

TEXAS SOCIETY OF GENETIC COUNSELORS PRESENTS THE

ANNUAL EDUCATION CONFERENCE

FEBRUARY 25, 2022 8:00 AM - 5:00 PM

PROGRAM BOOK AND RESOURCE GUIDE



	AGENDA
	For live virtual conference on February 25th
	ive presentations will <u>NOT</u> be available post-conference* Welcome Remarks
0.00 AM	Kelly Moyer, MS, CGC
	Olivia Trimmier, MS, CGC
8:15 AM	Overview, Diagnosis, and Treatment of Hereditary
	Pheochromocytoma and Paraganglioma
	Syndromes
	Patricia Dahia, MD, PhD
9:15 AM	Mio Kitano, MD, MS, FACS, FSSO Affirming and Respectful Treatment of
	Transgender Patients in the Prenatal and Cancer
	Clinic
	Rosalba Sacca, PhD, MS, CGC
	Andy Cantor, MS, LCGC
10:15 AM	Break
10:30 AM	Keynote Lecture: New Treatments for Genetic
	Conditions
	Joseph Ray, MD
11:30 AM	Family Panel: Families Receiving Treatment for
	Genetic Conditions - CF, SMA, Achondroplasia
	(Pre-recorded with live Q&A)
	Moderators: Kelly Moyer, MS, CGC and Olivia Trimmier, MS, CGC Speakers: Julia P, Makenzie R, Amer H and Munira S
1:30 PM	Lunch
1:55 PM	Grant Winners
2:05 PM	TSGC DEI Initiative
2:15 PM	Fetal surgery: Now and the Future
	Kenneth J. Moise Jr., MD
3:15 PM	What Matters in the End: Empathy and Ethics in
	End-of-Life Care
	Nadia Tremonti, MD
4:15 PM	Closing Remarks
4:30 PM	TSGC Business Meeting
5:00 PM	Adjourn
	`//////////////////////////////////////

ON DEMAND CONTENT

The links to the following pre-recorded presentations are accessible via <u>www.tsgc.org/virtual-aec.</u>

To receive CEUs, evaluations must be completed by **Friday**, *March* **25th (one month post live conference)**.

Our speakers did a phenomenal job with this format - we hope you enjoy the content!

What We Are Learning from Cystic Fibrosis Modulators Karen Raraigh, MGC, CGC

Rapid Clinical Genomics: Testing Options for the Critically-Ill Patient *Sponsored by GeneDx*

Jane Juusola, PhD, FACMG

Molecular Diagnosis from Genetic Testing Guides Clinical Management of Epilepsy and Helps Improve Patient Outcomes

Sponsored by Invitae Dianalee McKnight, PhD, FACMG

Genetics of Chronic Kidney Disease: Advancing

Treatment and Outcomes

Sponsored by Natera Trudy McKanna, MS, CGC

Insights from the Lab: Tips for Clinical Genetic Counselors

Sponsored by Sema4 Rachel Doyel, MS, CGC

WE ARE EXTREMELY THANKFUL TO OUR GENEROUS SPONSORS FOR SUPPORTING THIS CONFERENCE AND THE TSGC!

<u>Genome Level</u>





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l/yriad genetics

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Gene

Exome Level









Panel Level





SNP Level







A FEW HOUSEKEEPING ITEMS

How to obtain CEUs

- Listen for and write down the attendance codes (the next page will help you keep track), feel free to print it!
- Go to <u>www.tsgc.org/virtual-aec</u> to complete your CEU evaluations (codes required) before Friday, March 25th.
- Don't forget to complete your CEU evaluations for the prerecorded content.
- Please don't ask for codes in the chat. We will announce codes at the end of each speaker.



Block your work calendar



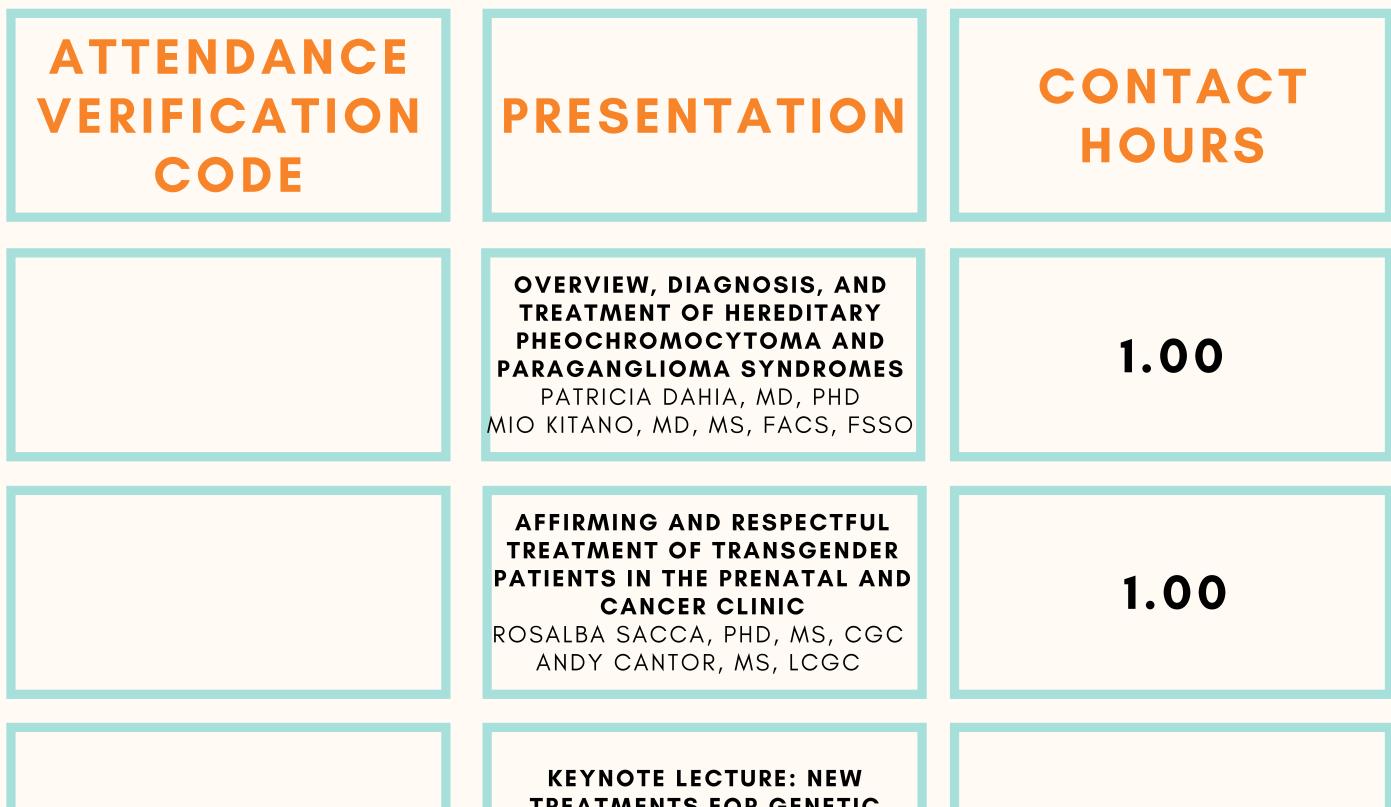
- Put your phone on silent (or out of reach!)
- Close your email and any apps with notifications
- Set your workspace up for success!

Engage!

- Use the Zoom reactions
- Contribute to the virtual chatter via the chat button
- Ask questions of our presenters using the Q&A feature!



LIVE CONFERENCE CEU TRACKING SHEET



TREATMENTS FOR GENETIC CONDITIONS JOSEPH RAY, MD	1.00
FAMILY PANEL: FAMILIES RECEIVING TREATMENT OF GENETIC CONDITIONS - CF, SMA, & ACHONDROPLASIA	1.50
FETAL SURGERY: NOW AND THE FUTURE KENNETH J. MOISE JR., MD	1.00
WHAT MATTERS IN THE END: EMPATHY AND ETHICS IN END-OF-LIFE CARE NADIA TREMONTI, MD	1.00

YOUR PATIENTS WITH ACHONDROPLASIA MAY WANT YOU TO KNOW THIS

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SPEAKER BIOS Patricia Dahia, MD, PhD



Dr. Dahia is currently a tenured Professor of Medicine at the University of Texas Health Science Center at San Antonio. Her career has been dedicated to research on the genetics of cancer, with emphasis on inherited endocrine tumors and discovery of cancer susceptibility genes (including PTEN, SDHB, KIF1B, EPAS1/HIF2A, TMEM127, etc). For the past two years, she has served as the medical director of the UTHSCSA Clinical Translation Science Award (CTSA) activities to promote innovative clinical trial operations at UTHSCSA, and across CTSAs.

Mio Kitano, MD, MS, FACS, FSSO

Dr. Kitano is an Associate Professor in the Division of Surgical Oncology & Endocrine Surgery in the Mays Cancer Center at UT Health San Antonio. She is board-certified in both General Surgery and Complex General Surgical Oncology. Dr. Kitano's clinical interests are focused on management of upper GI malignancies, including gastric and pancreas cancer, and neuroendocrine tumors of the gastroenteropancreatic tract as well as surgical treatment of benign and malignant endocrine disorder.



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- Lincoln S. et al. Genetics in Medicine 23, 1673-1680 (2021)
- As of November 2021. View ClinVar contributors at: http:// www.ncbi.nlm.nih.gov/clinvar/docs/submitter_list
- Cummings et al. Sci Transl Med. 9:1-11, 2017.
- Abramowicz & Gos. J Appl Genet. 59:253-268, 2018.

Truty et al. Am J Hum Genet. Accepted.

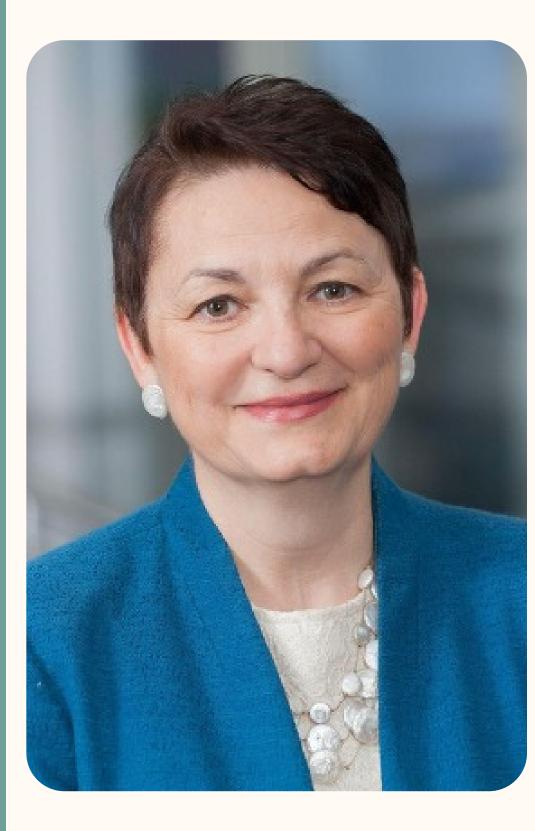
Lee et al. Genet Med. 22:490-499, 2020.

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- Nykamp et al. Genet Med 19, 1105-1117 (2017).
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10.Sim NL et al. Nucleic Acids Res. 2012;40:W452-W457. 11. Adzhubei IA et al. Nat Methods. 2010;7(4):248-249 12.Loannidis NM et al. Am J Hum Genet. 2016;99[4]:877-885 13.Invitae data on file

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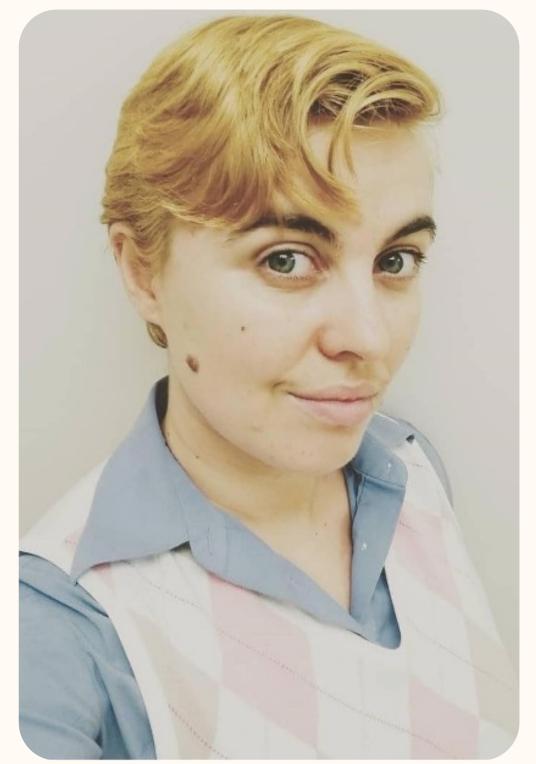
SPEAKER BIOS Rosalba Sacca, PhD, MS, CGC



Dr. Sacca is a Senior Genetic Counselor in the Clinical Genetics Service at Memorial Sloan Kettering Cancer Center and also worked in the Cancer Genetics and Prevention program at the Dana-Farber Cancer Institute. She is currently a member of the NSGC Cancer SIG Gender-Inclusive Subcommittee and a board member of the State of Connecticut Regenerative Medicine Research Advisory Committee.

Andy Cantor, MS, LCGC

Andy is a prenatal genetic counselor at Integrated genetics. They are a non-binary person and a certified trainer on gender inclusive care who co-presented the wellreceived educational breakout session "Gender-Inclusive Genetic Counseling: What Have We Learned and How Do We Incorporate it into Our Practice?" at the NSGC's Virtual Annual Education Conference in 2021.



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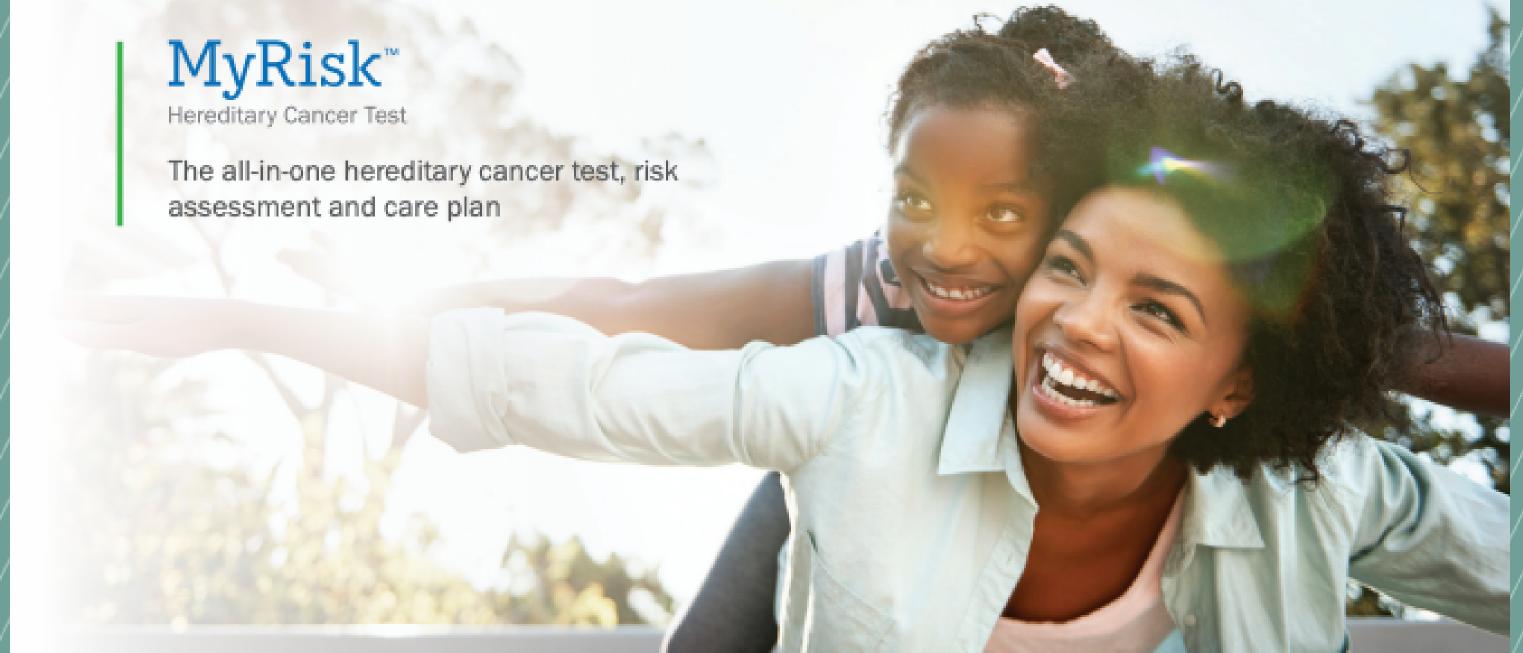
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1. Hancock et al. (2019) Clinical experience across the fetal-fraction spectrum for a noninvasive prenatal screen with low test-failure rate. UOG. doi/pdf/10.1002/uog.21904. 2. Hogan et al. Validation of an Expanded Carrier Screen that Optimizes Sensitivity via Full-Exon Sequencing and Panel-wide Copy Number Variant Identification. Clinical Chemistry 2018; doi:10.1373/clinchem.2018.286823

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SPEAKER BIOS

Joseph Ray, MD

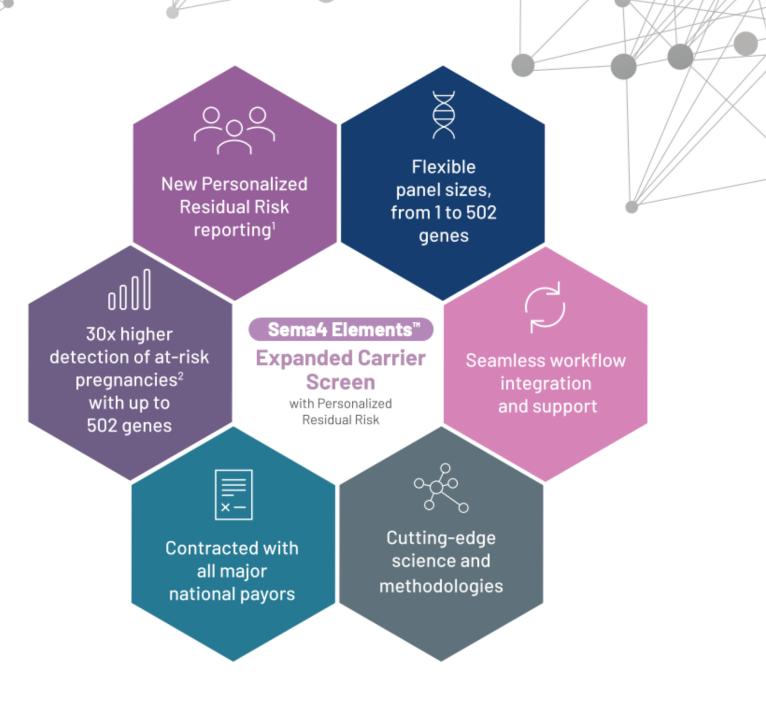


Dr. Ray specializes in medical genetics and sees adult and pediatric patients with genetic conditions through the University of Texas Medical Branch. He is known as an excellent physician and teacher, and has a special interest in treating genetic conditions.

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 Expanded Carrier Screening (502 genes) with Personalized Residual Risk [White Paper]. Sema4 (2021).
 With 502-gene panel compared to traditional carrier screening of CF and SMA.

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FAMILY PANEL BIOS

Julia P



Julia's son Coleman (4yo) is about to enter part B of a Phase 3 trial for Trikafta (granules in food twice per day, Vertex) in patients from 2-6 yo for the treatment of Cystic Fibrosis. Julia has been a board member of the South Carolina chapter of the Cystic Fibrosis Foundation for the past three years, and their fundraising team, "Coleman for the Cure", has raised \$150,000 for the Cystic Fibrosis Foundation since 2018. She wants attendees to learn from her family about the successes and challenges which can accompany new treatments for young

children.

Amer H and Munira S

Amer and Munira's son Ahmin (13yo) is approaching four years taking Voxzogo for Achondroplasia as part of a clinical trial through Biomarin. This San Francisco Bay area family was instrumental in creating support for this and other medications for this condition through their nonprofit organization Growing Stronger Research Fund http://www.growingstronger.org/aboutgrowing-stronger.html. As a couple they have been very proactive in providing their son and others with Achondroplasia treatment options which are targeted to reduce surgical and painrelated complications of the disorder. They would like our attendees to know some of the triumphs and challenges that families face with a genetic diagnosis and a new treatment.







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FAMILY PANEL BIOS

Makenzie R

Makenzie's son Blaise (2 years) is participating in a clinical trial for Zolgensma (One time infusion, Novartis gene therapies) for Spinal Muscular Atrophy type I. He also takes Evrysdi (Oral daily med, Genentech). She wishes to advocate for the importance of early diagnosis and treatment for these genetic diseases, and how this can change the quality of life.





FAMILY PANEL RESOURCES

CF

Coleman for the Cure

<u>https://fightcf.cff.org/site/TR/GreatStrides/116</u>
 <u>South Carolina Charleston?</u>
 <u>team id=104234&pg=team&fr id=9166</u>

SMA

Cure SMA

<u>www.curesma.org</u>

• Fighting for Kaiden

- Raises money for equipment for SMA families
- <u>www.fightingforkaiden.org</u>

ACHONDROPLASIA

- Growing Stronger Foundation
 - Raises money and provides information regarding treatment for Achondroplasia

o <u>www.growingstronger.org</u>

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GRANT WINNER: KATIE HUANG



Barriers Experienced by Underrepresented Minorities in Becoming A Competitive Genetic Counseling Applicant

Katie Huang, MS, CGC. 1,4 Advisory Professor: Claire N. Singletary, MS, CGC. 1,2,3

 1 - The University of Texas MD Anderson Cancer Center UTHealth Graduate School of Biomedical Sciences, Houston, TX
 2 - Department of Obstetrics, Gynecology and Reproductive Sciences, McGovern Medical School at the University of Texas Health Science Center at Houston, TX

3 – Department of Pediatrics, UTHealth, Houston, TX

4 - Clinical Cancer Genetics, The University of Texas MD Anderson Cancer Center, Houston, TX

Introduction: Representation for both racial/ethnic and gender identity minorities in genetic counseling (GC) remains the lowest among similar healthcare professions. Barriers that underrepresented minority (URM) individuals face in becoming a competitive GC applicant have not yet been described. Academic capital (AC) is a theoretical framework describing the social processes necessary for individuals to navigate and succeed in higher education.

Aims: This study aimed to characterize barriers experienced by applicants who self-identify as underrepresented and to explore how AC could identify areas for intervention.

Methods: Prospective GC applicants for the 2021/2022 cycles were recruited to complete an anonymous online survey in Qualtrics (HSC-MS-20-0653) through a multi-pronged snowball recruitment method across social media, email, and websites. In total, 156 eligible participants completed the survey which included background information, application process experiences, and validated Academic Capital Scale (ACS). Responses were analyzed through STATA v14.0 for descriptive statistics and ATLAS.ti for open-ended responses.

Results: Top identity aspects in which participants felt underrepresented were race/ethnicity (49%), first-generation college student (37%), socioeconomic status (31%), multicultural background (26%), sexual orientation/LGBTQ+ (26%), and English as a second language (21%). On the ACS, there was a significant difference between participants who identified with one underrepresented group compared to more than one group (p = 0.018) suggesting intersectionality should be studied. Areas for focus based on ACS were concerns about costs, navigation of systems, and college knowledge. Shadowing a genetic counselor, costs, and personal statements were rated as the most challenging application components. The top three factors impacting where to apply were rotations, financial assistance, and cost. The National Institute of Health definition of disadvantaged emerged as a possible proxy to use in holistic admissions.

Conclusions: These findings suggest that interventions at the institutional or social network level, such as partnerships with URM institutions or centralized mentorship/shadowing programs, would improve GC-specific AC formation. Financial assistance from training programs or state/national organizations could reduce cost barriers. Overall, creating a successful URM GC pipeline requires interventions targeted to URMs and modifying the definition of a competitive applicant through holistic admissions.

GRANT WINNER: CHELSEA WAGNER



Investigating the Efficacy of an Electronic Pre-Test Genetic Counseling Tool for Routine Prenatal Screening

Chelsea Wagner, MS, CGC 1; Blair Stevens, MS, CGC 1; Jacqueline Harkenrider, MS, CGC1; Samantha Montgomery, MS, CGC 1; Aarti Ramdaney, MS, CGC 1; Shannon Mulligan, MS, CGC 1

1 Department of Obstetrics, Gynecology, and Reproductive Sciences, McGovern Medical School at UTHealth, Houston, TX, USA.

Abstract: All pregnant persons should be offered screening for genetic conditions including aneuploidy and autosomal recessive conditions. Patient decision making should include counseling that enables informed consent and includes the option to decline all testing. Time constraints may limit an OB/GYN's ability to provide such comprehensive counseling, while work force and scope of practice concerns limit the ability for genetic counselors to perform pre-test counseling for all pregnant persons or persons considering pregnancy. For these reasons, patients and providers need an unbiased interactive tool to facilitate informed patient decision making that can identify individuals who would benefit most from traditional in-person genetic counseling in lieu of traditional counseling methods.

Aims: 1. Develop an interactive web-based tool to facilitate informed patient decision making for routine aneuploidy and carrier screening decisions. 2. Assess the knowledge, attitude, and deliberation patterns of participants who use an electronic educational module to facilitate informed decision making regarding routine prenatal genetic testing using a validated Multi-Dimensional Measure of Informed Choice (MMIC) tool. 3.Compare informed decision making metrics to patients who undergo in person genetic counseling with a genetic counselor.

Methods: Aims will be achieved through a cross sectional descriptive study evaluating the two service delivery models (in person genetic counseling and the interactive online tool). A diverse set of participants will be eligible if they are over the age of 18, Englishspeaking, and have obstetric care at one of our designated UTPhysicians clinics. Participants will be offered a voluntary survey following their genetic counseling appointment or completion of the online module. At the conclusion of the survey, participants will have the opportunity to share their email address to receive compensation for their participation in the form of an \$20 electronic gift card, which was made possible through the Texas Society of Genetic Counselors Grant Award. Data collected from the survey will be evaluated for all appropriate variables using the statistical analysis program, STATA. Tests will be considered to be statistically significant at p < 0.05. Descriptive statistics will be used to characterize the demographics, satisfaction, and elements of the MMIC tool. Comparisons will be made between individuals who used the electronic module and those who received traditional in-person genetic counseling. Inferential statistics will be used to describe differences in uptake, knowledge, attitude, and deliberation regarding informed decision making for prenatal genetic screening options between service delivery models.

Society of Genetic Counselors

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References

 Rafalko J, Caldwell S, Soster E et al. Application of mosaicism ratio to multifetal gestations. *PLoS One*. 2021;16(3):e0248467. doi:10.1371/journal.pone.0248467.
 Internal data
 Palomaki GE, Deciu C, Lambert-Messerlian GM, et al. DNA sequencing of maternal plasma to detect Down syndrome: An international clinical validation study. *Genet Med*. 2011;13(11):913-920.

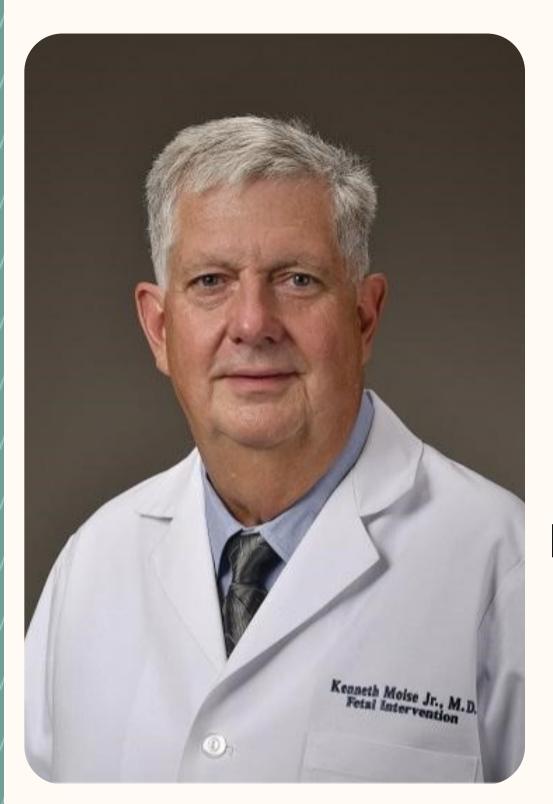


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SPEAKER BIOS Kenneth J. Moise Jr., MD



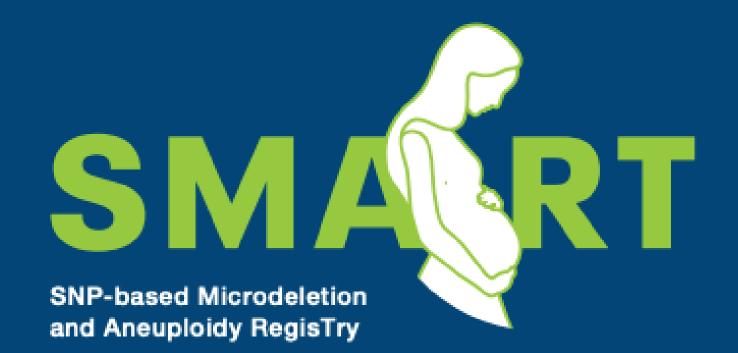
Dr. Moise, director of the new Comprehensive Fetal Care Center at Dell Children's Hospital in Austin, is the former President of the International Fetal Medicine and Surgery Society. He is also a founding member of the North American Fetal Treatment Network (NAFTNet). He currently serves as the principal investigator of an international clinical trial sponsored by Jansen Pharmaceutical, Inc. to study the use of a novel monoclonal antibody to negate the need for intrauterine transfusions in

the treatment of severe HDFN.

Nadia Tremonti, MD

Dr. Tremonti is double-board certified in pediatrics and hospice and palliative medicine. She works to ensure that terminally ill children receive quality end of life care. With limited financial and staffing resources, she began the palliative care program at Children's Hospital of Michigan in 2007, and was honored with the 2014 Hastings Center Cunniff-Dixon Physician Awards in the early career category. Since then, the program has grown to serve more than 500 families.





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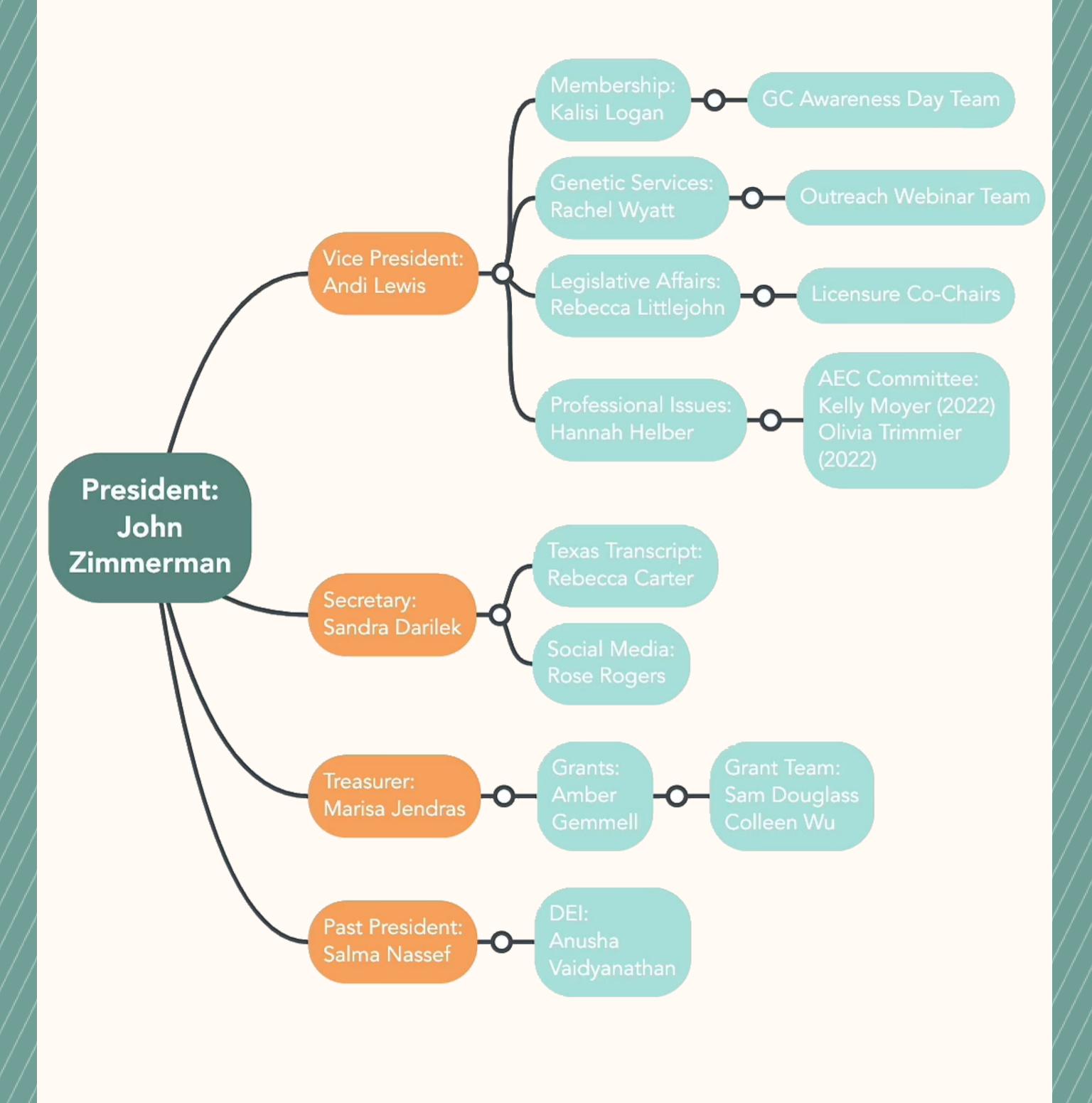
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CHROMOSOME-LEVEL DETECTION in cfDNA



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INTERESTED IN A TSGC LEADERSHIP POSITION?

The following Committee Chair positions are open for self-nomination for the 2022-2023 term. Please contact the TSGC President (John Zimmerman; John.Zimmerman@UTSouthwestern.edu) if you are interested in one of these positions:

Licensure Committee Co-Chairs (2): The Licensure Committee leads genetic counseling state licensure efforts under the direction of the Legislative Affairs Chair. This includes drafting a state licensure bill; facilitating communication between TSGC, the Texas Medical Association (TMA) and other stakeholders; and coordinating efforts of our membership surrounding ongoing licensure activities.

Diversity, Equity and Inclusion (DEI) Committee Co-Chair (1): The DEI Committee is tasked with developing outcomes-based initiatives aimed promoting diversity, equity, and inclusion within the Texas genetic counseling community.

Additional volunteer opportunities:

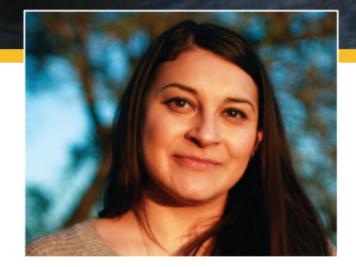
GC Awareness Day Team: Contact Membership Committee Chair, Kalisi Logan; Kalisi.Logan@austin.utexas.edu Grants Committee: Contact Grants Committee Chair, Amber Gemmell; Amber.Gemmell@UTSouthwestern.edu Nominating Committee: Contact Vice President, Andi Lewis; andi.lewis@invitae.com Outreach Webinar Team: Contact Genetic Services Committee Chair, Rachel Wyatt; wyattr1@uthscsa.edu Texas Transcript Team: Contact Editor-in-Chief, Rebecca Carter; rebecca.d.s.carter@gmail.com

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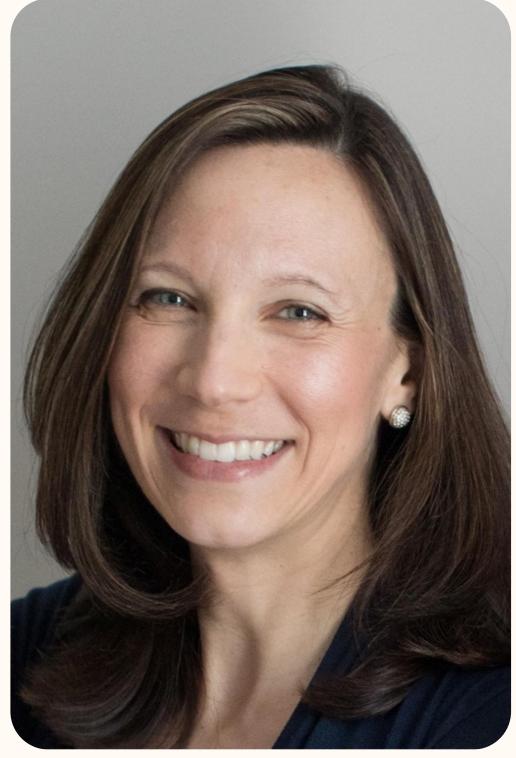






Karen Raraigh, MGC, CGC

Karen is an experienced pediatric genetic counselor and researcher with a special interest in genotype/phenotype correlation in Cystic Fibrosis. She has served as an integral team member of the CFTR2 website which uses information from the 88,000 patients in their database to provide clinical information about specific CFTR variants and genotypes (variant combinations) to researchers, patients with cystic fibrosis, and their families. She has previously given many talks to local and national GC audience.

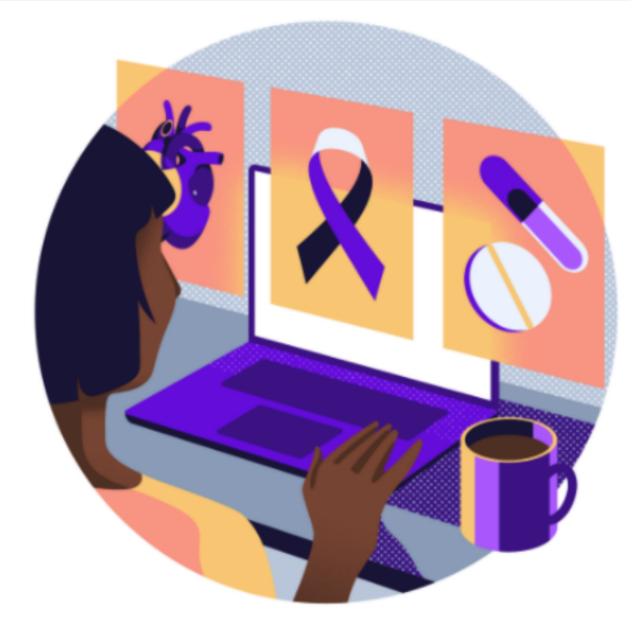


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BOTH PARENTS REQUIRED	\$2,590	N/A
FAMILY - DUO		
Patient + one additional family member	\$3,890	\$4,790
FAMILY - TRIO		
Patient + two additional family members	\$4,990	\$5,990
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Sequencing of additional family member beyond trio - \$1,390

For Family - Duo and Trio, a report is issued for the patient only

Report for additional family member - \$490

PGxome Whole Exome Sequencing DIAGNOSTIC

Patient Only	PGxome	Rapid PGxome	Prenatal PGxome	
Patient Only Patient (Proband)	\$1,890	\$2,290	\$2,860	
Patient PLUS BOTH PARENTS REQUIRED Patient + Targeted, Variant Testing of Parents	\$1,990	N/A	N/A	
Family - Duo Patient + one additional family member	\$2,290	\$2,690	\$3,360	
Family - Trio Patient + two additional family members	\$2,490	\$2,890	\$3,590	

When is the last time you went shopping FOR AN EXOME OR A GENOME?

If it's been awhile, what you find may surprise you!

			LAB NAME		LAB NAME		LAB NAME	
TEST	PRICE	Turn Around Time (TAT)	Price	TAT	Price	TAT	Price	TAT
EXOMES								
Exome Proband	\$1,890	4 weeks						
Exome Trio	\$2,490	4 weeks						
Rapid Exome Proband	\$2,290	9 days*						
Rapid Exome Trio	\$2,890	9 days*						
GENOMES								
Genome Proband	\$2,490	4 weeks						
Genome Trio	\$4,890	4 weeks						
Rapid Genome Proband	\$2,990	9 days*						
Rapid Genome Trio	\$5,990	9 days*						

* Verbal test results.



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22FEB04

SPEAKER BIOS

Jane Juusola, PhD, FACMG



Dr. Jane Juusola is Vice President of Medical Affairs at GeneDx, a genetic testing laboratory based in Maryland. She is board-certified in Clinical Molecular Genetics and has worked at GeneDx since 2012. Jane has co-authored >80 articles in molecular and human genetics, including publications on rapid clinical genomic testing and identification of novel disease genes.

Trudy McKanna, MS, CGC

Trudy is an experienced genetic counselor who recently became the Director of Medical Education – Transplant and Renal Genetics for Natera. She worked as a preceptor for small group discussions in Medical Ethics, Human Behavior and Development, and Medical Humanities for first and second year medical students at Michigan State University, and is a former president of the Michigan Association of Genetic Counselors.



SPEAKER BIOS

Dianalee McKnight, PhD, FACMG



Dee McKnight, Ph.D., FACMG is a board-certified clinical molecular geneticist who has been in the genetic diagnostic industry for over 10 years with expertise in pediatric and rare disorders. She was previously the Director of the Neurogenetics Testing Program at GeneDx, and currently serves as Director of Medical Affairs at Invitae.

Rachel Doyel, MS, CGC

Rachel Doyel is a board certified and licensed genetic counselor with extensive clinical and industry experience. She is currently the Director of Clinical Support and Education for Women's Health and Senior MSL at Sema4. Rachel is active in the Indiana Network of Genetic Counselors and is a member of the National Society of Genetic Counselors. She is also active in educational efforts for genetic counseling graduate students and is currently a member of the Indiana State University Genetic Counseling Program Advisory Board.



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CO-CHAIR BIOS

Kelly Moyer, MS, CGC



Kelly is Director of Genetic Counseling Services and an Instructor for Women's Health at Dell Medical School in Austin. She has the privilege of performing prenatal and pediatric genetic counseling through Ascension hospitals and clinics, specializing in Maternal fetal medicine, Fetal care, Cystic Fibrosis, and Clefting conditions. She is really excited about the new treatments available for people with genetic conditions and wants everyone to know about them.

Olivia Trimmier, MS, CGC

Olivia Trimmier graduated from the University of South Carolina genetic counseling program, and worked as a prenatal genetic counselor for Integrated Genetics in Georgia, South Carolina and Texas. After 6 years with Integrated Genetics, she moved to Blueprint Genetics, a rare inherited disease start up, to work as a Genomic Testing Consultant. Since 2019, Olivia has been a women's health Clinical Science Liaison for Invitae, and provides support for sales team members and physicians across 16 states.



PUZZLE TO PRIZES

Complete the following to be entered into our raffle of five \$50 Amazon giftcards! Email the filled out PDF or answers to texasGCaec@gmail.com by EOD on Friday, March 25th. Happy Puzzle-ing! **Individuals who participate in puzzle to prizes consent to sharing their contact information with our sponsors.**

1. Amicus Therapeutics is committed to improving the

lives of patients and families affected by what

category of diseases?

- 2. What is the name of the NIPS offered by Billion to One?
- 3. Biomarin offers treatment for what genetic disorder?
- 4. Color guides choices for health and... ?
- 5. How many genes does Fulgent offer on their test menu?
- 6. Invitae is the largest contributor to what public variant database?













- 7. LabCorp has experience with MaterniT in how many patients?
- 8. Myriad Genetics' myChoice ® CDx is for patients with what cancer type?
- 9. What is the name of Natera's newest publication, which is the largest prospective NIPS study to date?
- 10. What is the name of PerkinElmer's test which offers enhanced coverage of large CNVs?
- 11. Prevention Genetics offers two rapid tests, what are they?
- 12. How many genes are offered in Sema4's expanded carrier screen?

