<http://depts.washington.edu/labweb/Divisions/MolDiag/MolDiagGen/index.htm>

BROCA Testing is performed with next-generation sequencing technology, which detects all classes of disease-causing mutations. The Department's clinical test is based on the research of Drs. Tom Walsh and Mary-Claire King, who first developed this technology to study hereditary risk of breast and ovarian cancer and reported it in 2010. UW-OncoPlex™ is a multiplexed gene sequencing panel that detects mutations in tumor tissue in 194 cancer-related genes for cancer treatment, prognosis, and diagnosis.

Vendor-Sponsored Presentations

Thursday, September 18, 2014

11:30am - 12:00pm

Personalis, Inc.

The ACE Clinical Exome™ Test: An Advanced Diagnostic Test for Genetic Disease
Sarah Garcia, Ph.D., M.S., C.G.C.

In this workshop, we will discuss the highly accurate exome sequencing and interpretation that underlie Personalis' diagnostic services for Mendelian disorders. Topics include, our approach to finishing the medical exome using ACE Technology, improving the sensitivity and specificity of structural variant calling, correcting the human reference sequence to enable better alignment and variant calling, and our biomedically-driven approach to variant filtration. Clinical examples demonstrating the impact of Personalis' accuracy improvements on the sensitivity to detect causative variation in Mendelian disorders will be presented.

12:15pm - 12:45pm

Myriad Genetic Laboratories

myRisk™ Hereditary Cancer Panel: Clinical Data Supporting a Pan-Cancer Panel Approach to Hereditary Cancer Testing

Jennifer Saam, PhD, MS, CGC, Job Title TBD, Company TBD

Myriad's myRisk Hereditary Cancer panel evaluates 25 cancer susceptibility genes associated with 8 cancers. This product theater will summarize recent data which demonstrate the clinical overlap of hereditary cancer genes and supports the utilization of a pan-cancer panel over targeted or disease-specific panels to identify clinically actionable mutations.

1:15pm - 1:45pm

Edimer Pharmaceuticals

EDI200: Potential Treatment for X-linked hypohidrotic ectodermal dysplasia (XLHED)

Neil Kirby, Ph.D., President and CEO of Edimer Pharmaceuticals

X-linked hypohidrotic ectodermal dysplasia (XLHED) is a disorder of ectodermal development causing hypohidrosis, hypodontia, and hypotrichosis. EDI200, a novel protein therapeutic under clinical investigation, initiates the development of the ectoderm potentially providing lifelong benefit with a single course of therapy—an approach that may have clinical implications for other disorders.

Friday, September 19, 2014

12:45pm - 1:15pm

Ambry Genetics

The Panel Results Came Back. Now What Do I Do? Cancer Panel Case Presentations with Your Peers.

Laura Panos, MS, CGC, Product Manager, Company TBD

Many questions exist about counseling following next-gen cancer panel results, particularly when unexpected mutations do not fit the patient's phenotype and with mutations in less well-described genes. In this session, Ambry genetic counselors will interview clinicians regarding such cases from their practice and the post-test counseling and recommendations given.

1:45pm - 2:15pm

BioMarin Pharmaceutical Inc.

Diagnostic Challenges in the Lab and in the Clinic: Morquio A and MPS VI

Laura Pollard, PhD, FACMG, Assistant Director, Biochemical Genetics Laboratory, Greenwood Genetic Center

Brooke Smith, MS, CGC, Certified Genetic Counselor, Greenwood Genetic Center

Rare genetic diseases such as Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome) and IVA (Morquio A Syndrome) are under recognized and challenging to diagnose. This presentation will include an overview of diagnostic strategies and recommendations and a clinical case study as an example of the challenges in diagnosis.

[illegible]

Supporting the National Society of Genetic Counselors

Your Clinical Practice
Advanced



Leaders in Genetic Testing

- Cardiology
- Neurology
- Whole Exome Sequencing



Please visit Booth #334 to learn more about our services.

- Comprehensive analysis for accurate diagnosis
- Patient advocacy for limiting out-of-pocket costs
- Consultation with genetic experts
- Patient education services



For more information call: **877.274.9432**
Visit our website: **labs.transgenomic.com**